Leistungsgerechte Vergütungsansätze und zielgruppenorientiertes Informationsmanagement im Bereich seltener Erkrankungen

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Zusammenfassung

Der Begriff seltene Erkrankungen bezeichnet eine Gruppe von heterogenen und zumeist komplexen Krankheitsbildern, von denen in ihrer Gesamtheit ca. 4 Millionen Menschen in Deutschland betroffen sind. Zu ihren gemeinsamen Besonderheiten zählt neben der geringen Anzahl Betroffener u. a. ein überdurchschnittlich hoher Kosten- und Zeitaufwand für die Behandlung und Beratung von Betroffenen, der unzureichend vergütet wird. Auch ein Mangel an umfassenden, qualitätsgesicherten und verständlichen Informationen für Menschen mit seltenen Erkrankungen, ihre Angehörigen und ärztliches Personal kann festgestellt werden. Damit sind seltene Erkrankungen aus gesundheitsökonomischer und versorgungspolitischer Perspektive hoch relevant. Im „Nationalen Aktionsplan für Menschen mit Seltenen Erkrankungen“ wurden verschiedene Maßnahmenvorschläge formuliert, um die Situation von Betroffenen zu verbessern und eine hochwertige Versorgung sicherzustellen. Dazu zählt u.a. die Entwicklung adäquater Vergütungssysteme (Handlungsfeld Versorgung und Zentren) und der Aufbau eines krankheitsübergreifenden, qualitätsgesicherten Informationsportals (Handlungsfeld Informationsmanagement). Um diese Maßnahmen umsetzen zu können, bedarf es Kenntnissen über die Präferenzen der Beteiligten im Hinblick auf zukünftige Vergütungsinstrumente und spezifische Informationsbedarfe.

Das Ziel der Dissertation liegt in der Analyse und Entwicklung leistungsgerechter Vergütungsansätze und eines zielgruppenorientierten Informationsangebots im Bereich seltener Erkrankungen. Im ersten Modul wird die Angemessenheit bestehender Vergütungsmöglichkeiten diskutiert und Ansätze für eine leistungsgerechte Vergütung entwickelt. Anschließend werden die Bedarfe und Präferenzen von Betroffenen, ihren Angehörigen und medizinischen Versorgern an unterschiedliche Informationsangebote analysiert und Anforderungen für ein zentrales Informationsportal abgeleitet. Zusätzlich werden Qualitätskriterien für Onlineinformationen zu seltenen Erkrankungen erarbeitet, die Qualität vorhandener Informationsangebote im Internet analysiert und Möglichkeiten für ein Informationsmanagement im Spannungsfeld zwischen Qualitätssicherung und Zielgruppenorientierung eruiert.

Im Handlungsfeld Versorgung und Zentren kann zunächst eine deutliche Unterdeckung von Kosten in der Versorgung von Menschen mit seltenen Erkrankungen krankheitsübergreifend und über verschiedene bestehende Vergütungsformen hinweg festgestellt werden. Aus Sicht von Vertretern spezialisierter Versorgungseinrichtungen, gesundheitspolitischen Akteuren und Patientenvertretern besteht unmittelbarer Handlungsbedarf, um die im Nationalen Aktionsplan erarbeitete Zentrums- und Versorgungsstruktur nachhaltig zu erhalten und eine hochwertige Versorgung zu gewährleisten. Als präferierte Form der Vergütung kann eine Sonderpauschale für

seltene Erkrankungen identifiziert werden, die den besonderen zeitlichen Bedarf in der Versorgung Betroffener berücksichtigt. Die Pauschale sollte nach verschiedenen Schweregraden differenziert und zusätzlich zur regulären Vergütung erstattet werden.

Im Handlungsfeld Informationsmanagement kann darüber hinaus ein erheblicher Bedarf nach onlinebasierten Informationen zu seltenen Erkrankungen festgestellt werden, die über einen zentralen Zugangspunkt erreichbar sein sollten. Abhängig von Indikation, Erkrankungsstatus und weiteren personenbezogenen Faktoren variieren die spezifischen Informationsbedarfe. Übergreifend können jedoch zentrale Informationskategorien, wie beispielsweise psychosoziale und sozialrechtliche Beratung, aktuelle Veranstaltungen und Selbsthilfeangebote, identifiziert werden, die Patienten, ihre Angehörigen und Versorger gebündelt auf einem krankheitsübergreifenden Informationsportal präferieren. Um den Umstand zu berücksichtigen, dass Informationswebseiten zu seltenen Erkrankungen häufig nicht spezifischen Qualitätskriterien genügen, für Suchende dennoch bedeutsam sein können, sollte bei der Auswahl der Informationsbasis ein gleichermaßen qualitäts- und bedarfsorientiertes Vorgehen gewählt werden, dass die Qualität der angebotenen Informationsseiten transparent darstellt.

Insgesamt ist festzuhalten, dass in beiden Handlungsfeldern übergreifend für die Gesamtheit seltener Erkrankungen präferenzbasierte Ansätze für eine leistungsgerechte Vergütung und ein zielgruppenorientiertes Informationsmanagement entwickelt werden konnten. Es bedarf nun weiterer Anstrengungen, um die entwickelten Ansätze in der Praxis zu implementieren, beispielsweise anhand modellhafter Rahmenvereinbarungen über eine leistungsbezogene Sonderpauschale zwischen Leistungserbringern und Kostenträgern für ausgewählte seltene Indikationen. Um im Bereich Informationsmanagement eine Verbesserung der Qualität der Informationslandschaft zu fördern, sind u.a. Untersuchungen zu Professionalisierungpotentialen der relevanten Anbieter notwendig.

Schlagwörter: Seltene Erkrankungen, Vergütung, Informationsbedarfe, Informationsmanagement, Informationsqualität, Informationsportal

Abstract

The term rare diseases refers to a group of heterogeneous and mostly complex clinical pictures that affect approximately 4 million people in Germany. In addition to the small number of people affected by each rare disease, their common characteristics include the above-average costs and time required for the treatment and counselling of those affected, which is insufficiently remunerated. A lack of comprehensive, quality-assured and comprehensible information for people with rare diseases, their relatives and medical practitioners can also be identified. Thus, rare diseases are highly relevant from a health economic and care policy perspective. In the National Action Plan for People with Rare Diseases, various proposals for action were formulated to improve the situation of those affected and to ensure high-quality care. These included, among others, the development of adequate remuneration systems (field of action: care and centres) and the establishment of a cross-disease, quality-assured information portal (field of action: information management). In order to be able to implement these measures, knowledge is needed about the preferences of those involved with regard to future remuneration instruments and specific information needs.

The aim of the dissertation is to analyse and develop performance-based remuneration approaches and a target group-oriented information offer in the field of rare diseases. In the first module, the appropriateness of existing remuneration options is discussed and approaches for performance-based remuneration are developed. Subsequently, the needs and preferences of patients, their relatives and medical providers for different information services will be analysed and requirements for a central information portal will be derived. In addition, quality criteria for online information on rare diseases will be developed, the quality of existing information offers on the internet will be analysed and possibilities for information management in the area of conflict between quality assurance and target group orientation will be explored.

In the field of action of care and centres, a clear undercoverage of costs in the care of people with rare diseases across diseases and across different existing forms of remuneration can be identified. From the point of view of representatives of specialised care facilities, health policy actors and patient representatives, there is an immediate need for action in order to sustainably maintain the care and centre structure developed in the Action Plan and to ensure high-quality care. A special flat fee for rare diseases can be identified as the preferred form of remuneration, which takes into account the special time requirements in the care of those affected. The flat rate should be differentiated according to different degrees of severity and reimbursed in addition to the regular remuneration.

In the field of information management, a considerable need for online-based information on rare diseases can also be identified, which should be accessible via a central access point. Depending on the indication, disease status and other personal factors, the specific information needs vary. However, central information categories, such as psychosocial and socio-legal advice, current events and self-help offers, can be derived across the board, which patients, their relatives and providers prefer bundled on a cross-disease information portal. In order to take into account the fact that information websites on rare diseases often do not meet specific quality criteria, but can nevertheless be significant for those looking for information, a procedure should be chosen for the selection of the information base that is equally quality- and needs-oriented and that transparently presents the quality of the information sites offered.

Overall, it should be noted that in both fields of action, preference-based approaches for performance-based remuneration and target group-oriented information management could be developed for the entirety of rare diseases. Further efforts are now required in order to implement the developed approaches in practice, for example by means of model framework agreements on a performance-related special flat rate between care providers and insurance providers for selected rare indications. In order to promote an improvement in the quality of the information landscape in the area of information management, studies on the professionalisation of the relevant providers are necessary, among other things.

Key words: rare diseases, remuneration, information needs, information management, information quality, information portal

Inhaltsverzeichnis

1 Motivation und Zielsetzung

Schätzungsweise 30 Millionen Menschen in Europa leben mit einer seltenen Erkrankung [1]. Dazu zählen in der Europäischen Union Krankheiten, die weniger als einen von 2.000 Menschen betreffen. Heute sind mehr als 6.000 verschiedene seltene Erkrankungen bekannt [2]. In den meisten Fällen handelt es sich dabei um komplexe und schwerwiegende Erkrankungen, denen eine chronische Ursache zugrunde liegt und die sich bereits im Kindesalter manifestieren [3, 4]. Trotz der Vielzahl und Heterogenität seltener Erkrankungen sind ihnen in der Versorgung spezifische Herausforderungen gemein:

Die Suche nach einer Diagnose ist für viele Patienten ein langwieriger Prozess. In der Regel stellt der Hausarzt oder ein anderer Primärversorger die erste Anlaufstelle für Betroffene dar. Aufgrund mangelnder Kenntnisse im Bereich der seltenen Erkrankungen vergehen durchschnittlich bis zu fünf Jahre bis die Betroffenen eine gesicherte Diagnose erhalten. Die diagnostische Unsicherheit bei Ärzten ist besonders hoch, wenn verschiedene Organsysteme beteiligt sind und sich die Symptome nicht eindeutig einem Fachgebiet zuordnen lassen [5, 6]. Über viele seltene Erkrankungen liegen zudem nur begrenzte Informationen vor, was die Informationssuche für Betroffene, Angehörige und Leistungserbringer erschwert. Die vorhandenen Informationsangebote sind darüber hinaus schwer auffindbar, da sie über verschiedene Quellen im Internet verstreut und teilweise unbekannt sind [4, 7].

Auch wenn eine Diagnose sicher gestellt werden konnte und Patienten den Weg in die spezialisierte Versorgung gefunden haben, stehen in vielen Fällen keine Therapien zur Verfügung, da die Ursachen der Erkrankungen oftmals unerforscht sind. Auch für symptomorientierte Behandlungsansätze, die darauf abzielen, den Krankheitszustand zu verbessern, Zugang zu psychosozialer Unterstützung zu ermöglichen und die Lebenserwartung zu erhöhen, bedarf es spezialisierter Versorger. Bedingt durch die Komplexität vieler seltener Erkrankungen und ihrer Auswirkungen auf verschiedene Organsysteme kann eine qualitativ hochwertige Behandlung nur durch besonders qualifizierte Fachkräfte erreicht werden [8, 9]. Die dafür notwendigen Mindestfallzahlen für Qualität in der Versorgung sind ausschließlich in spezialisierten Einrichtungen zu erzielen. Zudem braucht es eine multiprofessionelle Versorgung, bei der die einzelnen Fachgruppen bestmöglich koordiniert und Informationen zwischen den verschiedenen Schnittstellen geteilt werden. Bedingt durch die Seltenheit der Erkrankungen sind diese oftmals nicht wohnortnah, sondern lediglich in zentralisierten klinischen Strukturen für die Betroffenen erreichbar. Im Vergleich zur Versorgung von Menschen mit häufigen und gut erforschten Erkrankungen ist sie für die sogenannten Spezialambulanzen und Zentren für seltene Erkrankungen mit einem erhöhten Zeit- und Vorhalteaufwand, z.B. im Hinblick auf Expertise und Gerätschaften, verbunden.

Der überdurchschnittliche Zeitaufwand resultiert u.a. aus umfangreichen Aktenrecherchen, nationaler und internationaler kollegialer Zusammenarbeit und Austausch, Fortbildungsbedarfen etc. [4, 10].

Aus gesundheitsökonomischer Perspektive sind seltene Erkrankungen damit in vielfacher Hinsicht bedeutsam: Geht man davon aus, dass es zu den Hauptaufgaben eines Gesundheitssystems gehört, (a) eine stabile und hochwertige Versorgung der Bevölkerung zu gewährleisten, (b) allen Menschen einen bedarfsgerechten Zugang zu Gesundheitsleistungen zu ermöglichen und (c) die zur Verfügung stehenden Ressourcen zum größtmöglichen Nutzen der Patienten einzusetzen, ergeben sich besondere Herausforderungen im Hinblick auf seltene Erkrankungen. So bedingt eine verzögerte Diagnostik eine Zunahme gesundheitsbezogener Leiden sowie eine Unter- und Fehlversorgung Betroffener. Zwar haben Technologien auf Basis von Genomsequenzierungen die diagnostischen Möglichkeiten in den vergangenen Jahren stetig erweitert; diese sind jedoch mit hohen Kosten verbunden und finden mehrheitlich in der spezialisierten Versorgung Anwendung [11].

Auch die Vergütung der Versorgung birgt ökonomische Herausforderungen: Aufgrund der niedrigen Patientenzahlen sowie der Schwere und Komplexität der Erkrankungen herrscht Konsens darüber, dass eine zentralisierte Versorgung in den so genannten Zentren für seltene Erkrankungen sinnvoll und notwendig ist. Es liegen jedoch zahlreiche Hinweise vor, dass die Versorgung in den Zentren nicht kostendeckend erbracht werden kann. Dies belegen u.a. Untersuchungen zur Mukoviszidose, einer der häufigsten seltenen Erkrankungen, wonach ca. die Hälfte der Versorgungsaufwendungen nicht durch die gesetzlichen Krankenkassen refinanziert werden. Die überdurchschnittlich hohen Kosten sind u.a. auf kostenintensive routinemäßige Untersuchungen und den hohen spezialisierten Personalbedarf zurückzuführen. Zu ähnlichen Ergebnissen kommt eine Analyse der Vergütungssituation bei der ambulanten Behandlung des Marfan-Syndroms. Auch in dieser Indikation konnte eine erhebliche Unterdeckung des angefallenen Ressourcenaufwands identifiziert werden [12, 13, 14].

Seltene Erkrankungen stellen somit große Herausforderungen für Gesundheitssysteme dar, deren Ziel darin besteht, allen Betroffenen unter Berücksichtigung der strukturellen und ökonomischen Einschränkungen eine ausreichende, zweckmäßige und wirtschaftliche Versorgung nach dem allgemein anerkannten Stand der Forschung zukommen zu lassen. Bis vor einigen Jahren verfügte das deutsche Gesundheitssystem jedoch kaum über Strategien, um auf diese Erkrankungen angemessen reagieren zu können. Erst in den vergangenen zwei Jahrzehnten haben Politik und Wissenschaft verstärkt Maßnahmen ergriffen, um die Versorgung von Menschen mit seltenen Erkrankungen in Deutschland und Europa zu verbessern [15].

Zunächst stufte der Rat der Europäischen Union seltene Erkrankungen 2009 aufgrund der hohen Gesamtzahl betroffener Menschen und der Schwere und Komplexität der Erkrankungen als Gesundheitsbedrohung für die Bürger der EU ein und empfahl u. a. die Einrichtung von spezialisierten Zentren und die Ausarbeitung von Plänen zur Steuerung von Maßnahmen auf dem Gebiet der seltenen Erkrankungen [16]. Für Deutschland wurden im selben Jahr in einem vom Bundesministerium für Gesundheit in Auftrag gegebenen Forschungsbericht zu „Maßnahmen zur Verbesserung der gesundheitlichen Situation von Menschen mit seltenen Erkrankungen“ zahlreiche dringliche Handlungsfelder identifiziert und Vorschläge zur Optimierung der Versorgungssituation formuliert [4]. Im darauffolgenden Jahr wurde ebenfalls auf Initiative des Bundesministeriums für Gesundheit das „Nationale Aktionsbündnis für Menschen mit Seltenen Erkrankungen“ (NAMSE) gegründet. In Kooperation mit dem Bundesministerium für Bildung und Forschung (BMBF) und der Allianz Chronischer Seltener Erkrankungen (ACHSE e.V.), einem Netzwerk von Selbsthilfeorganisationen, wurde damit ein deutschlandweites Koordinierungs- und Kommunikationsgremium geschaffen. Um der Empfehlung des Rates der Europäischen Union für eine Maßnahme im Bereich der seltenen Erkrankungen nachzukommen, hat das NAMSE darauf aufbauend einen Vorschlag für einen „Nationalen Aktionsplan für Menschen mit Seltenen Erkrankungen“ erarbeitet, der 2013 verabschiedet wurde [17]. Mit dem Ziel, die Lebens- und Versorgungssituation von Betroffenen zu verbessern, wurden 52 Maßnahmenvorschläge in vier Handlungsfeldern erarbeitet, die bestehende Initiativen bündeln und ein gemeinsames, koordiniertes und zielorientiertes Handeln aller Akteure bewirken sollen. Die Handlungsfelder umfassen: „Versorgung und Zentren“, „Forschung“, „Diagnosesicherung“ und „Informationsmanagement“.

Wesentliches Ziel des ersten Handlungsfeld lag darin, die Bildung von Fachzentren zu fördern und finanzielle Rahmenbedingungen für deren nachhaltiges Bestehen zu schaffen. Dafür wurde der Aufbau einer Zentrenstruktur in drei arbeitsteilig gegliederten und miteinander vernetzten Ebenen angestrebt. Im Mittelpunkt stehen dabei Typ-A- oder Referenzzentren mit übergeordneten, z. T. organisatorischen Aufgaben sowie Typ-B- oder Fachzentren, in der die meist ambulante Versorgung von Menschen mit einzelnen seltenen Erkrankungen erbracht wird. Ergänzt werden diese durch Typ-C- oder Kooperationszentren, die an Praxen oder Kliniken angesiedelt sind und konkrete Versorgungsleistungen bei Patienten mit gesicherter Diagnose wohnortnah sicherstellen sollen [18]. Zur Finanzierung der Zentrumsstruktur sollten die bestehenden kollektivvertraglichen Finanzierungsmöglichkeiten genutzt werden. Notwendige Mittel, die nicht aus den bestehenden Regelvergütungen getragen werden können, sollten von den Kostenträgern erbracht werden.

Im Handlungsfeld „Informationsmanagement“ stand u.a. die Verbesserung der Informationssituation im Bereich seltener Erkrankungen im Fokus. Grundsätzlich wurde davon ausgegangen,

dass Betroffene ein hohes Bedürfnis nach Informationen zu ihrer Erkrankung haben. Sowohl bei Patienten, die neu diagnostiziert sind als auch bei Betroffenen, die ihre Diagnose schon längere Zeit kennen, können sie als Orientierungshilfe dienen bzw. Behandlungs-, Therapie- und Vernetzungsmöglichkeiten aufzeigen. Zwar existierte zu vielen seltenen Erkrankungen eine breite Informationsbasis, oftmals wurde jedoch nur unsystematisch von Betroffenen, Angehörigen und Versorgern darauf zurückgegriffen. Es lagen zudem Hinweise vor, dass sich die Qualität der Informationsangebote sehr heterogen darstellt. Die Maßnahmenvorschläge zielten daher darauf ab, die Qualität der Informationslandschaft zu prüfen, das Angebot auszubauen und den Zugang zu bereits vorhandenen Informationsquellen zu erhöhen. Dafür sollte ein krankheitsübergreifendes Informationsportal als zentraler Zugangspunkt zu bestehenden qualitätsgesicherten Informationen zu seltenen Erkrankungen konzeptualisiert werden. Das Portal sollte insgesamt zu einer Verbesserung der Informationsangebote hinsichtlich Qualität und Quantität beitragen [18].

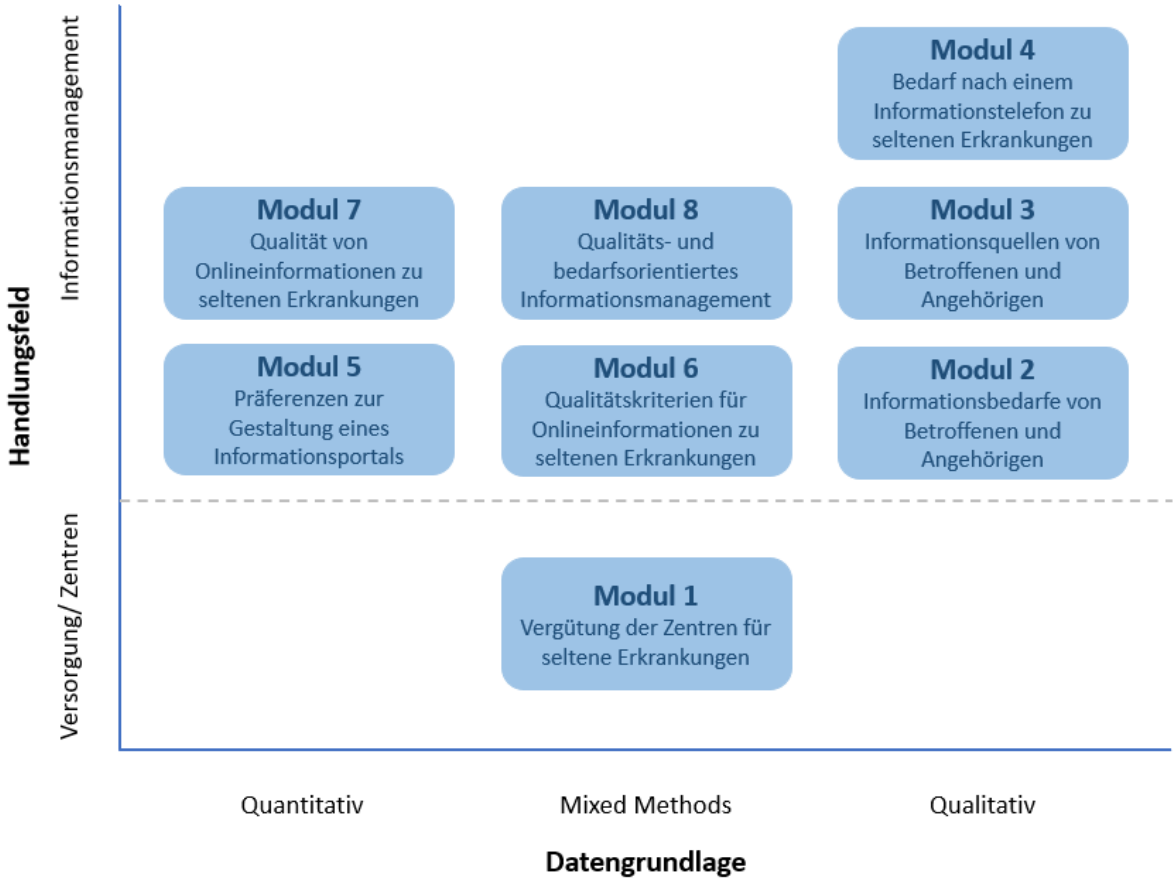
Um diese Maßnahmen umsetzen zu können, bedarf es Kenntnissen über die Präferenzen der Beteiligten im Hinblick auf zukünftige Vergütungsinstrumente und spezifische Informationsbedarfe. Das Ziel der Dissertation besteht folglich darin, in den beschriebenen Handlungsfeldern „Versorgung und Zentren“ sowie „Informationsmanagement“ des „Nationalen Aktionsplans für Menschen mit Seltenen Erkrankungen“ innovative Ansätze für eine Verbesserung der Vergütungssystematik und ein zielgruppenorientiertes Informationsangebot zu entwickeln. Auf der Grundlage eigener qualitativer und quantitativer Untersuchungen werden Herausforderungen im Hinblick auf bestehende Versorgungs- und Vergütungsformen herausgearbeitet, Weiterentwicklungsbedarfe diskutiert und präferierte Vergütungsmöglichkeiten erarbeitet. Ebenso werden die Bedarfe verschiedener Zielgruppen an ein Informationsangebot zu seltenen Erkrankungen analysiert, Qualitätsaspekte im Zusammenhang mit Onlineinformationen aufgearbeitet und Möglichkeiten eruiert, ein zielgruppenorientiertes und qualitätsgeprüftes Informationsportal zu konzeptualisieren. Die Dissertation verfolgt dabei folgende Fragestellungen:

1. Welche Herausforderungen ergeben sich im Hinblick auf die bestehenden Versorgungs- und Vergütungsformen für B-Zentren für seltene Erkrankungen?
2. Wie kann eine leistungsgerechte Vergütung für die ambulante Versorgung in den B-Zentren für seltene Erkrankungen gestaltet werden, die den Präferenzen der beteiligten Akteure entspricht?
3. Welche Bedarfe und Präferenzen weisen Patienten, ihre Angehörigen und medizinische Versorger hinsichtlich eines Informationsangebots über seltene Erkrankungen auf und wie können diese in die Konzeptualisierung eines onlinebasierten Informationsportals einfließen?

4. Welche Qualitätskriterien können für Onlineinformationen zu seltenen Erkrankungen sinnvoll angewendet werden und von welcher Qualität ist das vorhandene Informationsangebot im Internet?
5. Wie können die ermittelten Bedarfe und Präferenzen unter Berücksichtigung von Aspekten der Qualitätssicherung in die Konzeptualisierung eines zielgruppenorientierten Informationsportals über seltene Erkrankungen münden?

Die vorliegende Dissertation gliedert sich in verschiedene Module (siehe Abbildung 1). Zunächst werden ökonomische Herausforderungen, die sich für die B-Zentren für seltene Erkrankungen im Rahmen der bestehenden Versorgungs- und Vergütungsformen ergeben, sowie präferierte Lösungsansätze für deren Weiterentwicklung aufgearbeitet (Modul 1). Anschließend werden Informationsbedarfe und Informationsquellen analysiert, die für Menschen mit seltenen Erkrankungen und ihre Angehörigen im Krankheitsverlauf bedeutsam sind (Module 2, 3).

Abbildung 1: Module der kumulativen Dissertation



Quelle: Eigene Darstellung

Der Bedarf nach einem auf ein Informationstelefon wird in Modul 4 erläutert. Es folgt eine Präferenzerhebung zur Relevanz verschiedener Informationskategorien auf einem Informationsportal (Modul 5). Eine Untersuchung, wie Qualitätskriterien für Onlineinformationen an die Besonderheiten von seltenen Erkrankungen adaptiert werden können, bildet das Modul 6. Die Qualität von Onlineinformationen zu seltenen Erkrankungen wird in Modul 7 systematisch analysiert. Die Dissertation schließt mit einer Betrachtung zu einem bedarfs- und qualitätsorientierten Informationsmanagement im Rahmen eines zentralen Informationsportals (Modul 8).

2 Beitrag der vorliegenden kumulativen Dissertation

2.1 Herausforderungen und Lösungsansätze für eine leistungsgerechte Vergütung von B-Zentren für seltene Erkrankungen

Um sich einer bedarfsgerechten und von den am Versorgungsprozess Beteiligten präferierten Vergütungsmöglichkeit für die B-Zentren für seltene Erkrankungen anzunähern, ist zunächst eine Untersuchung der B-Zentrenlandschaft sowie der bestehenden Vergütungsmöglichkeiten und deren Weiterentwicklungsmöglichkeiten notwendig. Das Ziel der Publikation „Nachhaltige Vergütung der B-Zentren für Seltene Erkrankungen in Deutschland – Status quo und Lösungsansätze" (Modul 1) war es, die Versorgungs- und Vergütungsgrundlagen, auf denen die B-Zentren in Deutschland ihre ambulante Versorgung erbringen und abrechnen, zu ermitteln, die Angemessenheit der bestehenden Vergütungsmöglichkeiten zu ermitteln und nachhaltige Vergütungsansätze im Diskurs zu entwickeln.

In einem ersten Schritt wurde die Ist-Situation der aktuellen Versorgungs- und Vergütungsstrukturen in den B-Zentren für seltene Erkrankungen in Deutschland erhoben. Hierfür wurde gemeinsam mit Leitern unterschiedlicher B-Zentren ein quantitativer Fragebogen entwickelt, der an sämtliche B-Zentren verteilt wurde. In einem zweiten Schritt fanden zwei qualitative Fokusgruppen- und ein Experteninterview mit Vertretern der Zentren, der Kostenträger, der Gesundheitspolitik sowie Patientenvertretern statt, um Herausforderungen hinsichtlich der bestehenden Versorgungs- und Vergütungsformen, Weiterentwicklungsbedarfe sowie Ansätze für eine leistungsorientierte Vergütung zu diskutieren.

Insgesamt 39 B-Zentren beteiligten sich an der Fragebogenerhebung. Dabei zeigt sich eine deutliche Heterogenität hinsichtlich der Vergütungs- und Versorgungsstrukturen. 38 % der teilnehmenden Zentren werden ausschließlich über eine individuell verhandelte Pauschale für Hochschulambulanzen (HSA) vergütet, deren Höhe sich in Abhängigkeit der behandelten Indikationen stark unterscheidet. Eine Mischvergütung aus HSA-Pauschale und einer oder mehreren weiteren

Vergütungsformen, wie z.B. der persönlichen Ermächtigung von Krankenhausärzten, der Ermächtigung von Krankenhäusern oder der Ambulanten Spezialfachärztlichen Versorgung (ASV), liegt in 41 % der teilnehmenden Zentren vor. Insgesamt wird deutlich, dass oftmals unterschiedliche Möglichkeiten der Versorgung an Krankenhäusern parallel Anwendung finden und dafür verschiedene Vergütungen nach dem Einheitlichen Bewertungsmaßstab (EBM), der Gebührenordnung für Ärzte (GOÄ) oder als Pauschale erfolgen.

In den qualitativen Interviews berichten die an der Versorgung seltener Erkrankungen Beteiligten bei vielen Indikationen von einer Unterdeckung der ambulanten Kosten in den B-Zentren. Diese zeigt sich über verschiedene vorhandene Versorgungs- und Vergütungsformen hinweg. Insbesondere die interdisziplinäre Zusammenarbeit und der erhöhte Zeitaufwand bei der Versorgung von Menschen mit seltenen Erkrankungen würden über die bestehenden Vergütungsformen kaum abgebildet. Die unzureichende Vergütung gefährdet die Verstetigung der im NAMSE-Prozess entwickelten Zentrumsstrukturen, indem die Bereitschaft, sich im Bereich seltener Erkrankungen zu engagieren, durch fehlende monetäre Anreize gemindert wird. Auswirkungen auf die Versorgung von Betroffenen werden von Vertretern der B-Zentren und der Patientenvertretung benannt. Um die Versorgung von Menschen mit seltenen Erkrankungen nachhaltig sicherstellen zu können, wird die Notwendigkeit zur Weiterentwicklung der Versorgungs- und Vergütungsformen gesehen.

Die Teilnehmenden sprechen sich für eine einheitliche Regelung der Vergütung aus, welche die besonderen personellen und zeitlichen Aufwendungen, die mit der Behandlung der seltenen Erkrankungen verbunden ist, berücksichtigt. Die zeitlich aufgewendeten Ressourcen werden übergreifend als sinnvolle Grundlage erachtet, um den spezifischen Aufwand der B-Zentren zwischen Kostenträgern und Leistungserbringern monetär verhandeln zu können. Eine für verschiedene Schweregrade differenzierte „Sonderpauschale für seltene Erkrankungen“, die den besonderen zeitlichen Bedarf in der Versorgung von Patienten abbildet, wird als Vergütungsansatz präferiert.

2.2 Informationsmanagement im Bereich seltener Erkrankungen

Losgelöst von der Diskussion um eine leistungsgerechte Vergütung im Handlungsfeld der Versorgung und Zentren, ist die Frage nach der Ausgestaltung eines bedarfsgerechten Informationsangebots im Handlungsfeld Informationsmanagement. Auch im heutigen Informationszeitalter stellt die Suche nach validen und umfassenden Informationen über seltene Erkrankungen eine Herausforderung dar. Die Bedeutung von Information für Betroffene und ihre Angehörigen, die Identifikation von konkreten Informationsbedarfen sowie deren Implikationen für ein be-

darfsgerechtes und zielgruppenorientiertes Informationsangebot stehen im Mittelpunkt des Artikels „Information Needs of People with Rare Diseases - What Information Do Patients and their Relatives Require" (Modul 2).

In leitfadengestützten qualitativen Interviews wurden die Erfahrungen von Patienten mit seltenen Erkrankungen und ihren Familienangehörigen im Hinblick auf ihre Informationssuche und Informationsbedarfe erfasst. Hierfür wurde ein strukturierter Leitfaden entwickelt, mit der Zielgruppe pre-getestet und angepasst. Betroffene und Angehörige aus 11 Erkrankungsgruppen wurden rekrutiert, um eine hohe Heterogenität des Samples zu gewährleisten. Die Interviews wurden transkribiert und nach der strukturierten Inhaltsanalyse in Anlehnung an Mayring ausgewertet.

Die Auswertung der Interviews mit 55 Patienten und 13 nahen Angehörigen legt eine große Bedeutung von Informationen über den gesamten Erkrankungsverlauf dar. Eine Vielzahl von Informationsbedarfen kann festgestellt werden, die von medizinischen (Krankheitsbild, Therapie oder Forschung betreffend) und sozialrechtlichen Fragen über Selbsthilfe, psychosoziale Beratung und Hilfsmittel bis hin zu praktischen Fragen zum Umgang mit der Erkrankung im Alltag reichen. Ein Bedarf nach spezialisierten Ansprechpartnern für krankheitsbezogene Fragen wurde ebenfalls benannt. Insbesondere nach der Diagnosestellung, wenn der Wunsch nach Information besonders hoch ist, ist die Beschaffung von Informationen herausfordernd. Auch ein Mangel an laiengerechten und aktuellen Informationen wird berichtet. Häufig verbessert sich die Informationssituation mit Eintritt in die spezialisierte Versorgung. Ein onlinebasiertes Informationsportal, das verständliche Informationen zu den ermittelten Bedarfen bereitstellt und den Zugang zur spezialisierten Versorgung erleichtert, kann als präferierter zentraler Zugangspunkt für Betroffene mit seltenen Erkrankungen und ihre Angehörigen ermittelt werden.

Aufbauend auf den Erkenntnissen über die Wichtigkeit von Informationen für Betroffene und ihre Familien im Verlauf der Erkrankung und die spezifischen Informationsbedarfe, die übergreifend für unterschiedliche seltene Erkrankungen formuliert werden können, stellt sich nachfolgend die Frage nach bedeutsamen Informationsquellen für Menschen mit seltenen Erkrankungen. Um ein Informationsportal zu konzeptualisieren, das eine hohe Akzeptanz und Nutzung unter Betroffenen und Angehörigen aufweist, braucht es Wissen über die Bedarfe und Anforderungen, die Betroffene und Angehörige an die Informationsbasis stellen. Diese lassen sich erfahrungsbasiert über eine Reflektion der eigenen Informationssuche und der dabei genutzten Informationsquellen im Zeit- und Erkrankungsverlauf erfassen. Im Rahmen des Artikels „Use and importance of different information sources among patients with rare diseases and their relatives over time: a qualitative study" wurden diese Erfahrungen anhand einer qualitativen Erhebung krankheitsübergreifend zusammengetragen und ausgewertet (Modul 3).

Im Rahmen von 68 halbstrukturierten Interviews wurden fünfundfünfzig Patienten mit unterschiedlichen seltenen Krankheiten und 13 nahe Verwandte zu ihren Erfahrungen befragt (siehe hierzu auch die methodische Beschreibung von Modul 2). Die Teilnehmenden berichten, dass onlinebasierte Informationen die bedeutendste und am häufigsten genutzte Informationsquelle darstellen, insbesondere für Informationen, die zu Beginn der Erkrankung und der Informationssuche nachgefragt werden. Das Internet wird dabei als schneller und unkomplizierter Zugangspunkt zu Informationen wahrgenommen, wenngleich Schwierigkeiten im Umgang mit der webbasierten Suche benannt werden. Die gefundene Vielzahl an Informationen zu strukturieren und qualitativ zu bewerten wird insbesondere von neu diagnostizierten Menschen, die kaum über Vorwissen verfügen, als herausfordernd wahrgenommen. Schwierigkeiten werden außerdem von Älteren und Personen, deren Erkrankung durch einen sehr individuellen Verlauf geprägt ist, geäußert. Unterstützung erfahren die Befragten häufig durch Einrichtungen der Selbsthilfe, die den Patienten und Angehörigen helfen, die Informationen einzuordnen und zu verstehen. Der Selbsthilfe kommt auch im Hinblick auf die eigene Informationsbereitstellung eine große Bedeutung im Informationsprozess zu, als dass die zur Verfügung gestellten Informationen von den Nutzern als besonders vertrauenswürdig, hilfreich und gehaltvoll empfunden werden. Dies gilt insbesondere dann, wenn die Informationsbedarfe im Erkrankungsverlauf spezifischer werden. Neben Personen aus der Selbsthilfe werden auch spezialisierte Versorger und die Zentren für seltene Erkrankungen im Erkrankungsverlauf zu bedeutenden Ansprechpartnern für krankheitsbezogene Informationen. Viele Befragte berichten von einer hohen Qualität und Aktualität der Informationen. Aufgrund ihrer Nähe zur Forschung verfügen Fachärzte über aktuelles Wissen, das sie an ihre Patienten weitergeben.

Insgesamt kann festgehalten werden, dass Betroffene und ihre Angehörigen häufig nicht nur eine Informationsquelle nutzen, sondern bei ihrer Informationssuche auf verschiedene Medien und Informationsquellen zurückgreifen. Die präferierte Quelle hängt vom spezifischen Informationsbedarf sowie von anderen personenbezogenen Faktoren wie dem Vorwissen und Krankheitsstadium ab. Die Ergebnisse stützen die Erkenntnisse des ersten Moduls, dass ein zentrales Informationsportal für seltene Krankheiten ein geeigneter Zugangspunkt sein kann, um die Informationssuche von Patienten und ihren Angehörigen zu verbessern und sie mit spezialisierten Versorgern und anderen Betroffenen zu verbinden.

Neben einem webbasierten Informationssystem zur Verbesserung der Bereitstellung von Gesundheitsinformationen wurde in viele nationalen Aktionsplänen für seltene Krankheiten auch ein telefonischer Dienst als Zugangsmöglichkeit zu krankheitsbezogenen Informationen diskutiert. In diesem Zusammenhang untersucht die Studie „Telephone health services in the field of rare diseases: a qualitative interview study examining the needs of patients, relatives, and health

care professionals in Germany“ den Bedarf nach einem telefonischen Informationsangebot für Menschen mit seltenen Erkrankungen, ihre Angehörigen und Ärzte, welches ein ergänzendes Informationsangebot darstellen könnte (Modul 4).
Die Einzelinterviews wurden mit Hilfe eines strukturierten Leitfadens durchgeführt. Zusätzlich zu Patienten und Angehörigen aus Modul 2 und 3 wurde ärztliches Personal in die Untersuchung einbezogen. Dazu zählten Ärzte in hausärztlicher Versorgung, Ärzte in fachärztlicher Versorgung und Krankenhausärzte. Die Interviews wurden im Anschluss inhaltsanalytisch ausgewertet.
Insgesamt 107 Personen nahmen an der Interviewstudie teil. Achtundsechzig Personen, die an einer seltenen Krankheit leiden oder mit einer betroffenen Person verwandt sind und 39 Angehörige von Gesundheitsberufen beteiligten sich an der Untersuchung. Von 68 Patienten und Angehörigen und 39 Ärzten sprachen sich 52 bzw. 18 für die Einrichtung eines Telefondienstes für seltene Krankheiten aus. Die Befragten erwarten von einem telefonischen Angebot fachkundiges Personal, eine hohe Erreichbarkeit, Beratung zu medizinischen und psychosozialen Themen sowie Unterstützung bei der Einordnung der oftmals empfundenen Informationsflut. In Abgrenzung zu anderen, eher unpersönlichen Informationssuchsystemen wird insbesondere von Patienten und Angehörigen die Bedeutung des persönlichen Kontakts genannt. Für ältere Menschen und Informationssuchende ohne Affinität zum Internet wird ein telefonisches Informationsangebot als geeignete Alternative zum onlinebasierten Zugangspunkt genannt. Die Angehörigen der Gesundheitsberufe betonen die Bedeutung medizinischer Beratungsthemen, wie z.B. Differentialdiagnosen und Überweisungen in die spezialisierte Versorgung. Der Bedarf nach einer zentralen telefonischen Beratungsstelle für seltene Krankheiten kann den Ergebnissen zufolge abgeleitet werden.

Nachdem in verschiedenen qualitativen Untersuchungen eine Vielzahl an Bedarfen ermittelt werden konnte, gilt es nun zu untersuchen, wie diese bestmöglich in die Konzeptualisierung eines webbasierten Informationsportal über seltene Erkrankungen einfließen können. Vor dem Hintergrund begrenzter Anzeigemöglichkeiten von Informationen auf einem Portal war das Ziel der Publikation „Shaping an Effective Health Information Website on Rare Diseases Using a Group Decision-Making Tool: Inclusion of the Perspectives of Patients, their Family Members, and Physicians“, die Präferenzen von Betroffenen, Angehörigen und Versorgern für die Umsetzung der identifizierten Bedarfe in einem zentralen Informationsportal über seltene Krankheiten zu erfassen (Modul 5). Ebenso sollen Unterschiede zwischen den Präferenzen für bestimmte Informationsbedarfe von Patienten, Angehörigen und Ärzten herausgestellt werden.
Im Rahmen eines Analytic Hierarchy Process (AHP) wurde die Relevanz verschiedener Gesundheitsinformationen sowie die Struktur und Verteilung dieser Unterschiede bei Menschen mit

seltenen Krankheiten, ihren Familienangehörigen und Ärzten erhoben. Die einbezogenen Kriterien wurden durch eine systematische Internetrecherche und qualitative Befragungen verifiziert (s. Modul 2). Zu den identifizierten Informationsbedarfen gehörten: „Medizinische Fragen“, „Forschung“, „Soziale Beratungs- und Hilfsangebote“ sowie „Aktuelle Veranstaltungen“ (Hierarchieebene 2). Für drei Kategorien wurden folgende Unterkategorien formuliert (Hierarchieebene 3): „Diagnosefindung“, „Therapie“, „Allgemeines zum Krankheitsbild“ („Medizinische Fragen“), „Anlaufende Studien“, „Studienergebnisse“ und „Register“ („Forschung“), „Psychosoziale Beratung“, „Selbsthilfe“ und „Sozialrechtliche Beratung“ (Soziale Beratungs- und Hilfsangebote“). Mit Hilfe einer Sensitivitätsanalyse wurden Stabilität und Verteilung der Ergebnisse, mit t-Tests die Signifikanz der Ergebnisse bestimmt.

Insgesamt nahmen 120 Patienten, 24 Familienangehörige und 32 Ärzte an der Studie teil. Die Rangfolgen und Präferenzgewichte stellen sich sehr heterogen dar. Lokale und globale Rangfolgen von Patienten, Familienmitgliedern und Ärzten sind in Klammern angegeben: medizinische Fragen (3/4, 4, 4), Forschung (3/4, 2/3, 3), soziale Hilfsangebote (1, 2/3, 2) und aktuelle Ereignisse (2, 1, 1); Diagnose (6, 8, 9), Therapie (5, 9, 7), allgemeines Krankheitsbild (9, 4/5/6, 6), aktuelle Studien (7, 4/5/6, 3), Studienergebnisse (8, 7, 8), Register (4, 1, 5), psychosoziale Beratung (1, 2, 4), Selbsthilfe (3, 3, 2), und sozialrechtliche Beratung (2, 4/5/6, 1).

Die Ergebnisse zeigen, dass über die unterschiedlichen Hierarchieebenen hinweg die Informationskategorien zu seltenen Krankheiten sehr unterschiedlich gewichtet werden, was zu subgruppenspezifischen Ranking-Ergebnissen führt. Obwohl „Medizinische Fragen“ von allen Untergruppen als am wenigsten wichtig eingestuft werden, gibt es bei keiner der anderen Informationskategorien einen allgemeinen Gruppenkonsens. Signifikante Unterschiede zwischen den befragten Personengruppen werden bestätigt durch t-Tests, welche die subgruppenspezifischen lokalen Gewichte für die folgende Gegenüberstellungen verglichen: die Gewichte von Patienten und Angehörigen in den Kategorien „Therapie“, „Allgemeines zum Krankheitsbild“ und „Studienergebnisse“ unterscheiden sich signifikant. Außerdem zeigen Patienten und Ärzte signifikante Unterschiede in den Kategorien „Diagnosefindung“, „Allgemeines zum Krankheitsbild“, „Anlaufende Studien“, „Psychosoziale Beratung“ und „Aktuelle Veranstaltungen“. Auch der Vergleich der Ergebnisse von Ärzten mit denen von Familienmitgliedern zeigt für die Kategorien „Therapie“ und „Register“ statistische Signifikanz.

Die Ergebnisse der Studie legen nahe, dass die Bedeutung von Informationen über seltene Erkrankungen von Betroffenen, Angehörigen und Ärzten unterschiedlich wahrgenommen wird. Eine zielgruppenspezifische Darstellung von Informationen auf einem Informationsportal erscheint sinnvoll. Können nur begrenzt Kategorien von Informationen auf einem zentralen Informationsportal abgebildet werden, sollten die unterschiedlichen Präferenzen von Betroffenen,

Angehörigen und Versorgern berücksichtigt werden, um die Zielgruppenorientierung zu steigern.

Um sich der Konzeptualisierung und Umsetzung eines zentralen Informationsportals weiter anzunähern, spielen auch Anforderungen an die Informationsqualität eine bedeutende Rolle. Unter der Voraussetzung, dass ein Portal, welches Zugang zu Informationen zu einer Vielzahl an seltenen Erkrankungen bereitstellen soll, diese Informationen nicht selbst generieren kann, bedarf es eines Instruments zur Qualitätsprüfung sekundärer Informationsquellen. Bestehende Qualitätszertifikate für medizinischen Onlineinformationen werden jedoch nur selten im Bereich der seltenen Erkrankungen verwendet. Daher werden spezifische Qualitätskriterien benötigt, welche die Besonderheiten der Informationslandschaft zu seltenen Erkrankungen berücksichtigen. Die Entwicklung solcher Kriterien wurde in dem Aufsatz „Adopting Quality Criteria for Websites Providing Medical Information About Rare Diseases“ beschrieben (Modul 6).
Zunächst wurden relevante Zertifikate und Qualitätsempfehlungen für Websites mit Gesundheitsinformationen durch eine systematische Recherche identifiziert. Anschließend wurden sämtliche Qualitätskriterien der einzelnen Zertifikate und Kataloge zusammengetragen und thematisch strukturiert. Eine interdisziplinäre Expertengruppe diskutierte die identifizierten Kriterien und konsentierte ein Set für seltene Erkrankungen relevanter Qualitätskriterien.
Insgesamt 9 Qualitätszertifikate und Kriterienkataloge für Gesundheitsinformations-Websites mit 304 einzelnen Kriterien wurden erfasst. Daraus ergaben sich 163 verschiedene Qualitätskriterien, die jeweils einer der folgenden Kategorien zugeordnet wurden: inhaltliche Kriterien, technische Kriterien, Service-Kriterien, rechtliche Kriterien und Kriterien, die sich auf von Nutzern erstellte Inhalte beziehen. Schließlich wurde im Rahmen eines Expertenworkshops ein Konsens über 13 Qualitätskriterien für Websites mit medizinischen Informationen über seltene Krankheiten festgelegt. Von diesen Kategorien wurden vier („Datenschutzkonzept“, „Impressum“, „Erstellungs- und Aktualisierungsdatum“ und „Kontaktmöglichkeit zum Informationsanbieter“) als die wichtigsten für die Veröffentlichung medizinischer Informationen über seltene Krankheiten identifiziert. Die Erfüllung der weiteren Kriterien („Erstellungsprozess“, „Autoren“, „Quellen“, „Evidenz“, „Interessenkonflikte“, „Zielgruppe“, „Evaluation/Qualitätssicherung“, „Reviewverfahren“ und „Barrierefreiheit“) ist optional und kann hinsichtlich der Konzeptualisierung eines Informationsportals zur besseren Filterung der Anbieter herangezogen werden.

Nachdem zuvor Qualitätskriterien für Onlineinformationen zu seltenen Erkrankungen entwickelt werden konnten, gilt es nun, diese Kriterien auf bestehende Informationsangebote, die als Informationsbasis für ein Onlineportal zur Verfügung stehen, anzuwenden. Ziel der Studie „Rare Diseases on the Internet: An Assessment of the Quality of Online Information“ (Modul 7) war es,

die Qualität der Informationen im Internet über seltene Krankheiten zu bewerten. Außerdem sollte untersucht werden, ob die Qualität der Informationen über seltene Krankheiten zwischen den verschiedenen Kategorien von Informationsanbietern variiert.

Insgesamt 13 Qualitätskriterien für Webseiten, die Informationen über seltene Krankheiten anbieten, wurden in einen Fragebogen zur Selbstauskunft übertragen. Über eine Webrecherche identifizierte Anbieter von Onlineinformationen über seltene Krankheiten wurden gebeten, den Fragebogen auszufüllen. Dieser enthielt zusätzlich Fragen über den Informationsanbieter im Allgemeinen (z.B. Anbieterkategorie, Informationskategorie, Sprache, Verwendung von Qualitätszertifikaten und Zielgruppe). Für Informationsangebote, deren Anbieter nicht an der Selbstauskunft teilnahmen, wurde der Fragebogen anhand der zur Verfügung stehenden Angaben auf den Webseiten von den Forschenden ausgefüllt. Unterschiede zwischen verschiedenen Anbietertypen in den Subgruppenanalysen wurden mithilfe von t-Tests durchgeführt.

693 Websites mit Informationen über seltene Krankheiten wurden identifiziert. Insgesamt 123 Fragebögen (17,7 %) wurden von den Informationsanbietern vollständig ausgefüllt. Bei den übrigen identifizierten Informationsseiten (570/693, 82,3 %) wurden die Fragebögen von den Forschenden ausgefüllt. In vielen Fällen ist die Qualität der Webseiten verhältnismäßig niedrig. Eine Subgruppenanalyse zeigt keine statistisch signifikanten Unterschiede zwischen der Qualität der Informationen von Selbsthilfegruppen/Patientenorganisationen im Vergleich zu medizinischen Einrichtungen (P=.19). Die Qualität der Informationen durch Einzelpersonen (Patienten/Angehörige) ist signifikant schlechter als die der Informationen, die von Selbsthilfegruppen/Patientenorganisationen (P=.001), medizinischen Einrichtungen (P=.009) und anderen Verbänden und Fördereinrichtungen (P=.001) bereitgestellt werden.

Insgesamt ist die Qualität der Informationen im Internet über seltene Krankheiten gering. Qualitätszertifikate werden kaum verwendet und relevante Qualitätskriterien werden oft nicht vollständig erfüllt. Spezifische Informationsbedarfe, die von den verschiedenen Zielgruppen im Bereich seltener Erkrankungen besonders nachgefragt werden (siehe Module 2 und 5), sind unterrepräsentiert. Hierzu zählen beispielsweise Informationen über psychosoziale Beratung, sozialrechtliche Beratung und Familienplanung. Selbsthilfegruppen und Patientenorganisationen stellen den Großteil der Informationsseiten für seltene Krankheiten bereit und sind für Patienten mit seltenen Erkrankungen und ihre Angehörigen ungeachtet der Qualität eine bedeutende Informationsquelle.

Vor dem Hintergrund des aufgezeigten niedrigen Qualitätsniveaus von bestehenden Informationsseiten über seltene Erkrankungen auf der einen Seite und der ermittelten Bedarfe der unterschiedlichen Zielgruppen auf der anderen Seite, gilt es, geeignete Maßnahmen zum Informationsmanagement im Rahmen eines zentralen Informationsportals zu entwickeln.

Ziel der Arbeit „Zentrales Informationsportal über seltene Erkrankungen – Umsetzung eines qualitäts- und bedarfsorientierten Informationsmanagements“ ist es, die Bedarfe von Patienten und Angehörigen an ein onlinebasiertes Informationsangebot mit der qualitätsgeprüften verfügbaren Informationsbasis abzugleichen bzw. diese zusammenzuführen (Modul 8). Hierfür sind Vorschläge zu entwickeln, die ein gleichermaßen qualitäts- wie bedarfsorientiertes Informationsmanagement ermöglichen.

In einem ersten Schritt werden hierzu qualitative leitfadengestützte Interviews mit Patienten und Angehörigen durchgeführt, die anschließend inhaltsanalytisch nach Mayring ausgewertet werden (siehe hierzu auch die methodische Beschreibung von Modul 2). In einem zweiten Schritt werden über eine Webrecherche identifizierte Informationsseiten zu seltenen Erkrankungen anhand der Qualitätskriterien überprüft. Neben der Anzahl an Seiten, welche die Qualitätskriterien nicht erfüllen, wird außerdem untersucht, von welchen Anbietern diese stammen und welche Kriterien nicht erfüllt sind. Abschließend werden die Ergebnisse der qualitativen und quantitativen Ergebnisse zusammengeführt.

Angebote von Selbsthilfegruppen stellen für Betroffene und ihre Angehörigen oftmals eine bedeutsame Quelle bei der Informationssuche dar. Als besonders positiv wird die hohe Vertrauenswürdigkeit hervorgehoben. Darüber hinaus bieten sie im späteren Erkrankungsverlauf umfassende Information zu relevanten Informationsbereichen. Ein Großteil dieser Informationsangebote entsprecht jedoch nicht den definierten Qualitätskriterien. Daher wird ein Vorgehen empfohlen, bei dem die Qualität der bestehenden Informationsseiten über seltene Erkrankungen transparent dargestellt wird. Nicht qualitätsgesicherte Seiten können auf diese Weise aktiv angefordert werden, sind jedoch eindeutig kenntlich gemacht.

3 Beantwortung der Forschungsfragen und Ausblick

Aufgrund der erheblichen Bedeutung seltener Erkrankungen unter ökonomischen und gesundheitspolitischen Aspekten kommt der Analyse adäquater Vergütungsformen und eines krankheitsübergreifenden Informationsangebots große Bedeutung zu. Die in der vorliegenden kumulativen Dissertation eingebrachten Module leisten einen wesentlichen Beitrag zur

a) Weiterentwicklung bestehender Vergütungsmöglichkeiten hin zu einem präferenzbasierten Ansatz für eine leistungsgerechte Vergütung
b) Konzeptualisierung eines zielgruppenorientierten Informationsangebots über seltene Erkrankungen.

Im Folgenden werden die zentralen Forschungsfragen beantwortet.

1. Welche Herausforderungen ergeben sich im Hinblick auf die bestehenden Versorgungs- und Vergütungsformen für B-Zentren für seltene Erkrankungen?

In den B-Zentren für seltene Erkrankungen in Deutschland ist eine deutliche Heterogenität im Hinblick auf die Vergütung der ambulanten Versorgung zu erkennen. Entwickelt hat sich eine Versorgungs- und Vergütungslandschaft, in der eine große Anzahl unterschiedlicher Formen und deren Kombinationen vorzufinden sind. Neben einer Vielzahl an Zentren, die ausschließlich über eine HSA-Pauschale finanziert werden, kommen zahlreiche Mischformen unterschiedlicher Vergütungsformen vor. Bei Einrichtungen, die Auskunft über die Höhe der Vergütung erteilt haben, kann zudem eine deutliche Varianz in der Höhe der Vergütung festgestellt werden.
Eine hochwertige Versorgung für Menschen mit seltenen Erkrankungen aufrechtzuerhalten bzw. weiter auszubauen, scheint über die bestehenden Versorgungs- und Vergütungsformen nicht möglich zu sein. Die vom Gesetzgeber in den vergangenen Jahren geschaffenen Möglichkeiten zur Vergütung der ambulanten Versorgung weisen aus Sicht der beteiligten Akteure erhebliche Schwächen auf, die sich insbesondere in einer mangelhaften Berücksichtigung zeitlicher und personeller Aufwendungen zeigen bzw. in einem zu hohen administrativen Aufwand widerspiegeln. Diese sind über verschiedene Indikationen und sämtliche Versorgungs- und Vergütungsansätze hinweg erkennbar.
So handelt es sich bei der pauschalierten Vergütung im Rahmen der HSA häufig um eine für alle HSA-Patienten eines Klinikums einheitlich verhandelte Vergütung, die einerseits als zu niedrig erachtet wird, um die real anfallenden Kosten decken zu können und andererseits oftmals in der Ambulanz verbleibt, in der die Patienten als Erstes vorstellig werden. Auch die Vergütung im Rahmen der speziell für seltene Erkrankungen geschaffenen Ambulanten Spezialfachärztlichen Versorgung (ASV) wird für verschiedene Indikationen als unzureichend wahrgenommen. Ebenso erschweren ein komplexes Anzeigeverfahren und administrative Hürden, bedingt durch die notwendige Entwicklung indikationsspezifischer ASV-Richtlinien und Abrechnungssystematiken, eine erfolgreiche Anwendung. Insgesamt ist ein erheblicher Weiterentwicklungsbedarf der ambulanten Vergütungsstrukturen erkennbar.

2. Wie kann eine leistungsgerechte Vergütung der ambulanten Versorgung in den B-Zentren für seltene Erkrankungen gestaltet werden, die den Präferenzen der beteiligten Akteure entspricht?

Um eine bedarfsorientierte Versorgung und eine kostendeckende Vergütung nachhaltig gewährleisten zu können, sind aus Sicht der B-Zentren einheitliche rechtliche Rahmenbedingungen zur ambulanten Versorgung von Menschen mit seltenen Erkrankungen erforderlich. Neuere Re-

gelungen des Gesetzgebers, nach denen spezifische zuschlagsfähige Aufgaben von Einrichtungen der Spitzenmedizin zwischen Zentren für seltene Erkrankungen und Krankenkassen finanziell verhandelt werden können, sind nicht ausreichend, um eine Verbesserung der Vergütung für die Versorgung zentrumseigener Patienten zu erzielen. Um die aufwendige Versorgung Betroffener angemessen zu vergüten, bedarf es einer Anerkennung und Berücksichtigung der überdurchschnittlich hohen zeitlichen Aufwendungen. Ein Ansatz, der sich statt auf Kostennachweise auf Zeit- und Leistungsnachweise bezieht, wird auch aus Kostenträgerperspektive als Verhandlungsgrundlage für Vergütungen positiv bewertet.

Als praktikabelste Form der Vergütung wird grundsätzlich eine Pauschale bevorzugt, da die Erarbeitung einer eigenen Vergütungssystematik ähnlich dem DRG-System unter Berücksichtigung der Vielzahl an seltenen Erkrankungen kaum realistisch erscheint. Eine von der HSA-Pauschale losgelöste „Sonderpauschale für seltene Erkrankungen“ wird mehrheitlich präferiert. Um einen Anreiz für Zentren zu minimieren, bevorzugt leichte Fälle zu versorgen, wird eine nach Schweregraden differenzierte bzw. gewichtete Pauschale als geeignete Ausgestaltungsmöglichkeit festgehalten.

3. Welche Bedarfe und Präferenzen weisen Patienten, ihre Angehörigen und medizinische Versorger hinsichtlich eines Informationsangebots über seltene Erkrankungen auf und wie können diese in die Konzeptualisierung eines onlinebasierten Informationsportals einfließen?

Zunächst kann bei allen Zielgruppen (Patienten, Angehörigen und medizinischen Versorgern) der mehrheitliche Bedarf nach einem telefonischen Beratungsangebot festgehalten werden. Neben einem besseren Zugang von spezifischen Personengruppen, die über andere Informationsangebote, wie z.B. onlinebasierte Angeboten oder spezialisierte Einrichtungen, kaum zu erreichen sind, wird in diesem Zusammenhang der persönliche Austausch mit einer zentralen Ansprechperson bewertet. Diese sollte über entsprechende Expertise zu den Themen verfügen, die über ein Informationstelefon präferiert nachgefragt werden. Hierzu zählen aus Betroffenen- und Angehörigenperspektive insbesondere eine medizinische und psychosoziale Beratung. Eine Informationstiefe, die über allgemeine Beschreibungen der Krankheitsbilder hinausgeht, wird als notwendig erachtet. Eine zentrale telefonische Anlaufstelle wird darüber hinaus als Möglichkeit gesehen, neudiagnostizierte Personen und Menschen mit Verdacht auf eine seltene Erkrankung bei der Informationssuche und -verarbeitung, sowohl im Sinne eines Informationsdefizits als auch eines Informationsüberflusses, zu unterstützen und die Betroffenen in die spezialisierte Versorgung weiterzuleiten.

Zusätzlich zu einem Informationstelefon kann ein hoher Bedarf nach einem onlinebasierten Informationsangebot festgestellt werden. Für viele Patienten und ihre Angehörigen stellt das Internet den bevorzugten Zugangsweg zu krankheitsbezogenen Informationen dar. Insbesondere für neudiagnostizierte Personen, die noch nicht den Weg in die spezialisierte Versorgung gefunden haben, bietet das Internet die Möglichkeit, auf unkomplizierte Weise nach Informationen über ihre Erkrankung zu suchen. Trotz des einfachen und schnellen Zugangs sowie der sofortigen Verfügbarkeit ist der Umgang mit Onlineinformationen für Menschen mit seltenen Erkrankungen und ihre Angehörigen oftmals herausfordernd. So verfügen die Suchenden anfangs nur selten über ausreichend Kenntnisse und Erfahrung, die zum Teil große Anzahl an gefundenen Informationen einordnen zu können. Ebenso haben die Betroffenen Schwierigkeiten, die Qualität der gefundenen Onlineinformationen zu beurteilen, da Möglichkeiten, die Vertrauenswürdigkeit und Zuverlässigkeit nachzuprüfen, häufig unbekannt sind. Ein Bedarf nach validen qualitätsgesicherten Informationen kann daher festgestellt werden (siehe hierzu Frage 4).

Während zu Beginn vor allem Informationswebseiten eine große Rolle spielen, gewinnen im späteren Verlauf vor allem Selbsthilfeeinrichtungen und Experten in den Zentren für seltene Erkrankungen zunehmend an Bedeutung im Informationsprozess. Insbesondere die Möglichkeit zum persönlichen Austausch und die hohe Verlässlichkeit der Informationen werden sehr positiv bewertet. Für viele Betroffene stellt insbesondere der Austausch mit anderen Patienten einen Wendepunkt in ihrer Informationsrecherche dar, der erstmals eine Kommunikation auf Augenhöhe ermöglicht.

Im Hinblick auf spezifische Themen und Inhalte zeigt sich bei Menschen mit seltenen Erkrankungen und ihren Angehörigen eine große Bandbreite an Informationsbedarfen. Die meisten Betroffenen haben einen langen Diagnoseweg und viele Arztkontakte hinter sich, ehe eine seltene Erkrankung sicher diagnostiziert werden konnte. Der Wunsch nach Informationen, sobald eine Diagnose vorliegt, ist mehrheitlich sehr hoch. Diese reichen von Informationen zum Krankheitsbild, zu Therapie- und Forschungsmöglichkeiten, über psychosoziale Beratungsangebote, Selbsthilfeeinrichtungen hin zu alltagspraktischen und sozialrechtlichen Fragen. Um diese Bedarfe zielgruppengerecht in ein Informationsportal über seltene Erkrankungen überführen zu können, bedarf es einer Gewichtung hinsichtlich der Wichtigkeit der identifizierten Bedarfe und Präferenzen der einzelnen Zielgruppen im Hinblick auf deren Darstellung. Medizinische Fragen werden von Patienten, Angehörige und Ärzten übergreifend als am wenigsten wichtig eingestuft, bei den restlichen Informationskategorien kann kein allgemeiner Gruppenkonsens festgestellt werden. Während für Betroffene Informationen zu sozialen Beratungs- und Hilfsangebote vor aktuellen Veranstaltungen und Forschung von Bedeutung sind/gerankt werden, sind für Ange-

hörige aktuelle Veranstaltungen relevanter als soziale Beratungs- und Hilfsangebote und Forschung. Ein ähnliches Bild zeigt sich bei Ärzten. Die unterschiedlichen Präferenzstrukturen für Informationen über seltene Krankheiten bei Patienten, Familienmitgliedern und Ärzten zeigen sich auch bei Betrachtung der globalen Rangfolgen für eine tiefere Hierarchieebene mit ausdifferenzierten Informationskategorien. Für die Umsetzung eines Informationsportals bedeutet das, dass eine zielgruppenspezifische Strukturierung sinnvoll sein könnte. Bei begrenzten Anzeigemöglichkeiten von Informationskategorien zu seltenen Erkrankungen auf einem Onlineportal, sollten die unterschiedlichen Präferenzen von Betroffenen, Angehörigen und Versorgern berücksichtigt werden, um die Zielgruppenorientierung und somit die Nutzung und Akzeptanz zu erhöhen.

4. Welche Qualitätskriterien können für Onlineinformationen zu seltenen Erkrankungen sinnvoll angewendet werden und von welcher Qualität ist das vorhandene Informationsangebot im Internet?

Unter Berücksichtigung der Besonderheiten seltener Erkrankungen erscheint die Entwicklung spezifischer Qualitätskriterien für Onlineinformationen sinnvoll. Ein Großteil der Informationswebseiten zu seltenen Erkrankungen wird von Selbsthilfeeinrichtungen betrieben, die nur selten über die personellen und finanziellen Ressourcen sowie das notwendige Wissen verfügen, ihr Informationsangebot entsprechend bestimmter Qualitätssiegel und Kriterienkataloge zu gestalten. Um ein krankheitsübergreifendes Informationsportal über seltene Erkrankungen zu konzeptualisieren, welches auf vorhandene Informationsseiten im Internet verweist und dem Bedarf nach qualitätsgesicherten Informationen entspricht, sind Kriterien erforderlich, mit denen die Qualität bestehender Onlineinformationen geprüft werden können. Basierend auf einer Webrecherche und einer Gruppendiskussion mit Experten konnten 13 Kriterien als relevant festgehalten werden, die sich auf vier Anforderungsbereiche aufteilen: Anforderungen an die Erstellung von Inhalten, Anforderungen an Inhalte, Anforderungen an die Evaluation der Inhalte und Anforderungen an die Webseite. Von den 13 Kriterien wurden vier ausgewählt, die für eine Aufnahme ins Informationsportal zwingend zu erfüllen sind (Datenschutzkonzept, Impressum, Erstellungs- und Aktualisierungsdatum sowie Kontaktmöglichkeit). Die übrigen Kriterien werden als optionale Kriterien überprüft, die zu einer erhöhten Transparenz und besseren Filterbarkeit des verlinkten Informationsangebots beitragen können. Insgesamt kann festgehalten werden, dass die Verwendung von Qualitätskriterien für Webseiten mit medizinischen Informationen über seltene Krankheiten dazu beitragen kann, deren Akzeptanz unter Patienten, ihren Angehörigen und Ärzten zu erhöhen.

Eine Analyse bestehender Informationswebseiten zu seltenen Erkrankung anhand der identifizierten Qualitätskriterien ergibt, dass der Großteil des untersuchten online verfügbaren Angebots von niedriger Qualität ist. Keine der Webseiten erfüllte alle Qualitätskriterien und die entsprechenden Indikatoren vollständig. Während mehr als 90 % der Informationsanbieter die verpflichtenden Qualitätskriterien „Kontaktmöglichkeit“ (99,1 %) und „Impressum“ (92,1 %) erfüllt, werden Informationen zum „Datenschutz“ lediglich von 68,4 % und ein „Erstellungs- oder Aktualisierungsdatum“ von 67,4 % der identifizierten Informationsanbieter aufgeführt. Signifikante Unterschiede in der Qualität der Informationsseiten können zwischen verschiedenen Anbietern festgestellt werden. Grundsätzlich ist festzustellen, dass die Mehrheit der Informationsseiten von Patientenorganisationen oder Selbsthilfegruppen angeboten werden (38,8 %). Weitere wichtige Anbieter sind medizinische Einrichtungen (26,8%), andere Verbände und Trägerorganisationen (9,4 %), sowie Einzelpersonen (7,5 %). Die Qualität der Informationen von Einzelpersonen (z.B. Patienten/Angehörige) ist signifikant niedriger als bei Informationen von Selbsthilfegruppen/Patientenorganisationen, medizinischen Einrichtungen und anderen Verbänden und Trägerorganisationen. Keine statistisch signifikanten Unterschiede sind hinsichtlich der Qualität der bereitgestellten Informationen von Selbsthilfegruppen/Patientenorganisationen im Vergleich zu medizinischen Einrichtungen erkennbar. Informationen von der Anbietergruppe der anderen Verbände und Trägerorganisationen weisen statistisch signifikante Unterschiede im Vergleich zu solchen von Selbsthilfegruppen/Patientenorganisationen und medizinischen Einrichtungen auf. Die Qualität der Informationsseiten von dieser Anbieterkategorie ist signifikant höher.

5. Wie können die ermittelten Bedarfe und Präferenzen unter Berücksichtigung von Aspekten der Qualitätssicherung in die Konzeptualisierung eines zielgruppenorientierten Informationsportals über seltene Erkrankungen münden?

Es kann festgehalten werden, dass lediglich die Hälfte des identifizierten Angebots an Informationsseiten den festgelegten (zwingend erforderlichen) Qualitätskriterien für Onlineinformationen über seltene Erkrankungen entspricht. Rund ein Drittel der Informationsangebote, welche die erforderlichen Qualitätskriterien nicht erfüllen, wird von Patientenorganisationen oder Selbsthilfegruppen bereitgestellt. Diese Angebote werden von Patienten und Angehörigen als besonders bedeutsam wahrgenommen. Zum einen handelt es sich, insbesondere bei sehr seltenen Erkrankungen, häufig um die einzig verfügbare Informationsquelle/-basis. Zum anderen enthalten diese Webseiten im Vergleich zu Informationsangeboten anderer Anbieter am häufigsten die Informationen, die von Betroffenen und Angehörigen von großer Wichtigkeit sind (z.B. psychosoziale und leistungsrechtliche Beratung sowie Austausch mit anderen Betroffenen). Ebenso

bieten sie eine Informationstiefe, die vor allem in einem fortgeschrittenen Erkrankungsstadium, wenn allgemeine Beschreibungen des Krankheitsbilds nicht mehr ausreichend sind, als wertvoll geschätzt wird. Folglich ist festzuhalten, dass Patienten und Angehörige Bedarfe hinsichtlich der Informationsbasis eines zentralen Informationsportals haben, welche mit einer Informationsbereitstellung, die ausschließlich auf der Einhaltung spezifischer Qualitätskriterien beruht, nicht gedeckt werden können.

Eine Schmälerung der Informationsbasis eines Portals durch den Ausschluss von Angeboten, die nicht den zwingend erforderlichen Qualitätskriterien entsprechen, könnte die Nutzung und Akzeptanz unter Menschen mit seltenen Erkrankungen und ihren Angehörigen verringern. Notwendig sind daher Maßnahmen, die ein gleichermaßen an der Qualität wie auch den zielgruppenspezifischen Bedarfen orientiertes Informationsmanagement ermöglichen. Daher wird ein Vorgehen gewählt, welches die Qualität verlinkter Informationsangebote transparent darstellt. Die Suchenden bekommen nach Eingabe eines Suchbegriffs zunächst solche Webseiten angezeigt, die den erforderlichen Kriterien entsprechen. Darüber hinaus haben sie die Möglichkeit, aktiv weitere Informationsangebote abzurufen, welche die Kriterien nicht erfüllen, aber dennoch hochwertige Informationen enthalten können. Die Suchenden erhalten zusätzlich den Hinweis, dass sich die Qualitätskriterien vorwiegend auf Merkmale beziehen, die die Qualität der Webseite sicherstellen sollen. Es wird explizit darauf hingewiesen, dass die zusätzlich abrufbaren Informationen inhaltlich von hoher Qualität sein können.

Betroffene und Angehörige können somit informiert darüber entscheiden, sich weitere, möglicherweise relevante Informationen anzeigen zu lassen. Durch dieses Vorgehen soll gewährleistet werden, dass Menschen mit seltenen Erkrankungen und ihre Angehörigen über das Informationsportal Zugang zu einer qualitätsgeprüften und möglichst umfassenden Informationsbasis erhalten, die an ihren Bedarfen orientiert ist.

Abschließend kann festgehalten werden, dass die kumulative Dissertation im Hinblick auf das Handlungsfeld „Versorgung und Zentren“ Ansätze für eine Weiterentwicklung bestehender Versorgungs- und Vergütungssystematiken hin zu einer leistungsorientierten Vergütung hervorgebracht hat. Es bedarf nun weiterführender Untersuchungen, in denen für exemplarische Indikationen die tatsächlich/real aufgewendeten Leistungen und Zeiten systematisch erfasst werden. Auf diese Weise kann eine Datengrundlage und Verhandlungsbasis geschaffen werden, um sich einer Sonderpauschale für seltene Erkrankungen weiter anzunähern. Ziel sollte sein, modellhafte Rahmenvereinbarungen für bestimmte seltene Erkrankungen zu treffen, anhand derer eine leistungsgerechte pauschalierte Vergütung entwickelt und verhandelt werden kann. Die Bereitschaft zur Unterstützung eines solchen Vorhabens konnte von den am Vergütungsprozess beteiligten Leistungserbringern und Kostenträger eingeholt werden. Eine solche Untersuchung

könnte die entwickelten präferierten Ansätze in eine praktische Umsetzung/Anwendung bringen und somit einen notwendigen Schritt für die Implementierung eines innovativen Vergütungskonzept erreichen.

Auch im Handlungsfeld „Informationsmanagement“ kann die Entwicklung eines zielgruppenorientierten Ansatzes für die Konzeptualisierung eines Informationsangebots festgehalten werden. In diesem Zusammenhang ist auf die bedeutende Rolle der Selbsthilfe bei der Informationsvermittlung im Bereich seltener Erkrankungen hinzuweisen. Angebote der Selbsthilfe vermindern die wahrgenommene Isolation Betroffener und ihrer Angehörigen, ermöglichen einen Austausch von Wissen und Erfahrungen auf Augenhöhe und bieten tiefgehende Informationen zu sämtlichen Themen, die für Informationssuchende im Verlauf ihrer Erkrankung wichtig sind. Um Einrichtungen der Selbsthilfe in ihrer zentralen Funktion bei der Informationsgewinnung zu stärken und das Angebot der Informationslandschaft weiter zu verbessern, bedarf es gemeinschaftlicher Anstrengungen von Gesundheitspolitik und Selbsthilfeorganisationen und -netzwerken zum Auf- und Ausbau von Selbsthilfeangeboten und für deren nachhaltige finanzielle Unterstützung. Es kann angenommen werden, dass mangelndes Wissen und Erfahrung sowie unzureichende personelle und finanzielle Ressourcen dazu führen, dass hochwertige Informationen nur selten spezifischen Qualitätskriterien genügen. Handreichungen und finanzielle Unterstützung für Selbsthilfeeinrichtungen sowie Untersuchungen zu Professionalisierungspotentialen könnten dazu beitragen, die Qualität der Informationsangebote nach messbaren Kriterien zu verbessern. Darüber hinaus bedarf es weiterer Forschungsanstrengungen zur Qualität von Informationen über seltene Erkrankungen in einem internationalen Kontext. Um ein Informationsangebot zu schaffen, welches auch für Menschen mit sehr seltenen Krankheiten, für die kaum Informationen verfügbar sind, einen Nutzen hat, ist ein internationaler Forschungskontext unerlässlich.

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Modul 1

Nachhaltige Vergütung der B-Zentren für Seltene Erkrankungen in Deutschland – Status quo und Lösungsansätze

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Nachhaltige Vergütung der B-Zentren für Seltene Erkrankungen in Deutschland – Status quo und Lösungsansätze

Zusatzmaterial online

Zusätzliche Informationen sind in der Online-Version dieses Artikels (https://doi.org/10.1007/s00103-022-03562-7) enthalten.

Einleitung/Hintergrund

In Deutschland leben rund 4 Mio. Menschen mit einer seltenen Erkrankung [1]. Dabei handelt es sich häufig um schwerwiegende und chronische Systemerkrankungen, die einer komplexen Diagnostik und Therapie bedürfen. Um eine hochwertige spezialisierte Versorgung der Betroffenen sicherzustellen, haben sich seit dem Jahr 2009 zahlreiche Zentren für seltene Erkrankungen (ZSE) gebildet [2, 3]. Diesen Zentren kommt im Versorgungsprozess eine zentrale Rolle zu, da sie Kompetenzen und Strukturen bündeln, zielgerichtete Therapieangebote vorhalten und Forschungsaktivitäten vorantreiben.

Um die Zentrenbildung nachhaltig zu fördern, wurde im Rahmen des Nationalen Aktionsplans für Menschen mit seltenen Erkrankungen im Jahr 2013 ein sog. Zentrenmodell etabliert, welches die Strukturen und Aufgaben der ZSE konkretisiert und die interdisziplinäre und multiprofessionelle Versorgung in Typ-A-, -B- und -C-Zentren unterscheidet [4]. An der Spitze der ZSE stehen demnach die Typ-A-Zentren oder „Referenzzentren", die Menschen mit gesicherter seltener Erkrankung in geeignete Versorgungsstrukturen überführen. Werden Patient:innen mit unklarer Diagnose, aber Hinweisen auf ein seltenes Leiden vorstellig, können diese Verdachtsfälle ebenfalls im Typ-A-Zentrum abgeklärt werden.

Angegliedert an diese übergeordnete Struktur sind mehrere Typ-B-Zentren [5]. Hierbei handelt es sich um Fachzentren, die über ein interdisziplinäres Versorgungsangebot für bestimmte Erkrankungen und Erkrankungsgruppen verfügen. Die Typ-C-Zentren agieren dabei als Kooperationszentren für spezifische Erkrankungen oder Krankheitsgruppen und stellen für Patient:innen mit gesicherter Diagnose oder konkreter Verdachtsdiagnose die ambulante Versorgung sicher. Typ-C-Zentren können bspw. niedergelassene Schwerpunktpraxen, Gemeinschaftspraxen, Medizinische Versorgungszentren (MVZ) oder Krankenhäuser sein.

Eng verknüpft mit der Zentrenbildung ist die Frage der Finanzierung der Versorgung in den Zentren. Die Vergütung der Versorgungsleistungen ist dabei grundsätzlich im Fünften Buch Sozialgesetzbuch (SGB V) geregelt und richtet sich nach der Versorgungsform, auf Grundlage derer die Behandlung der Patient:innen stattfindet. Der Gesetzgeber hat in den vergangenen Jahrzehnten im Rahmen diverser Reformen verschiedene Möglichkeiten zur ambulanten Versorgung im Krankenhaus geschaffen und somit einen Beitrag zur Sicherstellung einer spezialisierten und hochwertigen Behandlung für Patient:innen mit besonders komplexen und schwerwiegenden Erkrankungen geleistet [6]. Jedoch hat sich damit eine große Vielfalt und Heterogenität der Versorgungs- und Vergütungsformen herausgebildet (Tab. 1). Diese sind immer schwieriger zu durchschauen und bedingen teilweise als ungerecht empfundene Unterschiede bei der Vergütung teils identischer oder ähnlicher Leistungen.

Ambulanzen, Institute und Abteilungen der Hochschulkliniken, sogenannte Hochschulambulanzen (HSA), können nach § 117 SGB V ermächtigt werden, Patient:innen zu behandeln, die aufgrund der Art, Schwere oder Komplexität ihrer Erkrankung einer Behandlung bedürfen. Zu den relevanten Patientengruppen zählen auch Patient:innen mit seltenen Erkrankungen. Die Hochschulambulanzen erhalten für ihre Versorgungsleistungen je Behandlungsfall eine pauschalierte Vergütung. In Abhängigkeit von der Hochschulambulanz und dem Behandlungsaufwand können für jede Klinik bis zu 50 Pauschalen vereinbart werden, welche die unterschiedlichen Behandlungskosten adäquat abbilden sollen [7].

Auch *Krankenhausärzt:innen*, die über eine abgeschlossene Weiterbildung verfügen, können nach § 116 SGB V zur Teilnahme an der vertragsärztlichen

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Tab. 1 Rechtliche Grundlage für die Leistungserbringung und Vergütung von verschiedenen Versorgungsformen für seltene Erkrankungen im Fünften Buch Sozialgesetzbuch (SGB V)

§ SGB V	Inhalt
§ 73c a. F. (alte Fassung)	Besondere ambulante ärztliche Versorgung
§ 95	Medizinische Versorgungszentren
§ 116	Ambulante Behandlung durch Krankenhausärzt:innen
§ 116a	Ambulante Behandlung durch Krankenhäuser bei Unterversorgung
§ 116b	Ambulante Spezialfachärztliche Versorgung
§ 117	Hochschulambulanzen
§ 119	Sozialpädiatrische Zentren
§ 119c	Medizinische Behandlungszentren
§ 120 Abs. 1a	Pädiatrische Spezialambulanz
§ 140a	Besondere Versorgung

Versorgung ermächtigt werden. Voraussetzung hierfür ist jedoch, dass eine ausreichende Versorgung der Patient:innen ohne die besonderen diagnostischen und therapeutischen Methoden der/des Krankenhausärzt:in nicht anderweitig sichergestellt werden kann. Die Vergütung ist gemäß § 120 SGB V aus dem Budget der Vertragsärzt:innen im niedergelassenen Bereich zu finanzieren und führt somit zu einer Verringerung der vertragsärztlichen Einnahmen. In der Regel werden daher nur sehr begrenzt Ermächtigungen vom Zulassungsausschuss erteilt. Der Umfang der Versorgungstätigkeit ist vertraglich auf spezifische Ziffern des EBM (Einheitlicher Bewertungsmaßstab[1]) festgelegt.

Neben einzelnen Krankenhausärzt:innen können gemäß § 116a SGB V auch *Krankenhäuser im Falle einer Unterversorgung* oder bei lokalen Versorgungsbedarfen eine Ermächtigung zur vertragsärztlichen Versorgung erhalten. Diese gilt für 2 Jahre und entspricht im Hinblick auf den zu gewährenden Versorgungsumfang und die Vergütung der persönlichen Ermächtigung. Gemeinhin wird die Verknüpfung eines Versorgungsauftrags an eine Person gegenüber einer Institution von den Zulassungsausschüssen bevorzugt, um eine hohe Behandlungsqualität sicherzustellen. Dies führt jedoch gleichzeitig zu einer verminderten Kapazität und Flexibilität in der Versorgung, weshalb Kliniken *Institutionsermächtigungen* auch vor dem Hintergrund einer teamorientierten Therapiekoordination bevorzugen.

Des Weiteren ist mit § 116b SGB V die Möglichkeit für die *Ambulante Spezialfachärztliche Versorgung (ASV)* spezifischer seltener Erkrankungen geschaffen worden. Bereits im Jahr 2004 wurden Kliniken für die ambulante Versorgung komplexer Erkrankungen geöffnet. Der Gemeinsame Bundesausschuss (G-BA) wurde damit beauftragt, die seltenen Erkrankungen, Erkrankungen mit besonderen Krankheitsverläufen und hochspezialisierten Leistungen zu konkretisieren, welche ambulant erbracht werden können. Im Rahmen des GKV-Versorgungsstrukturgesetzes 2012 hat der Gesetzgeber den Geltungsbereich um vertragsärztliche Leistungserbringer:innen erweitert. Bislang liegen Konkretisierungen für 8 seltene Erkrankungen vor, darunter Mukoviszidose und Sarkoidose. Die Vergütung der ASV ist einheitlich für Klinik- und Vertragsärzt:innen und richtet sich derzeit nach festen Preisen des EBM bzw. bei Leistungen, die nicht Bestandteil des EBM sind, nach der Gebührenordnung für Ärzte (GOÄ). Langfristig soll eine eigene Vergütungssystematik entwickelt werden.

Ist eine ausreichende Versorgung über Frühförderstellen nicht sichergestellt, können auch *Sozialpädiatrische Zentren (SPZ)* zur ambulanten Behandlung von *Kindern und Jugendlichen* nach § 119 SGB V ermächtigt werden. Die Ermächtigung wird abhängig vom Bedarf zeitlich begrenzt erteilt. Die Grundlage der Leistungsfinanzierung ist in § 120 Abs. 2 SGB V geregelt, der eine Pauschale vorsieht, die unmittelbar von den Krankenkassen vergütet wird.

Ambulanzen, die ausschließlich Kinder versorgen, haben außerdem die Möglichkeit, gemäß § 120 Abs. 1a SGB V als *Pädiatrische Spezialambulanz* vergütet zu werden. Hierfür werden zwischen den Landesverbänden der Krankenkassen und den Ersatzkassen einheitliche einrichtungs- oder fallbezogene Pauschalen vereinbart, die unmittelbar durch die Krankenkassen vergütet werden. Auch *Einrichtungen der Behindertenhilfe* können nach § 119a SGB V vom Zulassungsausschuss zur ambulanten Versorgung von Menschen mit geistiger Behinderung zugelassen werden. Ihre Vergütung ist wie für die Pädiatrischen Spezialambulanzen in § 120 SGB V geregelt und erfolgt anhand einer pauschalierten Vergütung durch die Krankenkassen.

Daneben bieten *Medizinische Versorgungszentren (MVZ)* nach § 95 SGB V auch stationären Leistungserbringer:innen die Möglichkeit, an der ambulanten vertragsärztlichen Versorgung teilzunehmen. Bei MVZ handelt es sich um fachübergreifende ärztlich geleitete Einrichtungen, in denen Ärzt:innen, die in das Arztregister eingetragen sind, als Angestellte oder Vertragsärzt:innen tätig sind. Die Vergütung orientiert sich an der Honorierung anderer ambulanter Leistungserbringer:innen. So bekommt jede/jeder Ärzt:in im MVZ ein individuelles Regelleistungsvolumen (RLV) und ein qualifikationsgebundenes Zusatzvolumen (QZV). Gemeinsam bilden sie das Budget für kassenärztliche Leistungen des MVZ. Des Weiteren können Einzelleistungen nach dem EBM bzw. privatärztliche Leistungen (z. B. IGeL) gemäß der GOÄ abgerechnet werden.

Trotz der vielfältigen gesundheitspolitischen Bemühungen, angemessene Rahmenbedingungen für die ambulante Leistungserbringung für Menschen mit seltenen Erkrankungen durch stationäre Einrichtungen und stationär tätige Ärzt:innen sowie deren Vergütung zu schaffen, liefern Studien Hinweise darauf, dass in ZSE häufig keine kostendeckende Erbringung von Leistungen möglich ist. Dies ist insbesondere darauf zurückzu-

[1] Vergütungssystem der vertragsärztlichen bzw. vertragspsychotherapeutischen Versorgung in Deutschland.

Zusammenfassung · Abstract

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Nachhaltige Vergütung der B-Zentren für Seltene Erkrankungen in Deutschland – Status quo und Lösungsansätze

Zusammenfassung

Hintergrund. Um eine spezialisierte Versorgung von Menschen mit seltenen Erkrankungen (sE) zu gewährleisten, wurden zahlreiche Zentren für Seltene Erkrankungen (ZSE) gegründet. Für die ambulante Behandlung von Betroffenen in Krankenhäusern steht dabei eine Vielzahl von Versorgungs- und Vergütungsformen zur Verfügung. Studien zu einzelnen sE ergaben bereits Hinweise auf Defizite in Bezug auf eine kostendeckende Vergütung der ZSE.
Ziel der Arbeit. Untersuchung der aktuellen Versorgungs- und Vergütungsstrukturen in den ZSE und die Entwicklung von Ansätzen für zukünftige nachhaltige Vergütungsstrukturen.
Material und Methoden. Mittels Fragebogenerhebung wurden zunächst ZSE in Deutschland zu ihrer Versorgungs- und Vergütungsform befragt. Im Rahmen zweier Fokusgruppen- und eines Experteninterviews mit Vertreter:innen der ZSE, der Kostenträger, der Gesundheitspolitik sowie Patient:innen wurden im Anschluss die aktuellen Versorgungs- und Vergütungsformen, Möglichkeiten der zukünftigen Gestaltung der Versorgung von Menschen mit sE sowie Ansätze für eine leistungsorientierte Vergütung diskutiert. Das Material wurde inhaltsanalytisch nach Kuckartz ausgewertet.
Ergebnisse und Diskussion. 39 Zentren beteiligten sich an der Fragebogenerhebung. 38 % dieser Zentren werden über eine Pauschale für Hochschulambulanzen (HSA) vergütet, deren Höhe stark variiert. 41 % weisen eine Mischvergütung aus HSA-Pauschale und weiteren Vergütungsformen auf. In den Interviews wurde eine Unterdeckung der Kosten in den ZSE mit Auswirkungen auf die Patientenversorgung benannt und zur Sicherstellung einer nachhaltigen Versorgung Handlungsbedarf zur Weiterentwicklung der Vergütungsstrukturen festgestellt. Eine „Sonderpauschale für sE", die den besonderen zeitlichen Bedarf in der Versorgung von Menschen mit sE abbildet, wurde als möglicher nachhaltiger Vergütungsansatz präferiert.

Schlüsselwörter

Finanzierung · Pauschalierte Vergütung · Fokusgruppen · Qualitative Inhaltsanalyse · Ambulante Versorgung

Sustainable reimbursement of the B-centres for rare diseases in Germany—status quo and solution approaches

Abstract

Background. To ensure specialized care of patients with rare diseases, numerous centres for rare diseases were funded over the past few years. The reimbursement of patients' ambulatory care in hospitals, however, is characterized by a plurality of forms of care and payment. There is some evidence of deficits in the reimbursement of care of patients suffering from a rare disease from studies on individual rare diseases.
Objectives. To investigate current forms of care provision and reimbursement of centres for rare diseases and to develop future approaches for sustainable compensation.
Materials and methods. Initially, centres for rare diseases in Germany were asked to provide information about their forms of care and reimbursement using questionnaires. Subsequently, two focus group interviews and one expert interview with representatives from centres for rare diseases, health insurance, health politics and patients were conducted to discuss current and future meritocratic forms of care provision and reimbursement. The data were evaluated using content analysis.
Results and conclusions. Thirty-nine centres for rare diseases participated in the questionnaire survey. Of those, 38% receive a flat fee/allowance for university outpatient departments, the amount of which varies notably, and 41% obtain a mixed payment comprising an allowance for university outpatient departments and other forms of reimbursement. An under-recovery of costs in centres for rare diseases and its impact on patient care were mentioned in the interviews. In this context, a need to further develop forms of care provision and reimbursement has been identified. Participants prefer a special flat fee/allowance for rare diseases that covers the time-consuming care for patients with rare diseases.

Keywords

Financing · Flat fee · Focus group interviews · Qualitative content analysis · Ambulatory healthcare

führen, dass die Vergütungssysteme die Komplexität und den überdurchschnittlichen Versorgungsaufwand seltener Erkrankungen nicht ausreichend abbilden. So konnte beispielsweise eine Untersuchung zur ambulanten Versorgung von Mukoviszidose in Deutschland zeigen, dass nur rund die Hälfte der in den Ambulanzen real anfallenden Kosten durch die Vergütungen der Kostenträger gedeckt werden, wobei die zugrunde liegenden Daten jedoch teilweise lückenhaft waren [8]. Dies kann insbesondere auf den hohen zeitlichen und personellen Aufwand bei der ambulanten Versorgung von Mukoviszidose-Patient:innen in den Zentren zurückgeführt werden. Eine Analyse der Vergütungssituation bei der ambulanten Behandlung nach § 116b SGB V am Beispiel des Marfan-Syndroms konnte ebenfalls eine deutliche Unterdeckung des angefallenen Ressourcenaufwands feststellen [9]. Im Rahmen der Untersuchung wurden ebenfalls die Kosten der medizinischen Behandlung und die Erlössituation verglichen. Es zeigte sich, dass Zentren, die als Hochschulambulanz tätig waren, eine Unterdeckung von 84 % aufwiesen. Bei Zentren, die ihre Patient:innen im Rahmen der ASV versorgten, lag eine deutlich niedrigere Kostendeckungslücke vor. Dennoch wurden auch über

die ASV 22 % der Leistungsausgaben nicht von den Krankenkassen vergütet.

Damit eine qualitativ hochwertige Versorgung von Betroffenen dauerhaft sichergestellt und die Versorgungs- und Vergütungsstrukturen weiterentwickelt werden können, sind zunächst Informationen über die aktuelle Istsituation der Versorgungs- und Vergütungslandschaft der B-Zentren in Deutschland nötig. Im Rahmen der vorliegenden Studie soll daher zunächst quantitativ eruiert werden, auf welcher rechtlichen Grundlage nach SGB V die Leistungserbringung und -vergütung stattfinden. Weiterführende qualitative Untersuchungen zur Angemessenheit der Vergütungsinstrumente aus Sicht von Leistungserbringer:innen und Kostenträgern sollen die bisher kaum untersuchte Thematik der finanziellen Situation der ZSE näher beleuchten. Abschließend werden Ansätze zur Sicherstellung nachhaltiger Versorgungsstrukturen für seltene Erkrankungen und Möglichkeiten einer leistungsorientierten Vergütung der Versorgung von Menschen mit seltenen Erkrankungen an ZSE aufgezeigt.

Methoden

Quantitative Erhebung der Versorgungs- und Vergütungsstrukturen in den B-Zentren (Ist-Situation)

In enger Abstimmung mit verschiedenen Leiter:innen von B-Zentren aus dem Bereich der genetischen Erkrankungen der Haut, des Auges sowie der angeborenen Störungen des Stoffwechsels in Deutschland wurde ein Fragebogen entwickelt, mit dem die Istsituation der aktuellen Versorgungs- und Vergütungsstrukturen abgebildet werden kann (Abb. Z1 im Onlinematerial). Bei B-Zentren handelt es sich um Fachzentren, die über ein interdisziplinäres Versorgungsangebot für bestimmte Erkrankungen und Erkrankungsgruppen verfügen. Angegliedert sind diese an Typ-A-Zentren oder Referenzzentren, die u. a. Patient:innen ohne Diagnose, die nicht an einer gesicherten seltenen Krankheit leiden, und Menschen mit gesicherter seltener Erkrankung in geeignete Versorgungsstrukturen überführen. Neben der rechtlichen Grundlage für die Versorgung nach SGB V umfasst der Fragebogen Angaben zur Ausgestaltung der Vergütungsform bei der Erbringung ambulanter Leistungen an B-Zentren. Der Fragebogen wird komplettiert durch Fragen zu zusätzlichen finanziellen Mitteln (z. B. in Form von Spenden, Stiftungsgeldern, Forschungsmitteln), zu Stärken und Schwächen der bisherigen Versorgungs- und Vergütungsformen, zu aktuell nicht vergüteten Leistungsbereichen sowie zu Ansätzen für zukünftig präferierte Versorgungs- und Vergütungsformen.

Nach einer Vorstellung der geplanten Erhebung bei der Arbeitsgemeinschaft der Zentren für seltene Erkrankungen (AG-ZSE), einem Zusammenschluss der ZSE, der gemeinsame Aktivitäten der Zentren und ihrer Mitarbeitenden koordiniert, erhielten im September 2019 sämtliche B-Zentren in Deutschland ($N = 321$) per E-Mail einen Fragebogen. Hierzu wurde eine vom Nationalen Aktionsbündnis für Menschen mit seltenen Erkrankungen (NAMSE) zur Verfügung gestellte Liste zu Ansprechpartner:innen der A- und B-Zentren in Deutschland herangezogen und über eine Handrecherche ergänzt. Da der Rücklauf der Fragebogen insgesamt sehr niedrig ausfiel, wurden sämtliche B-Zentren, die sich nicht beteiligt hatten, 2 weitere Male mit Bitte um Unterstützung angeschrieben. In den Sitzungen der AG-ZSE stellten die Beteiligten zudem den Projektstand regelmäßig vor und baten um Teilnahme an der Befragung. Nach Abschluss der Erhebung im Dezember 2019 erfolgte eine deskriptive Analyse der Daten.

Qualitative Erhebung für Ansätze zur Weiterentwicklung der Versorgungs- und Vergütungsstrukturen der ZSE (Soll-Situation)

Neben der Planung und Durchführung der quantitativen Erhebung erfolgte ab Mitte 2019 die Konzeption von Fokusgruppendiskussionen zur Entwicklung nachhaltiger Versorgungs- und Vergütungsstrukturen. Bei Fokusgruppen handelt es sich um ein häufig verwendetes Instrument, um im Rahmen von moderierten und fokussierten Diskussionsrunden die Meinungen und Ideen der Gruppenteilnehmer:innen zu einem vorab definierten Thema einzuholen [10]. Durch den gemeinsamen Austausch und die Notwendigkeit zur Begründung der eigenen Ansichten und Haltungen gegenüber den anderen Teilnehmer:innen kann eine intensivere Auseinandersetzung mit dem diskutierten Thema stattfinden als in Einzelinterviews. Geplant wurde eine Fokusgruppe mit Vertreter:innen der Zentren und eine Gruppe mit gesundheitspolitischen Akteur:innen sowie Mitgliedern der Patientenvertretung. Hierfür wurde zunächst ein Leitfaden entwickelt, der die Beteiligten dazu anregen sollte, über die Heterogenität der aktuellen Versorgungs- und Vergütungsformen sowie die Ansätze zur Weiterentwicklung der Versorgungs- und Vergütungsstrukturen zu diskutieren [11].

Aufbauend auf den vorläufigen Ergebnissen der Fragebogenerhebung fand im November 2019 eine Anpassung des Leitfadens statt, der zudem mit externen Expert:innen in qualitativer Forschung abgestimmt wurde. Nach Durchführung der ersten Fokusgruppendiskussion mit Zentrumsvertreter:innen erfolgte eine Ergänzung des Leitfadens für die zweite Fokusgruppendiskussion. Über bestehende Kontakte, einen Aufruf bei der AG-ZSE-Sitzung, eine Anfrage an die NAMSE-Steuerungsgruppe und die B-Zentren wurden Teilnehmer:innen für die Fokusgruppen rekrutiert. Im Anschluss an die Fokusgruppendiskussionen wurde eine teilnehmende Person, die kurzfristig nicht an der Fokusgruppendiskussion mitwirken konnte, im Rahmen eines Experteninterviews befragt. Bei Experteninterviews handelt es sich um ein etabliertes Verfahren, bei dem über strukturierte leitfadengestützte Interviews das organisatorische und institutionelle Wissen einer Fachperson erhoben wird. Gegenstand der Forschung sind somit nicht die individuellen Werte und Vorstellungen der interviewten Person, sondern deren spezifisches Fachwissen [12, 13].

Tab. 2 Standorte der B-Zentren für seltene Erkrankungen und die dort behandelten Erkrankungsgruppen ($N = 39$)

Bundesländer	Anzahl Zentren (*n*)
Bayern	11
Hamburg	3
Mecklenburg-Vorpommern	4
Niedersachsen	4
Nordrhein-Westfalen	6
Sachsen	7
Sachsen-Anhalt	1
Schleswig-Holstein	2
Behandelte Erkrankungsgruppen	Anzahl Zentren (*n*)
Angeborene Störung der Blutbildung	1
Angeborene Störung des Stoffwechsels	7
Genetische Erkrankung des Auges	3
Genetische Erkrankung der Haut	5
Genetische Erkrankung des Verdauungstrakts	1
Immundefizienzen	4
Mukoviszidose und verwandte Lungenerkrankungen	3
Neuromuskuläre Erkrankung	13
Skelettdysplasien	2

Die Fokusgruppendiskussionen und das Experteninterview wurden auf Tonband aufgezeichnet und anschließend transkribiert. Die Auswertung der Transkripte erfolgte inhaltsanalytisch in Anlehnung an Kuckartz unter Entwicklung eines umfangreichen Kategoriensystems mit Unterstützung der Software MAXQDA (Verbi Software, Berlin, Deutschland) [14]. Vor dem Hintergrund der Fragebogenerhebung konnten zunächst verschiedene Kategorien deduktiv erarbeitet werden. Ergänzt wurden diese um Kategorien, die induktiv aus den Fokusgruppendiskussionen und dem Experteninterview abgeleitet werden konnten. Hierzu wurden die Transkripte zunächst im Hinblick auf die Forschungsfragen durchgesehen und Oberkategorien definiert. Diese wurden im Rahmen einer zweiten Durchsicht spezifiziert, sodass ein System aus verschiedenen Ober- und Subkategorien erstellt werden konnte.

Ergebnisse

Ist-Situation: Aktuelle Versorgungs- und Vergütungssituation

Insgesamt 39 B-Zentren aus 8 Bundesländern haben sich an der schriftlichen Erhebung beteiligt, dies entspricht einer Rücklaufquote von 12 % (**Tab. 2**). In Anlehnung an die Einteilung seltener Erkrankungen nach Organsystemen [15] ist ein Drittel der Zentren auf die Versorgung neuromuskulärer Erkrankungen spezialisiert, weitere 18 % befassen sich mit angeborenen Stoffwechselstörungen. Es folgen Einrichtungen zur Therapie von seltenen Erkrankungen der Haut, Immundefizienzen, Erkrankungen des Auges sowie Mukoviszidose und anderen Lungenerkrankungen. 2 Ambulanzen für Skelettdysplasien sowie jeweils 1 zur Behandlung von seltenen Erkrankungen des Verdauungstraktes und der Blutbildung komplettieren die Stichprobe.

15 Zentren (38,46 %) erbringen ihre Leistungen nach § 117 SGB V als Hochschulambulanz und erhalten eine individuell verhandelte HSA-Pauschale. Die Höhe der pauschalierten Vergütung variiert stark zwischen 50 € und 1490 € pro Patient:in und Quartal (p. P./Q.) und liegt bei durchschnittlich 266,10 € (Spannweite 1440 €). Bei 16 Zentren erfolgt die Vergütung einerseits als Hochschulambulanz über eine HSA-Pauschale und andererseits auch über weitere Vergütungsformen. So verfügen 5 ermächtigte Hochschulambulanzen zusätzlich über eine persönliche Ermächtigung von Krankenhausärzt:innen nach § 116 SGB V. Die pauschalierte Vergütung über die Hochschulambulanzpauschale beträgt in diesen Zentren zwischen 110 € und 700 € p. P./Q. (Mittelwert 213 €; Spannweite 510 €). Die Vergütungshöhe der nach EBM abgerechneten Leistungen ist unbekannt, da keine abrechenbaren Ziffern oder beispielhafte Abrechnungen übermittelt wurden. Eine Übersicht zu Versorgungs- und Vergütungsstrukturen der teilnehmenden B-Zentren findet sich in Tab. Z1 (Onlinezusatzmaterial).

In jeweils einem Zentrum kommt neben der HSA-Ermächtigung und persönlichen Ermächtigungen eine Leistungserbringung nach § 116b SGB V ASV bzw. § 120a SGB V als Pädiatrische Ambulanz zum Tragen. Darüber hinaus berichtet ein Zentrum, zusätzlich zur HSA- und persönlichen Ermächtigung auch eine Ermächtigung des Krankenhauses gemäß § 116a SGB V, eine Anerkennung als Sozialpädiatrisches Zentrum gemäß § 119 SGB V und einer Pädiatrischen Ambulanz nach § 120a SGB V zu besitzen. 2 Zentren rechnen neben der HSA-Pauschale einzelne Leistungen gemäß EBM im Rahmen der ASV ab.

Insgesamt wird eine erhebliche Heterogenität der Versorgungs- und Vergütungsformen deutlich, wobei in der Praxis in den Zentren in vielen Fällen mehrere verschiedene Möglichkeiten der Versorgung an Krankenhäusern angewendet werden und dafür unterschiedliche Vergütungen nach EBM, GOÄ oder Pauschale erfolgen.

Zusätzliche Mittel in Form von Spenden von Pharmaunternehmen und Privatpersonen erhalten 6 B-Zentren, diese variieren zwischen 1000 € und 60.000 € jährlich. In 7 B-Zentren stehen darüber hinaus Forschungsgelder zwischen 2000 € und 50.000 € p. a. zur Verfügung. Über Stiftungsgelder zwischen 1000 € und 200.000 € jährlich verfügen 6 Zentren.

Ist-Situation: Wahrnehmung der finanziellen Situation und bisheriger Vergütungsstrukturen

Im Rahmen der Studie wurden 2 Fokusgruppendiskussionen durchgeführt. In

der ersten Diskussionsrunde fanden sich 4 Vertreter:innen verschiedener B-Zentren und Indikationen teils persönlich, teils per Video- bzw. Telefonzuschaltung zusammen. Die zweite Fokusgruppe bestand aus 9 Teilnehmer:innen, zu denen Vertreter:innen aus der Gesundheitspolitik, von Patienten- und Klinikverbänden und der Pharmaindustrie gehörten. Auch bei dieser Gruppe gab es neben persönlichen Teilnahmen zusätzliche virtuelle bzw. telefonische Zuschaltungen. Ein Experteninterview mit der Vertretung einer Krankenkasse komplettierte die Stichprobe. Ausgewählte Zitate aus den Fokusgruppen und dem Experteninterview zur Untermauerung der qualitativen Ergebnisse finden sich in Tab. Z2 (Onlinematerial).

Die Frage nach der Angemessenheit der Vergütung wird von den Teilnehmer:innen unterschiedlich diskutiert. Die Vertreter:innen der Gesundheitspolitik und der Kostenträger weichen einer expliziten Stellungnahme tendenziell aus oder verweisen auf den geringen Anteil der seltenen Erkrankungen an den Gesamtkosten eines Klinikums. Demgegenüber liegt aus Sicht der befragten Zentrenvertreter:innen bei vielen Indikationen eine mangelnde Kostendeckung bei der ambulanten Versorgung vor, welche die B-Zentren vor erhebliche Herausforderungen stellt. Interne Berechnungen der Kliniken spiegeln wider, dass die Ressourcenverbräuche für die erbrachten Leistungen nicht durch die Erlöse aus den verschiedenen Versorgungs- und Vergütungsformen gedeckt sind. Eine konkrete Einsicht in entsprechende Berechnungen wurde im Rahmen der Studie jedoch nicht gewährt.

Die Berechnungen der Kliniken, welche die Defizite in der Vergütung aufzeigen, sind gemäß der Zentrenvertreter:innen jedoch nicht geeignet, um eine stärker an den Aufwendungen oder Leistungen orientierte Vergütung mit den Krankenkassen auszuhandeln. Dies liegt nach Aussage der Kliniker:innen nicht an der Qualität der Daten, sondern an der mangelnden Bereitschaft aufseiten der Kostenträger, diese als Verhandlungsgrundlage anzuerkennen. Auch ein konzertiertes Vorgehen durch Zusammenschlüsse der B-Zentren auf Landesebene konnte bislang keine nachhaltigen Veränderungen der Vergütungssituation erwirken. Seitens vieler Zentrenvertreter:innen werden die Verhandlungen auf Ebene der Bundesländer als vergeblich wahrgenommen. Um die Situation langfristig zu verbessern, seien Petitionen über den Bundestag notwendig, um eine Verankerung im Gesetz zu erreichen. Auch Rahmenregelungen auf Bundesebene wären sinnvoll.

Nach Einschätzungen der teilnehmenden Zentren- und Patientenvertreter:innen hat die Vergütungssituation bereits Auswirkungen auf die Patientenversorgung und mindert nachhaltig die Bereitschaft, sich in der Versorgung von Menschen mit seltenen Erkrankungen zu engagieren und entsprechende Strukturen aufzubauen und zu erhalten. Kostenträger und gesundheitspolitische Vertreter:innen hingegen sehen grundsätzlich keine spürbaren Einschränkungen für die Versorgung von Patient:innen.

Die aus Sicht der B-Zentren vorliegende unzureichende Finanzierung führt zu Unzufriedenheit und Frustration. Dabei zeigt sich die Unzufriedenheit mit den Vergütungsstrukturen über verschiedene Versorgungsansätze hinweg.

Viele der Zentren werden ausschließlich oder in Kombination mit anderen Versorgungsformen über die HSA-Pauschale vergütet. Insbesondere die für die Versorgung der seltenen Erkrankungen bedeutsame interdisziplinäre Zusammenarbeit und der erhöhte Zeitaufwand bei der Versorgung von Menschen mit seltenen Erkrankungen werden aktuell nicht ausreichend abgebildet. Zum einen handelt es sich bei den vereinbarten Pauschalen häufig um eine nicht auf die Besonderheiten der seltenen Erkrankungen ausgelegte Pauschale, sondern um eine für alle HSA-Patient:innen eines Klinikums einheitlich verhandelte Vergütung. Zum anderen verbleibt die HSA-Pauschale meist dort, wo Patient:innen als Erstes vorstellig werden. Leistungen anderer Klinikbereiche, wie z. B. Laboruntersuchungen, können hierüber häufig nicht abgerechnet werden. Dies liegt zum Teil auch daran, dass der pauschalierte Betrag zu gering ist, um ihn auf verschiedene Einrichtungen aufzuteilen. Zwar konnten in den vergangenen Jahren Verbesserungen in der Höhe der HSA-Pauschale erreicht werden, diese entsprechen jedoch nicht dem tatsächlichen Ressourcenaufwand der Kliniken.

Auch die u. a. speziell für die Versorgung von seltenen Erkrankungen geschaffene ASV-Regelung wird aus Sicht der Teilnehmenden kritisch betrachtet und weist insbesondere Schwächen im komplexen Anzeigeverfahren auf. Auch kann der Erfahrung einiger Kliniker:innen nach die Versorgung mancher Indikationen im Rahmen der ASV nicht hinreichend vergütet werden.

Einige der Zentrumsvertretungen verfügen zudem über Erfahrungen mit persönlichen Ermächtigungen. Diese werden kritisch diskutiert. Während einzelne, im Rahmen der Ermächtigung festgelegte Leistungen erbracht und vergütet werden können, bleiben darüber hinausgehende unberücksichtigt. Ebenso werden die Abhängigkeit von der Ermächtigungserteilung durch den Zulassungsausschuss bzw. die Konflikte mit den Niedergelassenen und die damit verbundene mangelnde Planungssicherheit kritisiert.

Aufgrund der herausfordernden Vergütungssituation über die gesetzlich verankerten Versorgungsmöglichkeiten sind die Zentren mitunter auf weitere Finanzmittel angewiesen. Zum Teil können Forschungsgelder zur Querfinanzierung bestimmter Leistungen eingesetzt werden. Gleichzeitig machen die Zentren, die über viele Forschungsmittel verfügen, die Erfahrung, dass ihnen weniger Finanzmittel vom Klinikum zugeteilt werden, was zu einer eingeschränkten Planungssicherheit nach Auslaufen der Forschungsprojekte führt.

Insgesamt ist festzustellen, dass von den Zentren über verschiedene vorhandene Versorgungs- und Vergütungsformen hinweg eine Unterdeckung der Kosten für die Versorgung von seltenen Erkrankungen berichtet wird. Die Vergütungsdefizite zeigten bereits Auswirkungen auf die Bereitschaft zum Erhalt spezialisierter Versorgungsstrukturen. Ein Bedarf zur Weiterentwicklung der Versorgungs- und Vergütungsstrukturen, um die Versorgung von Betroffenen

nachhaltig sicherstellen zu können, wird ausdrücklich benannt.

Soll-Situation: Ansätze für zukünftige Versorgungs- und Vergütungsstrukturen

Um eine qualitativ hochwertige Versorgung und angemessene Vergütung von ZSE nachhaltig sicherstellen zu können, wäre aus Perspektive der B-Zentren eine einheitliche und leistungsorientierte gesetzliche Regelung der Vergütung ambulanter Leistungen in ZSE notwendig. Die vom G-BA beschlossenen bundeseinheitlichen Qualitätsanforderungen für die Übernahme von besonderen Aufgaben durch Einrichtungen der Spitzenmedizin und die Zentrumszuschläge stellten vorwiegend auf die stationäre Versorgung sowie zentrumsferne Leistungen für andere Einrichtungen ab. Eine Verbesserung der Vergütungssituation für die B-Zentren könne aus Sicht der Teilnehmenden dadurch nicht erreicht werden. Die fehlende Grundlage im SGB V führe ferner dazu, dass die Kostenträger ihrer im NAMSE-Prozess eingegangenen Verpflichtung zur Finanzierung der Zentren nicht nachkommen.

Aus Sicht der Teilnehmenden sollte eine vereinheitlichte Versorgungs- und Vergütungsstruktur angestrebt werden, die auf die besonderen personellen und zeitlichen Aufwendungen, die die Behandlung der seltenen Erkrankungen mit sich bringt, fokussiert.

Dabei werden die zeitliche Komponente und die damit verbundenen hohen Personalkosten übergreifend als wesentlicher Faktor für die überdurchschnittlichen Kosten der Versorgung seltener Erkrankungen verstanden. Kennzeichnend für die ZSE sei eine „Vorhaltemedizin", die wenig Patient:innen bei gleichzeitiger Vorhaltung aufwendiger Diagnostik und Therapie durch hochspezialisierte Leistungserbringer:innen bedeute. Ebenfalls handele es sich um eine „sprechende Medizin", bei der die Versorger:innen viel Zeit für die Kommunikation mit Patient:innen, Kolleg:innen und anderen an der Versorgung beteiligten Leistungserbringer:innen benötigen. Bei der Vergütung seltener Erkrankungen müssten diese zeitlichen Aufwendungen angemessen berücksichtigt werden.

Aufgrund der Erfahrungen der Kliniken aus den Vergütungsverhandlungen mit den Krankenkassen wird der Einsatz detaillierter Kostennachweise als Verhandlungsgrundlage kritisch diskutiert. Eine Darstellung der zeitlich aufgewendeten Ressourcen erscheint dagegen als sinnvolle Alternative, um den zusätzlichen zeitlichen Aufwand der Zentren abbilden zu können. Nach Ansicht der Kliniken verfügen die Zentren über die entsprechenden Informationen, um den Krankenkassen den personellen Aufwand sowie die Kontaktzeiten darlegen zu können. Auch aus Sicht des Kostenträgers wird eine solche leistungsbezogene Verhandlungsgrundlage präferiert.

Als besonders herausfordernd bezüglich der Herausarbeitung nachhaltiger Vergütungsansätze wird auch die Abwägung zwischen dem bürokratischen Aufwand und der Genauigkeit der Vergütung erachtet. Grundsätzlich teilen die Teilnehmenden die Auffassung, dass auch eine pauschalierte Vergütung das Risiko überhöhter oder zu niedriger Vergütungen beinhaltet, aber einen guten Kompromiss im Sinne einer „Mischfinanzierung" für die unterschiedlich aufwendig zu versorgenden Fälle darstellt. Die Vergütung der Leistungen in pauschalierter Form wird mehrheitlich präferiert, da die Erarbeitung einer eigenen Vergütungssystematik für die ambulante Versorgung an Krankenhäusern in Anlehnung an das DRG-System[2] – wie es in der ASV-Regelung vom Grundsatz her vorgesehen ist – aus administrativen Gründen als nicht realistisch eingeschätzt wird.

Als eine Möglichkeit wird die Etablierung einer „Sonderpauschale für seltene Erkrankungen" angeführt. Die Loslösung einer pauschalierten Vergütung für seltene Erkrankungen von der HSA-Pauschale hätte auch für B-Zentren, die nicht an ein Universitätsklinikum angegliedert sind, den Vorteil, diese zusätzlich abrechnen zu können. Aber auch eine differenziertere HSA-Pauschale, die gesondert nach Gruppen von Indikationen eine leistungsbezogene Vergütung ermöglicht, wird von den Teilnehmenden als positiv beurteilt.

Um verschiedene Schweregrade differenziert abbilden zu können, wird eine abgestufte Pauschale diskutiert. Hierdurch könnte der Anreiz für Krankenhäuser gemindert werden, vorwiegend leichte Fälle zu behandeln. Alternativ wird die Möglichkeit angesprochen, eine über verschiedene Schweregrade gewichtete Pauschale zu ermitteln.

Insgesamt ist ein erheblicher Bedarf an einer von den Zentrumsregelungen losgelösten Weiterentwicklung der ambulanten Vergütungsstrukturen festzustellen. Besonders berücksichtigt werden sollten hierbei die überdurchschnittlich hohen zeitlichen und personellen Aufwendungen in der Versorgung von Betroffenen. Eine Sonderpauschale für seltene Erkrankungen, die diese Besonderheiten einbezieht, wird als Möglichkeit für eine nachhaltige und leistungsorientierte Vergütungsform präferiert.

Diskussion

Mit der in der Studie durchgeführten quantitativen Erhebung zur Finanzierung der B-Zentren wurde erstmals angestrebt, eine flächendeckende Erfassung der Versorgungs- und Vergütungsstrukturen der ZSE in Deutschland vorzunehmen. Trotz der geringen Teilnahmequote der Zentren konnte eine sehr heterogene Versorgungs- und Vergütungslandschaft nachgewiesen werden. So konnte gezeigt werden, dass der Großteil der B-Zentren ihre Patient:innen als Hochschulambulanz versorgt, die eine Pauschale als Vergütung erhält. Ferner konnten zahlreiche Mischformen von HSA-Pauschalen mit weiteren Vergütungsarten bei anderen Versorgungsformen festgestellt werden. Insgesamt lässt sich darüber hinaus eine große Varianz in der Höhe der mit den Kostenträgern ausgehandelten Pauschalen erkennen. Da sich einige ZSE nicht beteiligt haben, kann jedoch nicht abgeschätzt werden, wie sich die Varianz innerhalb spezifischer Indikationen darstellt. Auch Aussagen über zusätzlich vergütete Leistungen sind aufgrund

[2] Klassifikationssystem für ein pauschaliertes Abrechnungsverfahren nach diagnosebezogenen Fallgruppen (Diagnosis Related Groups – DRG).

fehlender Angaben zu abrechenbaren EBM-Ziffern nicht möglich.

Die Ergebnisse der inhaltsanalytischen Auswertungen der Fokusgruppendiskussionen und des Experteninterviews belegen, dass die derzeitige Vergütung insbesondere von den Zentren und den Klinikverbünden als unzureichend wahrgenommen wird. Besonders die für die Versorgung von seltenen Erkrankungen notwendigen Vorhaltungen an spezialisiertem Personal und aufwendiger Diagnostik und Therapie würden über die aktuellen Vergütungsstrukturen nicht hinreichend abgebildet. Diese Wahrnehmung stimmt mit vorherigen Untersuchungen zur Erlössituation am Beispiel spezifischer Indikationen überein [8, 9]. Wenngleich das Ausmaß der mangelnden Kostendeckung von Politik, Versorger:innen und Kostenträgern unterschiedlich diskutiert wird, wird ein Handlungsbedarf übergreifend erkannt und die Notwendigkeit für alternative Finanzierungswege gesehen. Die Mehrheit der Teilnehmenden teilt dabei die Auffassung, dass die verschiedenen im Gesetz verankerten Versorgungs- und Vergütungsstrukturen nicht zur nachhaltigen Vergütung der B-Zentren geeignet sind. So wird die ASV u. a. als zu kompliziert im Anzeigeverfahren und nicht kostendeckend bewertet. Dies deckt sich mit Untersuchungen zur Umsetzung der ASV, die belegen, dass die ASV-Regelungsdichte ein bürokratisches Hindernis darstellt und das Potenzial als innovatives Versorgungskonzept nicht entfaltet hat [16, 17]. Obwohl im Bereich der HSA-Pauschale aus Sicht der Kliniken in den vergangenen Jahren deutliche Verbesserungen in der Vergütung erzielt werden konnten, ist auch diese nach Aussage der ZSE noch nicht kostendeckend.

Grundsätzlich präferieren die Befragten im Hinblick auf zukünftige Vergütungsformen eine pauschalierte Vergütung. In Anbetracht der Vielzahl verschiedener seltener Erkrankungen wird die Entwicklung einer eigenen Vergütungssystematik als unrealistisch und unter administrativen Aspekten als zu aufwendig eingeschätzt. Eine Sonderpauschale für seltene Erkrankungen, welche Versorger:innen zusätzlich zu ihrer regulären Vergütung erhalten, bzw. eine Erhöhung der HSA-Pauschale wird daher als positiv angesehen. Um die überdurchschnittlichen personellen, diagnostischen und therapeutischen Aufwendungen entsprechend abbilden zu können, soll dem Faktor Zeit bei der Kalkulation der Pauschale eine besondere Bedeutung zukommen. Eine Fokussierung auf zeitliche Aufwendungen und erbrachte Leistungen wird auch von Kostenträgerseite präferiert. Die indikationsspezifische Abstaffelung nach verschiedenen Schweregraden wird als sinnvoll erachtet, um den Zentren keine Anreize zu setzen, die Versorgung leichter Fälle vermehrt in den Fokus zu rücken.

Diese Studie unterliegt verschiedenen Limitationen. Zum einen konnte im Rahmen der Fragebogenerhebung nur ein geringer Rücklauf erzielt werden. Grundsätzlich wäre es möglich, dass der Rücklauf im Zusammenhang mit einer Unzufriedenheit der B-Zentren bzgl. der Vergütungssituation steht und eine geringe Beteiligung Ausdruck für eine überwiegende Zufriedenheit mit den derzeitigen Strukturen ist. Die Autor:innen führen die niedrige Rücklaufquote jedoch vielmehr auf knappe zeitliche Ressourcen, auf die Priorität anderer Thematiken im Versorgungsalltag und insbesondere auf eine mangelnde Bereitschaft, sensible Kostendaten zu Forschungszwecken zur Verfügung zu stellen, zurück. Wie die Analyse der Fokusgruppen und des Interviews gezeigt hat, werden die Vergütungen der Krankenkassen in der Regel innerhalb des Klinikums umverteilt und den Zentren nach Bedarf zugewiesen. Es scheint daher plausibel, dass entsprechende Informationen beim Klinikum verbleiben sollen. Darüber hinaus ist die Höhe der mit den Kassen verhandelten Vergütung das Ergebnis klinikinterner Verhandlungen, was ebenfalls zu einer eingeschränkten Bereitschaft der Fragebogenteilnahme geführt haben könnte. Die Ergebnisse geben somit einen Ausschnitt der Versorgungs- und Vergütungslandschaft wieder, ohne diese repräsentativ darstellen zu können.

Für die Fokusgruppendiskussionen und das Experteninterview liegen ebenfalls Limitationen hinsichtlich der Repräsentativität vor, da im Rahmen qualitativer Untersuchungen grundsätzlich keine repräsentativen Stichproben gezogen werden können. Daher wird durch theoretisches Sampling versucht, eine Reichhaltigkeit und Konsistenz der Ergebnisse zu erzeugen. Hierfür finden gleichzeitig mit der Datenerhebung erste Datenauswertungen statt, die Hinweise darauf geben, welche weiteren relevanten Fälle ausgewählt werden sollten, um den zu untersuchenden Gegenstandsbereich möglichst umfassend abbilden zu können. Das Ziel liegt somit nicht im Produzieren von Ergebnissen, die für eine breite Population repräsentativ sind, sondern darin, zuverlässige und gültige Konzepte zu entwickeln.

Fazit und Ausblick

Insgesamt kann festgehalten werden, dass eine erhebliche Heterogenität der Versorgungs- und Vergütungsstrukturen in den ZSE vorliegt. Um die spezialisierte Versorgung langfristig sicherzustellen, sollten eine „Sonderpauschale für sE“ sowie die notwendigen Rahmenbedingungen im Gesundheitswesen entwickelt werden. Dies ist auch aus versorgungspolitischer Sicht notwendig, um die Bereitschaft von Ärzt:innen und Einrichtungen, sich nachhaltig im Bereich sE zu engagieren, aufrechtzuerhalten. Es bedarf weiterer Untersuchungen zu einzelnen Leistungen und notwendigen Zeitaufwendungen am Beispiel ausgewählter Indikationen, um sich einer von den Teilnehmenden präferierten Pauschale anzunähern. Die teilnehmenden Leistungserbringer:innen und Kostenträger haben bereits ihre Bereitschaft zur weiteren Zusammenarbeit und Unterstützung erklärt und mögliche Erkrankungen definiert. Zukünftig sollen deshalb modellhaft indikationsspezifische Rahmenvereinbarungen einer leistungsgerechten Vergütung erarbeitet und verhandelt werden. Hierdurch könnte ein innovatives Vergütungskonzept angestoßen werden, welches die Versorgung von Menschen mit seltenen Erkrankungen nachhaltig sicherstellt.

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Modul 2

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Information Needs of People with Rare Diseases - What Information Do Patients and their Relatives Require?

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Abstract

Context: Even in today's information age, finding comprehensive and valid information on rare diseases is difficult for those affected. A "National Action Plan for People with Rare Diseases" was adopted in Germany in 2013, calling for patient oriented information systems. However, little remains known about what information patients with rare diseases and their family members require. **Objective:** The study analyzed the information needs of patients living with rare diseases and of their relatives, to ensure a patient suitable information system. **Methods:** Semi-structured interviews revealed patients' experiences regarding information needs and acquisition. The evaluation followed Mayring's structured content analysis. **Results:** Interviews with 55 patients and 13 close relatives were conducted and analyzed. Patients and their relatives reported information needs ranging from medical and social law issues to practical questions helping to deal with the disease daily. Furthermore, there is demand for competent contacts for all disease-related matters, such as disease surveillance or support in submitting applications. **Conclusion:** People with rare diseases and their relatives have many information needs. We identified various topics that are relevant for patients with different conditions. To improve people's knowledge about their diseases and enable access to specialized care, this information, if applicable, should be included in an information portal on rare diseases, enabling patients' and families' access to relevant information at a central point.

Keywords: Information needs; Rare diseases; Information portal; Qualitative interviews

Introduction

Approximately four million people in Germany have a rare disease [1]. Rare diseases, as adopted by the Community Action Programme on Rare Diseases 1999-2003, are those diseases with a prevalence of ≤ 1 per 2.000 persons in the European Union [2]. Although conditions might differ significantly, patients with rare diseases and their relatives often face similar challenges [3]. These include protracted diagnosis processes and a deficient information basis. In 2013, the federal government adopted the "National Action Plan" to improve patients' health situation. Establishing a patient-suitable information system is one component of a broader set of measures to achieve this goal [4].

Nevertheless, little is known about what information people affected by a rare disease require after diagnosis. In one of few related studies, patients with rare autoimmune disorders described early educational needs relating to the recipient, time, and mode of information delivered [5]. Moreover, they reported insufficient information transmission after diagnosis and how this influenced coping with the disease. Other studies indicate that information is important for coping with illness [6]. Understanding an illness's causes, symptoms, and impact is seemingly a precondition for living with the disease. Participants' specific information needs, however, have hardly been studied. Recent surveys show that very few patients with rare diseases feel that their information needs have been met completely [7, 8]. Often, patients and their family members search for information, since general practitioners and other health professionals, except for specialists, lack information about their medical conditions. Surveyed patients would prefer receiving more information on what to expect from the condition, dealing appropriately with it (example: managing symptoms) and treatment options. Moreover, social information (example: about respite and care), and information about patient organizations and support groups are needed. Similar results were obtained from a survey on the

information needs of patients with a specific rare autoimmune disorder [9]. Participants rated research, treatments, and future living with the disease as the most important areas of concern and interest, followed by symptoms and diagnosis, causes, types, and support groups. Based on these findings, a patient information website was developed, shown to significantly affect patients' disease knowledge. Further studies confirm that online information may help patients face the future confidently and facilitate health consciousness [10].

As part of the project, "Conceptualization of a Central Information Portal on Rare Diseases" (ZIPSE), patients' and relatives' disease related information needs were collected to facilitate the expansion of a database considering these. Based on the results, a target group specific central information portal on rare diseases was developed and is available at www.portal-se.de.

Methods

Due to insufficient data on people's information needs, a qualitative research design was chosen. Through qualitative methods, issues on the scant can be investigated with maximum openness. To capture all the information needed by patients and their relatives during the course of the disease, the authors conducted qualitative interviews. We developed a structured guide eliciting information about their medical history, diagnostic processes, living with the disease, and information searches **(Table 1)**. Individual information needs could be derived from this. To check the guide's suitability for identifying individuals' information needs, we piloted it on patients and family members. The guide was subsequently adjusted for patients diagnosed before or shortly after birth, with no memories of their diagnostic paths.

Table 1: Semi structured interview guide.

Set	Principal questions
Experiences with the disease (from patients who consciously experienced their diagnosis)	Please remember the beginning of your disease. What changes did you notice?
	How did diagnosis proceed?
	What happened after diagnosis?
	When imagining yourself in that position again, how did you feel?
Experiences with the disease (from patients who did not consciously experience their diagnosis)	Please tell me about your disease and how life has changed due to it.
	How does your disease affect your everyday life?
	Some people want to learn more about the diseases that they live with. How about you?
Information seeking and information needs	How was that, striving to find information about your disease?
	Do you remember any events that you associate with increased demand for information?
	Please tell me about situations in which it was easy to gather information.
	Please tell me about situations in which it was difficult to gather information.
	Which moments do you consider important in searching for information?
Type of access	Please imagine the many possibilities of modern and classic media to communicate. Please recall your own situation. Which media did you use when searching for information?
	Which medium would you prefer for accessing information?
Completion	Are there any other topics that you would like to talk about?

To ensure a broad and balanced representation of those affected, eleven groups of rare diseases were formed when this study commenced, representing a comprehensive variety of rare diseases. We aimed to interview six patients or family members in each group. Moreover, 10 interviews with patients, who had waited at least 10 years for diagnosis, were planned. Thus, the sample should comprise 76 patients and their family members. However, upon saturation of interview data, a smaller sample would suffice. Participants were recruited by the Freiburg Center for Rare Diseases (FZSE) at the University Medical Center Freiburg, University of Freiburg, Germany. Interviewed were only those, who gave their written informed consent prior to participation.

The interviews were analyzed according to structured content analysis developed by Mayring [11]. Each audio recording was verbally transcribed and processed using MAXQDA analysis software. Subsequently, two researchers perused the interviews independently, to mark all relevant text passages. To develop an extensive system of categories, a deductive-inductive procedure was used. Several categories could be derived from previous research on current information on rare diseases on the Internet (deductive approach). These were completed by inductive categories emerging from the text (inductive approach). This procedure was followed by a critical examination and, if necessary, modification of the original categories. Afterwards, the marked text excerpts were analyzed with regard to the research question. Thereby, repetitions, commonalities, differences,

and cross references could be found in the material. Extracted citations were translated by an external translation service, approved by a native speaker and then included in the paper. The following will accompany direct interview quotations: Gender ("M" for male, "F" for female), a consecutive number, age, and status as either a patient ("P") or relative ("R").

Further, the information needs found in the interviews were presented and discussed in focus groups, to enable consensual validation. Participants were recruited chiefly from the initial study sample, supplemented by consultants from patient self-help.

Results

Patient demographics

Sixty nine people affected by rare diseases were interviewed. One interview was excluded from analysis due to technical problems during recording. Overall, interviews with 55 patients and 13 relatives were evaluated and interpreted **(Table 2)**. Women (N = 45) were represented almost twice as often as men (N = 23). Participants' mean age was 50.5 years. The sample comprised patients with 44 rare indications and their relatives, who could be assigned to the above mentioned groups of rare diseases. Recruitment of patients with genetic diseases of the digestive tract (N = 2), cystic fibrosis and other lung diseases (N = 4), hereditary disorders of the eye (N = 4), and connective tissue diseases (N = 5) proved difficult because of limited access to them. Target patient numbers were not attained. Preliminary analysis showed that further interviews would apparently not expand subject knowledge. Therefore, we did not interview eight more people, as initially planned.

Themes

Analysis yielded different major themes and subthemes. The first main theme, "need for information and information acquisition," describes patients' view of the importance of information when diagnosed with a rare disease and reflects how interviewees gained access to information about everything relating to their rare diseases.

Table 2: Patient demographics.

Characteristics	Participants (n = 68)	%
Mean age (years)	50.5	
Gender (female / male)	45 / 23	66.2 / 33.8
Rare disease		
Genetic skin diseases	10	14.7
Skeletal dysplasia	7	10.3
Neuromuscular diseases	9	13.2
Genetic eye diseases	4	5.9
Connective tissue diseases	5	7.4
Genetic kidney diseases	6	8.8
Cystic fibrosis and pulmonary diseases	7	10.3
Congenital blood formation disorders	4	5.9
Immunodeficiency	7	10.3
Congenital metabolic disorder	7	10.3
Genetic diseases of the digestive tract	2	2.9
Status (Patients / relatives)	55 / 13	80.9 / 19.1

The second theme, "specific information needs on rare diseases," describes concrete issues arising from living with a rare disease. "Comprehensibility of information" (theme 3) related to respondents' preferences regarding formal information preparation.

Theme: Need for information and information acquisition

For many patients, receiving the correct diagnosis meant a long and often frustrating path. In many cases, patients and family members waited years before being certain about diagnosis. This long wait was often described as grueling, frightening, and debilitating. Some of those interviewed reported years of changing doctors and being branded as malingerers.

"For years, I've been visiting one doctor after the other to find out what I have. That is really unsatisfactory." (M15/48/P)

"Awful. I felt helpless. I felt that I'm not being taken seriously, and physically, I was not well." (F39/64/P)

"Yes, I would say that during the time when I didn't know what was wrong with me, I felt sort of lost. Because somehow, no one could tell me anything; it was a totally ridiculous

period. If one only knew what was going on, what was wrong, what adjustments one needed to make; that would simply have been better." (F23/58/P)

Accordingly, all interviewees reported a strong need for information after diagnosis.

"(The wish to inform oneself) was immediately there. Well, I'm also a person, actually. I always make sure that I get the information I need right away, so that I know what lies ahead for me and how I can deal with it. I mean, this 'not wanting to know' is not for me at all." (F18/47/P)

But along with diagnosis, new uncertainties arise. Patients and their families want to know how the disease may affect their normal routine and what it means to live with that condition every day.

"I would have liked for someone to have come and simply told me what was really going on. That someone had explained to me what it meant to have this disease. He needn't have explained to me that I might die. That's not really necessary. But that he had simply told me what I had to pay attention to, what could happen, and what they were going to do to try to get the better of it." (F13/58/P)

Participants often reported a lack of knowledge among doctors about rare diseases and, accordingly, insufficient information provision by health professionals. While few patients and family members contacted specialists at an early stage, most respondents reported that general practitioners and specialists with limited experience in rare diseases did not give the required information. Often people stated that they are the only patient with a rare condition in their doctor's office and that their doctor communicated openly that he has not dealt with this disease since medical school. Even though many participants expressed understanding regarding the ignorance of physicians about the multitude of rare diseases, this creates a feeling of being alone with the disease and inability to cope with information seeking.

"Inform yourself? Yes, of course, but where? (...) one is really alone with one's illness." (M11/72/P)

"And I have to say that one cannot expect anything different from doctors, all registered doctors. Of course they did not recognize it; they are not familiar with this disease." (M15/48/P)

"And the doctors, well, even the primary care physician has no idea. I am the only patient with this condition in his practice." (F23/58/P)

This sometimes deficient level of knowledge among non-specialized physicians also reflects in the communication between doctors and their patients. In some interviews patients and their relatives reported an inability on the doctor's side to inform and communicate about the disease. This relates to the transmission of the diagnosis as well as information on the disease, it's severity, disease progression, and treatment options.

"So, first I tried to speak to my primary care physician, as the diagnosis in my case was not so clear, regarding what type of the disease it is. Is it essential thrombocythemia or polycythemia vera? And then I tried to speak to her about it, and she asked me which one of the two is worse. Um, then I thought, why don't you just spare me your questions?" (F26/47/P)

Nevertheless, participants greatly appreciate practitioners' willingness to learn more about their illness and their commitment to deal with it more in depth. Some people reported that their family doctor or their specialist in private practice familiarized him- or herself with the specific condition and obtained access to all of their documents. This involvement also reflects in the quality of the relationship between health professionals and their patients. According to the interviews, in some cases, however, doctors were not willing due to time restrictions or the low probability to ever have a patient with this disease in the office again.

"The present primary care physician took over everything from (...), so we have a very good relationship. When I have questions or problems, he listens to me, so it could actually not be better, and he has all the documentation, he has read up on it." (F40/49/P)

"My gynecologist, for example, asked whether she could take the results with her? She said she needed to inform myself about it, because she didn't know anything about the disease. I thought that was good." (F37/66/P)

"(...) But I don't think that the doctor had any time, because I am the only one/ for example I am my neurologist's only patient with that condition. And I think, I can understand if she does not feel like reading through all the documentation." (F36/41/P)

According to our respondents some doctors communicated, that they are pleased with the fact that their patient searches for information on his- or herself. Especially in a later stage, when patients become their own expert on their illness, physicians put the responsibility, for example in regards to therapeutic decisions or referral to other doctors on the patient or the relatives.

"(...) Last week I went there and said again that we need a referral to the hospital. No one just gets a referral to the hospital when they go to the doctor, right. First of all he says he has to examine you. And then he says, sure. You know what you have, you know where to go, go for it." (F18/47/P)

In many cases the picture is different when it comes to specialists in rare diseases, which are often located at the centers for rare diseases. Interviewees reported great satisfaction regarding the specialist's level of information and felt that they are able to give patients and their relatives a comprehensive advice. Moreover, people stated that their attending specialist takes time to address questions that remained unanswered and is endeavored to transmit the latest information to them. One person, however, related that even physicians in specialized centers offer too little information by themselves due to daily routines.

"So, then he took the time to explain everything to me in detail, the things that I had not understood." (F6/49/A)

"Doctors seldom volunteer to start talking about it. Even in the case of specialists, if I want to know something, when I am in doubt, I have to enquire. Because they are not very talkative or do not want to give you any information about it. Because of course, they also fall into a routine. And they are not even aware of the fact that every new patient initially arrives with zero information." (M15/48/P)

Since only few patients and family members get in contact with specialists at an early stage and receive the required information, many interviewees search online for information. Some experienced difficulties, either with the large amount of online information or the inability to distinguish between "good" and "bad" information.

"And I can remember when we were confronted with the diagnosis, that we did an unbelievable amount of research on the Internet." (F2/46/R)

"As wonderful as the Internet is, there is just so much available and such a flood of information that you don't really know what can be applicable to specific individuals, because you just don't know what applies to whom." (M2/48/R)

Specific information needs regarding rare diseases

Clinical picture

Analysis of the interviews showed a series of specific information needs regarding rare diseases. These needs may differ due to the heterogeneity of rare diseases. Nevertheless, numerous patterns could be identified across patients' information needs.

Many people stated requiring information about their clinical picture, especially immediately after diagnosis. They perceived basic information on what it means to live with the disease as desirable.

"Well, if so, then it happened at the very beginning. I consider it to be the most important factor, that so .../ one already knows what it is when the diagnosis is made. They say something and I think, 'What is that?'" (F4/43/R)

By now, the genetic background of increasingly many rare diseases is apparent, so, aspects of genetic causes and inheritance patterns are important to participants. This concerns information on genetic interrelations in general, coupled with information on inheritance patterns for close relatives or future children. In this context, family planning proved important. Women and men wishing to have children considered knowing if they could bear children important, in spite of illness, or if deciding to bear children is morally acceptable. Patients reported needing detailed information for decision making. People are also interested in possible alternatives to childbearing.

"Yes, and for me personally, cystic fibrosis also means sterility for a man, and I read a lot about this problem on the Internet: What alternative possibilities there were to having children etc." (M16/46/P)

"That's just such a big question: Could I even conceive? Could it even be successful or not? Of course, you ask yourself such entirely existential questions. (F28/27/P)

Information needs concerning medical issues are also closely linked to disease progression and life expectancy. Most interviewees reported preferring to have been informed on expected developments within the following years and decades and life expectancy.

"I also wanted to know whether my son would eventually get going. I wanted to know how it would develop and then someone told me that they simply don't know. I also wanted to know; in 2006 it was really bad; I simply asked how long he was actually going to live. Typically, I would never ask that, but it was such an issue, and then someone told me that they just don't know." (F3/60/R)

Other patients or parents of severely affected children disagreed; arguing that being informed about potential developments is neither helpful nor necessary and might elicit fears. Dealing with such negative aspects of illness might reduce present well-being.

"Because why should I worry about what is happening to me, if I will perhaps only need a transplant in 20 years? Why do I already need to know today how that works, everything that will happen, how many people survive it, and how bad it is? That's not always good." (F26/47/P)

Some patients and relatives preferred basic information early on, describing what the illness is all about and how it will affect everyday life; later on, more detailed information needs would emerge.

"One didn't feel as though one was sufficiently informed. One always wants to know a lot about things that affect them. And there, we will never know enough. Even what I know today isn't really adequate." (F27/44/P)

Therapy

Closely connected with the clinical picture, are questions concerning therapy options. Our interviewees prioritized learning of any possibility of treating their diseases. In this context, questions emerge regarding the availability of drugs to successfully control the disease.

"Yes, to know about the illness in general, whether there is any chance of being cured or any developments in this direction. Therapies or something like that." (M9/66/P)

"Yes, I'm interested in receiving therapy. Whether I could (...) improve something with pharmaceuticals." (M13/71/P)

People with rare, treatable diseases require information regarding any associated side effects and if other patients can share their experiences with the drugs. Moreover, patients with a syndrome, dependent on different drugs, required information on interaction with other drugs.

"That was really the point at which I had this Transient Ischemic Attack and had to start taking medicine that I would have to take for the rest of my life. Where one becomes more intensely involved. What am I actually doing to my body, what

are the side effects, what effect does this have on the illness?" (F28/27/P)

"Oh well, I'd say, what is always difficult is the possibility of interactions of the medication with other medicines. That is also a little vague." (M3/50/R)

Almost all respondents cited large demand for information on specialists treating rare diseases. Most wished for information on specialists near their homes. The major issue, however, was interviewees' willingness to travel long distances, upon learning that specialized institutions or physicians are dispersed across Germany.

"Classic questions are about locations of treatment, about doctors. There aren't many doctors who really deal with this disease. One is so glad to get information about whether there are doctors or other points of contact, possibilities for treatment, a cure, centers, and if yes, where, in which neck of the woods." (F27/44/P)

Research

Generally, according to interviews, patients might be eager to participate in research. Patients and their family members reported interest in obtaining information about research efforts in rare diseases. Participation in scientific trials raised substantial hope among interviewees, particularly those whose illnesses cannot yet be treated surgically or pharmaceutically. Some participated in different studies, hoping to benefit from innovative therapies. Some reported altruistic motives and wishing to help others by supporting scientific studies. In this context, information on clinics, in which relevant studies are located, is of particular importance. People want details on inclusion criteria, new substances being tested, and actual participation procedures.

"Occasionally, there were a few therapies or therapy studies in which he could participate, hoping that they could possibly improve something. Naturally, there is also a need for information. Are there any studies available, does he qualify for these studies, how does it work, how do we get access to the study centers?" (M1/44/R)

"For people who have the disease, it's actually very important for them to be able to see that something is happening. They are doing a lot of research at the moment. And when I saw this, I thought, 'Super, I will probably be able to benefit from it, and will still be around to experience it.' This is really important information that one receives." (M15/48/P)

For scheduled studies, it is crucial that sustained improvement of people's health be expected from treatment options under investigation.

"In principle, I don't need to know everything about all the research that raises its head and then disappears from view in the next two years because it turns out not be all that promising after all. Instead, I would like to know about simple things that could lead to treatment, or which could bring medium or long-term improvement to patients." (M12/47/P)

Psychological counselling

Living with a rare disease can be a huge psychological burden and necessitate therapeutic support. Some patients reported that being affected by a chronic and yet incurable disease causes considerable fears and insecurities. Moreover, a rare disease can also place a heavy burden on the family system; for example, when siblings do not get the attention that they need or when resultant physical constraints lead to interdependence among partners. Therefore, some patients consider seeking professional help for themselves or their families. The following questions arise in this context: How can I access therapy? Up to how much and under which preconditions will costs be covered by statutory pension insurance? How does illness affect one's mental health?

"And then I asked in a one-on-one conversation, hmm, 'Who can you recommend?' And then I learnt that, yes, PERSON is totally, totally amazing and it was just my luck that (…) the costs of consultations with a neuropsychologist were also taken over by the health insurance. That was really, really nice, yes, just being able to find out what effects neurological problems can have on the psyche." (F30/40/P)

"What I tried, first of all, was to find a good psychologist, a good one, where I could get an appointment, but the retirement insurance turned it down, it was an appeal against it and so forth." (F33/52/P)

Interviewees needed the contact information of therapists with experience in specific rare conditions. Patients preferred therapists to know about disease effects and be able to address individual needs.

"Because I actually expected that there would be specialists for this illness, with regard to the psychological changes. But at the moment, it appears to me that that is only the case in the organic field. And not in the psychological field." (F7/45/R)

Some family members described different needs from those of patients with rare diseases, concerning psychological support. While patients urgently struggle with coping with the diseases, their possibly fatal outcomes, or have difficulties accepting physical changes, mental stress, in terms of caring responsibilities or relationship changes, was a major issue for relatives.

Social law and law governing benefits

Nearly all respondents described social law aspects as important issues regarding which information acquisition is very difficult, stating that initiative by many people is required for information acquisition. Furthermore, other people's experiences with social law and its effects on them are important in information procurement. We found a lack of central-contact partners offering support on such. Participants also reported that even if they know the authority responsible for specific social law matters, employees could not always provide information.

"You always have to depend on your own initiative, your own knowledge, or help from other affected people, and you get no information or help from elsewhere – you have to make a great effort to seek it out by yourself. One wishes that there were a possibility to contact someone locally. Officially, this is

supposed to be the case in the offices. There are actually officers responsible for the disabled in the local and integration offices, and so forth, but by the time you have found out whether these points of contact exist, the child is actually too old for that department." (M2/48/R)

Particularly with regard to submitting applications, interviewees reported high demand for information. In this context, the application for a disabled person's pass is quite significant. Many patients and family members criticized not having been informed about the possibility of submitting an appropriate request, in the first place.

"What I find discouraging is that no one tells you that you have the right to a severely disabled person's card, because you're not as productive and resilient as before. That also affects your working life. Or you have the right to more vacation time, because you really need it. Or you have a supplementary pension allowance or can retire earlier. Mm, you just never get this information; you have to work it out or learn about it yourself." (F29/36/P)

If their right to file an application has been clearly pointed out to them, people contend with whether owning a disabled-persons' card is beneficial or not. Therefore, people want to be informed about advantages and disadvantages of the issuing of a disabled person's card. Additionally, it is essential for them to know where and how an application can be successfully made. In this context, some interviewees cited uncertainty regarding regional differences, since regulations applying to one state were not necessarily applicable in others. Thus, it was difficult for them to estimate whether the information found on the Internet applied to them or not.

"Well, I think this entire area of social security law and social legislation is important. This game I have to play, in order to apply for a severely disabled pass. Should I go to the pensions office, the social assistance office, the local authorities, or where do I go to for something like this? I think that is difficult because it is regulated differently from state to state and even from district to district. So, that is something that would certainly be a help." (M8/39/P)

Closely related to this, is which contact persons or institutions may help complete the forms required. Physicians' and health professionals' role, however, is a moot point. Some interviewees reported substantial support by doctors, whose specific knowledge enabled a successful initial proposal; others claimed that due to weak financial incentives, physicians do not offer any support.

"They kept on writing reports for me and I also sent them on, until now, maybe there is a certain sentence that they need to write .../ and now I have to wait again. Yes, after four weeks I could file an objection, but in order to do so, I would have to get in touch with the doctors again, and so forth, and I don't have the time. Then, I simply wait and maybe resend it again next year with more recent doctors' reports, I don't know. (...) Now they want to think about who should do the application, because the skin clinic apparently can't do it, because it wouldn't be paid for." (F13/58/P)

Therefore, people mainly receive support from self-help groups, affected others from among own acquaintances, or local social workers.

Resources

Interviewees cited medical aid as another area with major knowledge gaps. As the interviews show, there exists hardly any disease-specific information on medical aids, such as wheelchairs, prostheses, or dressing material. Furthermore, most available information is provided by suppliers of medical aids. Therefore, some participants expressed concerns about the independence and objectivity of this information.

"Naturally, the medical aid supply sector - which is a disaster - gets involved. Typically, patients are counselled by their healthcare supply store, which recommends the most expensive equipment. (...) But this is surely an area that is sadly lacking in information, for both the doctors' and the patients' point of view." (M8/39/P)

Patients forming part of support groups often receive information about this from other affected members. Use of the right medical aid can often substantially increase peoples' quality of life and well-being.

"For years, I had a normal 0815 wheelchair that we all know from hospitals. A heavy contraption. No one told me that, at my age, I was entitled to a low wheelchair. I only found out about it from the federal association. (...) I'm coping very well with it! It didn't cost me a cent – the medical insurance had to pay for it. But if people don't know about it." (F27/44/P)

Some interviewees reported uncertainties regarding correct handling of their medical aids. Physicians reportedly gave little information on these issues. In contrast, others described independent centers for medical aids as useful institutions, where people have the opportunity to try them out, themselves.

"Yes, if you can inform yourself about the wheelchair, how to use it ... I had to teach myself everything, and I also fell on my face a few times. But there aren't any offers in this regard – you have to investigate them yourself, somehow." (F31/61/P)

Medical aid issues are also closely related to reimbursement issues. In this context, the role of health insurers was controversially discussed. Some interviewees reported support by their health insurance; others described making an application as a strength-sapping process made unnecessarily difficult. This makes it more complicated for people to cope with their diseases.

"Let me put it like this: I've actually had pretty good experiences. But I know many people who have had very bad experiences. My health insurance really consents to everything. After I sued them. But you have to fight a lot. That discourages and weakens you. You know it would be good for you, but you don't get it or you have to pay for it yourself." (F27/44/P)

Practical information for everyday life

Almost all interviewees cited great demand for information helping them cope with problems and challenges in everyday

life, on account of disease. However, practical information for everyday life issues is hard to find.

"Well, those are the special ones, but you have no access to them. And even the practical ones, as I always say, you have no access to them either. (...) Where you receive really practical advice." (F19/44/P)

From the interviews, we identified a wide variety of everyday issues that patients have insufficient knowledge of. For patients with a rare disease, with a visible impact on their outer appearance, clothing plays an important role. In this regard, patients want to know how physical changes, described as a psychological burden, can be covered with clothing. In other respects, physical conditions due to illness often do not permit wearing of regular clothing, but that responding to patients' disease-specific needs.

"Everyday stories, such as where you can find suitable shoes. This is really difficult. For example, rubber boots - basically, you can't buy them "off-the-shelf;" you somehow have to buy two sizes too big and cut them at the top, so that you can get into them." (M5/41/R)

Another question arising was that of foodstuffs, which might further rapid progression of patients' disorders. According to patients and interviewed relatives, information on how an optimal diet might look was transmitted by nutritionists, physicians, and other patients.

"(...) But what's important is knowing what factors I need to avoid, so that it doesn't get any worse for me than it already is. (F29/36/P)

"Information on what you need to pay attention to, with regard to nutrition; I saw it there; in principle, that was the most important. I think that was with the histamine... the diet then, that I searched the most, yes." (F23/53/P)

Information on sports activities that are feasible and conducive for people's health is of equal interest to those interviewed. The main focus here is the appropriate sport for one or one's relative, considering the disease, and where to find suitable courses and contact persons.

"You (...) ask yourself, what kind of a sport, and of course one that he enjoys, (...) at which he can also excel? Where he has the chance of keeping pace, and is nevertheless healthy, rather than unhealthy, that is in terms of joints and the spine. And where can he learn that? As far as this is concerned, there's a continual need for information." (M5/41/R)

Particularly for parents or other close relatives of children with rare diseases, the question arises as to whether the children need special tutoring in kindergarten, due to disease-specific deficits. In this context, people cite uncertainties regarding the appropriate types of kindergarten for the children, against their individual circumstances, and how to deal with their diseases in these settings.

"How does it work with kindergarten, traditional kindergarten, an integrative kindergarten, special support measures? These were always points regarding which information was lacking." (M1/44/R)

For parents, the impact of the child's disease on his/her schooling career is important to know. They reported the need for information on whether and how to communicate their children's diseases in the school setting and what possible advantages and disadvantages are for speaking for or against an open-minded approach.

"And (...) when he starts school, at the latest, as we learn that from parents who have children who are a little older, the question naturally is: What will they do at school? How will they explain his situation to the class, and how can we make sure early on that he won't be underestimated or bullied?" (M5/41/R)

At a later stage, the persons affected reported demand for information on professional career types and possible limitations, due to illness. In this context, patients with very rare diseases reported difficulties finding out more about professional development, because there are hardly any other affected persons who share their experiences.

"I would say that it is problematic in areas of work and further training, because since there are only a few people involved, there are also few experiences." (M6/32/P)

For some adults living with a rare disease, pursuing a regular job is an important prerequisite to dealing positively with their illness daily and maintaining some normality in their life.

"Because as I said, from Wikipedia, it's very medical, and devastating to an extent. And I find, if there's a chance for a person to survive it, and to survive it well, and also become professionally active again, that can really help someone get there, too. That's what I think, and it would have been nice if such information had been available." (F13/58/P)

In the professional context, dealing with the disease is important. Many patients reported rather cautious handling thereof, fearing rejection and indirect discrimination by employers. Some patients, for example, expressed their concerns with not finding a new job, if the employer is informed about the illness.

"For me, I think, this is crucial. Or, for example, if I have a job interview, how much information do I need to give my employer about my health, in the first place (...) these are things about which one is unsure." (M12/47/P)

"I finished my studies last year and am now on the threshold of a professional life. Then come the questions: Should I tell my employer or just not bring it up?" (F28/27/P)

Self help

For almost all our respondents, exchanging experiences and views with other people affected plays a significant role when gathering information. Substantial information received concerning medical, practical, or legal topics was provided by patient associations or other patients, in general. Our respondents classify the practical knowledge of people with the same disease and experiences as particularly trustworthy and helpful. Many patients described the dialogues with other patients as an exchange at eye level. Therefore, patients and

their relatives search for self-help groups shortly after diagnosis, to contact those affected.

"Well, the most information, what helped us the most, was really the self-help group. Here, the exchange can really begin between two people who are on the same level, where one doesn't need to start each time by explaining the symptoms and the whole background, but rather begin tackling the issue." (M2/48/R)

"They can ask all of that (at the self-help association). What have other people gone through; what is important? Where one can ask questions, if one has become involved. (…)You can trust your doctors, but it's always good to have someone who has already experienced it, whom you can ask how it was." (F27/44/P)

However, finding those groups and associations was not easy for all our interviewees. Particularly, patients and family members of people with very rare diseases told us about problems getting the desired information.

"Information about self-help possibilities and self-help groups should be more present. In particular, their activities, and what they can do for those who have this disease. This is a big problem. And, especially for patients in rural areas, it is important to create networking opportunities and to refer them to groups and to provide them with information." (M16/46/P)

Patients and relatives discuss various themes among themselves. Especially in the period following diagnosis, our respondents reported the desire for an experienced contact person helping them accept the new situation, and sorting the information heap. Moreover, they hope for patient-suitable information on basic disease-specific questions and first-hand advice. As the disease progresses, more detailed information is required. Patients, for example, want to learn from others on how to deal with the disease in different settings, doctors under recommendation, and how to proceed with legal matters.

Comprehensibility of information

Besides patients' and relatives' information needs concerning certain issues, as mentioned before, there are also needs regarding formal aspects of information. Often, for inexperienced and newly diagnosed patients and their relatives, it is challenging to understand the information available. What was criticized in this context was not linguistic expression or the length or structure of the text, but the use of unexplained foreign words. This does not only apply to printed or web-based information, but also that given in personal conversation with physicians. High demand for information in a simple and easily comprehensible language can, therefore, be assumed.

"Someone came in and started speaking technical jargon, and I said, 'Stop! I don't speak this mumbo jumbo, I speak German! So, please tell it to me plainly.' And then he explained it to me." (M17/60/P)

"(…) Since, in these first three weeks of life, we also researched a tremendous amount in the Internet, but naturally, it's difficult for laypeople - I mean, neither my husband nor I studied medicine or human genetics." (F8/32/R)

"Well, with the (…) classic informational media, that is, printed brochures that are also sometimes created by the doctors, informational content was very high, but the comprehensibility not that great. This is because doctors tend to use specialist terms that are quite obvious to them, but which need explaining to patients, in case of doubt." (M16/46/P)

Additionally, patients expressed the need for up-to-date information. As far as possible, patient information should reflect the latest developments in practice and science. Equally, up-to-date experience reports from other people affected are required. Particularly with regard to information brochures and scientific books and papers, which our interviewees sometimes rated as more reliable than online information, this demand cannot be covered completely, yet.

"(…) What's happening at the moment isn't up to date. They have a brochure, but it's not up to date." (M15/48/P)

"But then again, there is the problem that many reference books just aren't reprinted regularly, that they are no longer current. And that again is a disadvantage with a disease like this, which is not yet very established. Because until a new edition appears again, they simply limp along; surely that's more accurate than a lot of the stuff on the Internet." (F12/53/P)

Patients with very rare diseases argued that information was not always detailed. Many interviewees required detailed information at a later stage, for example, about their condition and therapeutic possibilities, but had problems finding these.

"Well, I find that very difficult. Now that I know what's wrong with me, I actually find that on Wikipedia, it's also very, yes, I wouldn't say superficial, but one just reads it, but they don't really go into it in detail. And the problem for those of us who have VHL, is just that the symptoms vary. For example, the kidneys are not discussed in detail on Wikipedia. If I think that there is really something wrong with my kidneys, then I want to find out something about it in a more targeted manner." (F18/47/P)

Discussion

Despite often diverging conditions, several issues relating to the relevant information needs of people with rare diseases have been identified. Soon after diagnosis, patients and their families need basic and easily understandable information about future health-related expectations and steps. Then, too extensive information on mortality, for example, might overstrain patients, rather than enable coping with the disease. Later, detailed information on, for example, medical aids, patients' rights, or research efforts is required. In most fields, support of other affected people has played a major role in information acquisition. Experiences and knowledge of other patients or their relatives were considered particularly

trustworthy. Findings are seemingly consistent with the results of few previous studies on the topic. In these investigations, issues on causes, symptoms, and treatment were areas of concern and interest. Moreover, patients want information about the meaning of living with a specific rare condition and its expected impact on future life.

To improve patients' knowledge on all their disease-specific matters and help them cope with their conditions, a patient-suitable information portal on rare diseases should be developed, considering all the identified information needs. This could integrate all relevant information at one central point, allowing easy access to information. It would enhance the health competence of those affected, which is largely determined by information quality, availability, and adequacy. Information presented particularly to newly diagnosed and inexperienced patients or relatives should ideally be suitable for laypersons, without foreign words. The contact information of self-help groups should be presented concisely, since they are one of the first contact points from which people seek information. To also enhance information transmission by doctors, whose information on rare diseases is insufficient, patients' needs should also be captured and respected. This way, patients' quality of life and care can be sustainably improved.

The study has some limitations. Due to the qualitative design, the results' applicability is limited; therefore, they are not generalizable to all people with rare diseases. This study aimed to extensively determine patients' and families' information needs during the disease course, which might enhance abilities to cope with the disease and optimize the management of everyday life. Nevertheless, generalizations cannot be made about whether and to what extent these identified needs occur among all affected people in Germany. This would require a follow-up quantitative approach, testing the findings on a representative sample. Another limitation is that information needs are difficult to remember, especially when the period to which statements are applicable was long ago. For some interviewees, diagnosis was decades ago; so, they could hardly remember and reflect on information demands.

Conclusion

Even though the information age allows online information-seeking, reliable distinction between high-quality and sub-quality information is difficult. Moreover, available data on many rare diseases are scarce. Therefore, it is important to consolidate quality information at a central point that is easily accessible and trusted by patients. The challenge is developing such a central information portal on rare diseases, to account for all the demands of different patient groups at different times.

A lot of information needs of patients with rare diseases and their relatives are still unmet. For many patients, personal contacts, via self-help associations / support groups and other patients, helped alleviate uncertainties and knowledge gaps. These information needs often reflected personal experiences with doctors, medical aids, and legal affairs. To meet patients' and families' information needs, identified issues and demands should be comprehensively presented / integrated in modern information systems. Implementation should also comprise reference to competent contacts for those issues, matters, and needs, which are difficult to meet on a web-based information portal. A well-designed information portal can help improve the conditions of those affected, by enabling informed decision-making and simplifying access to specialized services and providers.

Also for doctors, who should be aware that they have a particular role in serving patients who search for professional help, such an information portal can be a reliable source of information. Often general practitioners and specialists, who usually don't deal with rare conditions, are the first point of contact for patients in the healthcare system. Therefore, patients and their families appreciate when physicians provide appropriate information and guide their way to specialized care. But also in the further course when patients and their relatives are looking for a competent healthcare near to home, physicians are important contacts for those affected. Doctors should therefore raise their awareness of rare diseases and search proactively for information, when they care for patients with rare diseases or those who might be affected.

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Ethical Approval

All participants gave their written informed consent for participation. An ethics committee (Ethics Committee of the Hanover Medical School) approved the study, before commencement, ensuring that the study was in accordance with the ethical standards of the 1964 Helsinki Declaration and its later amendments or comparable ethical standards.

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Modul 3

Use and importance of different information sources among patients with rare diseases and their relatives over time: a qualitative study

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Use and importance of different information sources among patients with rare diseases and their relatives over time: a qualitative study

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Abstract

Background: Finding reliable information on one of more than 7000 rare diseases is a major challenge for those affected. Since rare diseases are defined only by the prevalence criterion, a multitude of heterogeneous diseases are included. Common to all, however, are difficulties regarding information access. Even though various quantitative studies have analyzed the use of different information sources for specific rare diseases, little is known about the use of information sources for different rare diseases, how users rate these information sources based on their experiences, and how the use and importance of these information sources change over time.

Methods: Fifty-five patients with a variety of rare diseases and 13 close relatives participated in qualitative interviews. For these interviews, a semi-structured guideline was developed, piloted, and revised. Data analysis involved a qualitative content analysis developed by Philipp Mayring.

Results: The participants considered internet as the most important and widespread information source, especially for early information. Although patients have difficulty dealing with information obtained online, they consider online searching a quick and practical option to gather information. During the course of the disease, personal contact partners, especially self-help associations and specialized doctors, become more important. This is also because information provided online is sometimes insufficiently detailed to answer their information needs, which can be complemented by information from doctors and self-help.

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Conclusions: People rarely use just one type of source, but rather refer to different sources and informants. The source used depends on the type of information sought as well as other person-related factors such as preexisting knowledge and the disease stage. To improve people's information searching and connect them with medical specialists in rare diseases, a central information portal on rare diseases might be a suitable access point to provide free and quality assured information for patients, caregivers, and physicians. This would allow not only patients but also doctors to find quality assured information on symptoms and therapies as well as patient associations and specialized doctors.

Keywords: Rare diseases, Information sources, Informants, Health information seeking, Qualitative research, Content analysis, Self-help, Online information, Written information

Background

In recent years, rare diseases have become an important issue. Although a uniform definition is still pending, rare diseases are globally characterized only by their low prevalence. In Europe, "rare diseases" is the umbrella term for diseases that affect less than or equal to 1 in 2.000 people. Although rare diseases can differ greatly in type, symptoms, and causes, affected people usually face similar challenges. These include insufficient information. On the one hand, this is because many rare diseases are so rare, that only little information exists. Beyond that, information is often widely dispersed and difficult to find in the vastness of the internet or literature, so that access is limited [1].

However, it is undisputed that information plays an important role in coping with illness [2–8]. Based on Antonovsky's concept of sense of coherence, perceiving the world as comprehensible, manageable, and meaningful enables an individual to cope with critical life events [9]. In this context, information can make a decisive contribution in helping individuals develop their sense of coherence [10]. Understanding an illness's causes, symptoms, and impact seems to be a precondition for dealing with the disease in everyday life and can increase people's quality of life. Accordingly, not being provided adequate information about the disease and its implications can lead to feelings of resignation and fear among patients and their families [5–8]. Moreover, information is an important prerequisite to know where help can be found and pave the way to specialized centers and providers [3]. Both, again, impact patients' health situations. However, the information needs of individuals with rare diseases and their relatives include not only medical knowledge regarding diagnosis, therapy, progress, and prognosis but also information on various other aspects of the disease. These include practical information for everyday life, psychological counselling and social law aspects [3, 11]. Therefore, knowing how patients as well as their family members, who can also be strongly affected by their relatives' disease, search for information is an important issue. To shape the information gathering process as well as possible and thus meet the patients' information needs optimally, knowledge is needed on how searching is done, what sources are used, and what relevance different sources have. Moreover, how the use and relevance of different sources change over time should be investigated.

Information searching patterns from patients suffering from chronic but not rare diseases have been extensively analyzed. Numerous studies revealed different sources of information, which are of importance to different groups of patients, but mostly cancer patients [12–18]. These range from physicians, who are often rated as one of the most favored and trusted sources [13, 17, 18], to information brochures, the internet, as well as non-medical professionals, such as pharmacists and nurses [14–16]. Other generally used sources of information include books, newsletters, and mass media sources. For patients with common diseases, family members and friends were also used to gather disease related information [14, 16]. Moreover, some factors have been identified that affect people's search for information. Female patients were reported to inform themselves more often and to use more sources than male patients. Additionally, younger patients and those with a higher education showed more frequent information seeking behavior than older patients and people with a lower educational background [15]. Regarding phase of illness, it was found that shortly after their diagnosis people favored written information while at a later stage relatives and friends become increasingly important [16].

Because of the specific characteristics of rare diseases, such as unpredictable courses, limited available knowledge, lack of exposure in the media, etc., it can be assumed that information seeking behaviors by people with rare diseases are not completely similar to those of patients with common diseases [19]. However, still little is known about how people affected by a rare disease and their families search for information. Previous studies may be outdated, have relied on few single sources,

focused on specific information needs only, and did not focus on rare diseases in general [19–24]. Teixeira et al. [22] conducted a questionnaire survey of 1019 patients with a rare blood disorder, which showed that medical specialists are of particularly great importance when it comes to sources that were widely used for information gathering. In this study, respondents who reported feeling sufficiently informed about genetic testing and its implications for their health mostly reported having received this information from medical specialists before family doctors and support groups. Even though medical specialists were also the source they most trusted, they would like to get more information from their family doctor. Furthermore, among patients who did not feel sufficiently informed, the majority answered that they would like to gain information from their family doctor. Additionally, non-medical sources, such as patient associations, websites, nurses, and printed information were of importance to the respondents. After medical specialists, patient associations were the most trusted information source. Due to the high level of knowledge possessed by patient associations, these are often called patient experts [25].

General practitioners also proved to be one source of information patients would like to receive significantly more information from, according to a study by Matti et al. [21]. They identified preferred sources of information based on responses from 30 patients with multiple sclerosis (MS) and found that there was a discrepancy between the amount of information people actually receive and the amount they would like to obtain. Moreover, eye specialists and neurologists were identified as sources they would like to receive more information from. Regarding MS patient associations and MS specialist nurses, patients reported an almost ideal amount of information that was being provided.

An older study from Lanigan and Layton [20] on 108 patients with a rare skin disorder drew similar conclusions. The results from their questionnaire survey also illustrate that medical specialists were the most used and preferred information source, followed by general practitioners. However, it must be considered that this study occurred before the arrival of the internet, so that its relevance for today's conditions is limited. Wibberly et al. [23] studied 16 patients with a rare lung disorder and identified various information sources by means of face-to-face interviews. These include primary healthcare physicians, patient information leaflets, as well as the World Wide Web, nurse specialists, and patient support groups. The most valuable information sources were medical specialists in rare lung diseases, nurse specialists, as well as patient support groups.

Carpenter et al. [19] also confirmed that physicians and the internet were the most used and credible sources for patients with vasculitis to obtain information on medication, followed by pharmacists, and other affected people. Based on an online survey of 232 patients, they also found that family and friends are not relevant sources of information, presumably because they do not hold much information on rare diseases. Additionally, gender differences were found. While male patients, unlike female patients, rated their spouses or partners, as well as nurses as fairly credible sources, female patients preferred medication package inserts and the internet as sources of information.

Molster et al. [24] conducted an online survey of 810 patients with different rare diseases and found that the most sought and preferred sources of information were medical specialists and patient organizations followed by friends and family members. Regarding non-personal sources, respondents stated that they prefer to be referred to an information website or social media. Other preferred types of information sources included printed media, such as leaflets and brochures, as well as journal articles.

To summarize, family doctors and medical specialists, the internet, and support groups are of great importance for patients with rare diseases when searching for information on their disease. However, limited studies have investigated the use and perceived credibility of information sources over time and if so, their reasons for it. Since existing studies are based mostly on quantitative methods, further qualitative research is needed to analyze how people with rare diseases assess different sources and on what experiences. Due to its open approach, qualitative research can achieve a deeper understanding of peoples' attitudes and causes. The aim of this study, therefore, was to obtain a holistic picture of the information sources used by patients with various rare diseases and their relatives; specifically, what relevance they attach to different sources and how this relevance changes during the course of the disease.

Methods

Due to the lack of substantial data on information sources in the field of rare diseases, the authors decided on a qualitative study design. Thus, it is possible to explore under-researched areas with maximum openness and reveal all aspects of importance for patients and their families concerning finding information. To detect patients' experiences regarding information acquisition and information sources used, semi-structured interviews were conducted. Therefore, we developed an interview guideline, stimulating people to tell us about their medical history and the way they searched for information (Table 1). Since the research team included young associates with mostly theoretical knowledge in qualitative research, this was done in close cooperation with an

Table 1 Semi-structured interview guide

Set	Principal questions
Experiences with the disease (from patients who consciously experienced their diagnosis)	Please remember the beginning of your disease. What changes did you notice?
	How did diagnosis proceed?
	What happened after diagnosis?
	When imagining yourself in that position again, how did you feel?
Experiences with the disease (from patients who did not consciously experience their diagnosis)	Please tell me about your disease and how life has changed due to it.
	How does your disease affect your everyday life?
	Some people want to learn more about the diseases that they live with. How about you?
Information seeking and information needs	How was that, striving to find information about your disease?
	Do you remember any events that you associate with increased demand for information?
	Please tell me about situations in which it was easy to gather information.
	Please tell me about situations in which it was difficult to gather information.
	Which moments do you consider important in searching for information?
Type of access	Please imagine the many possibilities of modern and classic media to communicate. Please recall your own situation. Which media did you use when searching for information?
	Which medium would you prefer for accessing information?
Completion	Are there any other topics that you would like to talk about?

external specialist at Hanover Medical School, who has long-time expertise in qualitative health research. The specialist conducted a workshop during which they shared the knowledge and skills required for planning, conducting, and analyzing qualitative interviews. In addition, extensive literature studies were carried out. Afterwards, the authors developed a first draft of the interview guideline and discussed it jointly with the specialist. A concerted version was then presented at a research workshop held at Hanover Medical School with several internal and external qualitative researchers, during which revisions were made to generate the final version. Individual sources of information and their usefulness could be derived from this. After pretesting the interview guideline with three patients with rare diseases, we observed that patients diagnosed before or shortly after birth found it difficult to answer the opening questions and narrate their diagnostic paths. An alternative conversation starter was then added, to ensure that it was suitable for such patients too.

To select a heterogeneous and balanced sample, several medical experts from the project consortium divided the total of rare diseases into eleven different groups of diseases in accordance with the affected organ systems. It was planned to conduct six interviews in each group as well as ten interviews with patients, who had to wait for at least 10 years until they received a correct diagnosis. Thus, a total sample of 76 patients was planned to be recruited. Nevertheless, upon saturation of interview data, a smaller sample would suffice. Participants were recruited consecutively over several months by a physician and GCP trained study investigator Freiburg Center for Rare Diseases (FZSE) at the University Medical Center Freiburg, University of Freiburg, Germany. Patients with rare skin diseases and their relatives were reached out directly through FZSE via personal approach during patient visits (gatekeeper sampling) and board notices (sampling by self-activation). To recruit other groups of rare diseases, more centers for rare diseases belonging to the consortium of rare diseases (AG-ZSE) were included as well as patient organizations.

Care was taken to establish a consolidated interview atmosphere with the participants. Therefore, researches allocated enough time and visited patients and close relatives at home whenever requested. Telephonic interviews were conducted only if participants requested for it. After making small talk, we explained our research project in detail and answered any questions. We emphasized that all data would be kept strictly confidential and that anonymity would be ensured, so that retroactive conclusions concerning the participants would not be possible.

The interviews were analyzed following the structured content analysis method by Philipp Mayring [26]. Each audio recording was verbally transcribed and read into MAXQDA analysis software. Subsequently, two researchers worked through the first three interviews and marked all relevant text passages. To develop an extensive system of categories (Table 2), a deductive-inductive

Table 2 Coding tree

Core categories	Sub categories
Print media	Books
	Brochures/leaflets
Television	
Helpline	
Internet	Non-specified
	Patient organization
	Medical facility
	Encyclopedia
	Social media
	Scientific database
	Email/newsletter
Personal contacts	Center for rare disease/specialized clinic
	Primary care doctor
	Pediatrician
	Specialized doctor in outpatient practice
	Parents
	Social worker
	Patient organization
	Other affected persons
	Congress

procedure was used. Several categories could be derived from the theoretical framework based on previous research on information sources for rare diseases, including medical specialists, patient organizations, and primary care doctors. These were completed by inductive categories if they appeared from the text. This procedure was followed by a critical examination and, if necessary, modification of the original categories. Afterwards, the marked text excerpts were analyzed with regard to the research question. After assessing the interview transcripts, the researchers conducted three focus groups with participants of the interviews and one focus group with patient representatives and members of the Alliance of Chronic Rare Diseases in order to discuss and validate the findings.

Extracted citations were translated by an external translation service, approved by a native speaker and then included in the paper. The following will accompany direct interview quotations: Gender ("M" for male, "F" for female), a consecutive number, age, and status as either a patient ("P") or relative ("R").

Results

We interviewed a total of 55 patients and 13 close relatives between March and December 2014 (Table 3). There were almost twice as many women ($N = 45$) as men ($N = 23$). Participants' mean age was 50.5 years. The interviews lasted 10–143 min, with 68 min on average.

Based on the evaluation of the interviews, a multitude of different information sources used by patients and their relatives for gathering information on rare diseases was revealed. The authors disclosed four main themes that were of importance in nearly all interviews. These main themes include the "internet as the first source of information" (theme 1), which describes the relevance of online searches for those affected. The second theme highlights the role of patient organizations and other patients in the information retrieval process, which allow for communication at peer level. Doctors and their perception as a source of information by persons affected is illustrated in theme 3. Lastly, theme 4 deals with written information.

The internet as the first source of information on rare diseases

Many of those interviewed reported in detail about their struggle to receive a correct diagnosis. Often this meant a long-lasting and emotionally charged odyssey. The need for information, once a diagnosis has been made, was accordingly high. In this context, for almost all the respondents, the internet and especially search engines such as Google were one of the first sources to search for information on their own or their relatives' disease. According to the interviewees, this allowed them quick and uncomplicated access to information. In this context, different approaches to how to proceed when searching online for information were identified. Most of the participants simply googled their disease and clicked in a more or less unstructured or unskilled manner through the provided information websites, while others advanced more systematically. In many cases, it was possible to establish a connection between people's searching approach and their prior knowledge. Patients or family members, who work in the health sector and are familiar with medical terms, demonstrated a more targeted and satisfied approach to research online than those without medical backgrounds. Moreover, people who are familiar with online searches reported fewer difficulties.

> *"It was when everything was new. We took in all the information we could."* (M47, 59 years, P).

> *"Well, the information is primarily shared over the internet."* (F14, 57 years, P).

> *"(...) I enter it into the internet and then find the information. It would now be the easiest and*

Table 3 Participant characteristics

Patients		
Age	Gender	Group of rare disease
23	female	Genetic skin disease
32	male	Cystic fibrosis and pulmonary disease
32	male	Immunodeficiency
39	male	Skeletal dysplasia
66	male	Genetic skin disease
85	female	Connective tissue disease
70	male	Connective tissue disease
72	male	Genetic kidney disease
47	male	Congenital metabolic disease
50	female	Immunodeficiency
53	female	Genetic skin disease
58	female	Genetic disease of the digestive tract
54	female	Cystic fibrosis and pulmonary disease
57	female	Immunodeficiency
44	female	Neuromuscular disease
43	female	Cystic fibrosis and pulmonary disease
47	female	Neuromuscular disease
71	male	Neuromuscular disease
44	female	Genetic skin disease
53	female	Connective tissue disease
72	male	Genetic skin disease
48	female	Immunodeficiency
54	female	Genetic skin disease
58	female	Congenital metabolic disease
72	female	Immunodeficiency
48	female	Genetic kidney disease
47	female	Congenital blood formation disease
44	female	Skeletal dysplasia
27	female	Congenital blood formation disease
36	female	Genetic kidney disease
40	female	Congenital metabolic disease
61	female	Neuromuscular disease
48	male	Congenital blood formation disease
44	female	Genetic eye disease
52	female	Genetic eye disease
46	male	Cystic fibrosis and pulmonary disease
60	male	Neuromuscular disease
62	female	Neuromuscular disease
48	female	Genetic eye disease
61	female	Connective tissue disease
66	female	Congenital metabolic disease
18	female	Congenital blood formation disease
64	female	Congenital metabolic disease
37	male	Cystic fibrosis and pulmonary disease
49	female	Congenital metabolic disease
59	male	Genetic kidney disease
70	male	Connective tissue disease
45	female	Genetic kidney disease
51	female	Genetic kidney disease
62	female	Genetic eye disease
39	female	Neuromuscular disease
51	male	Immunodeficiency
40	male	Skeletal dysplasia
74	male	Cystic fibrosis and pulmonary disease
69	female	Immunodeficiency
Relatives		
Age	Gender	Group of rare disease
44	male	Neuromuscular disease
48	male	Skeletal dysplasia
28	female	Genetic skin disease
46	female	Genetic skin disease
60	female	Skeletal dysplasia
50	male	Neuromuscular disease
43	female	Skeletal dysplasia
46	male	Congenital metabolic disease
40	female	Genetic skin disease
49	female	Cystic fibrosis and pulmonary disease
45	female	Genetic skin disease
32	female	Genetic disease of the digestive tract
41	male	Skeletal dysplasia

quickest way for me." (F17, 47 years, P).

"Well, when I am looking for something like this, I will look at Wikipedia first, because I think it's great and well-structured. Yes, then I do not know anymore. Then you land somewhere at large. What just/whichever link appeals to one, but I cannot recite it now." (F67, 45 years, R).

Even though, the internet was perceived as providing easy and quick possibilities for information seeking, most respondents did not report satisfaction with the search results at the beginning of their research. Dissatisfaction, for example, arose when only little information was available. This was particularly the case when people were affected by very rare diseases with only a few sufferers or few research efforts. Otherwise, finding a multitude of information was also challenging for

searchers. Interviewees, who told us that there was a wide range of information, often felt they are not enough of an expert to manage these amounts of data. Moreover, people suffering from diseases that proceed differently in each individual case recounted problems comprehending what information is correct for specific cases. Younger persons and people who use the internet on a regular basis reported fewer difficulties with large quantities of information than those who are unskilled in online searches and of older age. Moreover, it could be seen that people reported fewer difficulties as the disease progressed and their expertise grew.

> *"I also think that it is better, I think it sucks when there are several million websites when you look up cancer or the like. I also think that if someone gets diagnosed with cancer, he immediately wants to know what impact it will have. If there are then a thousand websites, you will go completely crazy."* (F17, 47 years, P).

Another challenge reported in connection with online searches was that of dealing with information that is perceived as frightening. Many interviewees told us that when they started searching, they found information on the internet that was shocking, for example regarding life expectancy, severe courses of disease, etc. This information was so dreadful that some of our interviewees did not continue their online research. In this regard, some patients criticized being left alone with their findings and worries and wished for greater support from their doctors. Being alone with this information, in their opinion, could incite panic or despair. The results suggest that when people start searching they do not have enough expertise or support by others to put information into its proper context and assess it correctly. Our interviewees, in this connection, expressed the need for a closer support, especially by their doctors.

> *"Well, I was only on Wikipedia. What I read there shocked me, because it sounded extremely bad. After that, I never went onto the internet again."* (M60, 46 years, R).

> *"You stand there alone, and that is, that is the problem, when you stand alone with your illness. Err. Meanwhile you think about it and say: Mhh. And now?"* (M38, 60 years, P).

Furthermore, peoples' perceptions of the utility and credibility of the information found online varied greatly. This became obvious in regard to who is behind the information (website), what information is communicated, and how. Since most patients and their relatives barely know about their own or their relatives' disease shortly after diagnosis, the assessment is based partially on who is the websites' operator rather than on the contents of the information itself. Many of our interviewees first encountered Wikipedia when they started searching online. Some of them rejected this website, since the information offered there was too generic for them. Others criticized Wikipedia because it does not control its information, which can be changed arbitrarily by anyone at any time. In contrast, other patients and family members expressed positive views about it. From their point of view, especially in the beginning, Wikipedia is a good source of information to get an idea of the disease, its causes, symptoms, and progression. It was also highlighted that this information, compared with others, was clearly structured as well as quickly and freely available. Looking back, some people who now have an extensive knowledge on their disease rated the quality of the information offered there as good or high.

> *"(…) and then, after the appearance, one decides what is serious, yes, who is behind it, (…) are the err, here mmhhh Alliance of the chronic/well, the ACHSE associated, NAMSE associated, yes."* (F35, 44 years, P).

> *"Yes, I had, of course, I have. I then do not want useless information, because of my job I also have reasonable/ well, I would never at Wikipedia, we already had it."* (F19, 44 years, P).

> *"I just entered it and then usually ended up at Wiki. Wikipedia. It was the most reliable for me."* (F14, 57 years, P).

Medical databases on the internet, such as PubMed, were hardly used. Often only interviewees with medical backgrounds reported knowing these sources of information. This was described as an advantage in relation to other patients who do not have medical backgrounds, due to its high quality and current information.

> *"I therefore rather checked at PubMed or so, but it was of my advantage, because I have been active in the field myself."* (M65, 40 years, R).

Patient organizations and other affected persons – information sharing at peer level

In many cases searching the internet for information helped patients or their family members to contact patient organizations at an early stage. No interviewee reported being informed by their doctor about this way to receive support and information. Almost all our respondents who used a patient association

website valued their supply of information highly. One person, however, criticized that their information was not comprehensive and current enough regarding new developments and findings. Another patient, who visited a website that was not specialized on one disease but a group of diseases also reported lower satisfaction, since there was detailed information only on the more common rare diseases. Other interviewees praised their relevant and helpful information. In particular, concerning information on issues in everyday life, such as finding medical specialists near to home, dealing with the disease in family and working environments, etc., self-help organization websites provided crucial hints. One person especially emphasized that his patient association helps to make the latest findings accessible to the general public by translating English scientific articles into German and displaying them on the website. Thus, patient organization websites contribute to knowledge transfer and access. For many of our respondents, patient association websites provided the most reliable and high-quality information, so that after identification, no further websites were used.

> *"Well the main information, the thing that helped with our progress the most, was the support group. The exchange actually starts there, when you join in on the conversation at eye level (…). "* (M58, 48 years, R).

> *"It strengthens one, when you sometimes think you are insane. (LAUGHS) Yes, because everything changes and one thinks, yes why am I feeling so bad, why am I always tired and hurting? But when you have the opportunity to exchange stories err, then you can put your mind at rest, because you learn that, ok, it is normal."* (F31, 36 years, P).

> *"No. I never looked it up, because I have to say, up to three years ago we regularly participated at the annual meeting of the support group or the regional meeting in LOCATION and therefore the information actually was sufficient."* (F51, 62 years, P).

Interviewees particularly valued the close personal contacts made with those committed to self-help. When a rare disease leads to similar and severe progressions and is accompanied by comparable restrictions and challenges as those of affected individuals, patient organizations play a key role in information gathering. While there is sometimes too little time for patients in the medical setting, in the self-help field patients with rare diseases and their relatives often feel that people take a lot time for their issues and needs. It was often reported that the personal contact resulted in a close and strong contact between existing members of the patient organization and the interviewees for years. Furthermore, people see information from patient organizations as an opportunity to gain practical knowledge that goes beyond the perfunctory information they receive from the internet. Since rare diseases often show an individual progression, online information is perceived as too generic, while self-help contacts meet the demands for more specific information.

> *"I then called the chairman myself and he immediately took an hour of his time and answered everything, the questions, that I already had and more (…). "* (M64, 46 years, R).

> *"And those are the information, which the doctor does not give you, how I deal with everyday life, when I need what."* (F22, 72 years, P).

> *I: "How do you judge the quality of the information?"*
> *P: "That however is good, well only the information about the support group, nothing else."*
> *I: "And the information, that you found on other sites in the internet?"*
> *P: "No. It was too general, unmeaningly."* (F10, 50 years, P).

Nevertheless, some patients feel no need for personal exchange or even reject the principle of self-help. This is based mainly on the assumption that it only serves the purpose of commiserating with each other. This can be noticed, in particular, among people who have trusting relationships with persons outside patient organizations, such as medical specialists in hospitals, who are available to answer any questions. However, individuals who are reserved about the idea of self-help due to this assumption often have no practical experience with self-help at all. Others see no additional benefits since disease progression differs too greatly from one person to the next. Moreover, people with a mild disease course sometimes do not make contact with patient organizations, since their need for information and exchange is low. They reported being able to cope with their situation and pointed out that they get along. Furthermore, meeting with patients with serious disease progression is perceived as discouraging.

> *"Whining does not help; therefore, I do not sit down and moan. I do, however, understand the people that complain in the support group. Yes, I do not know if it helps them."* (F39, 62 years, P).

Physicians, basic health care provider and highly specialized experts

During their medical care process, patients and their families often met many different physicians. Although some patients reported receiving a quick diagnosis and were referred to specialized care from the very beginning, such as patients with cystic fibrosis, which can be easily diagnosed shortly after birth, many respondents first consulted their family doctor when searching for a diagnosis and did not attend a medical specialist until a later stage. Even in the further course of treatment, not only medical specialists, but also primary care doctors play an important role due to community care provision. The experiences with doctors outlined by the patients and their families are, however, very heterogeneous.

> *"I was lucky to be under the care of a very experienced orthopedist from an early stage on (…). "* (M04, 39 years, P).

> *"Yes, I was not amused about it, but also not depressed. Every time I was told that it was not it, we somehow made new attempts to get a diagnosis. I have also been to a lot of so-called experts on muscles."* (M18, 71 years, P).

Preference for commitment and support instead of knowledge transfer from general practitioners

Many of the participants, who first contacted their general practitioner (GP), feel dissatisfied regarding information provided by their doctor. Many of our interviewees criticized that their doctor gave too little or even no information on their disease. Especially when patients received their diagnosis they complained about too little and barely patient friendly information. Even though patients and their relatives understand that doctors, who do not deal with rare diseases on a regular basis, cannot hold information about all rare diseases, they would wish for more transparent dealing with that lack of knowledge.

> *"I have to say that, when it comes all doctors, (…) you cannot expect anything else from them, they did not identify it, do not know this disease, that is to say, if you go there, here, my hemogram is not in order, standard things get asked (…). A good doctor can recognize that a level is out of the norm, but that was of course also a little stupid, sort of, that he did not think to look into the other direction too."* (M34, 48 years, P).

> *"Yes, and there I was the one time, err, with my telephone and thought, yes, maybe the doctor will say something about it, but no, it was done for her! She had the diagnosis and it was over. I am supposed to look for someone, who mhm, yes look for a doctor."* (F28, 47 years, P).

Patients expressed frustration and resignation with general practitioners who refused to seek assistance for their limited knowledge. Particularly, shortly after receiving a diagnosis, when specialized centers for rare diseases or contact partners had not yet been found patients felt left alone and helpless.

Nevertheless, other patients reported high satisfaction with information transmission from family doctors. In many cases, this contentedness resulted less from an immediate and comprehensive offer of information on the GPs' part, but more from the commitment to learn more about their patients' conditions and go in search themselves. However, even if the GP did not acquire the knowledge by himself but through the patients or their relative, this was highly valued. From the interviews, it was found that in such cases GPs often became trusted informants, near to their homes, who played an important role in patient's health care provision.

Specialists and centers for rare diseases – trusted and current disease-related information

For almost all interviewees involved in specialized care, such as at centers for rare diseases or university hospitals, the doctors working in these institutions are an important information source regarding medical issues. Besides patient organizations, medical specialists in these centers were often described as key informants on disease specific information. After diagnosis, as well as in the course of the disease when the state of health deteriorates noticeably or treatment becomes necessary, the need for information sharing with specialized doctors arises. Many of our interview partners reported very high information quality and valued the fact that specialized carers are available for all kind of questions. The currency of the information was furthermore praised. Because of their proximity to research efforts and other experts, medical specialists have up-to-date knowledge that they pass on to their patients, which is highly respected. One interviewee, however, complained that he would have to claim medical specialists' information instead of doctors transmitting their knowledge by themselves.

> *"For me, it is enough to have the feedback from the very knowledgeable skin clinic."* (F01, 23 years, P).

> *"Professor PERSON always tried to share his knowledge and his research with his patients."* (F40, 48 years, P).

Of particular importance is also the fact that patients and their families normally have fixed individual

contacts in the centers for rare diseases, who are entirely familiar with their disease history and symptoms. In this context, people also positively highlighted not needing to repeatedly explain their condition, which was felt as a relief. Some people also discussed longstanding and trusting physician-patient relationships arising from that, allowing for low-threshold contact, as well as quick and personal answers to all medical concerns. From the interviews, it became clear that patients and their families also see medical specialists and centers for rare diseases as a (good) complement to the range of information offered from their patient's association. While those hold relevant and most trusted information on most issues beside medical issues, medical specialists are especially important regarding detailed aspects concerning therapy, diagnosis, etc.

Printed information – high quality, but not up to date information and sparsely used

Even though many of the patients and their relatives in our interview sample received information to a large extent from the internet or personal contacts, others, however, reported the wish for printed information. On the one hand, this is because people appreciated the possibility of holding something in their hands, where they can look things up again, when they feel like it. This was especially emphasized at earlier stages of disease progression.

> *"(…) I would rather need it in writing, to refer back to again."* (F31, 36 years, P).

Shortly after diagnosis, for example, information brochures are perceived as helpful sources, since they provide comprehensive and often comprehensible information. Moreover, people reported that brochures are well suited for bringing them on the day of doctors' appointments to give them a review of their disease. At later stages, however, brochures do not cover people's needs for more specific and detailed information.

> *"Well I also (…) got the booklet, how do I deal with it myself and where can I get help from. Very good information, yes."* (F43, 18 years, P).

> *"Here you go. My husband has brought me informative literature, because I knew that he (doctor) did not know it. I pushed it into his hands and told him to read about it."* (F31, 36 years, P).

Some of the interviewees found it helpful to read magazines offered by patients' associations. Additionally, for those who did not actively participate in regional meetings or did not look for personal exchange, this type of information provision was important. In this connection, patients especially highlighted experience reports from other affected patients and families as valuable information.

> *"The most important source of information was simply/ the newspaper of Glandula. Publicly displaying the personal experience reports that people wrote there, the stories of what they have been through, when they got diagnosed. That is what I realized and what I took in."* (M47, 59 years, P).

Additionally, specialist books were used for information gathering, but some of the interviewees put them aside, discouraged by the medical terminology. Especially in the time shortly after diagnosis, they exceeded the capabilities of patients and relatives. People also complained that books would often not be up to date, a fact that can be important when considering that specialist books often refer to medical issues such as therapeutic options, which could be subject to frequent amendment.

> *"(…) and that is anyway the medical terminology and how can you as a layman go and change it for yourself, or read it, it will not do, it does not work."* (F17, 47 years, P).

> *"As mentioned before, books, they definitely are not; they definitely do not have the latest insights."* (M55, 74 years, P).

Discussion

Different sources for different needs

Patients affected by a rare disease as well as their relatives use a variety of different sources to keep themselves informed. In accordance with previous quantitative studies of specific rare diseases, among others, especially the internet, patient associations as well as specialist doctors play an important role when gathering information [6, 20–23]. However, to date, the types of information sources used by patients with various rare diseases and their families, how they assess these information sources, and how their value changes over time have remained unclear.

From our interviews, it was shown that initially after diagnosis, when the need for information is very high, only few patients obtained detailed and profound information from their doctors. This is in line with a study by Molster et al. [24] who reported that almost three-quarters of the surveyed patients with a rare disease received little to no information at the time of diagnosis. A systematic review investigating experiences of patients

with rare diseases found that more than half of the included studies reported lack of knowledge among health professionals about patients' rare diagnosis [8]. Most patients and relatives therefore searched online for further information and were confronted with a flood of information. To assess the quality and relevance of such information and deal with frightening information is a difficult task for laypersons. Therefore, this first step of information search is often a frustrating and intimidating experience. Contact with other affected persons can help patients and relatives to find their way through the thicket of information by placing them into a proper context and thus, gain a deeper understanding of the disease. Additionally, doctors can contribute to successful information acquisition if they face the challenges that people with rare diseases bring to their care provision openly. This includes that doctors show willingness to become acquainted with their patients' diseases and do not leave them alone with information acquisition, especially in the initial time after diagnosis. This is underpinned by various studies [5, 11, 27–29]. Lack of involvement is common among health professionals when they lack experience in their patients' diagnosis [27]. Particularly, when medical professionals withdraw in such cases, it can lead to feelings of resignation and insecurity [5]. Efforts to mitigate their lack of knowledge, on the other hand, are highly valued by patients with rare diseases [11].

We were also able to show that the use of different sources is not stable, but can change over time. While, for example, people regarded the internet as an uncertain source of information due to information overload in the beginning, at a later stage their perception changed as they learned a more targeted approach to search and carefully choose which websites to use. Thus, our results indicate that the importance of different sources varies depending on, among other things, the state of disease progression and the state of knowledge.

Great potential for patient associations

The interviews have shown that patient organizations play a major role in people's information acquisition. Previous studies of different rare diseases have confirmed the importance of patient organizations and knowledge sharing with other people experiencing the same condition [11, 19, 21–25]. As a contact partner at peer level, they can help people to cope with their disease by offering comprehensive and comprehensible information as well as guide their way to specialized care by helping patients and families to find competent caregivers from the very beginning. This way, time-wasting detours in information searches can be avoided. As described by the interviewees, patient associations can close the gap of information offered by medical specialists in rare diseases, by not only providing medical information, but information relevant to everyday life. Huyard [11] reported a similar finding among patients with one of the six rare diseases and their parents. They sought answers to questions regarding living with the disease in daily life, such as how to lead a happy life, from other affected persons [11]. Therefore, information from patient organizations should be regarded as an important supplement for information offered by patients' caregivers. However, despite very positive self-help growth, its potentials do not seem to have been completely realized. Nowadays, there are approximately 60, 000 self-help organizations with a health-related focus in Germany, but only a small number of those deal with rare diseases [30].

Although possibilities for participation have increased over the past decade, in the future self-help associations should be even more integrated to improve patients' health care. As we have shown, no patient or relative from our interview study was made aware of the possibility of contacting a patient organization by their doctor. Under the term of "self-help friendliness" different attempts to institutionalize relationships between carers from the in- and outpatient sector and self-help associations in Germany have been made [31]. In this context, a set of commitments has been agreed to sustainably integrate self-help on a collective level into health services [32]. In the stationary sector, for example, different quality criteria have been defined to ensure a close connection between hospitals and self-help. So far, however, few care facilities have joined these voluntary collaborations. In the future, carers in the field of rare diseases should also endeavor to collaborate with patient associations. Besides opening their medical care provision to knowledge and experiences from patient organizations, they could also strengthen contacts between their patients and self-help groups and thereby support their patients' coping processes.

However, to permanently secure patient organizations' work, sufficient funding is required. Even though, the funds approved by statutory health insurance recently increased due to the Prevention Act (PrävG) adopted in 2015 [33], it is still unknown whether patient organizations have sufficient financial resources to sustain their important work. Especially, for self-help in the field of rare diseases, which is often characterized by local groups with a limited number of members and low public visibility, sustainable funding to maintain their services seems to be endangered; hence, further research is needed. Moreover, still little is known about the economic potential of self-help groups. The study shows that patient organizations play a major role for patients to find highly specialized care units. This is also interesting from an economic point of view. Patient organizations do not only provide

information very efficiently at low cost but also provide a communication platform for patients to exchange their worries, fears, experiences, and observations. Until now, the role of patient organizations has not been assessed from a health economic point of view and, therefore, should be studied in the future.

Online sources for quick and easy information gathering and recommendation for a central information portal on rare diseases

Besides the great potential for self-help associations, it has been demonstrated that online information is currently of crucial importance for patients and their families to gather information. It especially enables newly-diagnosed patients to search for information quickly and easily. Additionally, in more advanced stages of the disease, people rely on online information in case they need information on current developments. Dissatisfaction, however, arose due to the unfiltered flood as well as the unknown quality of the information.

Therefore, new approaches for optimizing and developing user oriented information systems are preferable. For this reason, efforts have been made to establish and implement an information portal on rare diseases (ZIPSE) [34], where patients, their relatives, as well as medical professionals can access clearly presented and high-quality information from a central web based point. Since information provides the basis for coping with the disease and receiving specialized care [2–8], such a portal can help to improve patients' health situation sustainably. Besides increasing their quality of life, reduced doctor-hopping and targeted therapy can help to use limited financial resources more adequately. This also allows doctors, who cannot hold information on all 7000 rare diseases, to obtain information, for example, on treatment options, medication, or specialized medical colleagues when necessary. This could also help on the caregivers' part to make their patients' healthcare more efficient and compensate for the uneven level of information, which was often criticized in the interviews. Physicians should be conscious of their important role in people's health care and endeavor to better inform themselves on their patients' diseases, and give them specific assistance regarding which websites to use and where self-help contact partners can be found.

Strengths and limitations

The purpose of this study was to gain insights into how people affected by rare diseases experienced their search for information, which sources of information they used, and how they assess different sources. We conducted interviews with an extensive sample of patients, with a variety of rare diseases, and their relatives, revealing a wide range of attitudes and opinions. Unfortunately, not all aspects that have been mentioned in the interviews could be reproduced in detail in this manuscript due to lack of space. Rather, the main themes were presented as comprehensively as possible. Therefore, supplementary observations regarding information sources and their potentials should be a topic for further publications.

It must be noted that our sample included individuals who had been living with the rare disease for many years and whose information needs may not be as high as those who have been recently diagnosed. Hence, recall bias cannot be completely ruled out. Nevertheless, living with symptoms, finding a correct diagnosis, and searching for information on the disease represent phases of great significance for patients and their relatives; thus, a sufficient ability to recall could be assumed.

Due to the qualitative approach of this interview study, it is not possible to generalize the findings to patients with rare diseases and their relatives as a population. It must be kept in mind that findings from a qualitative survey must be embedded in their spatial and temporal context [26]. However, that does not mean that they are not transferable to other people and situations. The creation context, however, must be considered when applying the findings to a new context.

Moreover, it was not possible to conduct theoretical sampling due to limited access to patients and their families as well as time restrictions. Sample recruitment was carried out by the Freiburg Center for Rare Diseases (FZSE) at the University Medical Center of Freiburg, University of Freiburg, Germany. As this center specializes particularly in the treatment of people with rare skin disorders, it was difficult to gain access to patients with other rare diseases. Nevertheless, by covering most of the planned six interviews in each group and reaching a saturation point at a later stage of the interview process, a heterogeneous and balanced sample can be assumed.

It should be mentioned that the interviews were obtained from a study of the conceptualization and implementation of a central information portal on rare diseases. This study identified the information needs of people living with rare diseases, their families, and of health professionals to integrate them into the information portal. Nevertheless, the researchers evaluated the interviews regarding important information sources in an unbiased way and with maximum openness.

Conclusions

In our study, various information sources, such as the internet, self-help organizations, and doctors, have been confirmed as important access channels for people living with a rare disease and their families. Due to the qualitative approach, reliable statements on the reasons why, and how important they are to patients and their families have been made for the first time. Moreover, it was

possible to show how the importance of different sources changes over time.

For physicians, especially those who do not deal with rare diseases daily, this does not mean they must hold information on all 7000 rare diseases, but they do need to know where to get quality assured information when necessary. For them as well as patients and their families, a central information portal, such as ZIPSE, might be an option. Interested people can find here bundled high quality information on a large number of rare diseases, which makes searching for information easier. It can also raise awareness of services from patient organizations that are of particular importance for patients and their families as they help to bring them together with specialized partners and address their need for practical everyday information as well as share experiences.

Abbreviations

AG-ZSE: Consortium of rare diseases; F: Female; FZSE: Freiburg Center for Rare Diseases; GCP: Good Clinical Practice; GP: General practitioner; M: Male; P: Patient; PrävG: Prevention act; R: Relative; ZIPSE: Central Information Portal on Rare Diseases

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Authors' contributions

SL conducted interviews and analyzed and interpreted the data. MF, TH and JMS contributed to the methodological conception of the interview study. AB and DR conducted interviews and contributed to the interpretation of the results. FS recruited patients and relatives for participating in our study and contributed in conceiving the manuscript. SL drafted the manuscript and all authors were involved in its review and revision. All authors read and approved the final manuscript.

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Availability of data and materials

The datasets generated and analysed during the current study are not publicly available due to data privacy but are available from the corresponding author on reasonable request.

Ethics approval and consent to participate

This study received ethical approval from the research committee of the University of Freiburg in February 2014 (ref: 53/14).
Written informed consent was obtained from all individual participants included in this study.

Consent for publication

We have obtained consent to publish from the participant to report individual patient data.

Competing interests

The authors declare that they have no conflicts of interest.

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Telephone health services in the field of rare diseases: a qualitative interview study examining the needs of patients, relatives, and health care professionals in Germany

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Abstract

Background: Rare diseases are, by definition, very serious and chronic diseases with a high negative impact on quality of life. Approximately 350 million people worldwide live with rare diseases. The resulting high disease burden triggers health information search, but helpful, high-quality, and up-to-date information is often hard to find. Therefore, the improvement of health information provision has been integrated in many national plans for rare diseases, discussing the telephone as one access option. In this context, this study examines the need for a telephone service offering information for people affected by rare diseases, their relatives, and physicians.

Methods: In total, 107 individuals participated in a qualitative interview study conducted in Germany. Sixty-eight individuals suffering from a rare disease or related to somebody with rare diseases and 39 health care professionals took part. Individual interviews were conducted using a standardized semi-structured questionnaire. Interviews were analysed using the qualitative content analysis, triangulating patients, relatives, and health care professionals. The fulfilment of qualitative data processing standards has been controlled for.

Results: Out of 68 patients and relatives and 39 physicians, 52 and 18, respectively, advocated for the establishment of a rare diseases telephone service. Interviewees expected a helpline to include expert staffing, personal contact, good availability, low technical barriers, medical and psychosocial topics of counselling, guidance in reducing information chaos, and referrals. Health care professionals highlighted the importance of medical topics of counselling—in particular, differential diagnostics—and referrals.

Conclusions: Therefore, the need for a national rare diseases helpline was confirmed in this study. Due to limited financial resources, existing offers should be adapted in a stepwise procedure in accordance with the identified attributes.

Keywords: Rare diseases, Telemedicine, Health-seeking behaviour, Helpline, Health information

Background

Rare diseases (RDs) are predominantly very serious and chronic diseases as approximately 80% are genetic in origin. Therefore, they often have a negative impact on the life expectancy and quality of life of those affected. In particular, people suffering from very rare RDs occurring once among 100,000 people are in danger; 5000 to 8000 different RDs have been detected thus far, accounting for 6% to 8% of the population [1]. Therefore, approximately 350 million people worldwide suffer from an RD, and half of them are children. People affected often struggle to obtain a proper diagnosis as healthcare providers have little experience of these conditions, and there is limited research evidence available. In addition, treatments, which, when available, are very expensive. These hurdles trigger an odyssey through health service systems and, in this context, the search for helpful health information. However, useful,

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high-quality, and up-to-date information is often hard to find [2].

The following article examines the potential of telephone services in satisfying this desire and elicits the revealed health-seeking behaviour. It introduces 'helplines' as services solely offering telephone-based information. Different types of information are differentiated, such as references, counselling, and/or medical information. Comparing different information access points, helplines are currently often used after the Internet and booklets [3]. Per the findings of Mevissen et al. [4]. Internet information can be delivered in addition to telephone information but should not be seen as an adequate replacement. Highlighting the importance of helplines aligned with other information access points, Ekberg et al. [5] offer an explanation for these findings as they show that emotional support needs are often intertwined with information- or advice-seeking needs.

The case of helplines in the literature

To present helpline research, a brief indicative literature review was conducted by searching the MEDLINE database. The DIMDI (Deutsches Institut für Medizinische Dokumentation und Information, German Institute of Medical Documentation and Information) platform was used as a search tool. In all, 233 results were generated, including the keywords 'helpline' and 'help line' (search date: 27 May 2016). Results concerning animal research were excluded. Exclusion and inclusion criteria were set as displayed in Table 1.

Telephone services were often mentioned as being useful as a recruitment tool for participants of other health-related studies or the evaluation of health policies. These studies were excluded as they are not relevant in this context, leaving a total of 83 results. Findings are often based on the evaluation of caller statistics illuminating the profile of callers (65%, 54/83). Besides, many studies conducted a thematic analysis of telephone conversations. Questions were raised about caller satisfaction, perceived effectiveness, and support provision. Only five studies used interviews as a research method. Another five studies conducted structured literature reviews. Health professionals were rarely included. Most of the studies evaluated helplines addressing issues such as tobacco cessation (15%) [6–19], psychological problems (13%) [20–31], cancer (14%) [32–43], and family planning and sexuality (13%) [44–54]. Trials dealing with the specific concerns of RD patients and their physicians could not be determined. Building an argument for telephone services in general proves to be very difficult as helplines contribute to very heterogeneous health-related outcomes. Two studies, for example, use a successful referral to an appointment as an endpoint for the measurement of effectiveness of a helpline on sexuality and family planning [55, 56]. Tobacco cessation helplines with proactive counselling monitor the chance of quitting, [57] and psychological helplines the number of suicide preventions [58]. Other benefits are rather intangible and therefore difficult to measure, confronting helpline research with the criticism that little robust evidence is generated [59]. For example, general practitioner (GP) helplines offer access to the health care system after closing hours [60]. Furthermore, users of helplines for family planning, addiction, and violence perceive a telephone service as beneficial due to the ability to talk anonymously about delicate health issues [54, 61]. In this regard, helplines offer the chance to identify as well as bridge gaps between patients and health care service systems and, thus, play an important role in health care systems. Therefore, existing research suggests potentials of RD helplines; however, this hypothesis still needs verification. Ferreira et al. [62] report that helplines designed after patients' needs contribute to the overall satisfaction of citizens with health care systems and their effectiveness, therefore highlighting the need for further research on what exactly is needed. The presented literature suggests that there are differences in themes, staffing and structuring of helplines, which should be thoroughly thought of. Encouraged by these findings, we further investigated potentials of RD helplines, resulting from the gap between information offering and need, to further improve health-seeking processes of people affected by RD.

Table 1 Inclusion and exclusion criteria

	Inclusion criteria	Exclusion criteria
Population	All potential patients (not only focusing on rare diseases), family members and physicians	Studies regarding animals
Intervention	Telephone services	Email services, Internet platforms
Outcome	Examination or improvement of helpline service or design Evaluation of caller behaviour	Examination of helpline callers to examine their general health behaviour (not offering additional knowledge to helpline design)
Publication type	Caller statistics, interviews and reviews	Interventional studies
Language	English, German, and French	All other languages
Time frame	All publications up to May 27th, 2016	None

As the literature search did not reveal any RD specific publications, we added a targeted manual search aiming for literature on RD helplines. Only one paper from Houÿez et al. [63], summarizing caller statistics of the European Network of Rare Disease Help Lines (ENRDHL), was found. However, the retrospective design did not allow for any recommendations improving existing structures. Additionally, two oral presentations [64, 65] mentioning RD helplines as part of national information provision on RD were listed. Other initially identified literature reported on non-specific disease helplines. Besides, Iskrov and Houÿez [66] also analyzed ENRDHL callers. On the other hand, Mazzucato et al. [67] stressed the need for telephone services parallel to other information systems. They forward an argument for bundling RD helpline services at a national level, noting that feedback concerning the functioning of RD policies can be retrieved immediately. However, data on the operational realization has not been raised.

Political endorsement

Political processes have initiated national efforts targeted towards the improvement of the overall situation of individuals suffering from RD. In the EU, for example, policy proposals for the improvement of the overall situation were summarized in the European Commission Communication on RD in November of 2008 [68] and the European Recommendations to Member States by the Council of Ministers in June of 2009 [1]. Consequently, EU member states were encouraged to develop national plans to enact these requirements. Germany, for instance, published a National Plan for RD, the National Action League for People with RD (Nationales Aktionsbündnis für Menschen mit Seltenen Erkrankungen, NAMSE), in August of 2013 including 52 policy proposals [69]. Part of this action plan is the improvement of knowledge transfer through the expansion of disease-spanning, quality-orientated, and Internet-based information databases and systems. Towards this goal, the Central Information Portal for RD (ZIPSE – www.portal-se.de) was implemented. This is in line with an increasing international effort targeted towards the improvement of information structures. A growing number of national and transnational RD Internet platforms evolved [70].

Alongside an Internet-based information provider, the implementation of a telephone-based information service has been conceived as an alternative information access point. The Commission Communication also mentions the need for national RD helplines. To this end, the ENRDHL was named and created in the context of the European Rare Disease Solidarity Project (RAPSODY, September of 2006 to April of 2008). The focus of this initiative is the improvement of quality of services and providing a unified standard by sharing the experiences of European RD telephone helplines [71].

This demand is clearly highlighting the crucial point. Helplines do already exist, as an Orphanet list on international RD helplines [70], however, projects such as RAPSODY show that there are efforts necessary to set common standards. Besides, ENRDHL consists solely of members from eight countries, plus two countries in which helplines are still under construction [63]. Germany is not yet listed. However, NAMSE policy proposals 38 and 39 include the analysis of the implementation of a telephone service. NAMSE recommends to set up "[…] a pilot project to determine which target groups would make best use of such a hotline, what types of questions would most often be posed and what answers can best be delivered to these questions. This information would serve to determine the probable frequency and type of questions and how to plan to best meet these demands." [69].

The present article

The literature search shows how important telephone services are for health care service provision. Besides, there is little knowledge on RD helplines. Particularly, the perspective of potential callers has not been chosen in helpline research so far. This enables us to capture all relevant aspects for the design of a satisfying and effective RD helpline. The secondary aims of the underlying article were to add to the existing literature and to allow for substantiated decision-making in the political context aiming for the improvement of information provision for patients, family members and physicians. In this regard, the major aim of the study was to examine the needs of patients, relatives and health care professionals for a telephone based health service for RD in Germany triangulating perspectives of all potential callers, interviewing individuals suffering from RDs, their relatives, as well as health care professionals (HCPs).

Methods

Setting

The interview study was conducted as part of ZIPSE project, aiming for the implementation of an Internet platform for information on RD and considering a telephone service as an additional information access point. Patient and relative interviews were carried out between March and November of 2014 by three interviewers. HCP interviews were carried out by two interviewers between April and October of 2014. A qualitative setting was chosen as this design not only offers the opportunity to provide a first impression of a possible need structure but also drafts an RD helpline through the eyes of those interviewed. Forty Interviews

with patients and relatives were conducted face-to-face and telephone-based in 29 cases. One interview could not be evaluated as the record was not readable due to technical difficulties, leaving 68 recorded interviews from patients and family members. In the case of physicians' interviews, 39 interviews were conducted. Only seven interviews were carried out using the telephone to avoid long travel and scheduling on short notice. A change of interview medium was necessary due to the broad geographic coverage of the study within Germany.

Participants

Patients and relatives were recruited through the Freiburg Centre for RD located at the Department of Dermatology of the University Medical Centre at the University of Freiburg and through RD self-help groups. The equal coverage of the many disease groups summarized under the broad definition of RD was targeted. Therefore, six participants were equally chosen among genetic skin disorders, skeletal dysplasias, neuromuscular disorders, genetic eye disorders, disorders of the connective tissue, genetic kidney diseases, cystic fibrosis and lung diseases, inherent disturbance of haematopoiesis, immunodeficiencies, inherent metabolic disturbances, and genetic diseases of the digestive tract. However, interview results showed in nearly all cases a complex, polysystemic pattern of involvement. At least nine patients had experienced a long process of diagnosis with duration of search for a diagnosis of more than 10 years. Thus, adding 66 patient and relative interviews to 10 interviews with prolonged diagnosis, a total sample of 76 patients was planned to be recruited. Nevertheless, interim analysis showed that upon saturation of interview data, a smaller sample would suffice. Further interviewing was not performed as this would not have led to expanded knowledge on the research subject. The final sample contains 55 individuals living with an RD and 13 family members.

For HCP interviews, five different groups were incorporated: GPs, specialists, physicians working in a hospital and medical therapeutic practitioners (MTP). In this context, the term "clinicians" refers to those physicians working in a clinical surrounding. In Germany this subgroup needs to be distinguished from "specialists" who have settled in a private practice. RD guides differed in qualification (e.g. human geneticist, biologist, and physician) but were equally trained for the guidance of RD patients through the health care system. Participants were recruited by the Centre of Quality and Management in Health Care embedded in the State Medical Chamber of Lower Saxony in Hannover. All participants were recruited within the geographic region of Lower Saxony as this is regarded as representative of all areas of Germany. Only RD clinical guides were recruited all over Germany as they occur less frequently. The following selection criteria were employed: regional aspects were considered, differentiating professionals working in rural, urban, or metropolitan areas. Resident physicians were differentiated by whether their work was conducted in either single or joint practice. Regarding clinical doctors, the level of health care provision was considered, e.g. basic, regular, specialist, and maximum medical care. Finally, the hierarchy level of participants was considered, distinguishing between chief, senior, and assistant physicians.

Ethical considerations

A positive ethics committee vote was obtained for the interview study from the ethics committee of the Albert-Ludwigs-University Freiburg (number 53/14). Informed consent was obtained in writing from all participants.

Data generation and analysis

Semi-structured interviews were chosen as participants needed to be directed to the subject of interest. In some interviews, a narrative structure would have led to the extensive presentation of a single health issue, which was the focus of the person interviewed, not reflecting on other subjects, which were still important, though less so. Patient and relative interview flow was initiated by asking for experiences with diagnosis and treatment and important steps of their professional careers as well as experiences with RD patients on the side of HCPs. Then, interviewees were asked whether they saw a need for such a service. "How do you feel about the option to attain information by telephone?" If they were in favour of an RD helpline, they were encouraged to describe their mental picture of the helpline with particular reference to specific characteristics. A semi-structured interview guide was piloted during two interviews and afterwards adapted per interviewee needs. The HCP interview guide was developed in accordance with the structure of the guide for the people affected. However, some changes were necessary due to the different perspectives of HCPs on the topic. To ensure standardization, both interview guides were mutually discussed.

All interviews were recorded and later transcribed using the F4 transcription software. A standardized transcription guide was drafted for all interviews by three different interviewers. Transcripts were evaluated using MAXQDA, a programme for qualitative and mixed-methods data analysis. A structured content analysis was conducted following the guidelines provided by Mayring [72]. First, each interviewer formed categories inductively for three different interviews. Then, the chosen interviews were coded collectively to ensure inter-subjective or inter-rater reliability. Differences were addressed in the guide, clarifying a uniform coding strategy. Afterwards, attributes were extracted inductively by a single analyst to minimize

interpretation bias. Finally, results were discussed within the whole research group, and results from patients and families were triangulated with those of HCPs. All quotations were translated by an external translation service, approved by a native speaker, and then included in the paper.

To ensure the quality of evaluation, the quality criteria of Mayring [73] were complied with.

Results

Interviews were conducted until a high degree of saturation was achieved. No additional knowledge on RD information provision could be generated from further interviewing.

Following patients' reports on predominant complaints of their complex diseases, all RD-affected interview partners could be categorised within one of the predefined disease groups. Patients with diseases of the digestive tract ($n = 2$), cystic fibrosis and lung diseases ($n = 4$), genetic diseases of the eye (n = 4), and disorders of the connective tissue ($n = 5$) were difficult to represent in the sample because of limited availability and polysystemic patterns. Therefore, the following patients could be included: genetic skin diseases ($n = 10$), skeletal dysplasia ($n = 7$), neuromuscular diseases ($n = 9$), genetic eye diseases (n = 4), connective tissue diseases (n = 5), genetic kidney diseases ($n = 6$), cystic fibrosis and pulmonary diseases (n = 7), congenital blood formation disorders (n = 4), immunodeficiency (n = 7), congenital metabolic disorder (n = 7) and genetic diseases of the digestive tract (n = 2). Participants could indicate disease severity on a three-item scale. Table 2 shows a summary of socio-demographic variables for patient and relative interviews.

The table also shows socio-demographic characteristics of HCPs. One hundred and forty-one HCPs were invited to participate in the interview study. Of these, 39 candidates took part. Ensuring the diversity of participants, special regard was given to selection criteria concerning the structure of health care provision. Nine GPs, nine physicians, nine clinic doctors, six clinical guides, and six MTPs were included.

Table 2 Socio-demographic variables, patients, and relatives

Sample characteristics		
Parameters	Patients and relatives ($n = 68$)	Physicians ($n = 39$)
Sex		
Male	23	23
Female	45	16
Age		
Average	51	46
Maximum/Minimum	85/18	
Educational qualification		
Abitur/A-levels	13	
Secondary education	19	
Technical collage/University	19	39
Advanced technical college degree	12	
Secondary modern school qualification	5	
Age at diagnosis		
Average	34	
Maximum/Minimum	74/0	
Disease severity		
No specification	4	
Low	8	
Medium	28	
Severe	28	
Profession		
Employed	31	39
Housewife/Houseman	2	
Unemployable/Special circumstance	15	
Student/Scholar	2	
Pensioner	18	

Advocating for a rare diseases helpline

Of 55 RD patients, 41 advocated for the implementation of a helpline about RD. A smaller proportion ($n = 8$) objected to the service or described it as unnecessary as the telephone based information was already available online or through a physician. On the other hand, one interviewee interpreted the helpline as a tool for psychological counselling and neglected helplines in general for this reason. The remaining six participants did not offer a distinct answer. Regarding the interviews with relatives, 11 participants did endorse implementation. Two interviews could not be interpreted clearly as statements were given that were neither obviously for nor against an RD helpline. Therefore, a need for an RD helpline can be verified for some RD patients and relatives.

Of 39 HCPs, 18 endorsed the implementation of an RD helpline. Only four objected to the service or described it as unnecessary. An RD helpline was regarded as unnecessary whenever a suitable colleague was available. Physicians preferred a personal contact, they were already familiar with. A total of 17 participants offered an ambiguous or no answer. As physicians ($n = 27$) made up the largest portion of HCPs, these were evaluated separately as well. A total of 14 reported their endorsement of such a service, four claimed it to be unnecessary, and nine did not answer the question in a manner that could be definitively coded. Consequently, these findings verify for some HCPs a need for an RD helpline.

Expectations towards a rare diseases helpline

A detailed summary of the content analysis of patient and relative interviews brought forth the following necessary characteristics of a helpline. Quotations are labelled by interview code, age, and gender. The interview code consists of a letter and interview number. The following abbreviations were used: 'A' standing for relative or affiliated, 'P' for patient, 'GP' for general practitioners, 'S' for specialists, 'MTP' for medical technical practitioners, and 'G' for guides.

Staffing with professionals

Interviews demonstrated that patients and relatives perceived an RD helpline as helpful when their questions were answered by professionals. Ten of the participants explicitly highlighted this fact ($n = 10$). References to other access points, regardless of their profession, were experienced as rather unsatisfying.

> *'Currently it's like this / our people are annoyed about it – they call up the branch office of LOCATION and have to ask their questions, then they call up LOCATION in order to receive an answer, and then they have to call back the people who are involved; I can't conduct a conversation about my problems like this. I can't solve a problem with a question – that usually functions in the most..../ or somehow, we'll ask questions on Radio Eriwan, where the answers only consist of yes, no and occasionally perhaps.' (P11/53/f).*

Similar to what was observed in the patient and relative interviews, HCP participants generally asked for a professional contact at the other end of the phone ($n = 11$). In most cases, a physician was named. One participant indicated that a hotline should not be staffed with a data management employee, nurse, or secretary even though they can sometimes be of much help.

> *'If one of them is clever and can give me tips afterwards, then I thank God for it and .../ but this should always come from doctors [I1: OK], not a nurse or a secretary."* (GP02/37/m).

It was reasoned that only trained physicians could provide precise medical information. Therefore, an employee with substantive clinical experience was demanded. (S04/35/m) Expert knowledge of the person in charge was also highlighted. In the case of physicians, this meant extended training for one specialty. Nevertheless, biologists and laboratory experts were also mentioned in isolated cases. (GP05/61/m) Participants were also conscious of the difficulty of realizing this demand and therefore emphasized that an expert for each relevant medical field could not be demanded for an RD helpline. (S01/39/m).

Personal contact

Another highlighted aspect was the importance of personal contact in addition to other rather impersonal information research systems ($n = 10$). Specifically, a single point of contact was demanded. It was reasoned that this kind of contact could accelerate and ease information search. Consequently, it was seen as helpful with regard to orientation in the health care system.

> *'Then they'll surely sit down and study Internet sites and the brochures and information materials that are given out, but a human contact partner can sometimes expedite and simplify this search for information. Simply because one doesn't just enter questions onto a screen by himself, but rather because he initiates communication with someone. If this office, the information office, was really staffed with competent personnel and not just some students who are completing their internship and don't really know what it's all about, then it would be a good idea, but would then also mean that money would need to be exchanged.' (P37/46/m).*

On the other hand, psychosocial advantages were emphasised.

> *'Therefore, we have always sought out personal contact during the search and made use of it, simply because contact with a human being is much more pleasant and one can exchange information more effectively than when one simply calls up inflexible information from the Internet and then has to determine what is really applicable and what is not.' (A02/48/m).*

The importance of personal contact ($n = 7$) was also identified as a category during HCP interviews. As expected, the focus was laid on the exchange of medically relevant information. For example, psychosocial issues were not named as a reason for the demand for direct communication. Instead, personal contacts were preferred as patients could be quickly introduced, and immediate feedback could be generated.

> *'Where one can also have a telephone conversation, which, in any case, is better than sending emails back and forth, since one can then react directly, briefly introduce the patient with his symptoms and perhaps even give the patient an appointment promptly, so that he can be examined in detail.' (GP01/39/m).*

Frequently, an immediate contact and information receipt was required. (MTP02/35/f, MTP04/25/m, S04/35/m, GP01/54/f, GP03/48/f, C06/47/f) Furthermore, it was outlined that some medical issues cannot be described using predesigned web search masks given by Internet providers. Information can be searched only if previously made searchable. Fine nuances between blank facts cannot be depicted.

'So, to make a comparison once again; if I now say, as already mentioned in the example, I enter three things/ it's different to saying to a colleague: "Man, I have the feeling that he's really sick. And then it hurts somewhere on the left, sometimes more, sometimes less and so on". It doesn't make sense to enter this into a screen. [I1: Hm] And that's really important.' (GP02/37/m).

Availability

Participants ($n = 6$) expressed a wish for extended opening hours.

'And then, okay, if I have the office, let's look at the ACHSE as an example. Then that's also / and it's rather stupid, at the one, they only work a half-day and it's always … / so you always end up calling outside of business hours.' (P06/85/f).

Interviewees hope to avoid waiting periods and to receive contemporary answers. Waiting lines raise dissatisfaction and impatience (P17/47/f) similar to answering machines or automatic answers. (P51/62/f) It was reported that there should be at least enough human resource capacities to ensure a return call within an appropriate timeframe. (P14/57/f, P50/51/f).

HCPs referred to availability during four interviews (n = 6). HCPs did not highlight an uninterrupted 24/7 availability as important. In acute and/or life-threatening situations, an RD helpline would not be the first choice. In such a situation, an emergency call asking, for example, for a poison centre would be preferred. One GP mentioned that availability during regular office hours would be absolutely satisfying. Following the results of interviews with patients and relatives, it was also indicated that immediate availability is necessary, especially avoiding waiting lines.

'Personally, I find telephone conversations better, [I1: Hm] But I know how awful it is to be put on hold. [I1: Yes] [I2: Hm] Then one calls from here [I1: Yes] and tries to get connected. I know, I've had REALLY bad experiences there. If I want to reach anyone and I say to someone: I have five women here [I1: Hm] and then you get someone simply hangs on stubbornly. It can sometimes take HOURS. [I2: Hm, simply lay the receiver to the side] exactly! Lay the receiver to the side and wait until the call back comes through. That's useless. [I2: Hm] I can't afford to waste time like that here.' (S04/35/m).

This demand is in line with the demand for fast and immediate access to information. On the other hand, a dial-back system, collecting calls and answering them afterwards at a particular date, was also suggested by one participant (S13/50/m). Remarkably, this would contradict the demand for a fast access to information previously mentioned during interviews with patients, relatives, and HCPs.

Low technical barriers

The telephone is also mentioned as an alternative to web access ($n = 6$) that is also suitable for the elderly and information seekers with no affinity for or no available Internet access. Additionally, one interviewee noticed that some people with RDs are limited in their mobility through their disease. Consequently, these people are unable to reach personal contact partners such as physicians and other therapeutic personal.

'However, the problem is often those people who can't do it. We have a contact partner per telephone for those who are not mobile / great restrictions for the illness [AM]. Another example is the case of the DM 1 advanced stage, where the people are often no longer able to go places by themselves / they need so much strength and energy in order to cope with the few daily tasks, then they have something for it / but good, one always wants everything in any case.' (P11/53/f).

Asking HCPs for their opinion on the telephone as an alternative to the Internet as an information medium, results were heterogeneous. While younger HCPs preferred the Internet over a telephone and did not assign an important role to it, HCPs of higher age were rather indifferent or clearly preferred the telephone:

'Personally, I'm a big fan! [I1: Yes] So, the telephone— I would always give the telephone preference [LAUGHING], over some impersonal Internet site. But I think that's also really "old-fashioned"' (C07/42/f).

HCPs even align with the need for a low technical barrier for certain patient sub-groups such as the elderly.

'Yes, I believe that exactly those people who, let's say don't have Internet access or who lack the knowledge, we're talking about the older members of the public/. […]' (C03/46/m).

Topics of counselling

Further, patients and relatives described possible topics that were expected to be discussed on the phone. Psychosocial and medical aspects were predominately named. Interviewees described the following medical contents: They hoped for an explanation of their disease pattern and of symptoms at hand.

'[…] first of all, the symptoms of the clinical picture, of course, and how the people affected deal with them. And then, of course, also self-help groups.' (P52/39/f).

In particular, participants demanded answers not only of general questions but also of questions concerning specific sections of the disease as well as information on the genetic background. (P12/58/f, P51/62/f) Concerning disease development information, possible methods to stop or lessen the burden of disease were reported to be most relevant. (P53/51/m) This was found in combination with the demand for information concerning the application of medication dosages or therapy and behaviour in the case of emergency. (P50/51/f, P54/40/m) Aside from these, patients also wished to be informed about the status of research. (P07/70/m).

In addition, persons concerned also brought forward psychosocial aspects. (A12/32/f, P47/59/m) Patients and relatives reported that they do see a need for the resolution of general problems arising from disability as well as specific disease problems. (P25/58/f, A05/60/f) Furthermore, it was perceived as helpful to talk about diseases, learn how other patients handle their disease, and learn whether self-help groups already exist. (P52/39/f) The importance of practical information on everyday life was highlighted again at this point. (A05/60/f) Just one person explicitly negated such an offer, claiming to be in no need of a helpline where one can have a good cry. (P04/39/m) On the other hand, a contact person was seen as an opportunity to counterbalance the desperation of one's own situation with the prospect of being counselled and reserved when necessary. (P23/48/f) One interviewee noted that other sites did not take one seriously and hoped for an improvement. (A06/50/m) Similarly, when disorientated, a contact person was sought to aid with calming down, helping with the search, and coming up with concrete help.

'Yes, that one has a competent person on the other end of the line, so that one, for example, if he is doing badly or if he has any problems, that he receives the help he needs. In other words, that there is someone available for the moment. He doesn't have to bring everything back into tip-top shape immediately. Just perhaps someone who is there to say: "Yes", and "try to stay calm" for now, or, "I'll help you, I will sort it out, I'll do it" / "I'll check up on it" and so forth, so that one isn't simply/ yes, that one isn't turned away, but rather… / or be subjected to long waiting times.' (P21/53/f).

When discussing topics of counselling, some HCPs specifically mentioned the need for endorsement concerning medical issues ($n = 7$). In particular, medical cases were reported as needing to be discussed via telephone, describing symptoms and patient histories. Three HCPs specified this demand, highlighting the need for differential diagnostics or a demand for assistance with the differential diagnostic process of elimination. (GP03/46/f, MTP04/25/m, S04/35/m).

'I would also think that this could be useful for rare diseases, so that one could simply receive a differential diagnosis, a second opinion. So, I'll tell you what the symptoms are and you can tell me what it could be.' (MTP04/25/m).

Participants also demanded information on self-help groups. (GP05/61/m).

Guidance

Those polled also talked about the necessary functions of a helpline. Often, aiding orientation within an information overflow or during information undersupply seemed to be necessary. Additionally, the sample demanded a guide to lead the way through information chaos. (P09/47/m) Beyond that, advice for further research was seen as beneficial. (P52/39/f, P29/44/f, P38/60/f) Even a general reference suggesting that such information exists was perceived as helpful. (P13/54/f) Therefore, it is not surprising that the scope of available information was most commonly underestimated.

'Although sometimes one naturally also …/ one thinks he is well informed, and he has no idea that there is actually still much, much more information available or that a variety of other opportunities exist for him.' (P32/40/f).

This category did not occur during HCP interviews.

Referral

Another function that was additionally demanded was referral. For example, information about care facilities and physicians was cited (A06/50/m, P10/50/f), indicating that

this is of special importance at the beginning of a disease. (P47/59/m) Nevertheless, it was also highlighted that this was not the only task.

'A12: Yes, I find it good (info hotline). But, in my opinion, as I have just indicated, that would need to be a little larger. That psychosocial counselling services are referred to.

Interviewer: Yes, OK.

A12: and that one does more than to just say, "Yes, there's the doctor."' (A12/32/f).

HCPs also mentioned the need for referral in addition to medical counselling ($n = 3$). At this point, HCPs reported that they realize that it is impossible to make their wish for immediate specialist knowledge for each medical field come true. On the contrary, they realize the impracticability of this demand.

'That makes sense, yes. That makes sense. Well, I wouldn't expect to be able to call the medical association, for example, and say that I have someone on the phone who is experienced in this area. [I1: Hm] You can't expect that. But if you can call and say: 'Do you have a contact that is particularly responsible for such and such a disease pattern'? That makes sense.' (S01/39/m).

'Let me say, in order to be in a position to address his request, and I believe that this telephone opportunity is really good here, since it gives us the opportunity to shift and sort a little and [I1: okay] to say who belongs here and who doesn't.' (G07/31/f).

In this regard, HCPs emphasize that the number of referrals can and must be minimized to shorten odysseys through health care systems. (GP03/48/f).

Discussion

Patient and relative interviews showed that helplines are predominantly necessary due to the possibility of personal contact and low technical barriers. RD patients and relatives wish for a helpline run by professionals with extended availability. An RD helpline should offer information on medical and psychosocial issues. In addition, participants hope for guidance through information chaos as well as referral where needed.

In general, the need for an RD helpline from the perspective of HCPs was confirmed with some minor differences, even though no statements to the extent of the demand can be made to a comparably high percentage of unspecific answers. An RD helpline should be staffed with professionals. However, a medical professional was specifically demanded. Criteria for staffing should be broad knowledge of RD, a multidisciplinary orientation, and knowledge of differential diagnostic procedures. Personal contact was preferred as details of medical cases could be described, even if not put in words easily. HCPs also asked for additional referrals to other experts. Good availability was specified as reachability during office hours suggesting that a request surplus could be managed through a call-back system. As this proposal is not in line with patient and relative interviews, it is not considered for the final concept. Medical professionals recognized a low technical barrier as an important issue for themselves and people affected.

Many studies report the staffing of helplines with nurses [74]. In this study, patients, family members, and physicians particularly demanded the employment of professionals with a special emphasis on physicians. McKenzie, [75] for example, reported the successful commitment of GPs for the coverage of after-opening hours. In this regard, the broad knowledge of GPs seems to be a suitable qualification for the management of an RD helpline potentially incorporating the need to familiarize with various diseases across different medical disciplines. Besides, GPs are familiar with transferring patients and communicating with various medical professionals. GPs also add their expertise when it comes to long-term differential diagnostics. On the other hand, training can add to the necessary qualification spectrum especially when it comes to very rare diseases or psychosocial needs and seem to be quite suitable for the management of helplines even though a re-alignment of practice is necessary due to the interaction via telephone [75]. Advice has been obtained by the German Cancer Information Center which offers a German Cancer Helpline. In this context, psychologist or social workers are often added to the Team rendering advice to other counsellors [76–78].

The importance of personal contact was highlighted during the interviews. Even though interviews raised ease of information search as an argument for building a helpline, the psychological value of personal contact also needs to be stressed. Anderson [79] raised a high negative impact even on family members of children suffering from RD and demanded more psychological support. Helplines salvage the potential of reducing this distress [67] and therefore offer high psychological value.

Availability is a very subjective topic. As RDs mostly show a chronic pattern, emergency situations will not arise that often. Emergency calls can be addressed to emergency helplines. Nevertheless, the demand for extended opening hours remains unclear in its specification and requires further discussion.

Even though we live in a technically advanced digital age, there are still some EU households with

no Internet connection (32%). On the other hand, 98% have telephone access, either through a fixed or mobile device [80]. Adding to this, telephone services are always available in local languages while many websites on RDs are only available in English language, adding a language barrier to the technical one [67]. Nevertheless, it must be outlined that these barriers will be further reduced as technologic advancement progresses. Besides, there are already some translation programmes available online, which will most certainly be further developed.

Friedmann et al. [81] confirm the necessity of a multidisciplinary team for the coverage of inquiries of callers of a HCP helpline and therefore underpin the broadness of questions.

Originally, primary care physicians took on the role of guides when communicating and assessing health information. Therefore, it is not surprising that this category did not emerge within HCP interviews as they identify with this role. Nevertheless, Coumou and Meijman [82] state that GPs do have approximately 400 indications at hand. It is very likely that common indications are kept in mind rather than RDs, which are very unlikely to appear in their practice. These findings underpin the role of guides, which already exists as part of many RD centres and whose expertise is demanded in this case.

Tariq et al. [83] confirm the importance of referrals carried out by helplines. It quantifiably contributes to the effectiveness of health care systems. For example, an after-hours service helpline prevented 1363 people from unnecessarily attending an emergency department. Further, 228 individuals underestimating their conditions could be referred to an adequate health service provider.

Study significance

We suggest that our study has significance for the establishment of nationwide and centralized RD helplines worldwide due to shared problems such as long delays in diagnosis and dense RD health care infrastructure. In addition, the study broadens the perspective on RD telephone services rendered within the literature thus far by including potential users who have not yet called a helpline but would if services were adapted. In contrast, previous studies interviewed callers of existing helplines, focusing on affected people who were already interested in the service of the helpline [61, 64]. This new perspective offers a way to improve RD counselling, making it more attractive to the potential user pool and, therefore, extending its benefits to all those affected within society. Besides, many studies dealing with the question of health information provision do not include the possibility of different information access points. However, existing studies—for example, Mooney et al. [84]—found that patients suffering from antineutrophil cytoplasmic antibodies tended to reject detailed information on their disease and disease management when given the diagnosis through a physician as it was a lot to take in. At a later stage, truthful information was difficult to access, substantiating the benefit of a telephone service. Other studies analysing patients and families dealing with late stage cancer underpin the assertion that trained physicians may not communicate effectively due to missing knowledge of information needs of this patient group, [85] indicating the need for specialized and broadly available service providers.

Most heatedly discussed was the implementation of a central RD helpline considering all 5000 or 8000 very heterogeneous diseases. Implementing this kind of service necessitates an extensive financial budget. The estimation of necessary financial resources proves to be quite difficult as many assumptions and projections are necessary. A high-budget case with 60,589 estimated contacts per year necessitates an annual budget of 2.59 € million with 35 full-time employees (FTE).[1] As full case coverage requires extensive budgeting, the calculation is rather an indicator for what is already done for other diseases and could be done in the field of RD. However, a competent counselling service can be offered. In this case, the overall estimated need for RD information need cannot be covered. Costs for a base case scenario mount up to approximately 300.000 € (4 FTE) annually.[2] An evaluation of European telephone services by Houÿez et al. [53] shows similar results. According to the report, RD helplines should be staffed with a minimum of 1.5 (FTE), leading to annual costs of 150,000 to 300,000 €. Therefore, it can be suggested that, starting from this level, a stepwise implementation of the ideal scenario should be pursued.

As a solution for the shortage of monetary funds, a central telephone service offering referrals is often suggested. Such a service could bundle the heterogeneous landscape of existing RD telephone-based or disease-related information services in a similar manner to how ZIPSE is bundling web information. However, the implementation of such a service would contradict the results of the study as patients, relatives, and HCPs ask for direct contact with professionals. Therefore, a telephone service bundling all RD helplines and giving references cannot be suggested. Nevertheless, it can be suggested that existing RD-related helplines may be shaped following the results of this interview study. For example, the service of the Alliance of Chronic Rare Diseases (Allianz chronisch seltener Erkrankungen, ACHSE) can be further extended. Services of RD guides located at specialized centres for RDs can be adapted, bearing thoroughly in mind the wish of patients and relatives to not only be forwarded from one contact point to another.

Assumptions and limitations

This study was designed qualitatively to capture information needs, which could be served using a telephone service without guiding answers beforehand. Instead, participants were encouraged to give their own ideas on an RD helpline, assuming these to be of most relevance. Therefore, a limited number of patients, relatives, and HCPs could be interviewed. The qualitative design contributes to theory generation. The quantitative structure of interview results has been included to increase the transparency of result communication. To make projections and/or quantifiable statements, results need to be verified through a quantitative study.

Only 39 HCPs participated in the study from 141 invited. Studies show that physicians are more likely to respond when a small financial incentive is given. During this study, no financial resources were available for this purpose [86].

Many female individuals were interested in participating in the study. That is why the sample is biased towards women. Even though this should be kept in mind, studies show that health information providers are more often used by women as they are more likely to search for health information in general. Some providers report up to 97.5% female users [87].

The study was conducted against the backdrop of the German NAMSE process asking for the design of a national RD helpline. In order to minimized the bias towards favouring the establishment of a RD helpline interviewers first openly asked how participants feel about helplines to avoid putting neither negative nor positive words into the mouth of participants as suggested by Mayring 2002 [72]. Therefore, participants were not influenced towards a specific outcome.

Additionally, patient and relative interviews were conducted by three different interviewers. HCP interviews were held by two different interviewers partially conducting interviews together. Even though interview structure was discussed beforehand and interview guides were established and adapted after piloting, individual interview styles need to be recognized as an influential factor.

Interviews were not able to capture juridical topics during the questioning concerning the helpline. Therefore, it is obviously necessary to analyse why respondents did not include juridical or access to treatment matters even though experiences of other helplines show that people affected do not solely search for this via other media [63]. ACHSE user statistics (2011–2013; unpublished, based on private email communication) indicate that problems with cost takeover and other social legal problems are topics of counselling. Independent Patient Consultancy (Unabhängige Patientenberatung Deutschland, UPD) reports proportions of 66% and 67% [78, 79] medical-juridical questions within their annual patient monitor, pointing to the most likely reason for not mentioning juridical issues during the interviews. Obviously, they are closely linked to medical questions and not visible at first glance using structured content analysis.

Conclusions

Even though new technologies enable patients, relatives, and HCPs to access information rapidly, this study shows that there is still a point in making information accessible the 'old-fashioned way' via telephone. The telephone offers the unique chance to make professional insights directly available for all stakeholders, including exchanging medical and psychological issues. However, putting all desired aspects simultaneously into practice in an ad hoc implementation process with a central RD helpline offering information for all patients, relatives, and HCPs potentially calling the helpline would necessitate a huge financial budget. Therefore, a stepwise implementation is suggested. As a first step, it is suggested to improve major existing helplines to meet the identified needs. Afterwards, service availability can be extended. In the long run, existing services should be evaluated with regard to the fulfilment of these factors. The expertise from institutions as centres for RDs should be further included, bearing in mind the wish of patients and relatives to not be pushed from one information access point to another.

Endnotes

[1]In an ideal scenario, all identified relevant aspects should be put into practice. This first scenario demonstrates how many monetary resources could be put into a telephone-based information service when aiming for a service quality similar to that of the UPD and the KID. User statistics of the KID show that only 1.51% of the patient pool uses the information service, resulting in 23,024 contacts per year [78]. Extrapolating from this number, an RD helpline would need to expect 60,589 requests per year, making 35 employees necessary to answer all requests [76–78]. Average human resource costs of an interdisciplinary team are estimated to be 59,006.61 € per employee and year. Costs were calculated including monetary funds for rent, staffing, office equipment, publicity, and employee development. Calculating overall material costs makes an estimation of material cost per person possible, approximately 14,911.20 € in the initial year and 12,907.83 € for the following years. The calculation is based on German average wages of a mixed team consisting of physicians, social workers, and lawyers. Accordingly, an RD helpline makes funding of 2.59 € million in the first year necessary.

[2]Because of limited budgets, a low-budget case has been discussed as a second scenario. Following interview results, the most important aspect, 'quality of counselling by

experts' shall be obtained. Therefore, a multidisciplinary team shall be preserved, aiming for a minimum staffing by one physician, one social worker, one lawyer, and one temporary employee. Hence, a quality counselling service can indeed be offered, but, comparing both cases, the counselling team in the latter case is significantly shorthanded and therefore unable to cover all potential requests. In a base-case scenario, considering four full-time employees, overall material costs add up to 59,644.80 € for the initial year. For the following years, 51,631.30 € in costs are estimated. As previously mentioned, average human resource costs add up to a total of 236,026.44 €. Finally, the calculation results in overall costs of 295,671.24 € for the first year and 287,657.74 € for the following years.

Abbreviations

A: Affiliated or relative; ACHSE: Allianz chronisch seltener Erkrankungen, Alliance of Chronic Rare Diseases; C: Clinician; ENRDHL: European Network of Rare Disease Help Lines; FTE: Full-time employees; G: Guide; GMS: German Medical Science; GP: General Practitioner; HCP: Health Care Professionals; KID: Krebs Informations Dienst, German Cancer Information Service; MTP: Medical Therapeutic Practitioner; NAMSE: Nationales Aktionsbündnis für Menschen mit Seltenen Erkrankungen, National Action League for People with Rare Diseases; P: Patient; RAPSODY: European Rare Disease Solidarity Project; RD: Rare Diseases; S: Specialist; UPD: Unabhängige Patientenberatung Deutschland, Independent Patient Consultancy

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Availability of data and materials

The dataset generated and analysed during the current study is not publicly available as the anonymity of participants should be preserved; this anonymity is in danger due to the small prevalence of single rare diseases. However, the dataset is available from the corresponding author on reasonable request.

Authors' contributions

AB, MF, SL, and VL planned and designed the study. AB, SL, and DR conducted the interviews with patients and relatives. FS recruited patients and relatives. VL recruited and interviewed all HCPs. AB and FP conducted the cost analysis. HS and TH critically reflected on and analysed data on informatics. LB critically reflected on and analysed data on rare disease helpline management. AB coded the interview data. The acquired dataset was than discussed and verified by all researchers. AB drafted the manuscript. JMS, TOF, and MF revised the document for important intellectual content. All authors have read and approved the final manuscript.

Competing interest

The authors declare that they have no competing interest.

Ethics approval and consent to participate

A positive ethics committee vote was obtained for the interview study from the ethics committee of the Albert-Ludwigs-University Freiburg (number 53/14). Informed consent was obtained in writing from all participants.

Consent for publication

Not applicable.

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Modul 5

Shaping an Effective Health Information Website on Rare Diseases Using a Group Decision-Making Tool: Inclusion of the Perspectives of Patients, Their Family Members, and Physicians

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Original Paper

Shaping an Effective Health Information Website on Rare Diseases Using a Group Decision-Making Tool: Inclusion of the Perspectives of Patients, Their Family Members, and Physicians

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Abstract

Background: Despite diverging definitions on rare conditions, people suffering from rare diseases share similar difficulties. A lack of experience by health professionals, a long wait from first symptoms to diagnosis, scarce medical and scientific knowledge, and unsatisfactory treatment options all trigger the search for health information by patients, family members, and physicians. Examining and systematically integrating stakeholder needs can help design information platforms that effectively support this search.

Objective: The aim of this study was to innovate on the group decision-making process involving patients, family members, and physicians for the establishment of a national rare disease Internet platform. We determined differences in the relevance of health information—especially examining quantifiable preference weights—between these subgroups and elucidated the structure and distribution of these differences in people suffering from rare diseases, their family members, and physicians, thus providing information crucial to their collaboration.

Methods: The included items were identified using a systematic Internet research and verified through a qualitative interview study. The identified major information needs included *medical issues*, *research*, *social help offers*, and *current events*. These categories further comprised sublevels of *diagnosis*, *therapy*, *general disease pattern*, *current studies*, *study results*, *registers*, *psychosocial counseling*, *self-help*, and *sociolegal advice*. The analytic hierarchy process was selected as the group decision-making tool. A sensitivity analysis was used to determine the stability and distribution of results. *t* tests were utilized to examine the results' significance.

Results: A total of 176 questionnaires were collected; we excluded some questionnaires in line with our chosen consistency level of 0.2. Ultimately, 120 patients, 24 family members, and 32 physicians participated in the study (48 men and 128 women, mean age=48 years, age range=17-87 years). Rankings and preference weights were highly heterogeneous. Global ranking positions of patients, family members, and physicians are shown in parentheses, as follows: *medical issues* (3/4, 4, 4), *research* (3/4, 2/3, 3), *social help offers* (1, 2/3, 2), and *current events* (2, 1, 1); *diagnosis* (6, 8, 9), *therapy* (5, 9, 7), *general disease pattern* (9, 4/5/6, 6), *current studies* (7, 4/5/6, 3), *study results* (8, 7, 8), *registers* (4, 1, 5), *psychosocial counseling* (1, 2, 4), *self-help* (3, 3, 2), and *sociolegal advice* (2, 4/5/6, 1). Differences were verified for patients for 5 information categories (*P*=.03), physicians for 6 information categories (*P*=.03), and family members for 4 information categories (*P*=.04).

Conclusions: Our results offer a clear-cut information structure that can transparently translate group decisions into practice. Furthermore, we found different preference structures for rare disease information among patients, family members, and physicians. Some websites already address differences in comprehension between those subgroups. Similar to pharmaceutical companies,

health information providers on rare diseases should also acknowledge different information needs to improve the accessibility of information.

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KEYWORDS

rare diseases; decision making; health information needs; preferences; patients; relatives; physicians

Introduction

Worldwide, approximately 350 million people are affected by rare disease [1]. Despite diverging definitions, people suffering from rare diseases share common difficulties. Particularly, health care professionals have little experience with this patient group, and patients typically wait a long time from the first symptoms to diagnosis. Moreover, medical and scientific knowledge concerning rare diseases is scarce, and low research efforts often result in, if available, unsatisfactory treatment options. When there is a treatment option available, patients still often need to consider financial aspects. Patients also frequently experience difficulties with the cost absorption of expensive treatments. Furthermore, rare diseases are very serious and chronic. Severe symptoms result in high disease burden and can have a significant negative impact on one's quality of life. Above all, patients often face a shortened life expectancy [2]. Consequently, there is an urgent need for proper health information for this population.

The Internet offers a large pool of somewhat obscure information. In this context, this study examines how information on rare diseases can be presented in a more structured way. As a second step, we also examined whether stakeholder-specific websites presenting information in accordance with the information priorities of the targeted subgroups would be necessary. We hypothesized that the information structures of patients, family members, and physicians would be identical, as family members and physicians would generally search for information to fulfill patients' needs. This would consequently lead to a single platform incorporating the overall group consensus on information priorities and therefore information presentation.

The literature, however, has not yet addressed the differing information needs between patients, family members, and physicians. Health information helps to empower patients, enabling them to understand, treat, cope, and effectively manage their disease [3-5]. Rare diseases' patients are often called *experts* of their own illnesses because they gather health information consciously through Web searches or unconsciously through numerous consultations with different health care professionals [6]. Besides, doctors' assessments of patients' preferences appear to be critical for the outcome of health services [7]. In this regard, the dialogue between patients and physicians is critical. Therefore, health care professionals must be trained and prepared to listen to patients and discuss their experiences [8,9]. Furthermore, health information searches should be facilitated and encouraged, as they enable patients to be more effective in communicating with their physicians [5]. This study contributes and adds value to this existing literature and the underlying dialogue by eliciting the different perspectives of patients, family members, and physicians on the relevance of rare disease information.

Aside from the above points, little or no scientific knowledge exists for the 5000 to 6000 different indications summarized under the term rare diseases. Adding all diseases and all different information providers together creates a huge and obscure information pool. Indeed, information providers often fail to meet the information needs of patients and families searching social media and utilizing chat rooms to obtain information; however, they might be unaware of the low quality of this information [10]. On the other hand, obtaining knowledge of the many thousands of different rare diseases is well beyond the ability of physicians. Primary physicians are only familiar with approximately 400 different indications. Primary physicians can extend their knowledge through asking questions of colleagues and reviewing paper-based data sources [11]; however, even with the advent of electronic records, it remains highly time-consuming and difficult to search for the right terms and obtain appropriate evidence. Taken together, these facts suggest that effective health information presentation is exceedingly important. Collins et al suggest that information needs can be incorporated by capturing and embedding the relevance of information [12]. This study shows how this demand can be put into practice.

Literature shows that group decision-making tools are rarely applied when it comes to the establishment of health information portals. Health information needs are often met by retrieving information from historic user statistics or triggering retrospection. Stakeholders cannot actively participate [13,14]. However, by choosing the analytic hierarchy process as a group decision-making tool, we can actively involve patients, family members, and physicians to address their unmet informational needs. Furthermore, information categories that are underrated by stakeholders (ie, patients, relatives, or physicians) can be illuminated. A number of different models have already been applied during the establishment of effective cocreative business modeling [15,16]. However, until now, there have been no attempts to devise a similar model in a transparent manner for different stakeholders in relation to rare diseases.

The following study has been conducted against the backdrop of the conceptualization of a central website for rare disease information in Germany (ZIPSE, Zentrales Informationsportal über seltene Erkrankungen or central information portal about rare diseases) [17] connecting disease unspecific and specific information, as well as quality orientation for patients, their families, and health care professionals at a central platform [18]. As part of the German National Action Plan for Rare Diseases from 2013 (NAMSE, Nationales Aktionsbündnis für Seltene Erkrankungen) following the European council recommendations [19,20], knowledge transfer is improved

through the development of Internet information systems. Already existing Internet information is collected and organized to increase the visibility of rare disease knowledge [18]. Physicians, family members, and patients are critical to this process; they are the major beneficiaries and should profit by effective health information provision.

In this paper, we describe how patients, family members, and physicians can contribute directly to this process of effectively gathering and presenting health information. More specifically, we describe an innovative group decision-making process involving these individuals aimed at establishing a national rare diseases Internet platform. This study also examined the information preferences of these stakeholders to enable health care systems, decision makers, and other national and international rare diseases portals to appropriately structure information that patients, families, and physicians strive for. The relevance of information is crucial for stakeholders' ability to relate to each other within a strong network approach. In this regard, the study provides unique insights into the quantitative structure and distribution of information preferences for these stakeholders, answering the question on how information provision in the context of rare diseases should be structured.

Methods

Ethical Considerations

The questionnaire was distributed both Web-based and as a paper-based version. Accordingly, consent was obtained in written form. The paper-based version was distributed after qualitative interviews with patients and their relatives. A positive ethics committee vote was obtained for the interview study from the ethics committee at Albert Ludwigs University of Freiburg (number 53/14). The Web-based version allowed for collecting opinions anonymously without having participants disclose personal details at any time. An information sheet was presented to all participants describing the aim and scope of the study. All participants were informed that they could withdraw from the study at any time.

Analytic Hierarchy Process (AHP)

An analytic hierarchy process (AHP) was implemented for the collection of individual preferences, as this study was devised to contribute the decision-making processes implemented in the ZIPSE project. Saaty gives detailed information on the AHP methodology [21]. Two authors also give a detailed overview of its application in health care [22,23]. Lately, the Institute for Quality and Efficiency in Health Care in Germany discussed the AHP as a method for the inclusion of preference structures into early benefit assessment. Similar to conjoint analysis, AHP raises quantifiable weights that can then be used to combine multiple endpoints into an efficiency boundary [24,25]. AHP offers a direct approach, whereas conjoint analysis compares different attributes in combination, thereby leading to an indirect calculation of weights. Furthermore, it is more intuitive and easier to understand for inexperienced participants compared with other techniques (eg, the analytic network process [26] but more informative than other techniques, eg, best-worst scaling, ranking) [27]). Quantitative preference distances make extensive evaluation of preference structures possible [20,28]. Therefore, the major benefit to AHP methodology is that it raises not only ranks but also measurable distances between criteria weights, leading to a visible preference structure. AHP does not only give a clear-cut ranking, it also indicates what categories are weighted similarly. Therefore, attributes that are weighted similarly, but ranked differently, do not need to be excluded. The AHP is able to appreciate individual judgments adequately to thereby derive an overall group consensus [29] and offers a clear-cut preference structure that can be easily applied to the presentation of health information.

AHP is particularly interesting for the field of rare diseases as it is applicable independent of the size of the indication. Even opinions of very small rare disease subgroups can be raised and evaluated [20,28]. Moreover, AHP appreciates the heterogeneity of rare diseases, which because of its definition, summarizes quite diverging disease patterns, as subgroup specific opinions can be evaluated separately. Consequently, this study recognizes the value of AHP when examining rare diseases.

Hierarchy Definition

A total of 300 information websites addressing rare diseases were searched and scanned concerning available information on their home pages. Litzkendorf et al also collected and verified the items through a qualitative interview study [30]. Similar information categories have also been found by the Genetic and Rare Disease Information Center [31] and for other indications such as multiple sclerosis [32]. Accordingly, information categories were drafted and prestructured. Four experts in public health research and one expert in health economics research were chosen from the Center for Health Economics Research Hannover (CHERH). The major criterion for choosing these experts was a research focus on either rare diseases or patient-reported outcomes. Participants were addressed personally. An invitation for participation was forwarded via email along with an attached Microsoft Excel 2010 sheet containing the included items. Afterwards, the final definition of the items was discussed in a workshop scenario. As a result, the different information category descriptions address biases because of different interpretations of information categories. Definitions were finalized if they seemed closed to interpretation and easily understandable (see Multimedia Appendix 1). Thirteen items were chosen, which resulted in 15 pairwise comparisons. The final hierarchy is presented in Table 1.

Questionnaire Development

Other studies used computer-based programs that immediately reflected the level of consistency generated by the answer [33]. Then, corrections are initiated. However, in our study, we did not use an intelligent computer-based fill-out system, instead implemented a paper-based questionnaire. A first draft of the questionnaire was designed and pretested. The pretest revealed insufficient consistency. Therefore, the questionnaire was redrafted. A graphic showing the hierarchy structure was removed to allow space for a graphic demonstrating the exemplary filling out of one question on the questionnaire. Furthermore, a ranking task was integrated, which visualized the intrinsic priorities during the fill-out process. A research question was specified for each visual scale.

Table 1. Hierarchy for information on rare diseases.

Hierarchy level 1	Hierarchy level 2	Hierarchy level 3
Research topic	Parameters	Elements
Importance of health information on rare diseases	Medical issues	Diagnosis
		Therapy
		General disease pattern
	Research	Current studies
		Study results
		Registers
	Social help offers	Psychosocial counseling
		Self-help counseling
		Sociolegal advice
	Current events	

The end of a paragraph containing items from one hierarchy arm was highlighted to emphasize the beginning of a new category. A subsequent pretest revealed improved consistency. Before fielding the questionnaire, the usability and technical functionality of its Web-based version were tested by the authors and a collaborating institution (see Multimedia Appendix 2).

Sample

Patients, physicians, and family members were identified as the main users of health information on rare diseases [34] and a central rare diseases information portal [20]. Participants were recruited using three different recruiting strategies to ensure the adequacy of the sample. The Freiburg Center for Rare Diseases located at the Department of Dermatology of the University Medical Center, University of Freiburg contacted patients and family members using rare diseases self-help groups. Overall, 39 individuals were asked to complete the questionnaire. To participate in the study, patients had to be aged 17 years and older; if they were younger than 18 years, a close relative was invited for answering the questions instead. Interviews were predominately conducted via telephone. To ensure a broad and balanced representation of patients suffering from rare diseases, eleven groups of rare diseases were formed when this study commenced; this was believed to represent considerable variety in rare diseases. Patients were recruited in accordance with these groups. Physicians were recruited by the CHERH. First, physicians with experience in rare diseases and working for specialized rare diseases centers were recruited. Later, the target group was extended to include physicians not imperatively familiar with rare diseases. This seems legitimate, as opinions of physicians unfamiliar with rare diseases but also searching for information were included. Furthermore, a Web-based version of the questionnaire was devised. The link to the open Web-based version was stored on a website offering Web surveys and forwarded by Alliance for chronic rare diseases (Allianz chronisch seltener Erkrankungen, ACHSE) using a mailing list of ACHSE members. A short description of the study was included. All data were collected and stored anonymously. ACHSE checked the avoidance of identification of rare diseases' patients through disease characteristics. The study was initiated in August 2014, and data collection was finalized in August 2016. Overall, 112 questionnaires were answered online, and 64 paper-based questionnaires were completed.

Analysis

For each respondent, a consistency ratio (CR) was calculated. The CR was calculated in accordance with the following formula: $(\lambda_{max} - n)/(n-1)$. λ_{max}. The CR is a value which has been predefined by Saaty [21]. Following the threshold of Danner et al, we included all comparisons with a CR≤0.2; therefore, we assumed pairwise comparisons to be consistent up to this threshold [35]. Respondents with a higher CR were excluded. Individual priority vectors were calculated using the eigenvector method used in Saaty [21]. Afterwards, individual opinions were summarized using an aggregation of individual priorities method. As literature suggests that values must correspond to reciprocal values of individual participants, weights were aggregated choosing the geometric means calculation [27]. As priority values need to sum up to one, resulting local priorities were weighted accordingly. Then, local and global rankings were derived. The calculation was conducted using Microsoft Excel 2010 and R version 3.1.2 (R-project for statistical computing). Responses of patients, families, and physicians were compared. To compare differences between these three subgroups, a variance analysis should be conducted first. However, as we analyzed differences between each of the three groups, test statistics were calculated using a student *t* test. Only local weights were compared as global weights were derived from these. An analysis of sensitivity was conducted observing the stability of priority rankings. Typically, AHP studies conduct sensitivity analysis using expert choice and graphically altering the weights of decision criteria and observing how rankings of alternatives outcomes change. However, this study did not include a hierarchy level with alternative decision outcomes, only items. Therefore, we assessed the sensitivity by identifying outliers and excluding them. Thereafter, potential rank reversals were observed. The range of data was elicited by box plots.

Bootstrapping (N=1000) was conducted to assess the proximity of values in correspondence to the parameter of the population,

especially acknowledging small samples in the groups of family members and physicians.

Results

Sample Characteristics

The mean CR was 0.22 (median: 0.14, standard deviation, SD=0.24) for all 176 participants. Questionnaires with a CR above 0.2 were excluded. A mean CR was calculated for each subgroup. CR for all people suffering from a rare disease was 0.25 (SD=0.27), CR for families was 0.17 (SD=0.11), and CR for physicians was 0.14 (SD=0.10). Accordingly, the proportion of consistent answers was 56% for patients, 67% for relatives, and 83% for physicians, showing that most of the inconsistencies occurred in the patient subgroup. Solely regarding consistent answers, average CR for all participants was 0.09 (SD=0.05). Characteristics of all participants are shown in Table 2, including participants who answered inconsistently. Physicians were not asked about their civil status or the number of household members because this did not seem to serve our research question. Furthermore, disease severity and age of diagnosis were not applicable for two subgroup.

Information Priorities

Tables 3-5 show both global and local priorities of level 2 and 3 items for all participants interviewed. Standard deviations of local priority weights are presented. Resulting ranks are also listed. As bootstrapping showed that calculated geometric means systematically underestimated the weights of information category, weighted geometric means were calculated. Results are presented separately for each subgroup.

Sensitivity Analysis

The results range is displayed in Figure 1 and shows the potential sensibility of local weights to outliers. The ranking results were calculated based on the geometric means because the literature suggests that this procedure is more precise [27]. However, the following box plots show the range of results in a more intuitive manner, displaying the average mean, as well as the maximum and minimum local weights.

To test for potential rank reversal, we excluded outliers and observed whether rank reversals were of consequence. Figure 1 identifies the outliers visually. The patient subgroup displays only one outlier that results in a rank reversal for the category *research*. *Research* is consequently ranked last with a priority weight of .19. Family members show outliers for categories *medical information* (.09), *therapy* (.21), *diagnosis* (.19), and *general disease pattern* (.60). The exclusion of outliers does not cause rank reversal. For the last group, *physicians*, outliers were identified for the following items: *medical information* (.11), *diagnosis* (.22), and *research* (.17). No rank reversals were observed.

Significance of Results

To examine differences between groups, we conducted a student *t* test, assuming opinions were aggregated following the normal distribution within the population. The results are displayed in Table 6. The null hypothesis states that the importance of items is perceived equally; the alternative hypothesis states that the importance of information on rare diseases is perceived differently. Significant differences are marked.

Furthermore, bootstrapping with a 95% CI was conducted to examine whether sample results lay within specific ranges of the population regarded. The results are presented in Figure 2.

Table 2. Sociodemographic characteristics of patients, family members, and physicians (N=176).

Parameters	Patients (n=120)		Family members (n=24)		Physicians (n=32)	
	Included (n=67)	Excluded (n=53)	Included (n=16)	Excluded (n=8)	Included (n=25)	Excluded (n=7)
Sex						
Male	11	18	2	1	13	3
Female	56	35	14	7	12	4
Age						
Average	51	50	46	49	42	49
Maximum	85	87	62	62	69	56
Minimum	17	17	23	33	28	29
Civil status						
Married or cohabiting	43	37	8	7	-[a]	-
Single	11	11	3	0	-	-
Divorced	9	3	2	1	-	-
Widowed	4	2	3	0	-	-
Educational qualification						
Technical college or university degree	28	16	10	3	25	7
Abitur	9	5	3	1	0	0
Advanced technical college degree	6	5	0	1	0	0
Secondary education	17	19	3	3	0	0
Secondary modern school qualification	7	8	0	0	0	0
Members of the household						
Average	2	5	3	3	-	-
Maximum	5	2	5	5	-	-
Minimum	0	0	0	0	-	-
Age at diagnosis, years						
Average	37	37	4	15	-	-
Maximum	74	79	37	47	-	-
Minimum	0	0	0	0	-	-
Disease severity						
No specification	0	0	1	0	-	-
Low	6	3	0	0	-	-
Medium	32	21	7	5	-	-
Severe	28	29	8	3	-	-
Profession						
Employed	27	25	16	5	25	7
Unemployable	14	10	0	0	0	0
Pensioner	20	14	0	2	0	0
Student or scholar	1	2	0	0	0	0
Homemaker	1	1	0	1	0	0
Special circumstances (further education or provision of work)	4	1	0	0	0	0

Parameters	Patients (n=120)		Family members (n=24)		Physicians (n=32)	
	Included (n=67)	Excluded (n=53)	Included (n=16)	Excluded (n=8)	Included (n=25)	Excluded (n=7)
Medical rare disease experience	-	-	-	-	24	3

[a]The symbol indicates that data are not available.

Table 3. Ranking results of patients.

Parameters	Patients (n=67)				
	Local weight	SD	Global weight	Local ranking	Global ranking
Medical issues	.21	0.21		3 or 4	
Diagnosis	.34	0.24	.070	2	6
Therapy	.37	0.21	.076	1	5
General disease pattern	.30	0.19	.062	3	9
Research	.21	0.17		3 or 4	
Current studies	.32	0.22	.069	2	7
Study results	.32	0.20	.068	3	8
Registers	.36	0.26	.077	1	4
Social help offers	.30	0.19		1	
Psychosocial counseling	.35	0.22	.103	1	1
Self-help	.32	0.24	.095	3	3
Sociolegal advice	.33	0.21	.098	2	2
Current events	.28	0.22		2	

Table 4. Ranking results of family members.

Parameters	Family members (n=16)				
	Local weight	SD	Global weight	Local ranking	Global ranking
Medical issues	.13	0.18		4	
Diagnosis	.24	0.21	.031	2	8
Therapy	.20	0.18	.025	3	9
General disease pattern	.56	0.20	.071	1	3/4/5
Research	.22	0.20		2/3	
Current studies	.31	0.21	.071	2	3/4/5
Study results	.16	0.10	.037	3	7
Registers	.52	0.23	.117	1	1
Social help offers	.22	0.16		2/3	
Psychosocial counseling	.35	0.23	.075	1	2
Self-help	.33	0.27	.071	2	3/4/5
Sociolegal advice	.33	0.22	.070	3	6
Current events	.43	0.18	-	1	-

Table 5. Ranking results of physicians.

Parameters	Physicians (n=25)				
	Local weight	SD	Global weight	Local ranking	Global ranking
Medical issues	.13	0.17		4	
Diagnosis	.23	0.16	.029	3	9
Therapy	.37	0.17	.046	2	7
General disease pattern	.40	0.19	.051	1	6
Research	.18	0.14		3	
Current studies	.44	0.22	.078	1	3
Study results	.25	0.18	.045	3	8
Registers	.32	0.22	.057	2	5
Social help offers	.26	0.17		2	
Psychosocial counseling	.29	0.11	.076	3	4
Self-help	.32	0.20	.083	2	2
Sociolegal advice	.40	0.20	.104	1	1
Current events	.42	0.17		1	

Figure 1. Range of results (local weights) of consistent answers by patients, family members, and physicians. CUS: current studies; DIG: diagnosis; GDP: general disease pattern; MED: medical issues; THE: therapy; PSY: psychosocial counseling; REG: registers; RES: research; SOC: social help offers; SHE: self-help; SOL: sociolegal advice; STR: study results.

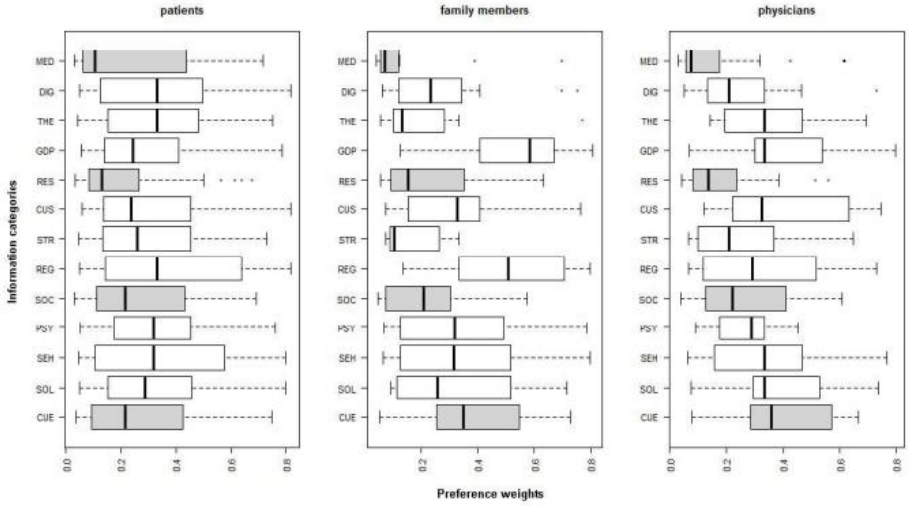
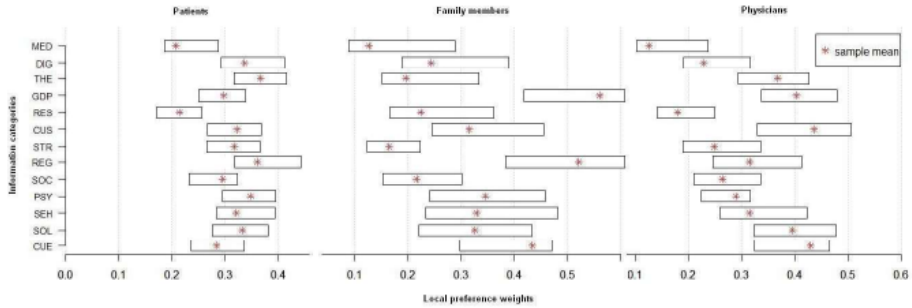


Table 6. Significance of differences between patients, family members, and physicians (n=108).

Parameters	Two-sample *t* test					
	Patients or families		Patients or physicians		Physicians or families	
	t statistic (degrees of freedom)	*P* value	*t* statistic (degrees of freedom)	*P* value	*t* statistic (degrees of freedom)	*P* value
Medical issues	1.60 (26)	.13	1.90 (55)	.06	0.04 (30)	.97
Diagnosis	1.43 (26)	.17	2.59 (62)	.01	−0.45 (26)	.66
Therapy	2.88 (26)	.01	0.07 (52)	.94	2.60 (31)	.01
General disease pattern	−4.26 (22)	<.001	−2.50 (39)	.02	−1.85 (32)	.07
Research	−0.65 (21)	.52	0.59 (54)	.56	−0.98 (24)	.34
Current studies	−0.26 (23)	.80	−1.98 (40)	.05	1.28 (34)	.21
Study results	3.99 (46)	<.001	1.20 (46)	.21	1.98 (38)	.06
Registers	−1.96 (25)	.06	0.87 (49)	.39	−2.44 (31)	.02
Social help offers	1.25 (27)	.28	0.19 (48)	.85	0.94 (34)	.35
Psychosocial counseling	0.01 (22)	.99	2.05 (78)	.04	−1.13 (20)	.27
Self-help	−0.12 (21)	.90	0.02 (48)	.98	−0.13 (26)	.90
Sociolegal advice	0.13 (22)	.90	−1.50 (44)	.14	1.17 (30)	.25
Current events	−1.98 (26)	.06	−2.52 (54)	.01	0.10 (31)	.92

Figure 2. The results of patients, family members, and physicians using bootstrapping and a 95% CI. CUS: current studies; DIG: diagnosis; GDP: general disease pattern; MED: medical issues; THE: therapy; PSY: psychosocial counseling; REG: registers; RES: research; SOC: social help offers; SHE: self-help; SOL: sociolegal advice; STR: study results.



Discussion

Principal Findings

This study shows that rare diseases information categories are weighted very differently, resulting in subgroup specific preference weight structures, distributions, and ranking results. Although *medical issues* were rated as least important by all subgroups, none of the other information categories showed an overall group consensus.

Significant differences between subgroups were confirmed by *t* tests comparing subgroup specific local weights for the following comparisons: the priority weight of patients and family members in the categories *therapy*, *general disease pattern*, and *study results* differed significantly. Moreover, patients and physicians showed significant differences within the categories of *diagnosis*, *general disease pattern*, *current studies*, *psychosocial counseling*, and *current events*. Comparing physicians' results against those of family members, *therapy* and *registers* showed statistical significance.

In quantifying these results, patients and family members showed diverging preference weights for 23% of the cases (3/13). On the other hand, patients and physicians showed different weights for 38% of the cases (5/13). Finally, physicians and family member's weights diverged only in two cases (15%, 2/13). These results indicate that patients and physicians show a comparably high percentage of diverging opinions on the importance of health information, weakening our initial hypothesis that physicians initiate their search strategy based on the patient-physician interaction. These results should be

discussed very carefully because the potential implications are hard to grasp. The statistical significance test was based on the local preference weight. However, the final result of the AHP was expressed as an absolute rank. Therefore, the results should be situated in the overall context. The local weights revealed significant differences in health information with regard to *therapy*. Specifically, patients put this category first (1) on the local level, whereas physicians put it last (3). Regarding the health information on *general disease patterns*, ranks were assigned inversely. Similar rank switches at the local level can be observed when comparing patients' and physicians' perspectives on information relating to *general disease patterns* and *psychological counseling*. Interestingly, *general disease patterns* were perceived as least important by patients (3), whereas physicians regarded it as most important (1). On the other hand, patients considered *psychosocial counseling* as the most important subcategory, whereas physicians considered it the least important.

Checking all subgroups for the sensitivity of results, a rank change could only be observed once. Therefore, we conclude that the results were relatively stable. These results are consistent with Danner et al [35], who interviewed patients while they were completing AHP questionnaires. Extreme values, which could lead to very unstable results, often go along with high inconsistencies. Per these findings, some extreme opinions could have been excluded because of the set CR threshold.

Theoretical Contributions

Interestingly, all subgroups prioritized information on *social help offers* and *current events* over hard facts such as *medical issues* and *research*. This is perhaps because certain medical topics can be discussed directly with physicians following a diagnosis. Unfortunately, we cannot directly compare these findings with the findings of other studies, as the study participants, information categories, and indications vary greatly. However, patients receiving genomic results outlined that they preferred filtering information to avoid information overload and to avoid learning what their future might look like [36]. This anxiety about the future might explain why patients rated medical information as less important, despite the fact that it was named as a main search item in studies such as that of Morgan et al [31]. On the other hand, Anderson et al [37], as well as Schwarzer [38] reported consistent findings with Australian families suffering from genetic metabolic diseases and children with anorectal malformations, emphasizing the importance of self-help groups in the long run and psychosocial counseling when self-help reaches its limits. Dellve et al [39] also highlight the importance of psychosocial counseling for family members, especially parents with a child suffering from a rare disease. These findings also quantitatively support the importance of not only research networks, as advocated for by, for instance, Aymé and Schmidtke [40], but also social networks in the field of rare diseases and inclusion of these networks within national and international rare diseases information platforms, reflecting the unique importance of self-help initiatives in the field of rare diseases. Common diseases often do not need the support of self-help groups because research and political action have already been largely implemented. On the other hand, for rare diseases, many initiatives and knowledge extensions originate from these self-help groups [6]. However, patient initiatives continue to be put at the end of the line. Given that research- and patient-oriented websites still primarily offer either websites for physicians or for patients, even though information valuable to all stakeholders are presented, this makes cocreation and the exchange of opinions even more important.

The information category *registers* was the most important category for families (at rank 1); patients regarded it highly as well, ranking it in 4th place immediately after *social help offers*. Only physicians attributed a high relevance to *current studies*. This statement emphasizes the importance of providing information on rare diseases registers and appreciates the worldwide effort put into the development of such strategies [41], mirroring the importance of longitudinal data acquisition and analysis as numerous rare diseases are connected to a genetic predisposition [19]. These results emphasize the considerable involvement of family members, as they are potentially also affected.

Relatively little interest in study results can be explained through the communication of the results itself. Long et al [42] report that participants of studies receive results only in 33% of the cases. Only half of respondents saw an opportunity to even request the results. However, in this case, almost all respondents demanded researchers to at least sometimes offer the results. The strengthening of the communication of study results can be seen as an opportunity to improve the inclusion of health innovations in health care systems.

The present health information survey among physicians and senior patients reveals some major problems when comparing these results to those of other studies. Specifically, the results vary widely, especially because the health information categories were outlined differently [43]. This indicates that further subgroup analysis can be performed while controlling for influential factors such as age and indication. However, it should also be emphasized that our study forms the basis for an Internet platform for rare diseases and therefore focused on the major relevant stakeholders for this disease category.

Besides, research has often focused on topics such as information access [44] or barriers to information access [45], which leaves the question of how information needs are specified unanswered [46]. Further research is necessary to examine this topic in more detail. Nevertheless, the results have potential for further improving the basis of physician-patient communication.

Practical Implications for Web-Based Health Information Provision

What do these results mean for rare diseases–related information providers such as ZIPSE? The differences between subgroups suggest that subgroup specific information is necessary. First, the ranking structure of rare diseases information categories can be translated, one-by-one, into website design by positioning topics in accordance with stakeholder priorities.

Besides, it seems advisable to considered Miller's Law to avoid information overflow. It appreciated that the whole load of rare diseases Internet resources cannot be processed at once [47].

Limited perception capacities of human brains make it indispensable to only display the most important information at first glance. Miller's Law states that the short-term memory of an average human brain can only absorb approximately 7 items at once, thus, limiting the effectiveness of Internet data processing. Moreover, considering Miller's Law and potential information overflow, only the most important seven items should be included. Therefore, the findings suggest that information categories such as *general disease information* (9), *study results* (8), and *current studies* (7) do not need to be presented initially. In the case of a website especially designed for family members, *current events*, *registers*, *psychosocial counseling*, *self-help*, *sociolegal advice*, *current studies*, and *general disease pattern* should be presented first. On the other hand, physicians prioritized information on *current events*, *sociolegal advice*, *self-help*, *current studies*, *psychosocial counseling*, *registers*, and *general disease pattern*.

Nevertheless, another perspective should also be thought of at this point. From an educational point of view, this study also presents information categories that currently seem undervalued. For example, patients do not perceive *current studies* (7) or *study results* (8) as important, even though these results might hold crucial information for their disease treatment or maintenance. Family members do not perceive *diagnosis* (8) and *therapy* (9) as very valuable. Group representatives often advocate for their children or partners who are suffering from a rare disease to treat these information categories as more important. Moreover, even though approximately 60% of patients see physicians as the primary source of information [14], physicians do not perceive information on *diagnosis* (9), *therapy* (7), and *study results* (8) as important. Therefore, it seems advisable to discuss whether information should be located to improve its visibility and to reflect its importance for the major stakeholder, the patient. Consequently, whether physicians' priorities should reflect patients' interests as an *information lobbyist* also requires examination. First of all, it seems advisable to not only include the underlying results into the design of information platforms on rare diseases but also to discuss information placement with experts in the field and to fully disclose information placement strategies. However, we strive for a high involvement of patients, family members, and physicians to realize efficiency potentials for health care systems. This can only be accomplished by respecting the outcome of the decision-making process translating results one-to-one.

Study Limitations

Data interpretation was a limitation. The AHP research sample size is still a topic of discussion. It has been highlighted that AHP does not require a particularly large sample size [48]. Other authors emphasized that there is no recommendation at all for AHP sample size [23]. Both sources base their statements on the fact that AHP reflects the opinion of the specific group and is thus a group decision-making tool. However, in this study, we raise preference weights, which should be representative for groups when an adequate sample size is achieved.

The quantitative aggregation technique shapes a clear-cut implementation structure for information categories. However, it must be acknowledged that the results illustrate the average opinions of rare diseases' patients, physicians, and family members.

Another issue that should be recognized when interpreting study results is the exclusion of inconsistent answers as part of the AHP methodology. Dolan [49] found that of 20 patients, 90% were willing and capable of completing an AHP. Danner et al [34] argued that extreme values are often chosen to emphasize answers that are not willingly contributed to inconsistencies. In our study, patients delivered inconsistent answers 44% of the time, whereas family members and physicians did so in 34% and 22% of the cases, respectively. However, these results were excluded to follow theoretical AHP requirements.

During pretests of the questionnaire's paper-based version, low consistency values were generated. Ranking cards were included as first choice assistive tools to mirror ranking results immediately. During interviews with patients and family members, this tool was very helpful and led to improved CR values. However, during interviewer-led AHPs, physicians refused to use it. Nevertheless, interviewers noted the shown ranking orders verbally. Finally, a ranking task was placed before each block of comparisons in the Web- and paper-based version.

Comparing physicians with patients, low participation rates are observed. VanGeest et al [50] stated that low participation rates are very common in physicians' surveys. Postal and telephone approaches seem to be more effective than Web-based strategies. Monetary incentives were found to be an effective strategy to increase participation rates. Nonmonetary incentives reflected little changes. Unfortunately, no monetary funds were available for this study.

As already indicated, a change of medium was necessary. Initially, a paper-based version was implemented. After the first recruitment period, a Web-based questionnaire was also introduced to broaden the target group. Several studies such as those of Hirsch et al [51] and Coons et al [52] found differences between participation for paper-based and Web-based surveys. Therefore, it is beneficial to combine both approaches considering representativeness, thus capturing both infrequent and frequent Internet users.

Finally, sociodemographic data show a relatively large proportion of female participants. Literature and other rare diseases Internet providers disclaim that health information on rare diseases are more often searched for by women than by men. For instance, Morgan et al [13] determined that 95.7 % of all inquiries to the Genetic and Rare Disease Information Center came from women.

Conclusions

This study describes an innovation in the involvement of patients, family members, and physicians in effectively gathering, structuring, and presenting health information in a world struggling with an information paradox, namely, health information overflow on the one hand and a major lack of information on rare conditions on the other. This innovation comes in the form of the chosen group decision-making tool, the AHP, which has helped transform individual qualitative

perceptions into a measurable scale. Accordingly, the strength of our study is its transparent quantitative demonstration of the information needs of physicians, patients, and family members, which makes direct comparisons and simple implementation possible. More specifically, this study provides unique insights into the quantitative structure and distribution of information preferences, as well as the validity of results. We were able to verify significant differences between preference weights of patients, family members, and physicians for some items, suggesting that the importance of rare diseases information is perceived differently in these subgroups. User-oriented information providers should seek to address these differences and provide stakeholder-specific websites in accordance with the relevance of health information. Furthermore, the importance of social help offers and current events as part of the information package might be underpinned, with a particular emphasis on the importance of social networks in the field of rare diseases. The finding that communication of study results is potentially undervalued can be seen as an opportunity to improve the inclusion of information on health innovations in health care systems. As we strive for a high involvement of patients, family members, and physicians to realize efficiency potentials for health care systems, the relevance of health information should be directly translated. Results must not only be considered when creating national rare diseases information platforms such as the ZIPSE but also when updating, redesigning, and implementing national and international rare diseases information platforms.

However, as part of the cocreation process, we solely focused on the subgroups interested in information on rare diseases as an explanatory variable for different information needs. We suggest that future studies examine other potential explanatory variables such as for instance gender, educational background, and civil status.

Finally, our findings might be helpful for improving communication between patients, legal guardians or partners, and health advocates, who are closely intertwined. This seems to have high potential because social and professional networks often remain separate within discussions of rare diseases. Promoting a discussion between stakeholders can help in combining forces within the backdrop of a networking approach, which has already been communicated and pursued through the implementation of national rare diseases plans. An understanding network that engages in successful collaboration can improve the quality of life of those affected by rare diseases, as well as lessen the perceived disease burden.

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Conflicts of Interest

None declared.

Multimedia Appendix 1

Description of rare diseases information categories.

[PDF File (Adobe PDF File), 22KB - ijmr_v6i2e23_app1.pdf]

Multimedia Appendix 2

Questionnaire.

[PDF File (Adobe PDF File), 155KB - ijmr_v6i2e23_app2.pdf]

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Abbreviations

ACHSE: Allianz Chronisch Seltener Erkrankungen
AHP: analytic hierarchy process
CHERH: Center for Health Economics Research
CR: consistency ratio
NAMSE: Nationales Aktionsbündnis für Seltene Erkrankungen
SD: standard deviation
ZIPSE: Zentrales Informationsportal über seltene Erkrankungen

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Original Paper

Adopting Quality Criteria for Websites Providing Medical Information About Rare Diseases

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Abstract

Background: The European Union considers diseases to be rare when they affect less than 5 in 10,000 people. It is estimated that there are between 5000 and 8000 different rare diseases. Consistent with this diversity, the quality of information available on the Web varies considerably. Thus, quality criteria for websites about rare diseases are needed.

Objective: The objective of this study was to generate a catalog of quality criteria suitable for rare diseases.

Methods: First, relevant certificates and quality recommendations for health information websites were identified through a comprehensive Web search. Second, all considered quality criteria of each certification program and catalog were examined, extracted into an overview table, and analyzed by thematic content. Finally, an interdisciplinary expert group verified the relevant quality criteria.

Results: We identified 9 quality certificates and criteria catalogs for health information websites with 304 single criteria items. Through this, we aggregated 163 various quality criteria, each assigned to one of the following categories: thematic, technical, service, content, and legal. Finally, a consensus about 13 quality criteria for websites offering medical information on rare diseases was determined. Of these categories, 4 (data protection concept, imprint, creation and updating date, and possibility to contact the website provider) were identified as being the most important for publishing medical information about rare diseases.

Conclusions: The large number of different quality criteria appearing within a relatively small number of criteria catalogs shows that the opinion of what is important in the quality of health information differs. In addition, to define useful quality criteria for websites about rare diseases, which are an essential source of information for many patients, a trade-off is necessary between the high standard of quality criteria for health information websites in general and the limited provision of information about some rare diseases. Finally, transparently presented quality assessments can help people to find reliable information and to assess its quality.

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KEYWORDS

rare diseases; self-help groups; Internet; health information exchange; quality indicators

Introduction

The European Union considers diseases to be rare when they affect no more than 5 in 10,000 people. It is estimated that there are between 5000 and 8000 different rare diseases, affecting nearly 30 million people in the European Union and 4 million people in Germany alone [1,2]. Consistent with this diversity, the quality of information available on the Web varies considerably. People searching the Web often find it very difficult to find the right information and to assess its quality [3,4]. With Orphanet [5], an information platform exists, which holds comprehensive and quality-tested information. However, the target group it addresses is potentially specialists rather than patients [6,7]. In keeping with the European Council's recommendations, Germany has published a National Action Plan for Rare Diseases in August 2013, which will guide and structure actions in the context of rare diseases within their health and social systems [8]. It includes 52 policy proposals. The national project ZIPSE (German: Zentrales Informationsportal über seltene Erkrankungen; English: Central Information Portal about Rare Diseases), initiated by the Federal Ministry of Health, deals with the realization of the plan's topics 37 to 39, which cover the subject of a central information portal [9]. Hereby, the health and well-being of people with rare diseases should be improved.

The aim of the ZIPSE project is to conceptualize and implement a central information portal about rare diseases in Germany. A centralized access point for quality-tested information appears to be very helpful for people with a rare disease, their relatives, and medical experts [9]. The portal itself does not contain primary information but refers to existing quality-assured information sources. The aim is the provision of an intelligent user guide to relevant and appropriate sources of information [10]. Web-based information and websites about rare diseases will be linked in the information portal. More precisely, a variety of quality-tested websites about rare diseases will be offered to all users. Furthermore, users will be able to search for disease-specific websites and to filter them by quality criteria. Therefore, a method to distinguish high- and low-quality websites needs to be established [10,11]. A number of quality certificates for websites dealing with medical information already exist. Websites with such a certificate demonstrate quality-tested content [3]. It can be hypothesized that existing quality certificates for websites with health information (eg, Health On the Net Foundation Code of Conduct, HONcode; DISCERN; and Stiftung Gesundheit) are rarely used by websites about rare diseases. It can be assumed that patient organizations often provide well-researched and reliable information about rare diseases, but they have limited resources in terms of time and money to present themselves as professionally as other information providers on the Web to fulfill the requirements of existing quality certificates. Furthermore, the providers' motivation to present themselves professionally is unknown. The quality control process of certificates such as HONcode can be costly and require significant effort owing to stringent requirements. Verifying websites providing medical information about rare diseases using quality criteria can help increase acceptance and signal trustworthiness to patients, relatives, and medical experts. Most existing quality certificates focused on medical information pursue different goals and contain a wide range of different types of quality criteria. Hence, specific quality criteria for websites about rare diseases are needed. The objective of this study was to generate a catalog of quality criteria suitable for rare diseases. Implementing these quality criteria will improve the evaluation and assessment of information about rare diseases for patients, health professionals, and other users of the information portal.

Methods

The method we adopted can be regarded as a process divided into 3 steps, as shown in the flowchart in Figure 1.

In step 1, a comprehensive Web search was performed to identify quality certificates and criteria catalogs for websites containing medical or health information. Although we focused on programs and catalogs active in Germany because of its implementation of the information portal about rare diseases, we considered several international sources as well. Quality certificates and criteria catalogs were only included if the quality criteria were published transparently. Furthermore, to be included the certificates and catalogs had to focus on Web-based resources containing medical or health information. Certificates, catalogs, and recommendations were therefore excluded if, for example, they focused only on printed medical information. Additionally, websites about rare diseases were analyzed to identify their quality criteria and their use of quality certificates. These criteria were added if they were not already identified through the Web search. Finally, all identified references were again checked for suitability.

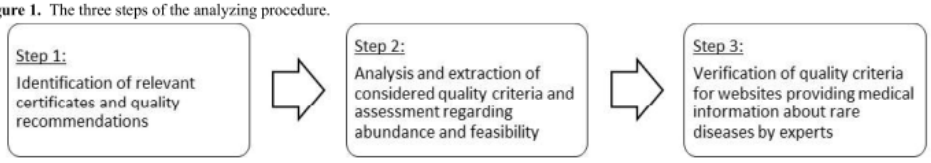
In step 2, the unique criteria of each certification program and catalog were examined, extracted into an overview table, and analyzed by thematic content. Thematic correlations between the criteria were pooled together with an inductive design into major categories. Experts on rare diseases were consulted on the construction of the major categories. Finally, each criterion was assigned to one of the following major categories: thematic, technical, service, content, and legal. Where feasible, the categories were broken down further into groups of criteria. Additionally, experts on rare diseases provided opinions and general information about the importance of each criterion and critical aspects of quality criteria for information about rare diseases. If a criterion was already present in the map, it was not reentered but marked as being part of another criteria catalog. In order to evaluate the importance of a single criterion, its repeated occurrence among different criteria catalogs was examined. Criteria appearing in several catalogs were considered

more important, whereas those that were part of a single catalog alone were considered less important. Thus, a hierarchy of the quality criteria appearing in the identified catalogs was constructed, ordered from the criteria appearing the most number of times to those appearing just once.

In step 3, the most important criteria were selected by the project group as preliminary quality criteria. Next, a workshop was held with various experts on website quality and other publications with medical content, experts on health economics and medical informatics, as well as medical experts in the field of rare diseases. A total of 27 experts participated in the workshop—4 of them were professors and 12 graduate doctors. These experts were invited to participate in the group discussion about quality criteria for websites providing medical information about rare diseases. Participants did not receive incentives to attend the workshop and discussion. The relevance and applicability of each quality criterion were discussed, evaluated, and verified by the expert group. The discussion with medical experts as well as experts on the quality of medical information focused on choosing the criteria that should be mandatory for websites offering medical information on rare diseases. Input from medical experts was equally valuable as input from experts on quality of medical information. At the end of the discussion, the experts were expected to arrive at a consensus on the importance of the different quality criteria. Finally, it was decided which of the quality criteria should be mandatory for these websites to be listed on the information portal about rare diseases. Experts from the following institutions participated in the workshop and group discussion:

- German Action Forum Health Information System (afgis e.V.)
- German Alliance of Chronic Rare Diseases (ACHSE e.V.)
- Agency for Quality in Medicine (ÄZQ)
- Federal Ministry of Health Germany (BMG)
- Charité Universitätsmedizin Berlin
- Center for Health Economics Research Hannover (CHERH)
- German Cochrane Center (DCZ)
- Frankfurt Reference Center for Rare Diseases (FRZSE)
- Institute of Medical Biostatistics, Epidemiology and Informatics (IMBEI), University Medical Center Mainz
- Institute for Quality and Efficiency in Health Care (IQWiG)
- Cancer Information Service Heidelberg (KID)
- Hannover Medical School (MHH)
- National Action League for People with Rare Diseases (NAMSE)
- Orphanet Germany
- Public Health Foundation
- Department of Dermatology, Medical Center University of Freiburg
- University Medical Center Hamburg-Eppendorf (UKE)
- Centre for Quality and Management in Healthcare, Medical Association of Lower Saxony (ZQ)

Figure 1. The three steps of the analyzing procedure.



Results

Identification of Relevant Certificates

A total of 9 quality certificates and criteria catalogs for websites containing medical or health information were identified. Of these certificates and catalogs, 2 were used internationally; 7 were verified only for German websites. The most common certificate for medical information websites was identified as HONcode [12]. Three further certificates verifying only German websites were identified: afgis Qualitätslogo [13], Stiftung Gesundheit [14], and Medisuch [15]. Additionally, several German, European, and international criteria catalogs were considered: afgis Checkliste für medizinische Websites [16], DISCERN [17], Gute Praxis Gesundheitsinformation [18], NAMSE Kriterien und Standards [19], and Patientenorientierte Krankheitsbeschreibung nach ACHSE-Kriterien [20]. Lastly, the results of a study identifying the most important quality criteria for medical information websites were analyzed [21]. All identified quality catalogs are described in Table 1. Furthermore, the development of quality criteria is an ongoing process, including more detailed quality assurance whereby recent quality catalogs take into account older catalogs and quality certificates. In summary, the identified quality catalogs, certificates, and recommendations show different thematic focuses on the criteria that are considered important to ensure a high quality of health information. Moreover, Table 1 shows that the process of determining the quality of information differs among the identified providers (self-reporting audits vs publicly available information).

Table 1. Quality catalogs and quality certificates.

Name	Description	Characteristics	Number of criteria (n=304)
NAMSE Kriterien und Standards[a] [19]	A discussion paper about quality criteria for enhancing patient information about rare diseases.	It contains requirements for the categories: creation process, results, implementation, and evaluation.	56
HONcode[b] [12]	As an international certificate, awarded by the Health On the Net Foundation located in Switzerland and established in 1995, it has held NGO[c] status at the United Nations since 2002. Since 1996, a free certificate for "trusted" sites with medical information was awarded. Since 2015, certification is provided as a paid service. The organization claims that about 8000 medical websites hold their certificate.	Its principles: Information must be authoritative—stating the qualifications of the author. Complementarity—information must supplement and help to support medical advice, not replace it. Privacy—compliance with confidentiality of personal data entered by a website visitor. Assignment—References to sources of information and dates must be present. Verifiability—treatments, products, and services must be supported by balanced, verifiable, scientific information. Transparency and contact information. Disclosure of funding—sponsorship, sponsors, and financial sources must be named. Advertising policy—separation of advertising and editorial content.	55
afgis-Qualitätslogo[d] [13]	The afgis Qualitätslogo is based on 10 quality categories for transparently provided information, whereby the verification is based on self-reporting audits.	It is based on 10 quality categories for transparently provided information: criteria for transparent information about providers, purpose and target group, authors and information sources, data release, timeliness, and planned maintenance of the information, possibility to give user-feedback, procedure of internal quality assurance, separation of advertisement and editorial contribution, financing and sponsoring, cooperation and networking, and data protection, data transmission, and use of data.	39
afgis Qualitätskriterien [16]	afgis Checkliste für medizinische Websites[e] is a guideline for providers that want to regenerate websites with medical information content.	It contains essential Web standards for the following categories: timeliness, data protection, design and navigation, medical information, legal aspects, service aspects, search engine, transparency, and access.	35
Gute Praxis Gesundheitsinformation[f] [18]	A catalog containing quality criteria for the development of health information with a requirement for evidence-based information.	It focuses on the development of health information with a requirement for evidence-based information, which is comprehensible given the expertise of the target group. Thus, the catalog contains different criteria for various target groups.	30

Name	Description	Characteristics	Number of criteria (n=304)
Stiftung Gesundheit[g] [14]	Awards a seal of approval after checking more than 100 issues, whereby the verification is based on information that is available on the website.	It awards a seal of approval after checking criteria out of the following categories: legal quality, publishing diligence, usability, and search engine optimization.	30
Patientenorientierte Krankheitsbeschreibung nach ACHSE-Kriterien[h][20]	Contains quality criteria grouped into 5 categories.	It contains quality criteria of the following categories: creation and formal aspects, medical-scientific data and information, disease management, establishment of contact and information about specialties of health professionals, and additional links and references.	28
DISCERN [17]	A tool to evaluate medical publications with a focus on patient information.	It focuses on the following: reliability of the publication and quality of information on treatment alternatives.	19
Medisuch [15]	Provides a certification process and is operated by the institute for quality and transparency of health information.	As a part of its certification process, information providers have to declare that the information provided on the website is not influenced by industrial offers.	12

[a] NAMSE Kriterien und Standards: NAMSE (National Action League for People with Rare Diseases) criteria and standards (in English).

[b] HONcode: Health On the Net Foundation Code of Conduct.

[c] NGO: nongovernmental organization.

[d] afgis Qualitätslogo: German Action Forum Health Information System (afgis) quality logo (in English).

[e] afgis Checkliste für medizinische Websites: afgis checklist for medical websites (in English).

[f] Gute Praxis Gesundheitsinformation: good practice health information (in English).

[g] Stiftung Gesundheit: Public Health Foundation (in English).

[h] Patientenorientierte Krankheitsbeschreibung nach ACHSE-Kriterien: patient-oriented description of disease by the criteria of ACHSE (German Alliance of Chronic Rare Diseases) (in English).

Analysis and Extraction of Quality Criteria

The number of criteria present in the quality certificates is listed in Table 1. The presented number can be higher (or lower) than the official numbers stated by the providers owing to a more detailed valuation of criteria by the project group. The number of criteria ranged from 12 to 56 in the catalogs analyzed. In total, we identified 304 single criteria items. Through this, we aggregated 163 different quality criteria into 5 major categories: thematic, technical, service, content, and legal. The thematic criteria category containing 90 criteria (90/163, 55.2%) was by far the largest, followed by the service category with 26 criteria (26/163, 16.0%), the technical category with 18 (18/163, 11.0%), the legal category with 15 (15/163, 9.2%), and the content category with 14 (14/163, 8.6%). The degree of detail varied among the different criteria catalogs, and while 66 criteria (66/163, 40.5%) were found in multiple catalogs, no criterion was found in all of the certificate definitions or criteria catalogs. The 2 most frequently occurring criteria appeared in 6 of the analyzed catalogs (6/9, 67%). Three criteria appeared in 5 (5/9, 56%) and 13 criteria in 4 of the catalogs (4/9, 44%), whereas 20 criteria appeared in 3 (3/9, 33%) and 28 criteria in 2 of the catalogs (2/9, 22%). The majority of 87 criteria were unique to a single catalog. With the exception of one catalog (Gute Praxis Gesundheitsinformation), each contains a criterion unique to itself. All identified quality criteria are presented in Multimedia Appendix 1. In summary, the number of criteria present in quality certificates and quality catalogs differs. Nevertheless, most catalogs contain a unique criterion not shown elsewhere. The number of quality criteria in each of the major categories varies widely.

Expert Verification

To assess the relevance of a quality criterion specific to websites offering medical information on rare diseases, different principles were applied. First, criteria appearing in many of the reviewed catalogs were considered more important to ensure a certain level of information quality. This resulted in initially selecting the two most abundant criteria (authors are mentioned and creation and updating dates of information are mentioned) as mandatory for websites to be listed in the information portal ZIPSE. Criteria appearing less often were only selected in consideration with their relevance and their applicability to rare diseases and the targeted websites. This relevance was assessed by checking several properties. If a criterion is applicable, it is to a certain extent defined by its feasibility. Criteria seemingly

important to the quality of general medical information may only be adapted to a limited extent. Finally, in the discussion workshop with 27 experts, quality criteria for websites offering medical information on rare diseases were defined. A consensus about the following 13 quality criteria for websites offering medical information on rare diseases was determined:

- Authoring information
- Mentioning of authors
- Mentioning of sources
- Mentioning of creation and update date
- Data security
- Declaration of evidence
- Marking of conflicts of interests
- Consideration of target group
- Evaluation of content
- Review of information
- Characteristics of the website (accessibility)
- Imprint
- Contact opportunity

A decision was made on the quality criteria that should be a mandatory requirement for websites about rare diseases for them to be listed in the information portal. As a legal requirement for all websites, an adequate *data protection concept* as well as an *imprint* is mandatory. Moreover, we identified the *creation and updating date* and the possibility to *contact the website provider* as very important categories for patients with a rare disease.

Discussion

Principal Findings

The literature review of quality catalogs, certificates, and recommendations for websites containing medical or health information showed different thematic focuses on criteria that are important for the quality of health information. Interestingly, the investigated certificates reveal a great variety of quality criteria used by the common certificates. There is also a wide range of quality criteria where the degree of detail varied among the different criteria catalogs. Furthermore, the process for determining the quality of websites differs among the identified providers (self-reporting audits, eg, [13] vs publicly available information, eg, [14]). The classification of the quality criteria into the major categories, thematic, technical, service, content, and legal, showed that the number of quality criteria in each category varies widely. The presence of a larger number of quality criteria in one category does not necessarily indicate a greater relevance of the category. It is rather an indication that this category can be investigated more thoroughly than categories with a smaller number of different criteria [12].

Defined quality criteria for websites about rare diseases were coordinated and verified by a multidisciplinary expert group to ensure the quality of the information provided. These quality criteria will be applied for registration of websites on the portal about rare diseases. Out of the 13 verified quality criteria for websites about rare diseases, 4 were identified to be mandatory for registration to the information portal. First, as a legal requirement for all websites an adequate *data protection concept* and an *imprint* are mandatory. Moreover, *creation and updating date* and *possibility to contact the website provider* were identified as very important categories for patients with a rare disease. The documentation of the creation and updating date of information is especially important owing to rapid advances in the development of information and to demonstrate the latest research findings [22]. The possibility to contact the website provider is also an important quality aspect for these websites. Particularly, if there is limited information elsewhere, patients, health professionals, and other users can offer the provider advice or suggestions for improvement or ask for more precise information about a rare disease [23]. These 4 categories are mandatory for registration to the information portal and for linking to medical information about rare diseases. Fulfillment of the remaining 9 categories is optional. Nonetheless, these categories are still important for quality-tested information about rare diseases. To achieve transparency, it would be beneficial to publish the degree to which the websites fulfill these categories. In particular, information on the characteristics of the website, such as its accessibility, is important for many patients [24]. Thus, the fulfillment of each single low-barrier criterion needs to be shown transparently.

Using quality criteria to verify websites providing medical information about rare diseases can help to improve their acceptance and signal trustworthiness to patients, relatives, and medical experts [3]. In further studies, all selected quality criteria will be transferred to a so-called self-disclosure questionnaire. These questions will then be used to assess the quality of rare disease websites. The results from the first evaluation of these can help to improve and adjust the quality assessment process of the information portal. Moreover, we can evaluate and test the assumptions made at the beginning:

- Do patient organizations provide well-researched and reliable information about rare diseases?
- Do they present themselves as professionally as other information providers on the Web to fulfill the requirements of existing quality certificates?
- Do websites with little content and a small editorial staff hold high-quality information?

A further problem for investigation is the availability of robust evidence of information on rare diseases. Providing evidence for the source of information is a requirement often sought to ensure a piece of information is well researched. However, with merely 5 in 10,000 people affected by rare diseases, it is almost impossible to collect sufficient data to statistically test a hypothesis. It could be argued that a single proven case is also a form of evidence, albeit a very thin one. However, as long as no other data exist, it is still the best evidence available [25]. There are also important implications for future research from analysis of those categories where we identified a lower number of different criteria. New detailed quality criteria on these categories may help improve the discussion on quality of websites providing medical information.

Limitations

Despite our focus on programs and catalogs active in Germany, we identified a large number and variety of different quality criteria. As with other quality catalogs, the defined criteria

cannot verify the thematic content of health information. These criteria simply verify factors influencing the thematic content, as well as the quality of the website itself. A more complex and expensive solution to verify the heterogeneous information about rare diseases would be for medical experts to verify and highlight single articles of listed websites about rare diseases in the information portal. The defined quality criteria for such websites were verified by the participants of a workshop. Although this workshop was held with 27 renowned and excellent experts on website quality and other publications with medical content, experts on health economics and medical informatics, as well as medical experts in the field of rare diseases, subjectivity in their decision-making process cannot be ruled out.

Conclusions

The relatively low intersection of criteria appearing in the different criteria catalogs shows that the opinion of what is important concerning quality of medical information differs. For the development of useful quality criteria for websites about rare diseases, a trade-off between the high standard of quality criteria for general health information and the provision of limited existing information about rare diseases, which is essential for many patients, appears unavoidable. Providing defined quality criteria for websites about rare diseases can help seekers to find reliable information and to assess its quality [3,4]. Accepted criteria for websites with information about rare diseases, which allow for a minimum of quality control while keeping the workload reasonable, have been defined. In summary, 13 categories with quality criteria were defined by a group consisting of medical experts as well as experts on the quality of medical information. Fulfillment of 4 of these categories (*data protection concept*, *imprint*, *creation and updating date*, and *possibility to contact the website provider*) was identified as being mandatory for registration to the information portal and for publishing medical information about rare diseases. With the help of these quality criteria, we can evaluate, for instance, the quality of information provided by rare disease self-help groups or other information providers.

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Conflicts of Interest

None declared.

Multimedia Appendix 1

Quality criteria for health information websites.

[PDF File (Adobe PDF File), 563KB-Multimedia Appendix 1]

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Abbreviations

ACHSE: German Alliance of Chronic Rare Diseases
afgis: German Action Forum Health Information System
BMG: Federal Ministry of Health Germany
e.V: registered society
HONcode: Health On the Net Foundation Code of Conduct
NAMSE: National Action League for People with Rare Diseases
ZIPSE: Central Information Portal about Rare Diseases

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Rare Diseases on the Internet:

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Original Paper

Rare Diseases on the Internet: An Assessment of the Quality of Online Information

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Abstract

Background: The importance of the Internet as a medium for publishing and sharing health and medical information has increased considerably during the last decade. Nonetheless, comprehensive knowledge and information are scarce and difficult to find, especially for rare diseases. Additionally, the quality of health or medical information about rare diseases is frequently difficult to assess for the patients and their family members.

Objective: The aim of this study is to assess the quality of information on the Internet about rare diseases. Additionally, the study aims to evaluate if the quality of information on rare diseases varies between different information supplier categories.

Methods: A total of 13 quality criteria for websites providing medical information about rare diseases were transferred to a self-disclosure questionnaire. Identified providers of information on the Internet about rare diseases were invited to fill out the questionnaire. The questionnaire contained questions about the information provider in general (eg, supplier category, information category, language, use of quality certificates, and target group) and about quality aspects that reflect the 13 quality criteria. Differences in subgroup analyses were performed using *t* tests.

Results: We identified 693 websites containing information about rare diseases. A total of 123 questionnaires (17.7%) were completely filled out by the information suppliers. For the remaining identified suppliers (570/693, 82.3%), the questionnaires were filled out by the authors based on the information available on their website. In many cases, the quality of websites was proportionally low. Furthermore, subgroup analysis showed no statistically significant differences between the quality of information provided by support group/patient organization compared to medical institution (P=.19). The quality of information by individuals (patient/relative) was significantly lower compared to information provided by support group/patient organization (P=.001), medical institution (P=.009), and other associations and sponsoring bodies (P=.001) as well.

Conclusions: Overall, the quality of information on the Internet about rare diseases is low. Quality certificates are rarely used and important quality criteria are often not fulfilled completely. Additionally, some information categories are underrepresented (eg, information about psychosocial counseling, social-legal advice, and family planning). Nevertheless, due to the high amount of information provided by support groups, this study shows that these are extremely valuable sources of information for patients suffering from a rare disease and their relatives.

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KEYWORDS

health literacy; rare disesases; quality indicators; health information exchange

Introduction

The quality of information provided on the World Wide Web has been highly discussed in the literature for the past few years (eg, [1-3]). In particular, regarding medical information, the provision of high-quality information is very important because misinformation can lead to serious health consequences for the affected patients. This is particularly relevant for information on the World Wide Web, where the information is used without the intervention of a medical professional, even though the related websites clearly state that this information cannot replace a medical professional's consultation [4-9].

In the field of rare diseases, information is scarce; it is difficult to find the right information as well as to assess the quality of the provided information in detail [10-12]. Additionally, only a few medical experts for specific rare diseases have comprehensive knowledge about the diseases. This limits the ability of patients to get access to high-quality information [13,14]. The definitions of rare diseases vary from 12:100,000 in Australia to 75:100,000 in the United States [15]. This study is set in Germany; therefore, it is based on the European Union definition that considers diseases to be rare when the prevalence is less than 50:100,000 [16]. It is estimated that there are between 5000 and 8000 different rare diseases affecting nearly 30 million people in the European Union and 4 million people in Germany alone [15,17,18].

A detailed description of the framework of this study can be found in the literature [19]. In brief, the aim of the project is to conceptualize and implement a central information portal about rare diseases in Germany, which refers to existing quality-assured information sources [20]. The distribution of information and knowledge about rare diseases is an important factor to improve the overall situation of people affected by a rare disease [17,21]. In this context, the Internet as a worldwide open-access medium has become more important during the last decade [22,23]. The Internet can improve the distribution of information about rare diseases to the general public and, in particular, to medical professionals, patients, and relatives of patients [22]. For the latter group, the Internet is one of the most frequently used information resources and often the primary source to search for information after getting a diagnosis [24]. Nevertheless, patients reported that they are often overstrained with the information they find on the Internet [25]. Information is often disordered and refers to different stages of the disease. Moreover, it is not possible to assess the quality of the information and to find the right information, such as social-legal advice [1]. For medical professionals, it is important to have access to the latest innovative research results and evidence-based therapeutic options as well as actual contact details of support groups [26].

The aim of this study is to assess the quality of information on the Internet about rare diseases. Additionally, the study aims to evaluate if information about rare diseases (eg, information provided by support groups) is as reliable as information provided by medical institutions by performing subgroup analyses. The assessment is based on 13 quality criteria for websites providing medical information about rare diseases [19].

Methods

We divided the methodological framework into several steps. First, as mentioned previously, 13 quality criteria for websites providing medical information were included to a self-disclosure questionnaire. The questionnaire contained questions about the information provider in general (eg, supplier category, information categories, language, use of quality certificates, and target group) and questions about quality aspects reflecting the 13 quality criteria (Textbox 1). The disclosure was not anonymous because the answers need to be checked by the authors. The questionnaire was verified and pretested by the patient organization Alliance of Rare Chronic Diseases Germany (ACHSE eV) and Orphanet Germany. Additionally, the verified version of the questionnaire was tested by selected rare disease information providers, which were randomly identified by an Internet search.

Second, information providers on the Internet were identified by an Internet search; all 8000 rare diseases, as listed in the Orphanet list of rare diseases and synonyms [27], were entered into the Google search engine by a number of research assistants from May 2015 to January 2016. This list included all registered rare diseases and their synonyms. For every disease, the first two hit lists, meaning the first 20 hits, were screened to identify information websites in the German language. A random check with 30 diseases showed that we could assume that a screening of the first two hit lists of each rare disease was sufficient to identify all relevant information websites. Websites that provided information about rare diseases were included in the database, whereas those that just presented contact data, for example, with no further information were excluded. Furthermore, websites providing information about several rare diseases were included into the database as a singular information provider. Third, all information providers were invited by email to fill out the self-disclosure questionnaire (September 2015 to March 2016). Then, these datasets were double-checked using the information available on the website. Data were checked for correctness (eg, does the website provide information about the stated information category?) and plausibility (eg, is the description of the process of systematic or literature research comprehensible?). For all information providers who did not fill out the questionnaire, the questions were answered by the authors based on the information available on the website. For that, authors checked the content and the characteristics of each identified website carefully. However, just 10 of 13 quality criteria could be answered by publicly available information. The remaining three quality criteria, representing the authoring information, evaluation of information, and review of information, were not reviewable by the authors. Consequently, for the main evaluation, these quality criteria were excluded. In the end, all datasets were evaluated. Microsoft Access was used for data storage. For data analysis, both Microsoft Excel and Microsoft Access (versions 2007) were used. Differences in subgroup analyses were performed using *t* tests.

Textbox 1. Quality criteria for websites about rare diseases.

Authoring information

- Do you perform a systematic (literature) search prior to providing information for your home page? If yes, then please describe this process.
- Are experts involved in providing information? If yes, then which field do they belong to?
- Do you document the process of providing information? If yes, then please describe the documentation process.
- Do you inform users about the process of developing information? If yes, please describe the process and provide the respective URL.

Authors

- Is general information about the authors mentioned?
- Are other persons who contributed to developing information mentioned?
- Is user-generated content distinguishable and labeled with a username?

Sources

- Does the information concern primary sources of information?
- If no, then do you quote external sources?

Creation or update date

- Is the creation date of information mentioned?
- Is the update date of information mentioned?

Privacy statement

- Is a privacy policy used to inform the user about the usage, storage, and disclosure of personal data?
- Do you inform the user in a prominent position about the storage of personal data for internal usage (eg, research) with an analysis tool and does the user has the opportunity to disagree?
- Does the user has to agree actively to the disclosure of personal data to third parties?

Declaration of evidence

- Is all medical information evidence-based and it is discernible on what basis points are made (eg, studies, expert statements)?
- Do you provide references to the limitations of the evidence and set out further evidence needs?

Marking of conflicts of interests

- Are advertisements marked as such plainly?
- Are sponsors named?
- Are targets and purposes of the home page published (eg, commercial interest)?
- Is the funding (except from self-financing) published?
- Are conflicts of interests mentioned?

Consideration of target group

- Is information presented target group-specific?
- Is it discernible to whom the information is addressed (eg, patients, doctors)?

Evaluation of information

- Does an archive with former or changed contents exist?
- Is all information checked consistently regarding correctness and accuracy?

Review of information

- Does an internal review process (content quality assessment) for the evaluation of contents exist? If yes, then please describe the process.

Characteristics of the website (accessibility)

- Did you check the website for accessibility through a BITV-Test? (The BITV-Test is a comprehensive accessibility evaluation instrument.) If yes, how many points has the website scored in this test?

- Is the font size of the website adjustable?
- Do you consider persons with color vision deficiency in the website coloration?
- Is the main menu selectable without a mouse?
- Information is available in a simple language (eg, according to the rules of the network Simple Language).
- Is the website's content readable by a software tool?
- Is it possible to subscribe to a newsletter?
- Is information available in a printed version?
- Are the contents shown in multimedia (eg, in terms of videos and photos)?

Imprint

- Is the imprint created according to § 5 TMG/§ 55 RStV following German law?

Contact facility

- Do users have the facility to provide feedback or to get in touch with the operator?
- Is a contact sheet easy to access?

Results

Overall, we identified 693 information suppliers on the Internet providing information about rare diseases in the German language or from German-speaking countries. A total of 123 questionnaires (17.7%) were completely filled out by the information suppliers. For the remaining identified suppliers (570/693, 82.3%), the questionnaires were filled out by the authors, omitting the questions referring to quality criteria representing the authoring information, evaluation of information, and review of information. A list of the identified information supplier is available from the corresponding author on reasonable request.

Most of the websites were located in Germany (632/693, 91.2%), Austria (21/693, 3.0%), or Switzerland (40/693, 5.8%); therefore, most of the sites were available in the German language (682/693, 98.4%). However, some were available only, or additionally, in the English language (108/693, 15.6%). The fact that websites can be available in more than one language has to be taken into account. The majority of websites were those of patient organizations or support groups (269/693, 38.8%). Other important providers were medical institutions (186/693, 26.8%), other associations and sponsoring bodies (65/693, 9.4%), and individuals (eg, patient/relative; 52/693, 7.5%). The three most frequent information categories of all information suppliers were information about disease patterns/symptoms (633/693, 91.3%), information about diagnostics (517/693, 74.6%), and information about medication, curative means, and aids (359/693, 51.8%). Little information was available about psychosocial counseling (49/693, 7.1%), in particular. As a target group, adults were most frequently addressed (662/693, 95.5%). All characteristics are shown in detail in Table 1.

Tables 2 and 3 show the comparison and distribution between supplier and information categories. For instance, it can be seen that information provided by individuals mostly focused on disease patterns/symptoms, wherby information provided by medical institutions additionally focused on diagnostics. Furthermore, information exchange with other patients and information about psychological counseling were mostly provided by support groups/patient organizations.

Table 1. Characteristics of information providers (N=693).

Item	n (%)
Supplier category	
Support group/patient organization	269 (38.8)
Medical institution	186 (26.8)
Other associations and sponsoring bodies	65 (9.4)
Individual (patient/relative)	52 (7.5)
Expert association	40 (5.8)
Individual (medical expert)	29 (4.2)
Pharmaceutical or medical technology company	26 (3.8)
Publishing or media company	21 (3.0)
Other	5 (0.7)
Information category (multiple answers possible)	
Disease pattern/symptoms	633 (91.3)
Diagnostics	517 (74.6)
Medication, curative means, and aids	359 (51.8)
Assistance for self-help	347 (50.1)
Information exchange with other patients	320 (46.2)
Other therapy options	317 (45.7)
Research	254 (36.7)
Personal advice	164 (23.7)
Training and continued education	128 (18.5)
Advice from doctors	116 (16.7)
Therapeutic guidelines	101 (14.6)
Desire to have children/family planning	93 (13.4)
Social-legal advice	86 (12.4)
Psychosocial counseling	49 (7.1)
Language (multiple answers possible)	
German	682 (98.4)
English	108 (15.6)
Country	
Germany	632 (91.2)
Switzerland	40 (5.8)
Austria	21 (3.0)
Target group (multiple answers possible)	
Adults	662 (95.5)
Children	235 (33.9)
Medical professionals	221 (31.9)
Self-disclosure	
Accomplished by the supplier	123 (17.7)
Accomplished by authors	570 (82.3)

Table 2. Comparison and distribution between supplier (individual-medical expert, individual-patient/relative, expert association, medical institution, and pharmacetuical or medical technology company) and information categories.

Category	Supplier									
	Individual (medical expert)		Individual (patient/relative)		Expert association		Medical institution		Pharmaceutical or medical technology company	
	n (%)	Supplier %	n (%)	Supplier %	n (%)	Supplier %	n (%)	Supplier %	n (%)	Supplier %
Medication, curative means, and aids	12 (3.3)	41.4	26 (7.2)	50.0	18 (5.0)	45.0	79 (22.0)	42.5	22 (6.1)	84.6
Information exchange with other patients	8 (2.2)	27.6	41 (12.8)	78.9	6 (1.9)	15.0	8 (2.5)	4.3	3 (0.8)	11.5
Diagnostics	22 (6.1)	75.9	27 (5.2)	51.9	30 (5.8)	75.0	158 (30.6)	85.0	21 (5.9)	80.8
Research	11 (3.1)	37.9	11 (4.3)	21.2	20 (7.8)	50.0	92 (36.2)	49.5	5 (1.4)	19.2
Training and continued education	6 (1.7)	20.7	3 (2.3)	5.8	13 (10.2)	32.5	46 (35.9)	24.7	0 (0.0)	0.0
Assistance for self-help	9 (2.5)	31.0	21 (6.1)	40.4	17 (4.9)	42.5	32 (9.2)	17.2	11 (3.1)	42.3
Desire to have children/family planning	5 (1.4)	17.2	6 (6.5)	11.5	1 (1.1)	2.5	14 (15.1)	7.5	5 (1.4)	19.2
Disease pattern/symptoms	28 (7.8)	96.6	47 (7.4)	90.4	32 (5.1)	80.0	165 (26.1)	88.7	23 (6.4)	88.5
Personal advice	6 (1.7)	20.7	2 (1.2)	3.9	7 (4.3)	17.5	41 (25.0)	22.0	5 (1.4)	19.2
Psychosocialcounseling	0 (0.0)	0.0	0 (0.0)	0.0	2 (4.1)	5.0	10 (20.4)	5.4	0 (0.0)	0.0
Other therapy options	16 (4.5)	55.2	27 (8.5)	51.9	15 (4.7)	37.5	98 (30.9)	52.7	11 (3.1)	42.3
Social-legal advice	1 (0.3)	3.5	2 (2.3)	3.9	3 (3.5)	7.5	13 (15.1)	7.0	4 (1.1)	15.4
Therapeutic guidelines	6 (1.7)	20.7	3 (3.0)	5.8	10 (9.9)	25.0	26 (25.7)	14.0	2 (0.6)	7.7
Advice from doctors	4 (1.1)	13.8	0 (0.0)	0.0	18 (15.5)	45.0	62 (53.5)	33.3	5 (1.4)	19.2

Table 3. Comparison and distribution between supplier (support group/patient organization, publishing or media company, other associations and sponsoring bodies, and other) and information categories.

Category	Supplier							
	Support group/patient organization		Publishing or media company		Other associations and sponsoring bodies		Other	
	n (%)	Supplier %	n (%)	Supplier %	n (%)	Supplier %	n (%)	Supplier %
Medication, curative means, and aids	148 (41.2)	55.0	15 (4.2)	71.4	36 (10.0)	55.4	3 (0.8)	60.0
Information exchange with other patients	227 (70.9)	84.4	3 (0.8)	14.3	24 (7.5)	36.9	0 (0.0)	0.0
Diagnostics	193 (37.3)	71.8	20 (5.6)	95.2	42 (8.1)	64.6	4 (1.1)	80.0
Research	76 (29.9)	28.3	4 (1.1)	19.1	33 (13.0)	50.8	2 (0.6)	40.0
Training and continuededucation	41 (32.0)	15.2	3 (0.8)	14.3	16 (12.5)	24.6	0 (0.0)	0.0
Assistance for self-help	223 (64.3)	82.9	4 (1.1)	19.1	29 (8.4)	44.6	1 (0.3)	20.0
Desire to have children/family planning	51 (54.8)	19.0	5 (1.4)	23.8	6 (6.5)	9.2	0 (0.0)	0.0
Disease pattern/symptoms	259 (40.9)	96.3	21 (5.9)	100.0	53 (8.4)	81.5	5 (1.4)	100.0
Personal advice	91 (55.5)	33.8	0 (0.0)	0.0	12 (7.3)	18.5	0 (0.0)	0.0
Psychosocial counseling	33 (67.4)	12.3	0 (0.0)	0.0	4 (8.2)	6.2	0 (0.0)	0.0
Other therapy options	108 (34.1)	40.2	14 (3.9)	66.7	24 (7.6)	36.9	4 (1.1)	80.0
Social-legal advice	54 (62.8)	20.1	0 (0.0)	0.0	9 (10.5)	13.9	0 (0.0)	0.0
Therapeutic guidelines	37 (36.6)	13.8	7 (2.0)	33.3	10 (9.9)	15.4	0 (0.0)	0.0
Advice from doctors	9 (7.8)	3.4	3 (0.8)	14.3	14 (12.1)	21.5	1 (0.3)	20.0

As a first investigation, all identified websites about rare diseases were analyzed for the use of quality certificates. The majority of websites about rare diseases did not use certifications or quality seals. Of the 693 websites analyzed, only 28 (4.0%) were certified by the international Health on the Net Foundation Code of Conduct (HONcode) [28]. Additionally, some were certified by the German certification programs German Action Forum Health Information System (afgis) [29] (7/693, 1.0%) or Medisuch [30] (8/693, 1.2%).

Table 4 shows the results for the evaluation of the quality of information on the Internet about rare diseases. The quality criteria authoring information, evaluation of information, and review of information were based on the datasets from the 123 questionnaires that were filled out by the information supplier. All other quality criteria were based on the datasets of all information providers. It was examined whether the information of websites satisfied the defined quality categories. For some categories, it was not necessary to meet every corresponding item; it was sufficient to fulfill a part of the corresponding items (eg, to fulfill the category sources, the website must contain either primary information or mention external sources, not necessarily both of them). None of the websites fulfilled all the quality criteria and the corresponding categories completely.

Table 4. Quality of information websites (N=693).

Item	n (%)
Quality criteria	
Authoring information[a]	102 (82.9)
Authors	376 (54.3)
Sources	229 (33.0)
Creation or update date	467 (67.4)
Privacy statement	474 (68.4)
Declaration of evidence	360 (51.9)
Marking of conflicts of interests	211 (30.4)
Consideration of target group	643 (92.8)
Evaluation of information[a]	99 (80.5)
Review of information[a]	47 (38.2)
Characteristics of the website (accessibility)	
BITV-Test (barrier-free information technology regulation)	0 (0.0)
Font size adjustable	692 (99.9)
Consideration of persons with color vision deficiency in coloration	396 (57.1)
User can have read out website's content	692 (99.9)
Main menu selectable without a mouse	689 (99.4)
Information in simple language	0 (0.0)
Newsletter	120 (17.3)
Printed version	218 (31.5)
Multimedia	299 (43.1)
Imprint	638 (92.1)
Contact facility	687 (99.1)
Use of quality certificates	
HONcode	28 (4.0)
Medisuch	8 (1.2)
Afgis	7 (1.0)
Stiftung Gesundheit	0 (0.0)

[a] Based on the datasets from the 123 questionnaires that were filled out by the information supplier.

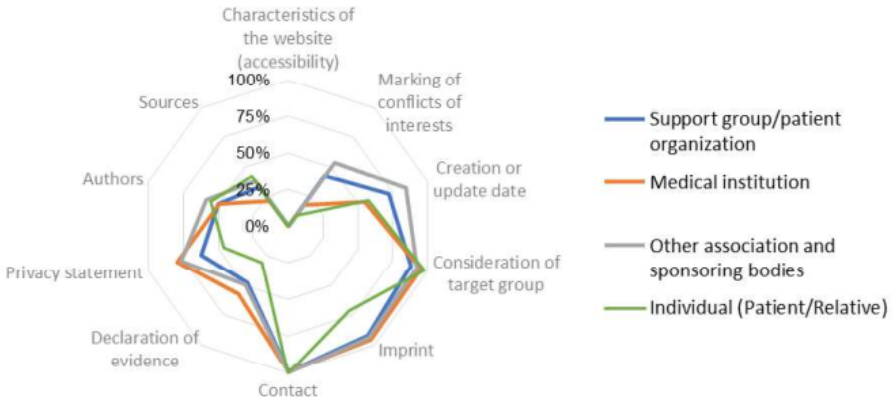
More than 90% of the information suppliers fulfilled the quality criteria of providing contact facility (687/693, 99.1%), imprint (638/693, 92.1%), and consideration of target group (643/693, 92.8%). Although important quality criteria for websites providing information about rare diseases, the criteria declaration of creation or updating date (467/693, 67.4%) and privacy

statement (474/693, 68.4%) were met by only approximately 70% of the identified information suppliers.

The information criteria about characteristics of the website (accessibility) can be divided into several aspects for more detailed analyses. For instance, 43.1% (299/693) of the websites provided the information with the support of multimedia, 31.5% (218/693) also provided printed information, and 17.3% (120/693) provided an email newsletter service. Moreover, 57.1% (396/693) considered persons with color vision deficiency in designing their websites. Detailed results are shown in Table 4.

Subgroup analyses were performed for the four most frequent information supplier categories: support group/patient organization, medical institution, other associations and sponsoring bodies, and individuals (patient/relative). Under the assumption that the fulfillment of every single quality criterion has equal weight, the quality of information of various information supplier categories were compared. On the basis of the 10 quality categories which could be evaluated for all information providers, statistically significant differences could be observed for the supplier category individuals (patient/relative) using a *t* test analysis. The quality of information by these suppliers was significantly lower compared to information provided by support group/patient organization (P=.001), medical institution (P=.009), and other associations and sponsoring bodies (P=.001) as well. No statistically significant differences were observed for the quality of information provided by support group/patient organization compared to medical institution (P=.19). Additionally, information provided by other associations and sponsoring bodies showed statistically significant differences compared to that provided by support group/patient organization (P=.007) and by medical institution (P=.001). The quality of information provided by other associations and sponsoring bodies was significantly higher. Figure 1 shows the distribution of fulfillment of quality criteria by information and supplier categories.

Figure 1. Fulfilment of quality criteria by information provider.



Discussion

Principal Findings

Information about rare diseases is scarce. In the German-speaking setting, 693 websites containing information about rare diseases were identified. In many cases, the quality of these websites, based on the defined quality criteria for websites containing information about rare diseases, can be assessed as insufficient. In addition, quality certificates are rarely used by information providers of rare diseases.

Particularly, the accessibility of the websites needs to be improved, although because of browser configuration, the adjustment of the font size, the selection of the main menu without a mouse, and the readout of website's content seems to be working for most of the websites without any problems. However, providing information by other means, such as email, newsletters, and printed versions, is offered only by some information providers. Support group/patient organizations and other associations and sponsoring bodies are more commonly among those who provide access to their information in various ways. None of the information suppliers provide information in simple language according to the official rules of the network of simple language [31]. Additionally, mentioning of sources of information and disclosing conflicts of interests are seldom stated, although these are important aspects for assessing medical or health information. Furthermore, because of rapid advantages in the development of information and to demonstrate the latest research findings, the documentation of the creation or updating date and the declaration of evidence should be stated more often. On the positive side, an opportunity to contact the website operator is provided in most cases.

Not all information suppliers provide an adequate imprint and privacy statement, even though this is required by German law. In particular, support groups/patient organizations and individuals (patient/relative) do not provide these kinds of

information, although their implementation should be rather straightforward. It can be hypothesized that ignorance and limited experience prevent these supplier categories presenting themselves as professionally as other information providers online. A guidance document for support groups/patient organizations and individuals could help to improve the website's quality.

By far, support groups and patient organizations provide most of the information websites for rare diseases. This reflects the importance of support groups for patients suffering from rare diseases and their relatives [32]. Due to limited knowledge about the diseases, the insufficient experiences of most of the medical professionals, and often limited therapeutic approaches, as well as the low number of affected patients, support groups for patients with rare diseases are important possibilities to share knowledge, experiences, and advice with other affected patients. Support groups and patient organizations for rare diseases constitute very important sources of information about rare diseases and contain high potential to solve upcoming research questions [32]. Moreover, the significant number of identified websites by individuals providing information about specific rare diseases shows that these persons feel isolated with the disease and that they want to make information about themselves public to get in touch with other people affected by the disorder.

Information about psychosocial counseling and the desire to have children and/or family planning are rarely presented on the websites containing information about rare diseases. Nevertheless, both are important information categories for patients suffering from a rare disease [26,33] and their relatives because 80% of all rare diseases have genetic causes [18]. Genetic questions are in line with questions about family planning and genetic theory. Moreover, because of the low number of affected persons and the feeling of being overstrained with the situation of being the only person suffering from this specific disease, psychosocial counseling constitutes an important role for all patients. For this, support groups and patient organizations already provide most of the available information in the categories of information exchange with other patients, assistance for self-help, family planning, personal advice, psychosocial counseling, and social-legal advice. Nevertheless, information and knowledge about psychosocial counseling and family planning in the field of rare diseases need to be extended.

Interestingly, there were no statistically significant differences identified between the quality of information provided by support groups/patient organizations and medical institutions. Only the quality of information provided by other associations and sponsoring bodies showed statistically better results than information provided by self-help group/patient organizations and medical institutions. Overall, cooperation and information transfer between all supplier categories can help to improve information quality and information access for patients suffering from rare diseases, their relatives, and medical professionals. Especially for rare diseases, cooperation activities can improve evidence-based clinical and health care research.

Future research on the quality of information about rare diseases must be considered in a more international context. Especially for ultrarare diseases, for which limited information is available and only a few people worldwide are affected, an international and intercontinental research context is indispensable.

Limitations

This evaluation of quality of information on the Internet about rare diseases is based on information websites available in the German language and/or hosted in Germany, Austria, and Switzerland. Information available on social media accounts were not included in the analysis [34]. The quality criteria cannot verify the actual medical content of health information. These criteria simply verify the factors influencing good thematic content, as well as the quality of the website itself. An evaluation of the quality of information about specific disease groups (eg, rare skin diseases) is not feasible due to the ambiguous classification of rare diseases provided by Orphanet.

Conclusions

The quality of information on the Internet about rare diseases was assessed based on 13 quality criteria for websites providing medical information about rare diseases. Overall, the quality of information on the Internet about rare diseases is insufficient, quality certificates are rarely used, and important quality criteria are often not fulfilled. Subgroup analyses have shown that information provided by support groups and patient organizations are as reliable as information provided by medical institutions. Additionally, there are some information categories that are underrepresented (eg, information about psychosocial counseling, social-legal advice, and family planning). These information categories need to be strongly addressed in future research on information on websites. Nevertheless, this study has shown that support groups are extremely important for patients suffering from a rare disease and their relatives.

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Conflicts of Interest

None declared.

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Abbreviations

ACHSE eV: German Alliance of Chronic Rare Diseases
afgis: German Action Forum Health Information System
HONcode: Health On the Net Foundation Code of Conduct

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Zentrales Informationsportal über seltene Erkrankungen – Umsetzung eines qualitäts- und bedarfsorientierten Informationsmanagements

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Zentrales Informationsportal über seltene Erkrankungen

Umsetzung eines qualitäts- und bedarfsorientierten Informationsmanagements

Einleitung/Hintergrund

In den vergangenen Jahren sind Menschen mit seltenen Erkrankungen und ihre gesundheitliche Versorgung zunehmend in den Blickpunkt von Politik und Öffentlichkeit gerückt. Ein vom Bundesministerium für Gesundheit (BMG) in Auftrag gegebener Forschungsbericht aus dem Jahr 2009 beleuchtete erstmals die Versorgungssituation von Betroffenen aus unterschiedlichen Perspektiven und identifizierte zahlreiche Lösungsszenarien zur Verbesserung ihrer gesundheitlichen Situation [1]. Obwohl unter dem Begriff „seltene Erkrankungen" sehr heterogene Krankheitsbilder zusammengefasst sind, sind die Betroffenen häufig mit ähnlichen Herausforderungen konfrontiert. So handelt es sich meist um genetisch bedingte und komplexe Erkrankungen, die sich oftmals bereits im Kindesalter manifestieren, aufgrund mangelnder Erfahrungen mit seltenen Erkrankungen von Ärztinnen und Ärzten jedoch häufig erst verzögert diagnostiziert werden. Auch wenn eine korrekte Diagnose gestellt werden kann, so sind Informationen über Therapiemöglichkeiten schwer auffindbar und spezialisierte Ansprechpartner regional ungleich verteilt, sodass eine optimale Versorgung erschwert ist.

Um die gesundheitliche Situation der Betroffenen langfristig zu verbessern, wurden die ermittelten Versorgungsdefizite im Rahmen des „Nationalen Aktionsplans für Menschen mit Seltenen Erkrankungen" in konkrete Maßnahmenvorschläge überführt [2]. Ein zentrales Handlungsfeld dieses Aktionsplans ist das Informationsmanagement im Bereich seltener Erkrankungen. Um die häufig weit verstreuten Informationen im Internet zu bündeln, wurde im Rahmen des Projekts „Zentrales Informationsportal über seltene Erkrankungen (ZIPSE)" die Konzeption und Implementierung eines Portals realisiert, über das Patientinnen und Patienten, ihre Angehörigen sowie medizinische, therapeutische und pflegerische Leistungserbringer zukünftig auf Informationen zugreifen können. Die Informationsbasis dieses Portals bilden jedoch keine selbsterstellten Informationen, sondern bereits online verfügbare Informationsangebote zu seltenen Erkrankungen.

Um eine hohe Qualität der verlinkten Informationen sicherzustellen, wurden zu Beginn des Projekts Qualitätskriterien erarbeitet [3], die sich vorwiegend an bestehenden Kriterienkatalogen und Checklisten für gute Gesundheitsinformationen orientierten [4–11]. Aufgrund der Vielzahl seltener Erkrankungen und der damit verbundenen Menge an Informationsangeboten im Internet wurden diese jedoch so angepasst, dass nicht die konkreten Inhalte der Informationsseiten, sondern verschiedene Attribute der Webseite als solche überprüft werden, die eine hohe Informationsqualität sicher erscheinen lassen. Gleichzeitig wurde bei der Implementierung des Portals auf eine hohe zielgruppenspezifische Ausrichtung des Informationsangebots geachtet. Im Laufe des Projekts haben sich Hinweise darauf ergeben, dass zu bestimmten seltenen Erkrankungen nur eine geringe Informationsbasis vorliegt und diese für die Betroffenen, ungeachtet der Einhaltung von Qualitäts- bzw. Webseitenstandards, von Bedeutung sein kann [12]. Ziel dieser Arbeit ist es daher, zu untersuchen, welche Anforderungen Patientinnen und Patienten sowie Angehörige an ein onlinebasiertes Informationsangebot stellen und ob nach der Anwendung der erarbeiteten Qualitätskriterien eine auf den Patientinnen- und Patienten- sowie Angehörigenbedarfen ausgerichtete Informationsbasis zu realisieren ist. Ggf. werden Maßnahmen abgeleitet, die ein gleichermaßen qualitäts- wie bedarfsorientiertes Informationsmanagement sicherstellen.

Methoden

Für die Überprüfung der Fragestellung kommt ein Mixed-Methods-Ansatz aus quantitativen und qualitativen Methoden zum Einsatz. Um zu überprüfen, welche Bedarfe Patientinnen und Patienten sowie ihre Angehörigen an ein webbasiertes Informationsangebot stellen und welche Informationsseiten aus ihrer Sicht

Published online: 07 March 2017

Tab. 1 ZIPSE Qualitätskriterien

Verpflichtende Kriterien	Erstellungs- und Aktualisierungsdatum
	Datenschutz
	Impressum
	Kontakt
Optionale Kriterien	Erstellungsprozess
	Autoren
	Quellen
	Evidenz
	Interessenkonflikte
	Zielgruppen
	Evaluation der Inhalte
	Review-Verfahren
	Barrierefreiheit

von Bedeutung sind, wurden qualitative leitfadengestützte Interviews durchgeführt. Diese sind durch ihr hohes Maß an Offenheit und Flexibilität dazu geeignet, die Wirklichkeit aus Sicht der Interviewpartner abzubilden [13]. Hierzu wurde ein thematischer Leitfaden entwickelt, der u. a. eine Erzählung über Erfahrungen bei der Suche nach krankheitsbezogenen Informationen sowie deren Bewertung initiieren sollte. Die Probandinnen und Probanden wurden demzufolge nicht direkt danach gefragt, was aus ihrer Sicht eine qualitativ hochwertige Informationsseite ausmacht; dies hätte eine Überforderung der Interviewteilnehmer bedeuten können [14]. Vielmehr sollten ihre Einstellungen hierzu indirekt aus ihren Erzählungen und Erfahrungen abgeleitet werden. Der Leitfaden wurde nach einer ersten Testung mit Patientinnen und Patienten sowie einem Angehörigen optimiert und erwies sich im Folgenden als geeignet, um die Betroffenen zum Erzählen über ihre Erfahrungen anzuregen.

Die Rekrutierung der Interviewpartnerinnen und -partner erfolgte über die Hautklinik des Universitätsklinikums Freiburg. Um ein möglichst heterogenes Sample zu erreichen, wurden zu Beginn des Projekts von medizinischen Expertinnen und Experten im Bereich seltener Erkrankungen elf Erkrankungsgruppen festgelegt, welche die Gesamtheit der seltenen Erkrankungen möglichst umfassend abbilden. Geplant waren sechs Interviews mit Betroffenen aus jeder dieser Gruppen, zuzüglich zehn Interviews mit Personen, bei denen die Diagnosestellung mindestens zehn Jahre dauerte. Hätte sich im Laufe der Datenerhebung gezeigt, dass keine neuen Erkenntnisse mehr aus den Interviews zu gewinnen seien, wäre ein vorzeitiger Abbruch möglich gewesen. Sämtliche Interviews wurden auf Tonband aufgezeichnet und anschließend wörtlich transkribiert.

Angelehnt an die inhaltlich strukturierende Inhaltsanalyse nach Philipp Mayring wurden die Transkripte anschließend ausgewertet [15]. Hierzu arbeiteten zwei Forscherinnen die ersten drei Transkripte zunächst unabhängig voneinander durch, markierten alle für die Fragestellung relevanten Textpassagen und entwickelten darauf aufbauend ein Kategoriensystem. Mittels dieses Kategoriensystems wurden die weiteren Interviews kodiert. Bei Bedarf wurde das System modifiziert bzw. um weitere Kategorien ergänzt, sofern sich diese aus den Interviews heraus ergaben. Die kodierten Textstellen wurden anschließend im Hinblick auf die Forschungsfrage ausgewertet, wobei Überschneidungen wie auch Unterschiede in den Interviews aufgedeckt werden konnten. Die Analyse erfolgte unter Zuhilfenahme des Programms MAXQDA.

In einem zweiten Schritt erfolgte eine deskriptive Auswertung der identifizierten und bereits im Portal registrierten Informationsseiten zu seltenen Erkrankungen (Stand: 23.09.2016) hinsichtlich der erarbeiteten Qualitätskriterien. Auf diese Weise konnte festgestellt werden, wie viele Seiten den gesetzten Qualitätskriterien entsprechen und welche Informationsbasis sich daraus für das Informationsportal (ZIPSE) ergibt. Die Identifizierung von Webseiten erfolgte über eine systematische Internetrecherche in gängigen Suchmaschinen, bei der nach sämtlichen seltenen Erkrankungen und deren Synonymen gemäß dem Orphanet-Verzeichnis gesucht wurde [16]. Hierbei wurden die jeweils ersten zwanzig Treffer gescreent und in eine Datenbank aufgenommen, wenn sie Informationen über seltene Erkrankungen enthielten. Anschließend erfolgte die Registrierung der Seiten im Portal sowie deren Qualitätsprüfung, im Zuge derer die Informationsseiten hinsichtlich der erarbeiteten Qualitätskriterien überprüft wurden.

Diese 13 Kriterien gliedern sich auf in vier Kriterien, deren Erfüllung zur Verlinkung im Portal zwingend erfüllt sein muss, sowie neun optionale Kriterien (**Tab. 1**). Zu den verpflichtenden Kriterien zählen neben der gesetzlich vorgeschriebenen Einhaltung der Impressumspflicht sowie der Angabe von Informationen zur Handhabung des Datenschutzes auch die Angabe eines Erstellungs- oder Aktualisierungsdatums der dargestellten Informationen und die Möglichkeit, mit dem Betreiber der Webseite in Kontakt zu treten. Unter den optionalen Kriterien sind Angaben zum Erstellungsprozess, den Autorinnen und Autoren sowie den verwendeten Quellen gefasst. Ebenso sind Angaben zur Evidenzgrundlage erwünscht. Darüber hinaus wird geprüft, ob Interessenkonflikte bestehen und benannt werden, ob ersichtlich ist, welche Zielgruppen angesprochen werden und ob die Inhalte der Webseite regelmäßig evaluiert werden. Ferner werden ein internes Reviewverfahren sowie Merkmale zur Barrierefreiheit überprüft. Die Überprüfung der optionalen Kriterien wurde für die vorliegende Untersuchung ausgeschlossen, da sie keine Auswirkungen auf die zur Verfügung stehende Informationsbasis haben. Aufbauend auf der Untersuchung der verpflichtenden Qualitätskriterien sind Aussagen darüber möglich, wie viele Seiten in ZIPSE verlinkt werden können und welche Seiten nach diesem Verfahren auszuschließen sind. Insbesondere wird analysiert, Seiten welcher Anbieter hierunter fallen und welche Kriterien zum Ausschluss führen.

Ein Abgleich beider Ergebnisse ermöglicht Aussagen darüber, ob die auf ZIPSE verfügbare qualitätsgesicherte Informationsbasis ausreichend ist, um den Bedarfen der Patientinnen und Patienten sowie ihrer Angehörigen gerecht zu werden. Gegenfalls sind Maßnahmen zu entwickeln, die ein gleichermaßen qualitäts- wie patientenorientiertes Informationsmanagement sicherstellen.

Zusammenfassung · Abstract

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Zentrales Informationsportal über seltene Erkrankungen. Umsetzung eines qualitäts- und bedarfsorientierten Informationsmanagements

Zusammenfassung

Hintergrund. Ein zentrales Informationsportal über seltene Erkrankungen wurde etabliert, über das u. a. Betroffene und Angehörige auf hochwertige Informationen zugreifen können. Hierfür wurden Qualitätskriterien entwickelt, die speziell auf Seiten über seltene Erkrankungen ausgerichtet sind. Gleichzeitig soll das Informationsangebot die Bedarfe von Betroffenen berücksichtigen.
Ziel der Arbeit. Zunächst werden die Bedarfe von Patienten und Angehörigen an ein onlinebasiertes Informationsangebot eruiert. Darauf aufbauend wird geprüft, inwiefern die gemäß den Qualitätskriterien verfügbare Informationsbasis diese Bedarfe decken kann. Ggf. sind Maßnahmen zu entwickeln, die ein ebenso qualitäts- wie bedarfsorientiertes Informationsmanagement sicherstellen.
Material und Methoden. Zunächst finden qualitative Interviews mit Patienten und Angehörigen statt, die inhaltsanalytisch ausgewertet werden. Anschließend erfolgt eine quantitative Auswertung von identifizierten Informationsseiten zu seltenen Erkrankungen. Untersucht wird, wie viele Seiten die Qualitätskriterien nicht erfüllen, von welchen Anbietern diese stammen und welche Kriterien nicht erfüllt sind. Es folgt ein Abgleich der quantitativen und qualitativen Ergebnisse.
Ergebnisse und Diskussion. Bei der Suche nach Informationen im Internet stellen Webseiten von Selbsthilfegruppen eine bedeutsame Quelle dar. Diese werden als sehr vertrauenswürdig wahrgenommen und bieten im späteren Erkrankungsverlauf tiefgehende Information zu relevanten Informationsbereichen. Häufig entsprechen diese jedoch nicht den ZIPSE-Qualitätsanforderungen. Daher wird eine transparente Darstellung der Qualität der ZIPSE-Informationsseiten gewählt. Nicht qualitätsgesicherte Seiten können nun aktiv angefordert werden, sind jedoch deutlich gekennzeichnet.

Schlüsselwörter

ZIPSE Informationsportal · Seltene Erkrankungen · Qualitätskriterien · Informationsmanagement · Mixed Methods

Central information portal on rare diseases. Implementation of quality- and needs-oriented information management

Abstract

Background. A central information portal on rare diseases (ZIPSE) has been conceptualized and implemented that allows patients, relatives and health care professionals to access quality-assured information. For this purpose, quality criteria have been developed specifically for rare diseases. At the same time, the information basis should take into account the specific needs of those interested.
Objectives. The needs of patients and relatives regarding online-based information are analyzed. Based on this, we examined to what extent the information basis, which is available according to the ZIPSE quality criteria, can cover these needs. If necessary, measures have to be developed to ensure quality- as well as needs-oriented information management.
Materials and methods. Qualitative interviews with patients and relatives were conducted, which were then evaluated using content analysis. Subsequently, a quantitative evaluation of the information on rare diseases in the portal was made. The research addresses how many websites do not fulfil the quality criteria, from which group of provider these websites originate and which criteria are not fulfilled. This is followed by a comparison of the quantitative and qualitative results.
Results and conclusions. When looking for information on the Internet, the websites of self-help groups represent a significant source. These are perceived as very trustworthy and in the later course of the disease, offer detailed information on important information areas. Information websites from self-help groups, however, often do not meet quality requirements. Therefore, a transparent representation is made regarding the quality of the ZIPSE information pages. Pages that are not quality-assured can be actively requested, but will be clearly identified.

Keywords

ZIPSE information portal · Rare diseases · Quality criteria · Information management · Mixed methods

Ergebnisse

Bedarfe an ein webbasiertes Informationsangebot aus Sicht von Patientinnen und Patienten sowie Angehörigen

Insgesamt 55 Patientinnen und Patienten sowie 13 Angehörige nahmen an den Interviews teil (Tab. 2). Dabei waren etwa doppelt so viele Frauen wie Männer im Sample vertreten (45 Frauen gegenüber 23 Männern). Das Durchschnittsalter der Befragten betrug zum Untersuchungszeitpunkt 50,5 Jahre. Aus allen im Vorfeld festgelegten Erkrankungsgruppen konnten Patientinnen und Patienten sowie Angehörige rekrutiert werden. Aufgrund eines erschwerten Zugangs zu einigen Patientengruppen wurde die angestrebte Zahl von sechs Personen je Erkrankungsgruppe jedoch nicht erreicht. Hierbei handelt es sich um die Gruppen „genetische Erkrankungen des Verdauungstrakts“, „Bindegewebserkrankungen“, „angeborene Störungen der Blutbildung“ sowie „genetische Erkrankungen des Auges“. Da im späteren Verlauf der Interviewdurchführung jedoch beobachtet werden konnte, dass weitere Interviews keine neuen Erkenntnisse mehr generieren, wurde die Rekrutierung weiterer Teilnehmerinnen und Teilnehmern eingestellt.

Insgesamt zeigen die Ergebnisse eine hohe Bedeutung von Internetinformatio-

Tab. 2 Patienten- und Angehörigencharakteristika

Variable	Gesamt ($n = 68$)	Prozent
Durchschnittsalter (in Jahren)	50,5	–
Geschlecht (weiblich/männlich)	45/23	66,2/33,8
Status (Patient/Angehöriger)	55/13	80,9/19,1
Erkrankung		
Genetische Erkrankungen der Haut	10	14,7
Skelettdysplasien	7	10,3
Neuromuskuläre Erkrankungen	9	13,2
Genetische Erkrankungen der Augen	4	5,9
Bindegewebserkrankungen	5	7,4
Genetische Erkrankungen der Nieren	6	8,8
Mukoviszidose und verwandte Lungenerkrankungen	7	10,3
Immundefizienzen	7	10,3
Angeborene Stoffwechselstörungen	7	10,3
Angeborene Störungen der Blutbildung	4	5,9
Genetische Erkrankungen des Verdauungstrakts	2	2,9

nen für die Betroffenen (Tab. 3). Die Befragten gaben an, insbesondere im Zeitraum kurz nach der Diagnosestellung, wenn der Bedarf nach Information besonders hoch sei, das Internet für ihre Suche nach Informationen genutzt zu haben. Die Erfahrungen, die die Patientinnen und Patienten in diesem Zusammenhang schildern, stellen sich jedoch sehr heterogen dar. Trotz der hohen Nutzung des Internets gaben nur wenige der Befragten an, mit den Ergebnissen der Onlinerecherche zufrieden gewesen zu sein. Einige berichteten, dass die Suche im Netz zu Beginn aufgrund der zum Teil angstauslösenden und schockierenden Fotos und Berichte ein traumatisches Erlebnis gewesen sei, das sie dazu veranlasst habe, nicht weiter online zu recherchieren. Dies wurde insbesondere im Zusammenhang mit Informationen zu schweren Erkrankungsverläufen und Lebenserwartung berichtet.

Andere berichteten von Unzufriedenheit im Zusammenhang mit der Menge an gefundenen Informationen. Einige Betroffene fühlten sich mit einer Flut an Informationen konfrontiert, die als Laie nur schwer zu bewältigen bzw. einzuordnen sei. In diesem Zusammenhang wurde die Möglichkeit eines Portals, auf dem Informationen gebündelt und übersichtlich dargestellt werden, positiv hervorgehoben. Andere kritisierten, dass zu manchen seltenen Erkrankungen gar keine oder kaum Informationen zu finden seien. Letzteres ist insbesondere vor dem Hintergrund von Bedeutung, dass Befragte mit sehr seltenen Erkrankungen angaben, dass zu Beginn jede Information, die man erhalten könne, von großer Wichtigkeit sei.

Zu einem späteren Zeitpunkt im Krankheitsverlauf äußerten die Teilnehmerinnen und Teilnehmer hingegen insbesondere Unzufriedenheit bezüglich der Tiefe der Informationen. Während kurz nach Diagnosestellung übersichtliche Informationen bevorzugt wurden, die einen Überblick über die Erkrankung bieten, so waren diese im späteren Verlauf für die Teilnehmer nicht mehr ausreichend, um ihre spezifischeren Informationsbedarfe, beispielsweise zu sozial- und leistungsrechtlichen Aspekten oder therapeutischen Möglichkeiten, zu decken. In diesem Zusammenhang wiesen die Interviewten auf Informationsseiten von Selbsthilfegruppen hin, die im Gegensatz zu Informationen z. B. in Enzyklopädien diese Informationstiefe bedienen könnten.

Die Bewertung der online gefundenen Informationen für die Betroffenen und ihre Angehörigen leitet sich teilweise daraus ab, welcher Anbieter für die Informationsseite zuständig ist. Da die Interviewten kurz nach Diagnosestellung kaum über Wissen zu ihrer Erkrankung bzw. der ihres Angehörigen verfügen, fehlen ihnen die Kompetenzen, die Informationen inhaltlich zu überprüfen. Eine qualitative Bewertung der recherchierten Informationen findet daher sehr selten statt. Vielmehr erfolgt die Bewertung auf Grundlage dessen, welche Personen oder Institutionen für die Seite und deren Inhalte verantwortlich sind. In diesem Zusammenhang beschrieben die Interviewteilnehmer insbesondere Webseiten von Selbsthilfegruppen als relevante Informationsquellen. Diese werden als besonders vertrauenswürdig erachtet, da sich die Interviewteilnehmerinnen und -teilnehmer mit den betroffenen Mitgliedern identifizieren können und ihnen eine besonders hohe Erkrankungskompetenz zugesprochen wird. Dies könnte auch darin begründet liegen, dass viele der Interviewpartnerinnen und -partner bis zu diesem Zeitpunkt keinen Ansprechpartner im Versorgungssystem gefunden hatten, der ihnen Informationen zu ihren Bedarfen bereitstellen konnte. So berichteten viele, dass sie einzig über Selbsthilfegruppen bedeutsame Informationen erhalten hätten und diese insbesondere im späteren Erkrankungsverlauf, wenn sich nur noch vereinzelt spezifische neue Informationsbedarfe ergäben, häufig die einzige Informationsquelle darstellten.

Des Weiteren bewerteten die Befragten die gefundenen Informationen danach, inwiefern diese ihre jeweils aktuellen Informationsbedarfe decken. Für neudiagnostizierte Patientinnen und Patienten sowie deren Angehörigen sind dies vor allem grundlegende Informationen, die ein Verständnis des Krankheitsbilds ermöglichen und Einblick in mögliche therapeutische Maßnahmen geben. Im weiteren Erkrankungsverlauf werden diese medizinischen Fragestellungen zunehmend spezifischer; hinzu kommen u. a. psychosoziale und leistungsrechtliche sowie alltagsrelevante Fragen. So gaben die Befragten u. a. an, Hilfe im Umgang mit ihrer Erkrankung im Alltag zu bedürfen. In diesem Zusammenhang wurden ebenfalls die Informationsseiten von Selbsthilfegruppen besonders positiv erwähnt. Während andere Webseiten häufig nur rein medizinische Aspekte der Erkrankung abdeckten, böten diese darüber hinausgehende Informationen zu alltagsrelevanten Themen.

Tab. 3 Belege aus den qualitativen Interviews mit Patientinnen, Patienten und Angehörigen

Aspekt	Zitat
Das Internet stellt für die Suche nach Informationen zu seltenen Erkrankungen eine bedeutsame Quelle dar	*I: „Es gibt ja heute relativ viele Möglichkeiten, die man zur Kommunikation verwenden kann. Was für Medien haben Sie genutzt bei der Suche nach Informationen?"* *P: „Google. Hauptsächlich den PC, Suchmaschinen. Heutzutage ist das glaube ich Standard."* (Patient, 32 Jahre, Interview 2)
Angstauslösende Informationen im Internet	*„Also ich war nur auf Wikipedia. Und was ich da gelesen hatte, das hatte mich halt geschockt, weil sich das ganz arg schlimm anhörte. Und danach war ich nie wieder im Internet."* (Angehöriger, 46 Jahre, Interview 4)
Bedarf nach detaillierten Informationen wird oft nicht gedeckt	*„Man findet immer so Informationshäppchen. Und das Schlimme ist, dass dann ganz viele Informationshäppchen im Internet sind, die dann ganz kurz die Krankheit vielleicht erläutern, (…) aber keine tiefen Informationen geben."* (Patient, 48 Jahre, Interview 60)
Nach Diagnosestellung sind sämtliche Informationen relevant	*„Es waren/als alles neu war. Da haben wir ja jede Information aufgesogen."* (Patient, 59 Jahre, Interview 47)
Tiefgehende Informationen über Internetseite von Selbsthilfegruppe	*I: „Und wie beurteilen Sie die Qualität der Informationen?"* *P: „Das ist wiederum gut, also nur das von der Selbsthilfegruppe, sonst nichts."* *I: „Und die Informationen, die Sie jetzt im Internet auf anderen Seiten gefunden haben?"* *P: „Ne. War für mich so allgemein, nichtssagend."* (Patientin, 50 Jahre, Interview 10)
Beurteilung von Webseiten erfolgt danach, wer für Informationen verantwortlich ist	*„(…) Und dann entscheidet man ja so ein bisschen nach dem Auftritt auch, was ist seriös, ja, wer steckt dahinter, (…) sind die ähhh, hier mmhhh Allianz der chronischen/ also, der ACHSE zugehörig, NAMSE zugehörig, ja."* (Patientin, 44 Jahre, Interview 35)
Bedeutsamkeit von Informationen anderer Betroffener	*„Oder wenn man/jemand der wirklich beschreibt, seine eigene Krankheit beschreibt. Man kann diese Parallelen sehen."* (Patient, 59 Jahre, Interview 47)
Internetseite von Selbsthilfegruppe bietet Informationen zu für die Betroffenen wichtigen Fragestellungen	*„Ja, ich kann immer nur wieder auf das MPN-Netzwerk zurückkommen. Weil es eigentlich so ziemlich mit die einzige wirklich ergiebige Quelle ist, sage ich jetzt mal. Wo man wirklich alle Sachen findet, die man wissen will."* (Patientin, 27 Jahre, Interview 1)

Informationsbasis gemäß ZIPSE-Qualitätskriterien

Die darauf folgende Untersuchung zu Qualität und Umfang der ZIPSE-Informationsbasis ergab, dass zum Untersuchungszeitpunkt (Stand: 23.09.2016) 664 Informationsseiten zu seltenen Erkrankungen im ZIPSE registriert waren, die im Folgenden einer Überprüfung unterzogen wurden. 339 dieser Seiten erfüllten die verpflichtenden Qualitätskriterien, wohingegen die Überprüfung bei 325 Informationsseiten negativ ausfiel. Dies bedeutet, dass nahezu die Hälfte (ca. 49 %) der identifizierten und im Internet verfügbaren Informationswebseiten mindestens eines der verpflichtenden ZIPSE Qualitätskriterien nicht erfüllt und somit grundsätzlich von einer Verlinkung im Portal auszuschließen wäre.

Eine genauere Auswertung der 325 Seiten, die den verpflichtenden ZIPSE-Kriterien nicht entsprechen, ergab, dass dies vorwiegend darauf zurückzuführen ist, dass kein Erstellungs- und Aktualisierungsdatum von Informationen (n = 210) angegeben wird oder keine ausreichenden Hinweise zum Datenschutz gem. § 13 TMG auf der Webseite zu finden sind (n = 206). Darüber hinaus fehlt zum Teil ein Impressum, welches gemäß Telemediengesetz (§ 5 TMG) und Staatsvertrag über Rundfunk und Telemedien (§ 55 RStV) gestaltet ist und seit 2001 verpflichtend ist (n = 51). Dieses gibt Auskunft über Name, Anschrift und Emailadresse des Anbieters sowie ggf. dessen Handels- oder Vereinsregistereintrag. In einigen Fällen ist auf den Webseiten kein von überall aus leicht zu erreichendes Kontaktformular eingerichtet, oder es fehlen Kontaktinformationen, welche dem Nutzer ermöglichen, mit dem Betreiber der Webseite in Kontakt zu treten, Feedback zu geben oder Fragen zu stellen (n = 5). Rund zwei Drittel aller negativ geprüften Webseiten erfüllen zwei oder mehr der vier verpflichtenden Kriterien nicht (63,07 %). Bei 120 Informationsseiten hingegen mangelte es nur an der Erfüllung eines Kriteriums.

Unter den Anbietern von Webseiten, die die verpflichtenden Kriterien nicht erfüllen, stellen Selbsthilfegruppen und Patientenvereinigungen mit mehr als einem Drittel (36,31 %) die größte Anbietergruppe dar. Dahinter folgen medizinische Einrichtungen (30,15 %) und Einzelpersonen in Form von Patientinnen und Patienten oder Angehörigen (10,46 %) sowie medizinischen Expertinnen und Experten (6,77 %). Sonstige Verbände und Trägerschaften (5,85 %) sowie Fachgesellschaften (5,54 %), pharma- oder medizintechnische Unternehmen (2,77 %) und Verlage und Medienunternehmen (1,85 %) sind weniger häufig unter den Anbietern zu finden. Ein Anbieter konnte nicht klassifiziert werden und wurde unter Sonstige vermerkt (**Abb. 1**).

Es kann folglich festgehalten werden, dass Patientinnen und Patienten sowie ihre Angehörigen Bedarfe an die Informationsbasis eines zentralen Informationsportals stellen, welche mit einer rein nach Qualitätsaspekten ausgerichteten Informationsbereitstellung nicht gedeckt werden können. Daher sind Maßnahmen erforderlich, die ein ebenso an der Qualität wie auch den Bedarfen ausgerichtetes Informationsmanagement gewährleisten.

Diskussion

Herausforderungen eines qualitäts- und bedarfsorientierten Informationsmanagements

Die Analyse der im Informationsportal (ZIPSE) registrierten Informationsseiten hat gezeigt, dass ungefähr die Hälf-

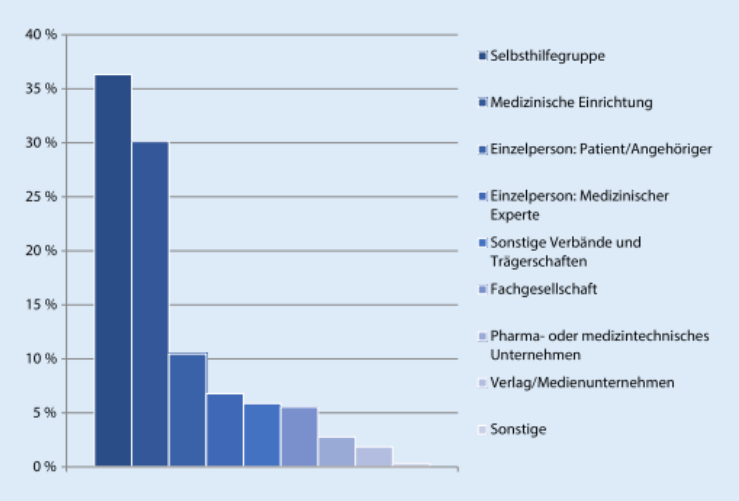

Abb. 1 ▲ Nicht-qualitätsgesicherte Informationsseiten nach Anbietern

te der zuvor bei einer umfassenden Recherche identifizierten Webseiten einer Anwendung der ZIPSE-Qualitätskriterien nicht standhält. Die den Nutzerinnen und Nutzern bereitgestellte Informationsbasis wäre somit der bei einer sonstigen Internetsuche gefundenen Informationsbasis um rund 50 % verringert. Insbesondere bei sehr seltenen Erkrankungen, zu denen häufig nur wenige Informationen verfügbar sind, kann dies die Zufriedenheit der Nutzerinnen und Nutzer eines solchen Portals einschränken. Die Auswertung der Interviews hat gezeigt, dass aufgrund der mangelhaften Informationsgrundlage besonders im Zeitraum kurz nach der Diagnosestellung, wenn die Informationsbedarfe sehr hoch sind, Informationen ungeachtet ihrer objektiven Qualität für die Betroffenen sehr bedeutsam sein können. Eine strikte Ausklammerung von Informationen, die festgelegten Qualitätskriterien nicht entsprechen, kann daher den Nutzen der Verwendung des Informationsportals (ZIPSE) für die Betroffenen und ihre Angehörigen vermindern.

Ein Großteil der Befragten machte keine konkreten Angaben dazu, was aus ihrer Sicht ein qualitativ hochwertiges Informationsangebot ausmache. Die Interviewteilnehmerinnen und -teilnehmer gaben jedoch an, welche Informationsseiten bei ihrer Suche von Bedeutung waren. Hierbei handelte es sich häufig um Webseiten von Selbsthilfegruppen, denen ein hohes Maß an Vertrauen entgegengebracht wird und die zu für die Betroffenen wichtigen Fragestellungen im Krankheitsverlauf tiefgehende Informationen bereithalten. Vor dem Hintergrund dieser Ergebnisse stellt sich u. a. die Frage, welche Bedeutung Betroffene seltener Erkrankungen, die im Hinblick auf die Informationssituation mit besonderen Gegebenheiten konfrontiert sind, etablierten Qualitätskriterien für Gesundheitsinformation beimessen. Es ist davon auszugehen, dass Menschen mit seltenen Erkrankungen aufgrund der erschwerten Informationssituation andere Anforderungen an Informationen stellen als Betroffene häufigerer Erkrankungen, zu denen eine höhere Informationsbasis vorliegt [17]. Diese Frage sollte in weiterführenden Studien näher untersucht werden.

Unter den in unserer Untersuchung überprüften Webseiten waren es häufig die Webseiten von Selbsthilfegruppen, die den ZIPSE-Qualitätskriterien nicht entsprechen und somit der Informationsbasis des Informationsportals (ZIPSE) entzogen würden. Eine mögliche Ursache hierfür ist, dass es sich bei Selbsthilfegruppen häufig um einen Zusammenschluss einiger weniger engagierter Betroffener oder Angehöriger handelt, die zwar über ein großes Wissen zu seltenen Erkrankungen verfügen, jedoch wenig Erfahrung in der zielgruppenspezifischen Aufbereitung ihrer Informationen haben. Darüber hinaus kann davon ausgegangen werden, dass Selbsthilfegruppen in der Regel nur über ein beschränktes Budget verfügen, welches eine professionelle Umsetzung von Webseiteninhalten erschweren kann. Dieses wurde bislang jedoch nicht wissenschaftlich untersucht. Unklar ist daher, inwieweit bestehende Leitfäden und Kriterienkataloge zu guten Gesundheitsinformationen bei in der Selbsthilfe tätigen Personen bekannt sind bzw. welche Gründe für eine Nichtumsetzung dieser Kriterien bestehen. Dies sollte in weiterführenden Studien untersucht werden.

Transparente Darstellung der Qualität verlinkter Informationsangebote

Um der Herausforderung bei der Betreibung des ZIPSE zu begegnen, einerseits Informationsseiten zu verlinken, die den ZIPSE-Qualitätskriterien entsprechen und andererseits Webseiten zu berücksichtigen, die für Suchende trotz nicht erfüllter Qualitätskriterien von Bedeutung sein können, wird ein qualitäts- und gleichzeitig bedarfsorientiertes Vorgehen gewählt. Nach Eingabe des Suchbegriffs in das Suchfeld erscheinen zunächst sämtliche qualitätsgesicherten erkrankungsspezifischen Informationsseiten in einer Listenansicht (**Abb. 2**). Zu diesen Webseiten können sich die Suchenden detaillierte Infos, wie z. B. eine Beschreibung des Informationsangebots und der Themenbereiche, anzeigen lassen oder direkt zum Informationsangebot wechseln. Unterhalb dieser Liste finden Nutzerinnen und Nutzer den Hinweis, sich weitere Seiten, die den ZIPSE-Qualitätskriterien nicht entsprechen, jedoch trotzdem inhaltlich hochwertig und bedeutsam sein können, nachgelagert anzeigen lassen zu können. Klicken die Suchenden dieses Angebot an, erscheint zunächst ein Hinweistext, den die Nutzerinnen und Nutzer aktiv bestätigen müssen. In diesem Hinweistext wird darauf hingewiesen, dass eine inhaltliche Überprüfung sämtlicher In-

Abb. 2 ▲ Darstellung der qualitätsgesicherten Suchergebnisse

formationen der im Portal verlinkten Webseiten nicht möglich ist und sich die Qualitätskriterien daher vorwiegend auf Merkmale, die die Qualität der Webseite sicherstellen sollen (z. B. Datenschutzkonzept, vollständiges Impressum etc.), beziehen. Gleichzeitig wird betont, dass die nachfolgend ergänzten Informationen somit inhaltlich eine hohe Qualität besitzen können, dies aber nicht durch die Redaktion überprüfbar sei. Dem Suchenden wird somit transparent dargestellt, welches Informationsangebot die ZIPSE-Qualitätsanforderungen erfüllt, und es wird die Möglichkeit eröffnet, selbst zu entscheiden, sich weitere, möglicherweise bedeutsame Informationen anzeigen zu lassen. In der Liste finden die Suchenden dann weitere Seiten, die optisch über ein Ausrufezeichen deutlich von den qualitätsgesicherten Seiten abgegrenzt sind (**Abb. 3**). Betreiber von registrierten Webseiten, welche die Qualitätskriterien nicht erfüllen, werden durch dieses Vorgehen außerdem dazu angeregt, ihre Informationsseite nachzurüsten, um zukünftig im sofort sichtbaren Bereich gelistet zu werden.

Das Informationsportal ZIPSE verfügt über weitere Schnittstellen zu bereits existierenden Datenbanken. Zusätzlich zu den im Informationsportal gelisteten Informationsseiten werden den Nutzerinnen und Nutzern des Informationsportals erkrankungsspezifische Informationen aus den Datenbanken von Orphanet, FindZebra und ACHSE e. V. angezeigt. Des Weiteren erfolgen eine erkrankungsspezifische Anzeige von Expertinnen und Experten sowie Selbsthilfegruppen, die im Versorgungsatlas über seltene Erkrankungen (se-atlas) gelistet sind, sowie eine Verlinkung zu wissenschaftlichen Publikationen aus der PubMed-Datenbank. Das Informationsportal ZIPSE fungiert somit als Metaplattform und als zentraler Zugangspunkt zu verfügbaren Informationen über seltene Erkrankungen im Internet.

Eine wichtige Limitation unserer Untersuchung ist, dass die Interviewteilnehmerinnen und -teilnehmer nicht direkt danach gefragt wurden, was aus ihrer Sicht Qualität im Hinblick auf Informationen zu ihrer Erkrankung bedeutet, was das In-Beziehung-Setzen der quantitativen und qualitativen Auswertungen erschwert. Hiervon wurde jedoch bewusst abgesehen, da belegt ist, dass zu direkte Erzählaufforderungen zu Interviewende überfordern können und eine Übersetzung der Forschungsfrage in eine indirekte Abfrage zielführender ist [14]. Daher wurden die Einstellungen der Teilnehmerinnen und Teilnehmer indirekt aus ihren Erzählungen über die Suche nach Informationen und deren Erleben abgeleitet. Als weitere Limitation lässt sich hinzufügen, dass bislang nur deutschsprachige Informationsseiten im Rahmen der quantitativen Auswertung berücksichtigt wurden. Zukünftig sollen auch englischsprachige Seiten im Informationsportal ZIPSE verlinkt werden. Ob sich die Qualität entsprechender Seiten und damit die ZIPSE-Informationsbasis anders darstellt, kann zum jetzigen Zeitpunkt jedoch noch nicht abschließend beurteilt werden. Ein Blick ins europäische Ausland und die USA verrät jedoch, dass entsprechende Bestrebungen zur Verbesserung der Informationsqualität zu seltenen Erkrankungen dort bereits z. T. seit Jahrzehnten zu verzeichnen sind. Etablierte Informationsdienste wie z. B. NORD oder GARD in den USA bieten Informationen zu einer Vielzahl seltener Erkrankungen und basieren in der Regel auf Primärinformationen, an deren

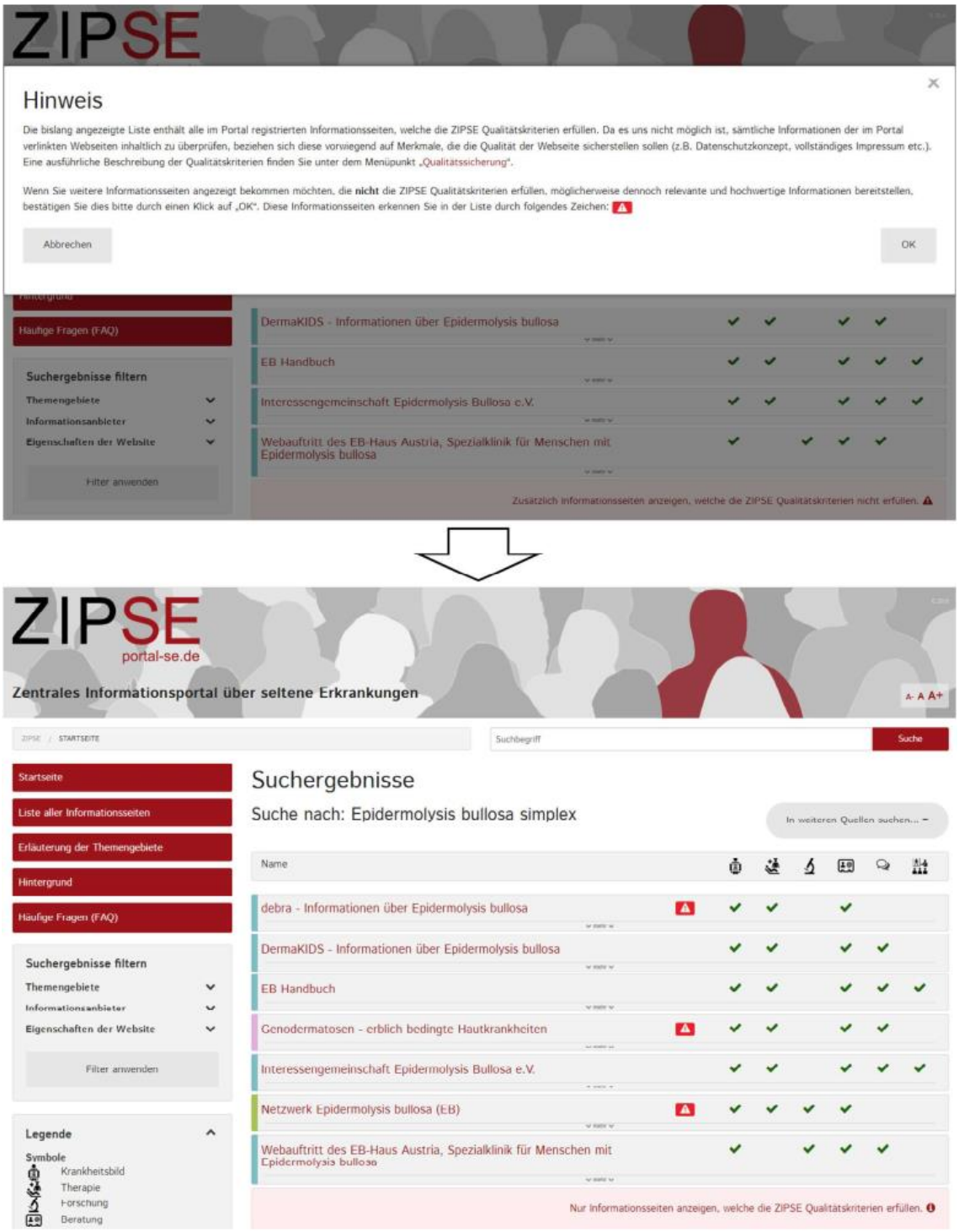

Abb. 3 ▲ Anforderung und Darstellung der nicht-qualitätsgesicherten Suchergebnisse

Erstellung hohe Qualitätsanforderungen angelegt wurden, sodass hier von einer allgemein hohen Informationsqualität ausgegangen werden kann [18, 19].

Fazit

Zusammenfassend lässt sich festhalten, dass Menschen mit seltenen Erkrankungen besondere Bedarfe aufweisen, wenn es um die Suche nach krankheitsbezogenen Informationen geht. Aufgrund der zum Teil mangelhaften Informationsbasis und des erschwerten Zugangs zu Informationen können für die Betroffenen Informationen von Webseiten, die nicht den etablierten Qualitätskriterien für gute Gesundheitsinformationen genügen, dennoch sehr bedeutend sein. So misst sich das Vertrauen in Informationen und somit auch deren Relevanz für Menschen mit seltenen Erkrankungen insbesondere daran, wer die Informationen erstellt hat und ob diese die sich im Verlauf der Erkrankung ändernden Informationsbedarfe ausreichend decken bzw. für den Umgang mit der Erkrankung im Alltag hilfreich sind. Um auf ZIPSE eine qualitätsgesicherte und gleichzeitig an den Bedürfnissen betroffener Nutzerinnen und Nutzer ausgerichtete Informationsbasis zu realisieren, werden daher ein transparenter Umgang mit der Informationsqualität sowie eine aktiv anzufordernde Bereitstellung nicht-qualitätsgesicherter Informationsseiten verfolgt. Auf diese Weise soll sichergestellt werden, dass die Betroffenen auf dem Portal auf eine an ihren Bedarfen ausgerichtete Informationsbasis zugreifen können.

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Einhaltung ethischer Richtlinien

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