What do you need? How do you cope?

Including everyday experience of patients and their professional caregivers in the development of suited health care programs for rare diseases

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BACKGROUND

Over 6.000-8.000 different rare diseases affect different aspects of patients' lives in different ways[1,2]. It seems practically impossible to provide scientific research and specialized support for each of these combinations.

Facing these limitations, we suggest mapping
1) patients' core needs and
2) individual coping strategies that are deemed helpful in everyday life.

Patients as well as their private and professional caregivers live and cope with the effects of a rare disease for many years on a regular basis.

As a result they have accumulated vast practical experience. Sharing this knowledge provides important orientation in self-help networks. However, this knowledge is not yet sufficiently and systematically used to inform scientific research, patient information and holistic, specialized health care programs[3].

REFLECTION/MOTIVATION

Do we fully listen to the patients' needs and perspectives?

Do we fully take their best practice strategies into account for the development of treatments?

OBJECTIVE

The study aims to develop appropriate research instruments to access everyday experience for rare diseases using the example of Williams Syndrome (WS, ORPHA number 904)[1].

Figure 1: Boy with Williams Syndrome

WS is a developmental disorder associated with a gene loss on chromosome 7. Most individuals with WS need lifelong support [4,5].

FOCUS AREAS

1 ***Needs***

2 ***Coping Strategies***

EXAMPLE QUESTIONS FROM INTERVIEW

Which are the challenges in the patients' life that put the greatest stain on you? Which problems most urgently need to be solved?

Which strategies did you find that allowed for a positive effect on the quality of life of the individual with WS?

METHOD

Design: *Concurrent Mixed-Methods Design* [6]

Sample: N=71, parents and professional caregivers (eg teachers, therapists) report on 65 individuals with WS, *Purposeful Sampling* [7]

Instrument: *Semi-structured qualitative interviews* including quantitatve ratings of everyday life challenges on two dimensions:
1) development over time (improvement, neutral, worsening), level of burden presented by challenge (5 point *Likert scale*)

Analysis: *Qualitative Content Analysis* [8], Inferential Statistics, Mixed Methods Analysis via MAXQDA

Current status: Data is currently being analyzed; besides scientific publications, results will be published as brochures for patients and healthcare guidelines

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Literature:

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[8] M. Schreier. *Qualitative content analysis in practice*. Los Angeles: Sage, 2012. **Figure 1**: The picture was provided courtesy of the Williams Syndrome Association (USA).