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

Svenja Litzkendorf[1], Ana Babac[1], Daniel Rosenfeldt[1], Franziska Schauer[2], Tobias Hartz[3], Verena Lührs[4], J Matthias Graf von der Schulenburg[1] and Martin Frank[1]

[1]Center for Health Economics Research Hannover, Leibniz University of Hannover, Germany

[2]Department of Dermatology, University Medical Center, University of Freiburg, Germany

[3]Institute of Medical Biostatistics, Epidemiology and Informatics, University Medical Center of the Johannes Gutenberg University, Germany

[4]Center for Quality and Management in Healthcare, Medical Association of Lower Saxony, Germany

Corresponding author: Svenja Litzkendorf, Center for Health Economics Research Hannover, Otto-Brenner-Straße 1, 30159 Hannover, Leibniz University of Hannover, Germany, Tel: 4951176214241; E-mail: sl@cherh.de

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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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