

## Facing ignorance: people with rare disorders and their experiences with public health and welfare services

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This qualitative study shows that people with rare disorders may experience barriers that can be related to the fact that the disorder is labelled 'rare' when they access health and welfare services. Service-providers within a range of sectors and services seemed to be reluctant to get involved in situations that imply diagnoses that are unknown to them. Further, they seemed to be reluctant to accept information offered to them by the user and they hesitated to seek up information by themselves. If the professionals got involved they tended to base their judgements on their personal assumptions, consequently followed by incorrect actions. The service-providers' negative responses represent a significant barrier for accessing adequate services and assistance for people with rare disorders. These experiences cut across the various disorders in the study and they cut across the various services.

**Keywords:** rare disorders; disability; health and welfare services

### Introduction

Research on rare disorders has attracted increased interest during the past decade. One reason for this is the legal and financial incentives that have been taken in many countries. As a result of these initiatives, knowledge and information about the latest developments in genetics and possible treatment are becoming increasingly frequent in the medical literature and on the Internet (OrphaNews 2010). Although it sometimes can be difficult to distinguish information that has gone through some type of quality assurance from other information on the Internet, information should now be easily accessible for both professionals and the public. Nevertheless, the attention has first and foremost been directed towards medical origin, progress, and possible treatment. We still have a lack of knowledge about how people cope with the rare disorder in their everyday lives (Bo Hansen and Ege 2007). Some studies have described that having a rare disorder relates to difficulties in obtaining a correct diagnosis, difficulties in finding relevant information about the diagnosis, and consequently the feeling of being alone with the problem (Berglund, Mattiasson, and Randers 2010; Gundersen 2011). Having a rare diagnosis has also been related to emotional and physical stress, and sometimes to a feeling of incompetence (Trulsson and Klingberg 2003; Dellve et al. 2006).

Rare disorders are life-threatening or chronically debilitating diseases that are of such low prevalence that special combined efforts are needed to address them. Many

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individuals live with a disorder that is rare. In Norway, a disorder is labelled 'rare' when it affects fewer than one in 10,000 inhabitants, which means fewer than 500 individuals with the condition in the country. It is estimated that about 30,000 Norwegian citizens suffer from one of the 720 different rare disorders that have been identified in Norway (Norwegian Directorate of Health, information provided on request, 15 April 2010).

Despite a certain ambivalence tied to diagnoses (Shakespeare 2006), receiving a diagnosis is considered a key stage in the lives of disabled people, whether positive or negative (Smith and Daughtrey 2000). Getting the correct diagnosis is decisive for access to adequate help and services. When physical or mental affliction and suffering is diagnosed the person may consequently be offered relevant treatment. Further, a diagnosis is essential for access to welfare benefits, assistance and support, as the system can then recognize the person's needs as legitimate, and release a variety of welfare services. Even though rare disorders do not necessarily imply either illness or disability (Nordenfelt 1983; Grönvik 2007), rare disorders in many cases will influence the person's health conditions and daily life in a disabling way. Similar to others who live with impairing health conditions, people with rare disorders can encounter barriers because of their symptoms and because of society's responses to their impairment (Thomas 1999; Priestley, Corker, and Watson 1999; Smith 2009).

### **The Norwegian health and welfare services and people with a rare disorder**

The founding principles of the Norwegian health and welfare system are maintained for all citizens regardless of type of disorder or impairment. The system emphasizes decentralized, locally based services. Decisions are based on professional knowledge, and services are executed by skilled professionals. In spite of high ideals and good quality in the health and welfare services, the services are characterized by ambiguity. Ideologically as well as legally, these services are expected to place a strong emphasis on individually adapted services. They are expected to promote user participation and empowerment, and similar to other European countries, patient education and self-managed treatment has become increasingly common (Santagostino, Gringeri, and Mannucci 2001; Buchbinder 2009; Nasjonalt kompetansesenter for læring og mestring 2010). However, in addition to a strong emphasis on user participation and empowerment, the system also grants the service-providers the power to frame the interaction with the user and the power to decide what should be the underlying objective of the interaction between the service-provider and the service user (Johansen 2005; Tjora 2008).

In spite of a high standard, research points to extensive coordination problems for individuals with chronic conditions connected to accessing multiple services (Lundeby and Tøssebro 2004; Myrvold and Helgesen 2005; Grut et al. 2007). Halvorsen (2008) points out that good quality of the relationship between the person that accesses services and the professional is fundamental in order to obtaining a good result. Whether the professional's actions are regarded as being of high quality depends on a number of aspects; in addition to technical and clinical knowledge and skills, emotional competence and reflective capacity are considered fundamental for receiving high-quality services (Potter, Gordon, and Hamer 2003; Murinson, Agarwal, and Haythornthwaite 2008).

### ***Counselling services on rare disorders***

Follow-up of users with rare conditions by the general services can be challenging because professionals as a rule may lack knowledge of the condition and the appropriate routines the first time they meet at person with a rare condition (Kemper et al. 2006; Berglund, Mattiasson, and Randers 2010). In acknowledgment of this, 16 state-financed National Resource Centres were established in Norway early in the 1990s. These centres do not offer medical treatment and follow-up. Their mandate is to act as a support to the general services in all sectors and on all levels. The centres provide information on specialist knowledge on rare disorders, offer counselling and information services to patients and their families, and to all kind of professionals working at all levels and in all sectors in the health and welfare services. In addition, the Norwegian Directorate of Health maintains a free telephone helpline for rare conditions as well as a website that offers relevant publications. Information, counselling and supervision on rare disorders should thus be easily accessible for all professionals in the health and welfare services.

### **Aim of the present article**

People with rare disorders may require a range of services across and within sectors and settings and they often have to relate to many different professionals (Miller et al. 2009). These relationships confront them with a complexity of structural, procedural, and attitudinal forces with which they have to deal. The present article aims to contribute to the understanding of how people that live with a rare disorder experience their contact with service-providers when they access public health and welfare services, and discusses whether they have certain common experiences in this regard that cut across the various disorders.

### **Materials and methods**

We used a qualitative semi-open approach to elicit the participants' narratives about living with a rare disorder. The interviews followed a semi-structured structure. The topics covered their experiences with health services, welfare and social services, their social network, family life, and their need for support and services. In the interviews, however, we used an open approach to explore the participants' experiences and opinions, opening the interviews by asking them to tell their story. The main point in the interviews was their story from when they first learned that they had a rare condition. During the interviews we had a particular focus on their past and present experiences with public health and welfare services. The experience of having a diagnosis that is labelled 'rare' was addressed in the closing part of the interview structure, but in most interviews it was introduced by the participant as a topic early in the interview.

Participants chose the time and place for the interview, which in most cases took place in their homes. The interviews lasted between two and three hours and were taped and transcribed with the participants' permission. In accordance with Patton (2002), the study was designed and conducted by a research team recognizing that multiple research participation may strengthen the study. The data collection was conducted by a team of three researchers and the analysis was done by the authors. The analysis followed an issue-focused open procedure (Thagaard 1998) with a contextual and

circular analytical approach. The two researchers that participated in the analysis read and discussed the interview transcripts, and as a part of the analysis both researchers read and discussed other relevant research and literature (Silverman 1993; Bury 2001).

### ***Data collection***

With the endorsement from the Committee for Medical and Health Research Ethics in Norway and the Norwegian Social Science Data Services the data collection was carried out from September 2007 to February 2008. We used a purposive sampling strategy. Potential informants were selected from the registers of eight National Resource Centres for rare disorders. A postal request, together with information about the study's scope, was sent via the centres. Those that agreed to participate did so by returning the letter of acceptance directly to the researchers. By permission, the present article includes anonymous quotations from the participants.

### ***Sample***

The sample was made up of 94 participants; 51 adults between 20 and 70 years of age with a diagnosis; 33 parents of a child between the ages of 3 and 16 years with a diagnosis or of an adult with a diagnosis and reduced ability to give informed consent; and 10 close family members of a person with a diagnosis (either adult siblings or spouses/partners). For children under 16 years of age (the legal age for making health-related decisions in Norway) and for adults with limited ability to give informed consent, their parents were interviewed. These parents participated both as a proxy for the child and because they were the ones that dealt with the services. Parents tended to encourage adolescents of about 14–16 years old to participate in the interview together with them. This was also the case for adults unable to give informed consent but whose parents regarded them as having a fairly high level of intellectual competence. Of the 51 adults with a diagnosis, 32 were female and 19 were male. All regions of Norway, and rural and urban areas, are represented. The sample included 32 participants that described themselves as having severe impairment, 37 that described themselves as having moderate impairment and 25 that described themselves as having minor problems owing to their rare condition. The number of participants in each diagnosis varied from 7 to 18.

The diagnoses that are represented in the present study are congenital medical conditions. They were chosen because they are rare, and because they represent diversity in symptoms and functional and medical consequences. This diversity was chosen because our research interest is directed towards obtaining knowledge that is not related to a specific diagnosis. The diagnoses have been well known in medical textbooks for many years, and all are under the responsibility of one of the National Resource Centres for rare disorders. Because of this we would expect that information about them should be easy to find. They represent intellectual, physical and sensory challenges, and some imply a combination of various impairments. All of them involve medical problems, latent or manifest, and they represent diversity in requirements for medical treatment and other adapted services.

Representatives of the National Resource Centres participated in the study design, together with representatives of the diagnosis-based organizations that were included in the study. This participation was organized through group discussions that addressed the scope and the topics for the study. The groups gave feedback on

the interview structure. In addition, the participants gave feedback to the research team on the interpretation of the findings in group discussions.

## **Results**

The participants in the present study experienced functional limitations because of their rare disorders and all of them related to a broad range of services that go beyond the medical services: adapted educational services; rehabilitation/habilitation; provision of assistive devices; social security services; and family support. Because of this we chose to use the term ‘user’ instead of ‘patient’ or ‘client’, which are terms that are associated with either health or social services, respectively. All participants encountered specific challenges because the service-providers acted in accordance to the fact that the diagnosis was rare. In the following subsections we will describe some of these challenges, and how the participants dealt with them.

### ***The professionals withdraw from unfamiliar situations***

The tendency for professionals to hesitate when the diagnosis is unknown or the problem is sensitive and emotionally difficult is known from previous research (Varvin 2002; Åsbring and Näränen 2003). According to Berglund, Mattiasson, and Randers (2010), lack of involvement is widespread when the diagnosis or the patient’s problem is unknown to the professional, despite the fact that the diagnosis is clear. The present study describes similar experiences related to situations where the diagnosis was clear and quality-assured information was available and supplied by the user.

In the present study the participants had experienced lack of involvement from many different service-providers. Experiencing lack of involvement was particularly difficult when medical professionals withdrew. In all, 80 out of the 94 participants were not followed-up by their general practitioner (GP) in the treatment or supervision of their rare disease because the GP had refused to be involved. Such experiences led either to endless searches for a GP that was reliable according to their understanding, or to resignation. Those that had no follow-up of their condition expressed insecurity regarding their health condition in general. Some of the participants were followed-up on a yearly basis by a regional or national hospital; these participants also tended to consult the same services for other healthcare needs they might have: ‘That doctor at the regional hospital is my GP, you could say’.

### ***The professionals make their guesses***

Other studies of patients with multiple chronic medical conditions have called attention to how medical personnel act without sufficient knowledge and base their decisions on assumptions and are inattentive to their patient’s worries about this (Grue 2008; Thommessen 2008).

The present study complies with this and shows that this applies to professionals within many sectors, even when the diagnosis is clear. The participants told of how professionals made decisions based on their personal assumptions about the condition. Professionals within many services and sectors were reluctant to relate to information offered by the user and they tended to refuse to take the effort to collect additional relevant information. The participants were uneasy about the

professionals' careless attitudes to their rare conditions and the responsibilities they demanded. Many of the children in the present study needed to be followed-up during the day in order to keep control over their symptoms and to prevent a worsening of the disease. This was, for example, the situation for children with cystic fibrosis, children with ichthyosis and children with bladder exstrophy. During the school day, the youngest children needed assistance in taking their medication and other necessary precautions, while the older children needed their school schedule to be adapted so that they could master this by themselves. In periods when the child did not show symptoms teachers assumed that follow-up during the school day was unnecessary.

For participants with conditions that showed changing or partly visible symptoms it could be difficult to explain to professionals that the consequences of neglect can be serious and that taking daily precautions is necessary even on days when the person is in good shape. A mother was told by the school headmaster that the school considered that helping her child during the school day was a matter of 'helping the child to be comfortable, which they did not consider to be their duty'. Another parent discovered that cystic fibrosis had been misunderstood as being 'something similar to asthma' and school staff had responded to the child as if asthma was the problem. The parents of children with cystic fibrosis were painfully aware that the condition can be life-threatening. When they discovered that the professionals who were responsible for their children during the day did not follow-up medication agreements this caused great concern.

On other occasions the professionals appeared to be familiar with the condition. They convinced the participants that they had handled similar conditions before, and that they were well acquainted with the condition. A mother of a child born with ichthyosis was told by the obstetrician that the conspicuous amount of skin in the amniotic fluid and the new-born's thick, scaly skin was quite common and the parents were told not to worry:

We have seen this many times before, they said, but I soon realized that none of them had ever seen anything like this before. We know now that the instructions they gave were quite the opposite of how we should have treated our child.

From a feeling of being taken care of this participant ended up with a feeling of being deceived.

Participants with symptoms that were only partly visible encountered problems because professionals tended to interpret the condition to be less impairing than the person explained. Even with a partial loss of hearing and sight, a person with Usher syndrome will be functionally blind and deaf in most social settings because both remote senses are affected. This situation leads to problems with credibility because the person does not appear to be blind and deaf at first sight—something that complicates the communication with service-providers in all sectors:

When I was hospitalized the hospital staffs was informed that I had Usher. I had told them that I needed assistance to fetch my food and to find my way down the corridor. They had not complied with my requests and the nurses just said to me 'the meals are served in the dining room. You can walk so you can go there'. They did not believe me. I look normal and my voice sounds normal. I did not look blind and deaf to them.

The participant's experience of not being believed when informing health personnel about the impairments and the connected needs added burden to the feeling of helplessness and vulnerability.

### ***Feeling vulnerable in the hands of the professionals***

The thought of having an accident or suddenly becoming ill was frightening for many participants because this would imply losing control, and in the worst case it could be life-threatening. In the hands of health professionals without adequate knowledge, many of them could risk medical malpractice. All of the participants were aware of the risk of possible maltreatment in connection to hospitalization. Erythropoietic protoporphyria (EPP), for example, leads to hyper-allergic reactions to special forms of light such as the light used in operating theatres or by the dentists. For patients with acute intermittent porphyria (AIP), a number of drugs, such as some commonly used anaesthetics, can cause severe attacks. Persons with myotonic dystrophy risk complications and death from general anaesthesia given during surgery, even if the disease is mild. The father of a child with myotonic dystrophy was painfully aware of this. When the child was admitted to the hospital for a planned operation the father did not trust the doctors to take the necessary precautions. He demanded to accompany the child all the way to the operating room and to talk to the surgeon before the operation: 'To make sure that everything was done right I had sent a scientific article with my child to call attention to the precautions with anaesthesia. Luckily I insisted on seeing the surgeon'. When seeing the surgeon the participant discovered that somebody had thrown the article away. The doctor did not know about the disease and the operation had to be postponed because the doctor was unprepared. The father's precaution was built on his lack of trust. However, his lack of trust prevented his child from having a possibly hazardous operation.

### ***Not being among the approved diagnoses***

Many different types of practical and financial support and coverage provided by the Norwegian Labour and Welfare Administration (NAV) can be helpful for people with rare disorders and can support their activity and participation in society and improve their quality of life. In this matter, the participants' situation is similar to many other disabled people. When applying for benefits, however, a common experience was that the application was denied by NAV. Participants had the strong impression that service-providers limited their services and their approval of benefits because of their lack of knowledge and familiarity with the rare disorder. Some participants described how staff at the local NAV office justified their denial by arguing that they had not heard about the disorder before and thus did not have routines for how to handle such cases. Others described how applications for subsidized treatments or financial support were denied because the particular disorder was not 'on the list of approved diagnoses'. In these cases, nobody had lobbied the authorities in order to make the disorder in question accepted as a condition that can benefit from certain types of support. Others had their applications denied because the service-providers claimed they lacked scientific documentation on the effectiveness of certain treatments or therapies. In no cases did the service-providers search for additional information about the diagnosis.

### ***Taking on the intermediary role***

In order to make the situation bearable the participants decided that professionals are not trustworthy until the opposite has been proved. Because of this, many of the participants placed the responsibility for acquiring adequate services on themselves by taking on the responsibility for providing information and knowledge to the professional. In this respect they exemplified the well-informed, 'modern', patient and service user (Stevenson et al. 2004; Buetow, Jutel, and Hoare 2009), a type of user that many professionals find challenging to relate to (Dieterich 2007).

They described how they constantly were searching for updated knowledge and documentation on their rare disorder. All of them had been provided with updated and quality-assured information by their National Resource Centre and they were eager to distribute this information to those professionals who they meet, whether these were health professionals, welfare service-providers or others. Some carried the information material with them wherever they went in case they met 'someone who needs to know', as one said. When they were in contact with service-providers whom they have not met before, some participants offered information about the diagnosis, relevant Internet sites and contact addresses for the National Resource Centres and/or other expertise. In some cases the participants interpreted the service-provider's reluctance to accept the information offered by them as the professional's fear of disclosing their ignorance and thus losing face *vis-à-vis* colleagues. One way to handle situations like these was to supply the professionals with Internet addresses, hoping that they would look-up the information without disclosing ignorance to colleagues.

In addition to playing an intermediary role by providing information about the diagnosis, participants also provided information about who else that carries knowledge about the diagnosis, and they followed-up to see to that the professionals acted according to the information they had provided:

We have to provide information, and we have to see to it that they read the information and relate to it. We have learned that we have to hang over their shoulders constantly. We have to check and double-check all the time.

To be the one that provided information about the rare diagnosis and other contact information was frustrating. When they also had to see to that the service-providers acted according to the information, this increased their feeling of insecurity and it expanded the place the diagnosis took up in their lives.

### ***The importance of a personal relationship with a professional***

In order to deal with situations where they are confronted with services providers' reluctance to accept information that was provided, many participants made great efforts to develop personal relationships with selected professionals, and they provided this professional with all the information and support they could get hold of. The participants who had a regular follow-up had taken specific initiatives to involve the professional. They put effort into establishing a good relationship with this professional. They provided this professional with the latest information regarding their diagnosis and informed about check-ups and tests that needed to be done. For them, the crucial point was whether the professional acknowledged the information and the initiatives they offered. Participants that described the follow-up

as ‘good’ emphasized the professional’s willingness to accept and relate to the medical information that they provided. From the participants’ point of view, the professional’s emotional and reflective competence was critical, and not first and foremost the technical knowledge. To them, a quality follow-up meant that the professional was willing to be supplied with information and guidance. This also implied that the professional acknowledged the participant’s opinions about the condition and about the necessary activities that need to be done.

Establishing a close relationship with a local professional meant that at least one professional had knowledge about the rare disease. This strengthened the feeling of security even if this support was offered by a single person rather than a team. However, when this person quit their job this competence vanished along with the support and follow-up that was given.

### ***The experience of being forced to disclose helplessness***

Several participants had gone to quite radical steps in order to create acceptance for their problems and an understanding for their situation, steps that they described as ‘not wished for’, as ‘unworthy’, and as ‘embarrassing’. People with ichthyosis took pictures of themselves when the condition was in a serious phase. They distributed the pictures to professionals in order to have proof of their problems: ‘it does not benefit you to try and keep up a good condition. You have to look bad to get credibility’. Others collected the enormous amount of clothes and bedclothes that were damaged because of the vast amount of ointment and lotions that needed to be applied several times a day, and brought this to the local service-providers for them to learn about the consequences of the condition. Parents had taken their children to the welfare office for the service-providers to see them: ‘It helps that they can see the impairment because they do not have the slightest idea about what this really is’. Even though the participants were the ones that had decided to go to these steps, they felt that the professionals’ responses to their situation had forced them to take these actions.

### **Discussion**

In the present article we have described how persons with rare disorders have experienced responses from health and welfare service-providers. The participants described common experiences that cut through the various disorders and challenges connected to the disorders. All participants in the present study told of a range of experiences with professionals within many sectors and services who were reluctant to look-up relevant information and reluctant to accept information provided by the user. Repeated experiences with professionals that withdrew from their case or that drew conclusions on what they assumed might be the problem and suggested actions according to this, led to a general feeling of insecurity and lack of trust, and consequently to scepticism towards professionals in general. The rarity of the disorder in itself seemed to influence the communication with the service-providers and the access to relevant services. This undermined the individual’s expectations of receiving good-quality services. Other studies have pointed to difficulties related to obtaining a correct diagnosis, lack of knowledge, and difficulties in finding relevant information as a barrier for people with a rare disorder. The present study shows that service-providers’ negative responses seem to represent a significant barrier for people with rare disorders. Service-providers within a range of sectors and services

seemed to be reluctant to get involved in situations that relate to diagnoses that are unknown to them. Professionals who did get involved tended to base their judgements on personal assumptions and consequently they suggested incorrect actions. These experiences cut across the various disorders in the study and they cut across the various services. As such, the experiences build a common feeling of being left out of the realm of knowledge and responsibility of the service-providers.

The narratives in the present study should be interpreted against the participants' long and sometimes exhausting diagnosis process, which Petersen (2006) has labelled a 'medical merry go round'. Many of the participants had lengthy and exhausting searches for a diagnosis and consequently a hope for a treatment. After having a correct diagnosis they expected services according to this. When they learned that they had a rare disorder, the participants found it understandable that professionals in the general services lacked knowledge about it, as most professionals encountered the rare disorder for the first time when they met one of the participants. However, the participants did expect professionals to relate to the information they were given and to offer services accordingly. They also expected the professionals to take the effort to collect the necessary information. When their expectations were not fulfilled, the participants developed a feeling of distrust. This distrust was built on repeated experiences with professionals that had refused to accept and acknowledge medical information from them and that had drawn conclusions from what they thought was right.

The findings of the present study should also be interpreted against a health and welfare system that emphasizes locally based individually adapted services with a strong emphasis on user participation. Within a modern health and welfare system, such as the Norwegian service system, service-providers have the right and obligation to act and to make decisions based on their professional education, knowledge, and formal position. When interacting with persons with a rare disorder, however, they seldom have the necessary and relevant knowledge on the rare disease to make the right decisions. On the contrary, the participants in the present study could provide information and knowledge on their disorder and they knew where counselling and supervision could be accessed, but they were not in the legal and formal position to release the necessary treatment and services.

For a person with a rare disorder, the management of such interactions over time tend to become a task in itself. Taking on the responsibility to provide information to professionals and also to follow-up the professionals they may encounter contributes to the identity of being a person with a rare disorder; a person that is placed outside of the professionals' curriculum. The unique rare experience comes into being when the individual is constantly confronted with a lack of knowledge within a service system that is supposed to represent professional support. This experience is amplified by the professionals' reluctance to engage in their situation. In this way the professionals' response to the rarity of the disorder in itself represents a significant barrier that underlines the feeling of being left alone. The services providers' reluctance to get involved may also lead to a consumption of more specialized health services than is necessary because the users also seem to turn to the professional that engages in their situation with health problems that could be dealt with by the GP or by other local health professionals.

When the health and welfare professional's decisions are based on personal assumptions and on the knowledge of other, more common disorders that may resemble the rare condition, this seems to create a user role that is characterized by a

strong responsibility to carry and provide knowledge and information. This also implies taking on the responsibility to establish contact and communication between themselves and the professionals they may encounter, as well as between the many professionals they need to relate to. Such experiences seem to be fundamentally different from the experiences of those individuals who meet a lack of knowledge once in a while, which is the case for most of the population. The collected sum of the many negative events, and the reflection upon them, contributes to the individual's identity as a person with a rare disorder.

The present study points towards apparent shortfalls in the ability of general services to relate adequately to individuals with rare disorders. The lack of knowledge and experience with rare conditions, combined with a limited ability to acknowledge this and act accordingly, is serious for the users and for the credibility of health and welfare professionals in the general services.

The results point to a need for acknowledging individuals with rare disorders as experts on their own condition, that the health and welfare professionals need to be conscious about the limits of their own competence, and the need for revising the patient–provider relationship. In essence, this implies a need for a shift in the balance of power between patients and providers. The findings further imply that there is a need for a closer collaboration between the specialized services that carry knowledge on rare disorders and the general services.

### **Concluding remarks**

As the present study looks at eight disorders with very different pathologies and with quite different impacts on the person's health and functional status, it offers a contribution to the understanding of experiences connected to accessing public health and welfare services for people with rare disorders beyond the experiences connected to a particular diagnosis. In most cases, having a rare disorder implies living with a chronic medical condition. There is a growing recognition that persons with chronic conditions often know more about their condition and the management of the condition than many of the professionals and that this knowledge may contribute to securing adequate treatment and follow-up (Petersen 2006). There have been increasing calls to make use of patients' knowledge in treatment. Therefore, it is imperative that service-providers recognize the particular experience and knowledge of those with a chronic condition—whether it is rare or of a more common kind.

### **Limitations of the present study**

This study was based on persons that are registered at a National Resource Centre for rare disorders and may have omitted experiences of people that are not in contact with such expert support and who may have other experiences. It is possible, therefore, that other points of view and experiences with services are not well represented. However, the strategy to recruit from a broad array of diagnoses should secure that the experiences described in the present article are representative beyond this sample and add insight into how it is to relate to public services for persons with rare disorders. Another possible limitation of the sample may be the demand for the participants to return a postal letter of consent. This may have led to a certain self-selection by those being active and vocal.

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