

Factsheet

Most People Living with a Rare Disease are Living with Disability

“Disabilities is an umbrella term, covering impairments, activity limitations, and participation restrictions. An impairment is a problem in body function or structure; an activity limitation is a difficulty encountered by an individual in executing a task or action; while a participation restriction is a problem experienced by an individual in involvement in life situations” [1, WHO].

“Disability is a universal human experience, in the sense that everyone can be placed on a continuum of functioning and either currently experiences or is vulnerable to experiencing disability over the course of their lives. This understanding of disability is the key to mainstreaming disability within the public discourse” [2].

Key policy references:

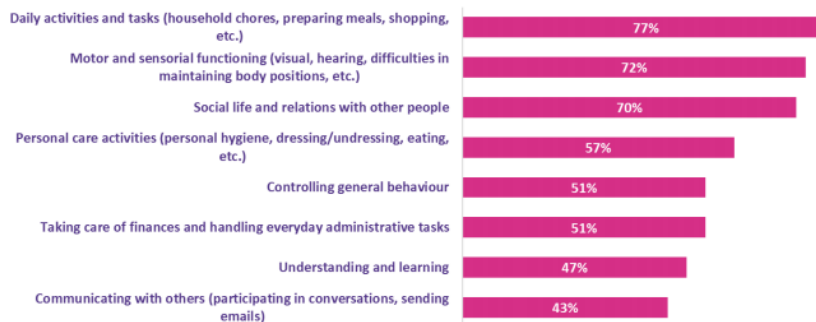
- The Commission Expert Group on Rare Diseases [Recommendations to support the integration of rare diseases into social services and policies](#), adopted unanimously by all Member States in 2016 [3], state that “rare diseases are often chronic, highly complex, severely disabling and frequently affect life expectancy” and recommend that “rare disease specificities should be integrated into national systems assessing a person’s level of functioning, in line with the United Nations Convention on the Rights of Persons with Disabilities;
- The European Rare Disease Task Force, in 2008 [4], underlined that rare diseases “all together account for a big part of the early-life deaths and life-long disabilities in the European population”.

Results of European survey on the social impact of rare diseases:

The survey “[Juggling care and daily life: The balancing act of the rare disease community](#)” [10] shows that rare diseases severely affect the everyday life of people living with a rare disease and their carers.

- **Rare diseases are disabling, limiting several Activities of Daily Living (ADLs):**

Graph. 1. To what extent does the person living with a rare disease have difficulties with... (Subtotal difficulty) (n=2689)



Some modalities have been grouped:
Difficulty=Completely unable + A lot of difficulty + Some difficulty

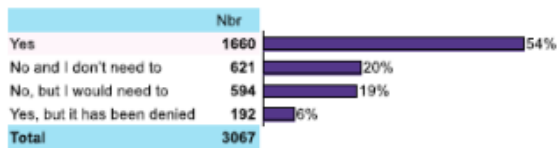
- Rare diseases **disabling aspects** are often invisible and may vary in time due to the fluctuation of symptoms:

“The difficulty lies in the impossibility of carrying a routine (...). The problem arises when one day you appear completely healthy, the next day you are sick, and two days later you appear completely normal again. Many people find it difficult to understand the disease and the process, and the absenteeism that entails”. Female, Spain

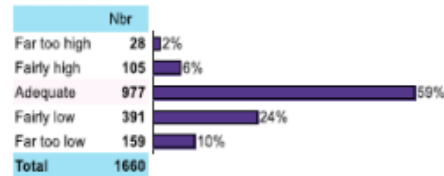
- **50% of people living with a rare disease have uncovered needs regarding the access to disability benefits:**
 - 28% of respondents expressed that they have access to disability benefits but not enough to cover their needs;

- 22% that they do not have access to these benefits but would need it.
- **People with a rare disease face challenges with disability assessment:**
 - 19% of respondents would need disability assessment, 6% have been denied one;
 - 34% of respondents who have been submitted to a disability assessment find the % of disability assigned to them to be fairly low or far too low;

Has the person living with a rare disease been submitted to an official assessment of the level of disability?



In your opinion, do you think that the percentage of disability officially assigned to the person living with a rare disease is...?



- **Rare diseases generate a significant and disabling time and care burden:**
 - 42% of people living with a rare disease and their carers spend more than 2h/day on illness-related tasks;
 - People living with a rare disease and their carers feel badly informed about their rights and feel that social services are badly prepared to support them;
 - Care pathways are complex and hard to manage e.g. need to visit different services in short time; lack of communication between providers;

"The disabled person has to deal with several different services to receive help and benefits. There are therefore often waiting times of around 6 months whilst dealing with each service or waiting for service A to send your paperwork to service B. During this time, you have not had time to employ someone and you are still not working". Female carer, France

- **Rare diseases lead to a strong impact on work-life balance** - absence from work, hampered professional activity, economic burden:
 - 70% of people living with a rare disease and their carers have to stop or reduce professional activity due to the disease;
 - 58% are absent from work over 15 days per year;
 - 69% face a decrease in income.

Key scientific references:

- Studies in different countries show that people living with a rare disease are often living with varying levels of disabilities [5]:
 - France: analysis of local registry with 1739 children shows that rare diseases often lead to different types of disabilities with different degrees of severity. In 26% of cases the RD was at the origin of cases of severe impairment [6];
 - Italy: study with 516 families with children with rare diseases: 70% of children also had a motor and/or intellectual disability [7];
 - Spain: In the region of Murcia, a study revealed that a third of the people living with a rare disease have obtained the official recognition of their disability, 25% of these in the highest degree of disability (over 75%) [8];
 - Australia: study with 46 families affected by rare diseases shows that 63% of respondents had some level of disability, of which 13% severe disability [9].

References

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- [4] Rare Diseases Task Force. Health indicators for Rare diseases: State of the art and Future directions. 2008.
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- [10] EURORDIS 'Juggling care and daily life: The balancing act of the rare disease community'. 2017 - European-wide survey with over 3000 respondents, conducted via Rare Barometer Voices in the scope of the EU-funded INNOVCare project.