

People with rare diseases

Common experiences

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Aim

To describe how people with a rare disease and their families experience public health and welfare services.

Method

A contextual approach based on conversation-like, semi-structured face to face interviews. Written consent and ethical clearance were obtained.

Participants

- 94 individual participants, all of which had been in contact with a nationwide resource centre:
 - Adults over 18 years of age with a rare diagnosis
 - Parents of children under 18 with a rare diagnosis
 - Parents of 'children' over 18 with a rare diagnosis and reduced informed consent
 - Spouse, partner or cohabitant, sibling and grandparent of a person with rare diagnoses
- Six group interviews, involving a total of 40 people, conducted at gatherings arranged by three support groups.

Results

The local society – common experiences

"I felt I was the only one in the world with these problems"

The most typical common experience was the feeling of being alone with ones problems and needs. Participants rarely met anyone of their own age with the same diagnosis in the area where they lived. Most participants therefore expressed a great need to meet others with the same diagnosis and that experienced the same challenges as themselves.

"They know nothing about my diagnosis"

Local service providers, such as health service professionals and social welfare officers, seldom had knowledge of the diagnosis or its consequences. Hence, participants seldom received optimal treatment or follow-up.

"They're not interested in learning about my disease"

While participants did not expect local service providers to have expert knowledge of their diagnosis, they did expect to be met with respect when they provided them with this knowledge. However, this was far too seldom the case. These service providers showed little interest in acquiring this knowledge because, as one said, *'it's a problem we're not going to meet again'*. Instead, service providers tended to make decisions and propose measures based on their own assumptions.

"I had to tell them everything"

Participants had to explain the implications of their diagnosis to their service over and over again. They also had to take the main responsibility for getting hold of and coordinating the necessary services. Sometimes service providers reacted negatively when the patient was the expert.

"As soon as they saw me, I got extra time for my exams"

Participants met differing responses depending on the visibility of the symptoms. Visible difficulties seemed to be more easily accepted by service providers when applying for assistance and support. Participants with less visible difficulties, on the other hand, seemed to be socially more integrated in society.



Experiences with the national resource centres

"For the first time, someone said that my baby was lovely, and I felt like I had come home"

Participants regarded the resource centres as contributing positively to their struggle to achieving a manageable everyday life. The centres provided participants with necessary information and knowledge that helped them gain control over their lives and experience a good quality of life.

- The participants saw the following factors as most important:
- The opportunity to have direct access to the centres (by telephone, e-mail or personal visits) without being referred by a physician.
- The experience of being treated with respect and empathy by the centre's staff.
- The opportunity to receive emotional support and practical assistance.
- The accessibility to updated knowledge of their particular diagnosis.
- The support and back-up they received from the centres in dealing with local service providers.
- The opportunity to participate in local meetings with centre staff.
- The close cooperation with diagnose-based support organisations that are within the centres' area of responsibility.
- The efforts of the centres and the support organisations to pave the way for persons with the same diagnosis and their families to meet each other.

Conclusion

A 'rare' experience is the feeling of being left out of the realm of knowledge of health and welfare professionals. This is both an exhausting and threatening feeling. This situation makes them vulnerable and dependent upon the few places in Norway which offer competent help.

Social and health policy in Norway has moved towards more individualisation and privatisation. One consequence is the demand for self-determination in matters that concern the individual. To be able to act as a citizen vis-à-vis the health and welfare services it is essential to be informed and to have knowledge. Most participants in this study receive support to act as citizens through their contact with a resource centre and a support organisation.

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Resource group

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Reference group

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Working groups

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