

# A survey about the living conditions of people with rare disabilities

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## 13 different disabilities

The Centre for Rare Diseases and Disabilities has conducted a survey in Denmark about the living conditions of people with rare disabilities. Through the evaluation of questionnaires and group interviews they've been able to provide comprehensive accounts of the challenges that are faced by people with rare disabilities. There are numerous portrayals of daily life throughout the survey that exemplify just how severely a rare disability affects everyday life.

The survey includes the following disabilities:

Type 1 Osteogenesis Imperfecta, Congenital immune deficiency diseases, Gilles de la Tourette's Syndrome, Rubinstein-Taybi's Syndrome (broad thumb-hallux syndrome), galactosemia, tuberous sclerosis, Ehlers-Danlos' Syndrome, Marfan's Syndrome, Angelman's Syndrome, Crouzon's Syndrome, dwarfism, Soto's Syndrome (cerebral gigantism) and Batten's disease (Spielmeyer-Vogt's Syndrome.)

## Questionnaire and interviews

The results stem from both the questionnaire and the group interviews. 891 questionnaire responses were collected through the patient associations with a typical response rate of 60-75%. The questionnaire consisted of a core of standard questions that were part of all of the surveys. Beyond these, there were questions tailored to the specific disabilities. Between one and three group interviews were conducted per survey.

## Unique conclusions – and similarities

When you study thirteen rather diverse disabilities, you end up with thirteen very unique conclusions, but there are also similarities.

- The diagnosis is essential within the social system. The waiting period up to the diagnosis is difficult on the parents.
- The public authorities lack knowledge about the diagnosis and especially about the prognosis.
- Often, the authorities fixate on the diagnosis description rather than the resulting restrictions on the individual

patient's ability to function in daily life.

- The type of support and services offered often depends on which commonly more known disability the rare diagnosis resembles most.
- The more invisible a disability is to the environment, the poorer the cooperation with the social authorities.
- If it is a controversial diagnosis (i.e. if the criteria for a diagnosis are uncertain or if they are evaluated in different ways) there'll be even more problems with the public authorities.

The most recent ten surveys (starting with Rubinstein-Taybi's Syndrome) contain an English summary. These can also be found at [www.csh.dk/projekter](http://www.csh.dk/projekter).

## Further Work: The common set of problems

We're planning on working on a dissertation that collects all of the findings and covers all of the thirteen surveys. This study is meant to shed light on the common set of problems that people with rare disabilities experience.



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