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Research Paper

Participation in society for people with a rare diagnosis

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Abstract

Background: Many rare diagnoses are syndromes that have complex consequences with a significant impact on the individuals' everyday life. Adults with rare diagnoses are a growing group and knowledge about their needs is often scarce.

Objective: The objective was to investigate the experiences of adults living with different rare diagnoses, how they perceived their difficulties, needs and participation in everyday life, and to identify some common issues and problems.

Method: Individuals with four different diagnoses were interviewed in four focus groups: Artrogryposis Multiplex Congenita AMC ($n = 9$), Dysmelia ($n = 11$), 22q11 deletion syndrome ($n = 10$) and Klinefelter syndrome ($n = 8$). The interviews focused on the following topics; education, working life, daily routines, sleep, housing, health care and society support system contacts. The study was conducted as qualitative research using content analysis.

Results: The participants described their needs and perceived consequences from not having their everyday needs met. A major theme covers most of the participants' experiences: Complex and varying consequences of the condition have an impact on their participation in education, working and everyday life, mostly due to contextual barrier factors.

Conclusion: The interviewees are affected by different rare conditions. Despite these differences they face similar challenges, due to the low prevalence of their condition and the resulting lack of both knowledge and holistic perspective of service providers. To gather, disseminate and implement information about rare conditions in society is thus a very important task. It is equally important to improve the ways to transfer information and to promote cooperation between service providers.

Keywords: Qualitative study; Rare diagnosis; Focus groups; Knowledge; Strategies; ICF

Adults with rare diagnoses are a growing group due to enhanced diagnostic methods and treatments. Many rare conditions are syndromes with complex and significant impacts on the individuals' everyday life.[1] Thus individuals with such conditions, in addition to health care, often need services from the community support system.[2] Due to the rarity of the condition and to the variation of symptoms and severity within each condition, knowledge about the condition and its consequent impacts are limited. Therefore individuals can face inadequate care.[3] Although significant progress has been made in research and public awareness during recent years, much remains to be done.[4] Mostly, focus is on medical and pharmaceutical issues. Social issues are sometimes discussed, but usually only regarding children with rare conditions and their families.[5,6] The experiences and needs of adults are therefore important to explore.

The definition of a rare diagnosis varies. In Sweden a rare diagnosis is defined as one with a population prevalence of less than 1/10,000.[7] The most common European definition is 1/2000,[8] and in the USA it is 1/1250.[9]

Each condition is rare, but taken together, many individuals are affected by a rare condition.[3] Many of these lead to comprehensive functional problems and activity limitations. Some conditions earlier considered very rare have gradually proved to be more common, since more children are diagnosed earlier in life due to enhanced diagnostic methods. The development of treatment and improved prognosis also lead to more individuals reaching adult age. The number of adults with rare diagnoses is therefore increasing.

The International Classification of Functioning, Disability and Health (ICF) is WHO's framework for describing and measuring health and disability from physical, individual and societal perspectives.[10] The ICF model takes into account both biological and contextual factors and their dynamic interaction, thus also showing the impact of environmental and personal factors on a person's functioning and level of participation.

This study was, with only general information, presented on a poster at the European Conference on Rare diseases & Orphan Products (ECRD) Conference in Berlin in May 2014.

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The traditional medical care model focuses on treatment and cure of impairments. Although considerable medical progress has been achieved during recent years, rare diseases are still incurable and mostly connected with lifelong impairments. Thus the medical condition must generally be regarded as permanent. Therefore the issue of contextual factors and how they can be improved, must be considered very important.

Aim

The aim of this study was to investigate the experiences of adults living with different rare diagnoses, how they perceived their difficulties, needs and participation in everyday life, and to identify some common issues and problems.

Method

Design

The study was conducted as descriptive qualitative research using content analysis. Data were collected through focus group interviews. Focus groups explicitly use group interaction as part of the method.[11]

A focus group guide was used and the interviews focused on the following topics: education, working life, daily routines, housing, sleep, health care and society support system contacts. The topics were based on the researchers' earlier experiences from meeting individuals with rare conditions, taking contextual factors in the ICF model into consideration.

Data collection

The four focus group interviews were held in an undisturbed room and the same researchers (GJ, AR) conducted all interviews.

The sessions lasted for approximately 2 h and were tape recorded.

Analysis

The interviews were transcribed verbatim and analyzed using manifest content analysis.[12] The contents were first read, to get an overview of the collected data. Then key words and text units were marked, patterns and common themes were identified. The text was then grouped into categories that were discussed by the whole research team until consensus was reached.[12] Citations presented in the result show the opinions of several of the interviewees.

Context

Ågrenska, situated in Gothenburg, Sweden, is a national center that provides programs for children, teenagers and adults with disabilities, many of them rare, their families and professionals. Most of the programs aim at enhanced coping for the participants.[13] One program offered is a three days course for adults with rare conditions. The course includes medical expert information about the condition, information and discussions about psycho-social factors, legislation and community support services, as well as health and life style factors. The course also provides a unique possibility for individuals with the same rare condition to meet others in a similar situation and exchange experiences. The purpose of the course is empowerment and enhanced coping for the participants. Another purpose is information gathering about the impacts of the conditions, as perceived by the participants. Therefore a focus group interview about the participants' everyday life and needs is performed at the end of every course.

Sample

All persons who attended an adult stay in 2006 and 2007 for people with AMC, Dysmelia, 22q11 deletion syndrome and Klinefelter syndrome were invited to participate in a group interview and all accepted. Nine women with AMC (aged 22–57), nine women and two men with dysmelia (aged 18–67), five men and five women with 22q11 deletion syndrome (aged 18–39) and eight men with Klinefelter syndrome (aged 17–69) participated.

Below are presented a few facts about each diagnose.

Arthrogryposis multiplex congenita (AMC)[14]

AMC is characterized by symmetrical contractures in more than two joints in different parts of the body. There are more than 150 known types of AMC. Some types are genetic and some have unknown causes. The most common type is Amyoplasia, classical arthrogryposis, symmetrically affecting hands, wrists, elbows, shoulders, hips, feet and knees. Frequently, the contractures are accompanied by muscle weakness. Amyoplasia is usually connected with severe mobility impairment. Another type of AMC is distal arthrogryposis. This type is characterized by joint deformities and contractures that restrict movement in hands and feet and can include permanently bent fingers and toes, overlapping fingers, clubfoot. People with AMC usually need a combination of multiprofessional therapies.

Dysmelia[15]

Dysmelia is an umbrella term for different types of congenital limb reduction differences. The condition is not hereditary and the cause of dysmelia is unknown. It is characterized by missing or foreshortened extremities. People with dysmelia usually need orthopedic surgery and measures from physio- and occupational therapists to acquire and retain best possible function.

22q11 deletion syndrome[16]

The 22q11 deletion results in abnormal development of many organs and tissues. Most individuals with the syndrome have a new deletion. Missing or underdeveloped glands as thymus or parathyreoidea cause immunological deficits with recurrent infections. Other symptoms are; cleft palate, dental anomalies, nasal and oral problems, heart defects, feeding difficulties and mildly characteristic facial features. Language and motor development may be delayed. Neurological abnormalities and cognitive as well as neuropsychiatric problems are described. Adults frequently develop psychiatric symptoms.

Klinefelter syndrome[17,18]

Klinefelter syndrome only affects males and is due to the presence of one or more extra X-chromosomes. Only about 10 percent of the affected men are estimated to get a correct diagnosis. All grown up men with the syndrome have small testes with little or no sperm production and low levels of testosterone. Autoimmune and cardiovascular diseases, osteoporosis and insulin resistance are common. Low levels of testosterone can also cause sleeping problems and aching joints, ligaments and muscles. Some degree of impaired cognitive development may be present e.g. language, reading and writing difficulties as well as reduced psychological drive. Motor development may also be delayed. Treatment includes testosterone replacement which can reduce some of the symptoms, and improve physical and psychological achievement.

Ethical aspects

An ethical discussion was performed together with external researchers.

All participants from these four diagnose groups were considered able to consent themselves, based upon given information. Before the course, the participants were *invited to participate in* an interview *and were informed about* the purpose of the interview, that it would be recorded, that their participation was voluntary, that their response would not affect their stay, and that they could change their minds at any time without explaining why. They were as well informed that the result was meant to be published in different reports. The same *invitation to participate* was repeated upon their arrival, and at the start of the interview. They did not receive any incentive for being a part of the focus group.

Results

The participants described their needs and perceived consequences from not having their everyday needs met. A major theme covers most of the participants' experiences: Complex and varying consequences of the condition have an impact on their activity and participation in education, working and everyday life. Eight categories were identified, all but one related to contextual factors. These categories are discussed below.

Impairments lead to lifelong consequences and activity limitations

This category is related to the health condition/bodily impairment and the participants in this study described that their symptoms, affected areas and needs had remained since childhood. Symptoms and needs sometimes presented themselves in a slightly different way due to development and demands during the years. New needs were also added during adulthood. Difficulties with concentration, planning, estimation and planning of time, all causing stress, were described as remaining since childhood for some participants.

> I had problems concentrating at school and I still have. If something more interesting occurs… then I drop my present focus… (case 20)

Some symptoms and their consequences got more prominent in adulthood, as demands increased by age, e.g. in parenthood and working life.

> …planning and structuring meals, it's really hard… when you've got children running around, you get completely distracted. (case 35)

> …learning to handle the machines at work was like hell. It was said to take six months to handle a new machine. For me it took one and a half years and I couldn't understand why. (case 27)

> …I was completely worn out when my daughter was having surgery… I had to take care of her, wash her and assist with everything. Nobody considered that I had the same disability as my child. (case 17)

> My healthy arm has become overstrained. (case 9)

Fatigue is a common problem, sometimes related to sleeping problems, e.g. problems to fall asleep or waking up several times once having fallen asleep. It is also a kind of general fatigue, not being related to sleep, work load or to what has been performed. This also leads to consequences in everyday life and a need for rest in daytime.

> You go to bed tired and wake up, just as tired. (case 25)

The following three categories are related to environmental factors.

Environmental knowledge and understanding affect activity and participation

Knowledge was recognized as a facilitating contextual factor and was seen to enhance understanding and

acceptance. This knowledge is of great importance for the correct support and adjustments and therefore affects the individual's possibilities for activity and participation. It was considered important that teachers and schoolmates, superiors and colleagues know about and accept the impairment. Often parents or the individual him/herself informed them.

> I just showed them 'My arm looks like this, therefore I take the paper with my mouth.' After that nobody asked any questions.… It's important not to be too different. (case 11)

> You should neither be favored nor disfavored… you should be treated as everyone else. (case 13)

> I told my teacher that reading is difficult… the exams were difficult because I couldn't understand the text and the questions.… Then I got a teacher who read the questions to me… and that worked out really well. (case 21)

> I've got some easier tasks now, because I have a very good boss, he has knowledge and is understanding. (case 8)

> …my colleague just asked 'What's the matter with your hands?' … I think it's good when people have the courage to ask. (case 16)

Feeling disregarded in education

Regardless of diagnosis, participants described that they needed an adapted school environment. For some individuals both attitudes and physical environment were in focus. Problems were perceived both in mainstream and in specially adapted schools but for different reasons. The interviewees reported that teachers in mainstream schools sometimes did not want to adjust physical tasks to the student's needs. Sometimes it was easier to exempt a student from an exercise than to adjust the task.

> The teacher did not adjust the exercise but excused me from the lesson instead … although I always wanted to participate as much as I could. (case 4)

> Physical education was a torment, actually. It depends on the teacher … I got another teacher who was interested … he saw my possibilities, I had my own tasks and set of appliances. (case 12)

Schools intended and adjusted for students with mobility problems sometimes accepted students with other needs as well, e.g. intellectual or neuropsychiatric.

> The National Upper Secondary School for Physically Disabled Students … for me it was a disaster … they accepted students with other problems as well, which, I mean, lowers the standard …. (case 15)

Needs related to cognitive abilities such as problems with learning, memory, concentration, planning and physical coordination were described by some of the interviewees. They had been offered varying help or support during their schooldays. For those being diagnosed late in age, as teenagers or young adults, neither themselves, nor their teachers, had understood their problems at school.

> I had to study a lot just to pass. When others studied as much as I did they could get the highest grade and I could never understand why. (case 34)

Lack of holistic view and coordinated environment

Participants described unmet needs for a holistic view, cooperation between different professionals, and coordination of measures. Better cooperation and transfer of knowledge between health care, the public insurance system and employers was asked for by a person needing reduced working hours to avoid strain injuries.

> They don't see the whole picture, when my healthy hand is worn out I'll be totally dependent on others. Then I can't continue my professional life. It's so stupid, that they don't have this perspective. (case 9)

Lack of cooperation within the same profession was also reported, e.g. that doctors do not see the bigger picture, nor understand the complexity or how symptoms can interact.

> Why don't we get adequate treatment, taking the whole syndrome into account? I don't get the right treatment; doctors just see a small part of my syndrome. (case 20)

> I haven't even once been offered to talk … about virility, of course you wonder yourself about the sterility and its consequences. (case 21)

Sometimes the persons have to take on the responsibility themselves, for which professionals to consult, at what intervals and what to ask for. Professionals don't take the initiative to offer anything.

> You have to be updated and know yourself what you need and want. (case 2)

> I think it's important to have someone who coordinates all actions, when you have this syndrome. (case 35)

Being discriminated and humiliated

This category can be regarded as related to both environmental and personal factors.

Participants reported feelings of discrimination and humiliation in different situations, sometimes because of prejudices, sometimes because of lack of knowledge and

understanding. This often resulted in participation restrictions.

> The worst thing is that there are so many assumptions about what is best for me – without asking me. (case 17)

> A vertically adjustable bed was not allowed to be installed into our double bed, if so, they threatened to take it back! (case 15)

> I had three young children and needed housing adaptation. Instead I was offered a flat in an old people's home! (case 17)

In addition to environmental factors personal factors e.g. individual preferences, play an important role for how a situation is perceived. Some participants stressed the importance of being like everyone else, not to have any special treatment positively or negatively. But this did not suit everyone.

> When all students are doing an activity they say 'and you can do it your way', then you are different from the start. (case 17)

> I studied at the Department of Social Work at University. It worked out OK, but everything was meant to be so natural …, nobody asked me anything, and I didn't feel comfortable that way either, because I'm not like everyone else. (case 14)

These last categories are related to personal factors.

Strategies to overcome difficulties

These examples below show the importance of personal factors like coping strategies and experiences as facilitating factors in everyday life.

Daily routines were found very important in order to function for some participants.

> I make lists! I write everything down… and then I just mark so I can see what I have completed. I have to book everything in advance, because just doing things when I feel like it, that doesn't work for me. (case 21)

It is important to think about your health in a long-term perspective, and not end up with unnecessary strain injuries.

> … having a water tap that you can lift and not have to turn … that is very good and preventive. (case 4)

Own reflections on choice of education and profession

The participants shared some thoughts and comments upon their vocational choices and the long-term consequences of these choices. It was obvious that age-related guidance from their environment would have been needed.

> I studied to become a preschool teacher and passed, but this work turned out to be too hard for me. … I've been trying several other jobs.

> … when I was younger I never imagined that I could get strain injuries. (case 1)

> I feel that I've not been responsive to advice … if somebody had told me what I could do and not do. (case 9)

> I think I would have needed more help. My parents always told me that I could do what I wanted to. I've thought about that. Should they have decided more for me? Talked about it more? (case 3)

Discussion

The interviewees are affected by four congenital, lifelong and different conditions. Despite the medical differences the interviewees often face similar problems concerning participation in everyday life, but for different reasons. This regards experiences in childhood, youth and adult life. Participation restrictions in everyday life are reported[19] and needs for environmental facilitators are discussed in other studies concerning rare conditions.[20]

There are two main problems perceived; one is lack of knowledge about rare conditions among health care and service providers and the other problem is organizational shortcomings and lack of cooperation on all levels. This is also reported from other studies.[3,21] What the interviewees reported in this study is also in accordance to the researchers' earlier experiences with individuals who have rare diagnoses.

In order to offer the right support and services, professionals need syndrome specific knowledge, in addition to general knowledge according to profession. Forming the basis for other syndrome specific knowledge, the medical facts about the condition have to be translated into other fields relevant for the individuals, e.g. educational and work-related. As most rare conditions are syndromes with lifelong consequences adults affected have multifaceted needs: medical, habilitation and social ones. Thus many service providers, all needing knowledge, will be involved. Therefore, knowledge must be considered one of the most important contextual factors. This is also shown from the result of this study. There are four categories mostly related to a need for knowledge; ''lifelong consequences and activity limitations,'' ''environmental knowledge and understanding affect activity and participation,'' ''feeling disregarded in education,'' and ''being discriminated and humiliated.''

Lack of knowledge can create more barrier contextual factors, e.g. attitudes that further prevent participation. This was described by the interviewees in school situations and when needing housing adaptation.

Lack of knowledge also lead to discrimination experiences for the interviewees in different situations, e.g. being excused from physical education lessons because the teacher did not know how to adjust the tasks.[22] On the other hand, informed employers and colleagues were described as enhancing factors for participation and independence.

There is also a need for correct and accessible patient information about the diagnoses, e.g. in order to enable individuals to manage their own health[23] and in order to enable them to make informed decisions.[2,24] Appropriate information is often insufficient.[3]

The need for patient information also became obvious in this study, e.g. when people make important decisions like choice of profession. Knowledge is necessary to balance possibilities in relation to the actual impairment and the person's expectations and wishes. Correct and accessible information can also help parents to give age-relevant guidance to their children. This affects the possibilities neither to be influenced only by attitudes of the environment, nor to make choices not sustainable in a lifelong perspective. Patient knowledge is also important in order to develop purposeful coping strategies. This is expressed primarily in the categories "strategies to overcome difficulties" and "own reflections on choice of education and profession." To be informed is also essential when describing medical and social needs to authorities in order to get access to the right care and services.

In this study, the second main problem perceived was related to the support system organization and lack of cooperation. Society of today is specialized. Provision of different services is the responsibility of different levels in society, i.e. municipal, regional and state levels. There are also different responsibility areas within the same level.

This was described, in the category "lack of holistic view and coordinated environment," by several interviewees. Lack of communication and cooperation was reported, both within the health care system and between medical professionals and e.g. the social insurance system. The importance of a holistic perspective was stressed. Poor coordination of services and lack of communication have been pointed out as one key experience among English patients with rare diseases.[2] Insufficient cooperation and poor communication lead to fragmentation of services.[25] Organizational shortcomings and fragmentation get severe consequences for people with rare conditions, often requiring multifaceted treatment and services.[26] Independently operating units without communication cause problems for people with common diseases as well.[25]

This can also be discussed in terms of the knowledge-translation concept (KT). The KT-concept stresses the importance of not only creating knowledge, but also transforming and transferring it between and within different units. It is a matter of using existing knowledge for different purposes in different contexts, thus bridging the gap between what is known and what is actually done and used for the benefit of patients.[21]

The ICF model stresses the dynamic interaction of biological and contextual factors affecting the outcome on an individual's functioning and participation. Placing the findings of this study into the ICF model, the first category identified is related to the participants' health condition and bodily impairment and all other categories are related to contextual factors. Contextual factors were described as having a significant impact on participation in everyday life and lack of knowledge, affecting many areas, is the most important barrier factor.

Limitations

The diagnoses for the interviews were chosen because they represent different physical limitations and problems. Common problem areas are seen despite the fact that four different groups were interviewed. Although these similarities were found it is possible that individuals with other rare conditions might have different perspectives. The interviews were performed at the end of the stay at Ågrenska, which may have positively influenced the willingness of the interviewees to participate in the interview.

Conclusion

Full participation in your own life situation is important for everybody. For individuals affected by rare diagnoses this can be difficult to achieve. People with rare diagnoses often face challenges, both due to the low prevalence of individuals with their diagnoses and due to system rigidity. This results in lack of knowledge among their service providers and official bodies. Origin and consequences of the conditions are closely linked to each other and all aspects important for the individual have to be taken into account.

To improve this situation, educational efforts are called for, within education as well as in the health-care system. To gather, disseminate and implement information about rare conditions in society is thus a very important task. Equally important is improving ways to transfer information and to promote cooperation between service providers. Thus, better knowledge and enhanced cooperation can improve the situation and enhance possibilities for participation for people with rare conditions.

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