THE IMPACT OF LIVING WITH A RARE DISEASE: BARRIERS AND ENABLERS OF INDEPENDENT LIVING AND SOCIAL PARTICIPATION

A Rare Barometer survey

April 2025

Rare Barometer is a survey initiative that robustly collects the experiences and opinions of people living with a rare disease and their close family members on topics that directly affect them. This programme is run independently by EURORDIS-Rare Diseases Europe and is a not-for-profit initiative. It conducts 1 to 3 studies annually and hosts a survey panel of more than 20,000 people who agreed to receive email invitations to participate in surveys and studies conducted by EURORDIS-Rare Diseases Europe.

EURORDIS-Rare Diseases Europe is a unique, non-profit alliance of over 1,000 rare disease patient organisations from more than 70 countries that work together to improve the lives of the 30 million people living with a rare disease in Europe. By connecting people living with rare diseases, families, and patient groups, bringing together stakeholders and mobilising the rare disease community, EURORDIS strengthens the voice of people with rare diseases and shapes research, policies and services. EURORDIS is financed by the European Union, member patient organisations, and AFM-Téléthon. EURORDIS also receives charitable donations, individual donations, and donations from corporate foundations and the health industry.

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EXECUTIVE SUMMARY

This report presents the results of a survey on independent living and social participation, conducted between July and September 2024, among people living with rare diseases and their family members. The objectives of the survey were: *i)* to estimate the level of participation in social activities such as education, work or leisure, *ii)* to identify barriers or facilitators in doing those social activities, *iii)* to understand preferences and needs regarding living arrangements and personal assistance, *iv)* to collect experiences of disability assessment, *v)* to identify the main difficulties in accessing social and disability rights. The report is organised in four parts:

Part 1 focuses on measuring disability. **8 out of 10 people with rare diseases lived with disabilities**: this figure comes from the measurement of disability rate using three instruments: functional difficulties (Washington Group Short Set – WGSS), activity limitations (Global Activity Limitation Index – GALI) and self-identification (asking participants if they consider themselves as a person living with disability).
Their **disabilities were diverse, complex, dynamic, and progressive** and ccouldan manifest through **pain or fatigue**. **70% of people with rare diseases considered themselves as a person with invisible disabilities** (or both visible and invisible disabilities).

Part 2 tackles disability recognition. **15% of participants had never undergone a disability assessment although they needed one**, which can impede their participation in society by hindering their access to the support they need.
Those submitted to disability assessment and recognition processes were still be **denied adequate disability recognition and support**. This was a consequence of barriers faced in disability assessment processes, such as a **dominant medical approach and insufficient consideration of the person's situation and needs.**

Part 3 delves into independent living, including difficulties in accessing social benefits, access to personal assistance, and living arrangement preferences. **Most participants found it difficult to access publicly funded support** for various reasons, including a lengthy and complex process. **62% of those who need a personal assistant did not have access to one** and, among those with access to one, **only 21% could choose their personal assistant**. More than 60% of personal assistants were family members, which was positively associated with time dedicated to assistance.
Although most participants were satisfied with their current living arrangements, **most people with rare diseases who lived in institutions prefered other living arrangements**.

Part 4 explores participation at work, school and in the community. **More than 1 out of 2 participants had already faced discrimination** because of their disease or disability. This includes discrimination in healthcare, public places, transport, employment and housing.
23% of the participants were unemployed or unable to work because of their disease. This is nearly four times the EU unemployment rate in 2023. Participants stressed the importance of workplace accommodations such as adequate work settings, flexible schedules, remote work, inclusive attitudes from colleagues, and adequate training or career counselling.
79% of pupils and students with rare diseases had limited participation in school (as measured by the Children and Adolescents Scale of Participation - CASP). Although having a disability was critically related to limited school participation, age, social environment, and country of residence also mattered.
84% of participants had limited participation in the community. This finding was determined by applying the community involvement module of the CASP to all respondents.

CONTENTS

INTRODUCTION

There are 30 million people with a rare disease in Europe. Each of them lives with a health condition that is present in less than 1 out of 2000 people. There are over 6000 distinct rare diseases, and their manifestations vary from condition to condition and person to person. While the rare disease community as a whole is inherently heterogeneous, people with different rare diseases face common barriers which keep them from fully accessing their health, social and independent living rights.

Guided by the *UN Convention on the Rights of Persons with Disabilities* and by the *UN Resolution on Addressing the Challenges of Persons Living with a Rare Disease and their Families*, EURORDIS-Rare Diseases Europe and its members envision a world where everyone with a rare disease can achieve their full potential. A world where their rights are respected and they access adequate care and support. A world where they are enabled to live independently, and to fully, equally and meaningfully participate in all areas of society.

This survey aims to support evidence-based advocacy to advance this vision by providing insights into the experiences and expectations of the rare disease community.

THE IMPORTANCE OF UNDERSTANDING THE DISABILITIES EXPERIENCED BY PEOPLE WITH RARE DISEASES

As stated in the *UN Convention on the Rights of Persons with Disabilities*, persons with disabilities are those who have long-term impairments which, in interaction with various barriers, may hinder their full participation in society on an equal basis with others.

Recognising that people with rare diseases live with disabilities and understanding these disabilities is crucial to guarantee their rights.

A 2017 Rare Barometer survey showed that people with rare diseases face functional limitations, care coordination barriers and limited work participation.[1] However, evidence on the prevalence of disability among the rare disease community at large is scarce, and there is a need for further insight using disability measurement instruments.

Furthermore, while the heterogeneity of rare diseases has been documented, evidence of the variety, complexity and severity of disabilities faced by people with rare diseases is also lacking.

DISABILITY RECOGNITION AND OTHER POTENTIAL BARRIERS TO OBTAINING ADEQUATE SUPPORT

Disability assessment and recognition processes are an entry door into obtaining social protection and various types of support. With disability assessments being traditionally medically based in multiple countries, it is tempting to assume that people with rare health conditions would face adequate assessments.

Findings of the 2017 Rare Barometer survey[1] showed that:

- People with rare diseases who needed disability assessments could not always access them.
- About one-third of those who had been through a disability assessment declared that their disabilities were underestimated.
- People with rare diseases had limited access to different types of support, including disability benefits and personal assistance.

Several issues remained to be understood regarding:

- The outcomes following disability assessments, both in terms of disability recognition and support obtained.
- The various barriers within the disability assessment process itself to appreciate why they are failing to capture and support the individual needs of people with rare diseases.
- Whether people with rare diseases have access to specific independent living support, including personal assistance.
- And, last but not least, as Europe advances with deinstitutionalisation measures, evidence is lacking on living arrangements within the community and, more importantly, on living arrangement preferences.

SOCIAL PARTICIPATION: SO MUCH LEFT TO UNCOVER

The World Health Organisation (WHO) defines social participation as involvement in various life situations, whether in the labour market, school, sports or recreational activities.[2]

Research on rare diseases and social participation has mostly focused on work participation among people living with rare diseases such as cystic fibrosis, haemophilia, or Marfan syndrome and problematics such as work participation rate, work disability rate, work participation determinants, productivity loss or individual' perspectives, mainly in a single country.[3]

Some studies on social participation among people with disability also focused on children and young people with rare and more common diseases.[4–12]

The 2017 Rare Barometer survey looked into some aspects of work participation, with two of its findings being that most people with rare diseases and family members had to stop or reduce professional activity due to the rare disease and were limited in their professional choices.[1]

These findings demonstrate the need for more evidence of various aspects of social participation for all people with rare diseases, using more sophisticated instruments, gathering answers from more participants and allowing considerations for national or regional contexts. [3]

CAPTURING THE VOICES AND EXPERIENCES OF THE RARE DISEASE COMMUNITY

In the face of the remaining evidence gaps, this survey looked into the disabilities that people with rare diseases live with and the challenges they face in obtaining disability recognition and independent living support.

It also explored the limitations they face in participating in society, including at school and work.

Across issues, the survey analysis also aimed at identifying possible variations between European subregions, differences for those with and without disability, gender disparities, and differences between adults and children and adolescents living with rare diseases.

1. DISABILITY

1.1. MOST PEOPLE WITH RARE DISEASES LIVE WITH DISABILITIES

8/10 people with rare diseases live with disabilities

Washington Group Short Set on Functioning (WG-SS): 87% of the participants had 'some difficulties', 'a lot of difficulties' or 'could not at all' see, hear, walk, remember/concentrate, selfcare (dressing or washing over) or communicate; Global Activity Limitation Index (GALI): 83% of the participants were limited or severely limited in performing activities that people usually do because of a health problem during the last 6 or more months; self-identification: 88% of the participants considered themselves as a person with a visible disability, an invisible disability or both. All participants (n=9591).

Eight out of ten people with a rare disease live with disabilities. As formulated by the UN Convention on the Rights of Persons with Disabilities (UNCRPD) and the World Health Organization (WHO), disability is an umbrella and multifaceted term. According to Article 1 of the UNCRPD, persons with disabilities include those who have long-term physical, mental, intellectual or sensory impairments which, in interaction with various barriers, may hinder their full and effective participation in society on an equal basis with others. The barriers faced can be, for example, participation restrictions and environmental factors.[13,14]

Different instruments can be used to measure disability prevalence in census or surveys, depending on the purpose of measurement (providing services, monitoring population functioning, equalising opportunities).

In this study, we have used three different instruments to measure disability prevalence (**Figure 1**):

- *The Washington Group Short Set on Functioning (WGSS)*, which measures disability through functioning difficulties. This report uses the WGSS as the primary indicator to compare people with and without disability in bivariate and multivariate analyses.
- *The Global Activity Limitation Index (GALI)*, which captures disability through participation restrictions. In this report, the GALI will be used to compare our sample and the general population of the EU, in alignment with EUROSTAT's European social surveys.
- *A self-identification (SI) approach* consisting of directly asking participants if they have a disability.

1.1.1. THE WASHINGTON GROUP SHORT SET ON FUNCTIONING (WGSS)

The WGSS measures functioning difficulties in six activities (or domains): seeing, hearing, walking or climbing stairs, remembering or concentrating, self-care, and communicating. Participants could select one of four response categories: *no difficulty*, *some difficulty*, *a lot of difficulty*, or *cannot do at all*.

In this survey, the disability prevalence as measured by the WGSS is **87% when we consider people with *some difficulty*, *a lot of difficulty*, or who *cannot do at all* in at least one domain.** It is 46% if we only consider those with *a lot of difficulty* or who *cannot do* at least one domain.

1.1.2. THE GLOBAL ACTIVITY LIMITATION INDEX (GALI)

GALI is used in European Social Surveys (ESS) as a proxy for measuring disability through restrictions in participation. It is a single-item instrument developed to measure long-standing health-related activity limitations. It has three response categories: *severely limited*, *limited but not severely*, and *not limited at all*. **83% of participants reported having moderate or severe activity restrictions**.

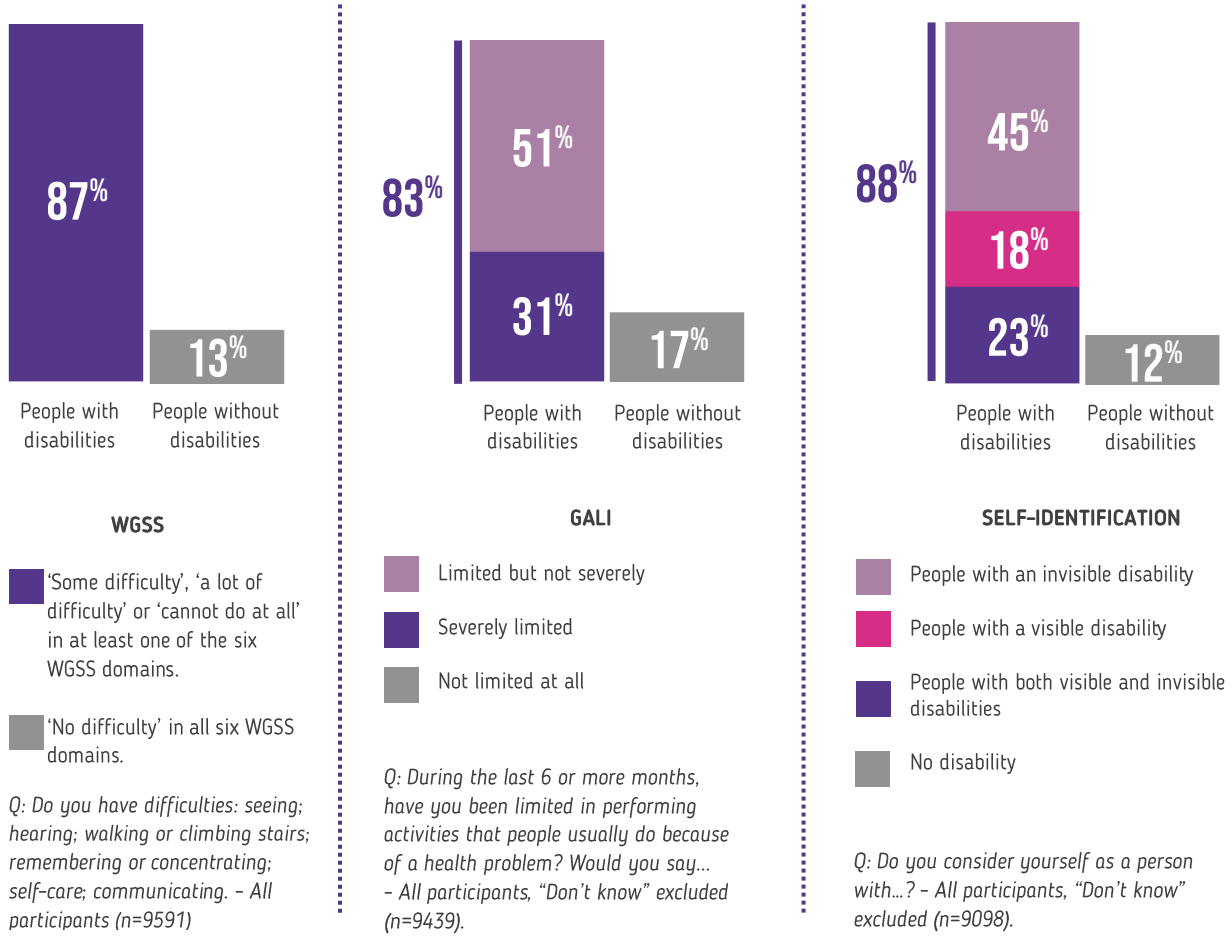
1.1.3. SELF-IDENTIFICATION

Lastly, participants were asked if they perceived themselves as a person with a disability and if their disabilities were visible, invisible, or both visible and invisible. In our survey, **88% of participants self-identified as living with a disability (invisible, visible, or both visible and invisible)**.

Self-identification is a subjective measure that, in some contexts, can lead to misestimating the prevalence of disability due to contextual, identity, and cultural issues that influence how a person may self-identify. To illustrate that, 29% (309/1051) of participants who did not self-identify as a person with disabilities have nonetheless declared living with functioning and/or activity limitations (as measured with the WGSS and the GALI). It is important to note that some people may consider disability as stigmatising. In addition, depending on their support networks, the advocacy campaigns they have been exposed to, and their hopes in their health and welfare systems, people with rare diseases may identify more as a person with a disability, more as a patient, or both.

Of those who self-declared a disability, 17% (1379/7961) were overlooked by the WGSS or GALI. This could be due to the limitations of these instruments in capturing societal barriers, as well as invisible or dynamic disabilities. For example, neither the WGSS nor the GALI fully capture pain and fatigue, while the GALI also focuses on the period of "the last six or more months". Lastly, the GALI focuses on limitations "due to a health problem", which puts a negative focus on health conditions with which some people may not identify.

Figure 1. Disability rates among people with rare diseases, according to the Washington Group Short Set on Functioning (WGSS), Global Activity Limitation Index (GALI) or self-identification question



***Source**: Rare Barometer survey conducted July-Sept. **Source**: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

1.1.4. DISABILITY RATES ARE HIGH ACROSS EUROPEAN SUBREGIONS

Disability rates, as measured by the WGSS, are quite similar across all European subregions (see the geographical distribution of the sample page 34), although they are slightly higher in Eastern and Central Europe and in Western Asia (Türkiye and Cyprus). However, the association between the two variables is weak (p-value=0.1; chi2=9).

There is a stronger association between the self-identification indicator and the European subregions (p-value<0.01; chi2=31.7). In other words, how people self-identify regarding disability is influenced by the European subregions they belong to. Self-declared disability is lower when measured in Eastern and Central Europe and Western Asia. In those two regions, people with rare diseases are less likely to self-identify as living with a disability, possibly due to historical, cultural and societal norms or attitudes (**Table 1**).

Table 1. Disability rates in European Subregions (UN Geoscheme – see p. 34)

	WGSS *Do you have difficulties: seeing; hearing; walking or climbing stairs; remembering or concentrating; self-care; communicating: 'some difficulties', 'a lot of difficulties' or 'cannot do at all'.*	**GALI** *During the last 6 or more months, have you been limited in performing activities that people usually do because of a health problem? 'Limited but not severely' or 'severely limited'.*	**Self-identification (SI)** *Do you consider yourself as a person with: "invisible disabilities', 'visible disabilities', 'both visible and invisible disabilities'*
Western Europe	87% (3107/3752)	83% (2946/3537)	**89% (3023/3405)**
Southern Europe	86% (2120/2467)	**78% (1895/2427)**	**89% (2106/2359)**
Northern Europe	88% (1436/1637)	**86% (1393/1618)**	**89% (1391/1557)**
Eastern and Central Europe	89% (936/1046)	84% (854/9591)	85% (825/973)
Western Asia	88% (344/392)	84% (309/367)	**82% (287/352)**
TOTAL	**87% (7943/9114)**	**83% (7397/8969)**	**88% (7632/8646)**

All participants who declared their country of residence; "Don't know" excluded: totals are not equal between categories because of the varying number of answers 'don't know'. Under-represented elements; over-represented elements. ***Source****: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

1.2. PEOPLE WITH RARE DISEASES LIVE WITH DIVERSE AND COMPLEX DISABILITIES

1.2.1. HETEROGENEITY

People with rare diseases live with different types of disabilities, as shown by participants having difficulties:

- 67% walking or climbing steps.
- 61% concentrating or remembering.
- 47% with self-care, such as washing all over or dressing.
- 42% seeing, even if wearing glasses.
- 33% communicating using their usual language, such as understanding or being understood.
- 20% hearing, even if using hearing aids.

In addition, **95% of participants experienced pain or fatigue** at least some days in the past three months, and **70% of the participants self-reported that they lived with an invisible disability**. Pain and fatigue can be manifestations of invisible disabilities, but they are common to all types of disabilities.

In our study, we found a positive relationship between the nature of the disability (invisible vs. visible) and the pain and fatigue experienced by people with rare diseases: 92% of people with visible disabilities, against 97% of people with invisible disabilities, experienced pain or fatigue at least some days (p-value<0.01; chi2=297.3).

1.2.2. COMPLEXITY

People with rare diseases live with diverse disabilities (e.g. sensory, motor, intellectual) which can accumulate with each other and with added manifestations, such as pain and fatigue, and other dynamic or progressive symptoms.

A majority of participants had difficulties with 2 or more activities, as measured by the WGSS:

- 72% of participants had difficulties with 2 or more activities. For example, 34% (3240/9591) had difficulties seeing and walking/climbing steps, and 9% (907/9591) had difficulties hearing and communicating using their language.
- 53% had difficulties in 3 or more activities.
- 35% had difficulties in 4 or more activities.

64% of participants had transient difficulties (that occurred during acute episodes, periodic crises or relapses) or worsening (progressive) manifestations. The percentage of participants with transient difficulties was the highest among people having difficulties remembering or concentrating (**Table 2**).

In total, 54% of participants had permanent difficulties, while 46% of participants with hearing difficulties and 43% of participants with seeing difficulties had permanent disabilities.

Table 2. Percentage of people with transient, worsening, permanent and improving difficulties per domain of WGSS.

	Transient difficulties (occurring during acute episodes, periodic crises or relapses)	Worsening (progressive) difficultuies	Permanent difficulties	Improving difficulties	Total
Walking or climbing stairs	25% (1580/6432)	31% (1976/6432)	39% (2515/6432)	6% (361/6432)	**100% (6432)**
Remembering or concentrating	37% (2133/5813)	21% (1210/5813)	38% (2216/5813)	4% (254/5813)	**100% (5813)**
Self-care	24% (1108/4542)	25% (1142/4542)	42% (1926/4542)	8% (366/4542)	**100% (4542)**
Seeing	29% (1196/4100)	25% (1036/4100)	43% (1760/4100)	3% (108/4100)	**100% (4100)**
Communication	30% (945/3199)	18% (580/3199)	40% (1294/3199)	12% (380/3199)	**100% (3199)**
Hearing	26% (498/1917)	26% (498/1917)	46% (887/1917)	2% (34/1917)	**100% (1917)**

***Question**: WGSS 'Do you have difficulties…' (in row) ; 'Is the difficulty…' (in column). **Source**: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

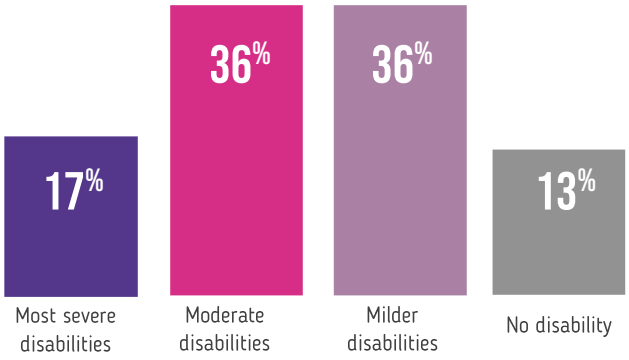
1.2.3. SEVERITY

Per the WGSS severity indicator, 15% of participants lived with severe disabilities.

Severity is captured here through the Washington Group Short Set (WGSS) by a score that simultaneously considers the difficulty level and the number of domains in which people experience those difficulties. Each difficulty level is attributed a score (no difficulty=0, some difficulties= 1, a lot of difficulties=6 and cannot do at all= 36), and those scores are summed up through domains. The total score is then classified in four categories (**Figure 2**):

- Individuals with no difficulties in any of the six functioning domains were labelled with **'No disability'**. Score = 0.
- Individuals with 1 to 4 functioning domains coded as *some difficulties* were labelled as living with '**Milder disabilities'**. Score = 1 to 4.
- Individuals with 5 to 6 functioning domains coded with *some difficulties* or up to 3 domains coded with *a lot of difficulties* were labelled as living with '**Moderate disabilities'**. Score = 5 to 23.
- Individuals with 4 or more functioning domains coded with a *lot of difficulties*, or with any domain coded *cannot do at all*, were labelled as living with **'Most severe disabilities**. Score= 24 to 216. For this group, we can observe that severity is a large spectrum: people labelled as living with severe disabilities had either four domains coded *a lot of difficulties*, or at least one domain coded *cannot do at all*.

Figure 2. Distribution of participants by level of disability severity according to the WGSS



All participants (n=9591). ***Source****: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

Box 1. Disability prevalence in the general European population and among people with rare diseases (GALI)

In the European Union (EU), disability-related data is collected by Eurostat during the European Health Interview Survey (EHIS) and the EU Statistics on Income and Living Conditions (EU-SILC) survey, using the GALI. Eurostat estimates that in 2023 in the EU population between 16 and 64 years old:

- **18.6% were living with a disability and 4.4% were living with severe disabilities**.
- Disability prevalence was higher among women (19.9%) than men (17.3%).

In our survey, **84% of participants between 16 and 64 years old were living with disabilities,** and **33% were living with severe disabilities** (GALI), which is significantly higher than in the general EU population.

2. DISABILITY RECOGNITION

Disability assessment and recognition processes are often necessary for people with disabilities to access different services, support and benefits that ultimately increase their participation in society.

An assessment does not necessarily imply adequate support. Even when the assessment is adequate, there might still be a gap between needs and benefits. In addition, disability entitlements are never really acquired and political changes or economic fluctuations may impact assessment rules leading to, such as changes in disability thresholds established for eligibility to certain types of support.

2.1. DISABILITY ASSESSMENTS ARE NOT PROVIDED TO ALL PEOPLE WITH RARE DISEASES WHO NEED THEM

Participants were asked if they had already undergone a disability assessment, which aims to assess and recognise disabilities. Overall, **15% (1435/9332) of the participants had never undergone a disability assessment, although they needed it**. Such lack of access to disability assessment may hinder their access to support.

Among the 57% (5364/9332) of participants who had already undergone a disability assessment, 61% (3268/5357) had multiple assessments.

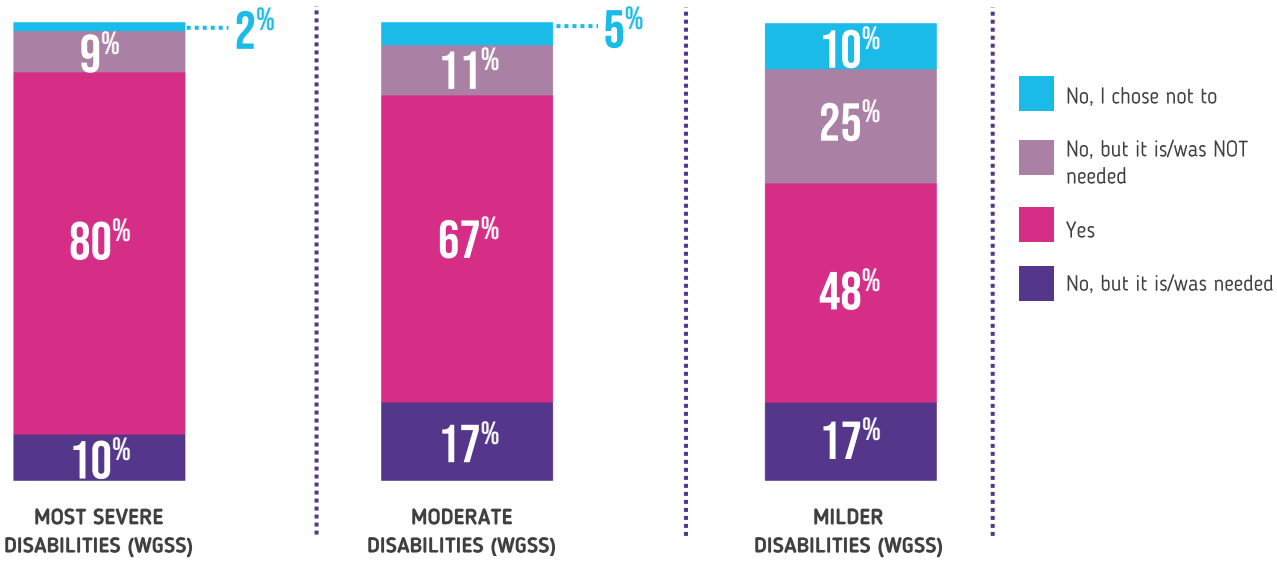
The assessments were conducted to determine participants' eligibility for various forms of support, including disability certificates, cash benefits, pensions, mobility aids, assistive technology, school or workplace adaptations, personal assistance, housing support, and referrals to community services.

People with more severe disabilities, as measured by the WGSS severity indicator, were more likely to have had disability assessments (p-value ≤ 0,01; Chi2 = 1,187.0): 80% of the participants with severe disabilities had undergone a disability assessment (**Figure 3**).

People with an invisible disability were less likely than those with a visible disability to have undergone a disability assessment: they were respectively 51% and 76% (p-value ≤ 0,01; Chi2 382.6).

Women with rare diseases were less likely to have undergone a disability assessment than men with rare diseases: they were 52% and 55%, respectively (p-value=0.0; chi2=11.3). Also, women were more likely than men to live with an invisible disability: they were 56% and 45%, respectively (p-value<0.01; chi2=105.7).

Figure 3. Access to disability assessment per level of severity of the disabilities (WGSS).



Participants with disabilities as per the WGSS (n=9332) ***Question****: Have you ever undergone a disability assessment, which aims to assess and recognise disabilities? Answers 'Don't know' excluded.* ***Source****: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

2.2. PEOPLE WITH RARE DISEASES FACE BARRIERS DURING DISABILITY ASSESSMENT

2.2.1. THE OUTCOMES OF DISABILITY ASSESSMENTS WERE BELOW EXPECTATIONS

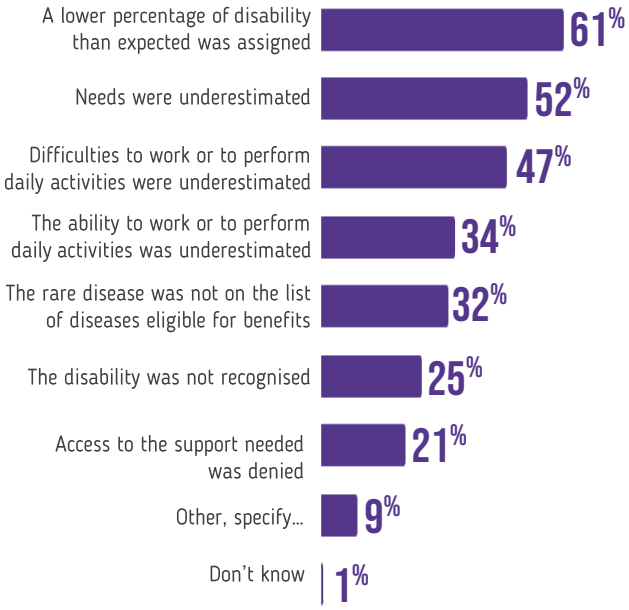
57% (5364/9332) of participants had already undergone a disability assessment. After the disability assessment, 16% of those were denied benefits and support, while 47% had only access to partial benefits.

31% of the participants submitted to disability assessments did not have the results they expected after the assessment (1607/5120), and the main three reasons were (**Figure 4**):

1. They were assigned a lower percentage of disability than expected (61%),
2. Their needs were underestimated (52%),
3. Their difficulties working or performing daily activities were underestimated (47%).

It is important to note that **25% did not have their disabilities recognised**.

Figure 4. Reasons why participants think they did not have the outcome they expected after the disability assessment



Participants who did not have the outcome they expected after the disability assessment (n=1607). ***Source****: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

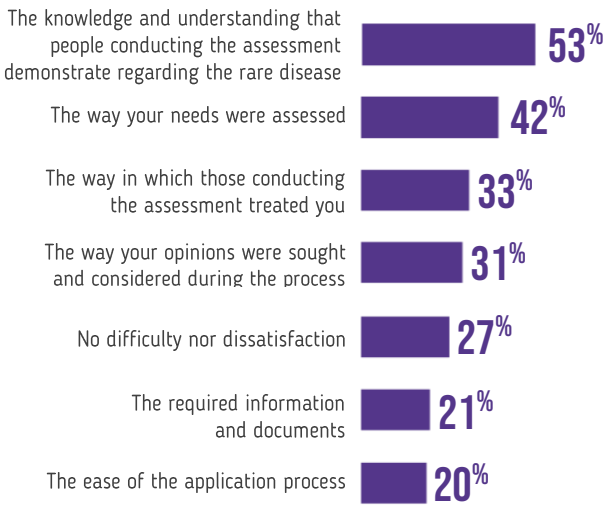
2.2.2. DISABILITY ASSESSMENT PROCESSES ARE COMPLEX AND LENGTHY

Only 27% of the participants who had a disability assessment encountered no difficulties or dissatisfaction with the disability assessment process. Figure 5 shows that the main difficulties or reasons for disatisfaction regarding their disability assessment were:

1. The assessors' lack knowledge regarding their rare disease (53%), despite medical information being the most requested evidence during disability assessment.
2. The way their needs were assessed (42%).
3. The way they were treated during the process (33%).

For those whose disabilities have been recognised as of today, **it may have taken several disability assessments and appeals to obtain such recognition**. This is shown in testimonials from the survey's open text question, 'Please tell us more about your experience with disability assessment: what went wrong? What went well and why? How could it have been better? What improvements would you suggest for the process?'.

Figure 5. Difficulty or dissatisfaction experienced during the assessment.



Participants who had a disability assessment (n=3626) ***Question****: 'During the assessment, did you/they have difficulty or dissatisfaction in:"* ***Source****: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

"*The correct classification, recommended by doctors and other specialists after rehabilitation, was rejected twice by the State Administration Office. I only received the correct classification after filing a complaint with the Social Court. The judge was appalled that the office had denied me the classification for several years.*"
Person living with a rare disease, Germany

"*The entire process lasted 15 months, included three appeals, all rejected, and ended with the establishment of the degree of disability by court order.*"
Person living with a rare disease, United Kingdom

"*The first review was negative. They only gave him 10%. We had to appeal, and he finally got 37%.*"
Family member, Spain

"*It was necessary to file an appeal, because the initial impulse is to reject the request, and only those who cannot give in and appeal have a chance.*"
Person living with a rare disease, Czech Republic

2.2.3. DISABILITY ASSESSMENTS ARE MAINLY MEDICALLY BASED BUT INSUFFICIENTLY INFORMED ON THE RARE DISEASE

Disability assessments experienced by participants were mostly medical, both in terms of the information they had to provide and of the professionals involved. **97% (5128/5306) of those who had a disability assessment were asked to provide medical evidence.** This is the most common evidence requested during the assessment and the most requested information in all countries.

The **dominant involvement of medical doctors and healthcare professionals** confirmed the medical approach of disability assessment (**Table 3**).

Table 3. Professionals involved in disability assessment by country.

What kind of professionals were involved in the assessment?	Countries where the answer is over-represented *Percentage of participants who selected the answer in the country*	Countries where the answer is under-represented *Percentage of participants who selected the answer in the country*	Total
Medical doctors only	Greece (82%), Portugal (82%), Austria (71%), Latvia (62%), Serbia (60%), Türkiye (58%), Belgium (55%), Italy (55%), Germany (52%)	Spain (24%), Finland (20%), Norway (19%), Ireland (14%), Denmark (13%), United Kingdom (10%), Sweden (8%)	43% (n=1972)
Several healthcare professionals (doctors, nurses, therapists…)	Russia (54%), Norway (51%), Ireland (40%), Finland (34%), Sweden (34%), France (27%), Romania (27%),	Portugal (12%), Spain (10%), Czech Republic (10%), Greece (8%), Croatia (5%)	19% (n=868)
Healthcare professionals (doctors, nurses, therapists…) and non-healthcare professionals (social workers…).	Spain (63%), Denmark (63%), Sweden (53%), Finland (45%), Croatia (45%), Czech Republic (43%)	France (29%), Belgium (26%), Italy (27%), Türkiye (24%), Germany (20%), Austria (13%), Latvia (11%), Greece (10%), Portugal (6%)	34% (n=1558)
Non-medical staff hired by private companies	United Kingdom (31%), Ireland (12%), Netherlands (7%)		3% (n=145)
Other			2% (n=92)

Participants who had a disability assessment and declared their country of residence, 'Don't know' excluded (n=4634). The relationship is very significant (p-value < 0.01, chi2= 1,465.6). ***Source****: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

Despite being medically based, disability assessments are not sufficiently informed about the person's rare disease and its manifestations. As shown in **Figure 5,** 53% of those who had a disability assessment experienced **difficulties with the knowledge and understanding the assessors demonstrated regarding the rare disease**. This difficulty is also visible in the responses to the survey's open question on participants' experiences with disability assessment.

> *"The evaluation staff knew almost nothing about my illness, and they didn't pay much attention to what I was saying."*
> **Person living with a rare disease, Italy**
>
> *"Doctors are unable to assess our daily life difficulties. They do not have training in rare diseases to carry out a correct assessment."*
> **Person living with a rare disease, Portugal**
>
> *"I was 18 years old. They looked at me and said, 'you are young, you have nothing'. They didn't see the x-rays and replied that they weren't orthopaedists and wouldn't understand anything. They didn't ask for a consultation with my doctor."*
> **Person living with a rare disease, Italy**
>
> *"The doctor told me that my file was too big to read and that since I was standing and had been able to come, I was not that sick."*
> **Person living with a rare disease, Belgium**
>
> *"Most of the time, my illness was unknown and equated with another illness that didn't fit at all."*
> **Person living with a rare disease, Germany**

Moreover, the **use of a list of diseases to determine disability status or eligibility for support creates barriers for people with rare diseases**. 32% of respondents submitted to a disability assessment stated that their rare disease was not on the list of diseases eligible for benefits when selecting reasons why they did not get the outcomes expected from the disability assessment (**Figure 4**). There are over 6000 rare diseases, many with very low prevalence or newly discovered, which makes it hard to ensure they are all included on such eligibility lists.

A medically based disability assessment might also create disparities between people who are diagnosed and those who are still on a diagnosis journey, which spans over 4.7 years on average for people with rare diseases.[15] Participants who were still undiagnosed were less likely to obtain recognition and to have full access to disability benefits after a disability assessment.

While there is a weak relationship between the diagnosis status and the access to disability assessment (p=0.1, chi2=7.6), we observed that compared to diagnosed participants, undiagnosed participants were less likely:

- To obtain disability recognition (45% vs. 24% for diagnosed participants, p-value<0.01, chi2=27.1).
- To obtain full benefit after an assessment (22% vs. 37% for diagnosed participants, p-value<0.01, chi2=31.3).
- To access any benefit after the assessment (24% vs. 16% for diagnosed participants, p-value<0.01, chi2=31.3).

The difficulties faced by people who are undiagnosed are reported in several testimonials from the survey's open text question on experiences with disability assessment:

> *"In the first evaluations, I had no medical reports with the diagnosis of my illness, despite my symptoms. In the last disability evaluation, I did have reports with the diagnosis; then the assessment was positive for 56% disability."*
> **Person living with a rare disease, Spain**

"The degree of disability was set at 30% because we have no diagnosis".
Family member, Germany

"First evaluation with unfavourable results - without diagnosis. Second evaluation was favourable - already with diagnosis."
Family member, Spain

Only **34% of participants who had a disability assessment reported the involvement of both healthcare and non-healthcare professionals**. **Table 3** shows the answer option that was the most selected by participants in each country regarding the type of assessors: participants in Sweden, Finland, Denmark, Poland, Croatia, Czech Republic, Switzerland, and Spain mainly reported multiple types of assessors.

2.2.4. PERSONAL SITUATION IS NOT SUFFICIENTLY CONSIDERED IN DISABILITY ASSESSMENTS

While most participants who underwent a disability assessment were asked to provide medical evidence, **only 60% (3176/5306) were asked about their daily activities, whereas only 53% (2834/5306) were required to provide information on their care and support needs,** and 28% (1524/5306) were asked to share information about their financial resources (the least requested information).

In addition, among those submitted to disability assessments, **42% reported having difficulties or dissatisfaction with the way their needs were assessed,** while 31% faced difficulties or dissatisfaction with the way their opinions were sought and considered during the process (**Figure 5**).

The lack of a holistic approach, considering the person's individual situation and environment, is also evidenced in the testimonials provided in response to the survey's open question on experiences with disability assessment.

"My opinion and my experience with the disease were not taken into account."
Person living with a rare disease, Spain

"There is a complete lack of consideration for the person, and therefore, the socio-cultural, psychological and holistic aspects are not taken into account."
Person living with a rare disease, Italy

"The assessment was too much based on legal procedures rather than on the person's own wishes and well-being."
Family member, Finland

"I was not personally seen or questioned for the disability classification. The decision was made based on the files."
Person living with a rare disease, Germany

"Experts are not interested in the fact that the child is unable to clean himself in the toilet, to fully dress, to wash himself. The experts see that his diagnosis is not on the list, and they do not care that the child remains helpless without care."
Family member, Latvia

It must also be noted that 33% of participants submitted to a disability assessment had difficulties or dissatisfaction with how those conducting the assessment treated them (**Figure 5**). Survey participants described various situations of impersonal, condescending and intimidating treatment:

"They didn't listen to me. They only looked at my file. They told me not to talk when I wasn't asked."
Person living with a rare disease, Greece

"They were rude, condescending, and didn't listen to my experience. They kept giving their uneducated version, even after multiple corrections by both me and my mum. They then used their inaccurate version of what they thought I could do instead of my actual ability to do things".
Person living with a rare disease, United Kingdom

"Everything is very impersonal, unfriendly and cold. You feel like an object. You are not informed about anything and do not know what is happening or how it works."
Person living with a rare disease, Germany

"People who evaluate do not hear the person. It could be improved if appraisers were more sensitive and reasonable, without prejudices and stereotypes."
Person living with a rare disease, Croatia

"The committee that was supposed to assess me was neither interested nor informed. The doctor was rude, unprofessional and humiliating."
Person living with a rare disease, Belgium

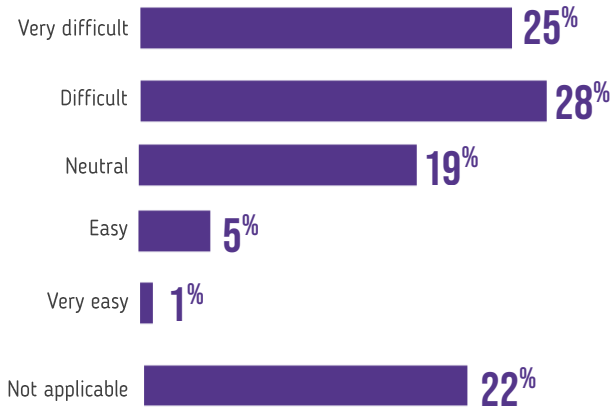
"Total lack of knowledge from the doctors. Impersonal, sometimes to the point of being intimidating."
Person living with a rare disease, Portugal

"The communication from the doctor was very condescending. Greeting: 'Just a little bit of voice, that won't get you a pension' and then 'your disease is so rare, why should you of all people have it?'."
Person with rare disease, Germany

3. INDEPENDENT LIVING

3.1. PEOPLE WITH RARE DISEASES FIND IT DIFFICULT TO ACCESS PUBLICLY FUNDED SUPPORT

Figure 6. How difficult do you find it to obtain state support such as attendant care, home support, financial support, assistive technology, mobility aids, etc.?



All participants? "Don't know" excluded (n=9308). ***Source****: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

53% (4956/9308) of participants found it difficult or very difficult to access publicly funded support (Figure 6), mainly because:

1. The application process was lengthy or complex (42%).
2. The disability was not considered severe enough (34%).
3. They lacked information about the application process (29%).

Results are similar for participants with disabilities: 55% found it difficult or very difficult to access publicly funded support for the same reasons: lengthy application process (43%), the disability not being considered severe enough (34%) and a lack of information about the application process (29%).

"I was made to feel that, despite being completely disabled, I was able to cope with my everyday life on my own. It is humiliating that I am not believed that I have to prove how bad I am, that I feel as if I am a welfare parasite."
Person living with a rare disease, Germany

"*I was in a relationship and lived with my partner and teenage daughter. They considered they could help me.*"
Person living with a rare disease, France

"I have been granted a disability certificate, but I still do not receive any kind of help. I have to work full time, even though it affects my health, and I have to force myself in my day-to-day life to be able to lead a normal life since nobody helps me."
Person with a rare disease, Spain

3.2. MOST PEOPLE WITH RARE DISEASES WHO NEED A PERSONAL ASSISTANT DO NOT HAVE ONE

Personal assistance refers to person-led human support available to someone living with a disability and is crucial to ensure that people can live independently in the community. Therefore, it is to be differentiated from provider-led assistance, characterised by discretionary decision-making regarding users' daily lives. Home care, or domiciliary care, for instance, differs from personal assistance.

32% (2957/9591) of the participants needed personal assistance, but most of them (62%) did not have one, while 38% of them had access to personal assistance.

3.2.1. PEOPLE WITH RARE DISEASES HAVE A LIMITED CHOICE OF PERSONAL ASSISTANTS, WHO ARE MOSTLY FAMILY MEMBERS

While the right to choose one's personal assistant is a pillar of independent living, **only 21% of the participants could choose their personal assistant** (**Table 4**). Among participants with personal assistance, that **role was ensured by a family member for 66%**.

Participants living with the most severe disabilities were more likely to have personal assistance: 34% of them had a personal assistant, against 18% of participants with milder or moderate disabilities (*p-value≤0,01; chi2=2062.7*).

Table 4. Do you have a personal assistant to help you live independently?

Yes	**12%** (1112/9458)
If yes, were you…	
…having a family member acting as a personal assistant?	66% (755/1,138)
…able to be the employer of the personal assistant using funding provided by public authorities?	28% (315/1,138)
…able to choose the personal assistant?	21% (244/1,138)
…having a personal assistant employed by public services?	12% (137/1,138)
…assigned a personal assistant?	8% (92/1,138)
…other, specify…	5% (55/1,138)
No, but it is/was needed	**20%** (1845/9458)
No, it was not needed / I chose not to	**68%** (6301/9458)

All participants (n=9458). ***Source****: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

3.2.2. PERSONAL ASSISTANTS ARE INVOLVED IN VARIOUS TASKS

Personal assistants support participants with a diverse range of tasks, mainly (**Table 5, column a**): household chores (76%), personal care (56%), social outings (55%), and transportation (49%). Those are also the main tasks that people who need a personal assistant but do not have one require support with, albeit with slightly different priority rankings (**Table 5, column c**).

45% (456/1006) of people with personal assistance declared they had no difficulties managing the personal assistant, while others **faced difficulties in managing the personal assistant:** 17% had difficulties due to their personal assistant being unable to work and not being replaced, 16% due to lack of training of the assistant, and 14% due to constant change of personal assistant.

3.2.3. PEOPLE WITH RARE DISEASES HAVE MORE HOURS OF SUPPORT WHEN THE PERSONAL ASSISTANT IS A FAMILY MEMBER

Half of people with rare diseases had at least 7 hours of personal assistance daily (median value) and an average of 10.9 hours per day (mean value). The number of hours depends on the type of personal assistance scheme (p-value≤ 0,01; Fisher= 22):

- 13 hours on average for participants whose family members ensured the role of personal assistant,
- against 7.8 hours on average when public services employed the personal assistant.

57% (529/924) of the participants who had personal assistance were satisfied or very satisfied with the number of hours, while 22% (200/924) were dissatisfied or very dissatisfied with the hours, and in particular:

- 19% of those who had family members as personal assistants were dissatisfied or very dissatisfied.
- 27% were dissatisfied or very dissatisfied among participants with publicly funded personal assistance.

Table 5. Tasks performed by personal assistants and support needs.

Access to personal assistance	Participants with personal assistants				Participants without personal assistance but who would need one	
Question	a. What tasks is the personal assistant currently assisting with? *(n=555)*		b. What tasks would you need assistance with and are not currently covered by the personal assistant? *(n=786)*		c. What tasks would you need assistance with? *(n=1789)*	
Tasks	Rank	Percentage	Rank	Percentage	Rank	Percentage
Household chores	**1**	**76% (424/555)**	**4**	**26% (205/786)**	**1**	**68% (1219/1789)**
Personal care	**2**	**56% (309/555)**	**3**	**27% (210/786)**	**4**	**48% (858/1789)**
Social outings	**3**	**55% (308/555)**	10	6% (49/786)	**2**	**51% (919/1789)**
Transportation	**4**	**49% (274/555)**	9	9% (71/786)	**3**	**51% (904/1789)**
Mobility around your place	5	37% (204/555)	6	16% (126/786)	8	21% (379/1789)
Medical care	6	35% (193/555)	**2**	**28% (223/786)**	5	39% (693/1789)
Other	7	13% (70/555)	7	12% (94/786)	10	10% (175/1789)
At the workplace	8	5% (25%/555)	5	25% (199/786)	9	13% (239/1789)
At school	9	4% (23/555)	9	9% (73/786)	6	23% (419/1789)
Extra school activities	10	3% (14/ 555)	**1**	**33% (259/786)**	7	23% (418/1789)

***Source**: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

3.3. MOST PEOPLE WITH RARE DISEASES ARE SATISFIED WITH THEIR LIVING ARRANGEMENTS

As emphasised in Article 19 of the Convention on the Rights of People with Disabilities (UNCRPD), persons with disabilities have the right to choose their place of residence on an equal basis with others, they have the right to choose where and with whom they live on an equal basis with other, and they should not be obliged to live in a particular living arrangement.

As such, our survey also aimed to assess the current living arrangements and, more importantly, the living arrangement preferences of people with rare diseases.

Table 6 shows that in the survey:

- 79% of participants lived with family, including parents, spouses, or partners; 17% lived alone; 1% lived in apartments with support services; and 1% lived in institutions.
- Among participants who were dissatisfied or very dissatisfied with their current living arrangements, 64% would have preferred to live with family, and 20% would have preferred to live alone.

Overall, 68% of participants were satisfied with their living arrangements, 14% were neither satisfied nor dissatisfied, and 18% were dissatisfied or very dissatisfied (this percentage is the same regardless of the living arrangements).

Most people living in apartments with support services (44%) had a severe disability (Table 7); those facilities were the ones with the highest proportion of people with severe disabilities, even more than in residential institutions (38%). Most of the participants living in residential institutions had moderate (40%) and severe disabilities (38%).

While they represented only 1% (143/9591) of the participants, **most people currently living in institutions would have preferred other living arrangements (68%),** such as living with family, alone or in apartments with support services. The remaining participants in institutions preferred their current living arrangements (32%).

Table 6. Current types of living arrangements, dissatisfaction and preferences.

Living arrangements	With family	Alone	Apartments with support services	Institutions	Other	Total
Current living arrangement	79% 7614/9591	17% 1624/9591	1% 63/9591	1% 143/9591	2% 147/9591	100% 9591
Preferred living arrangements among participants who are dissatisfied or very dissatisfied with their current living arrangement	64% 1074/1698	20% 334/1698	7% 118/1698	2% 26/1698	9% 146/1698	100% 1698

All participants, 'Don't know' not included (n=9591). ***Source****: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

Table 7. Percentage of participants living with disabilities and severe disabilities (as per WGSS severity indicator) per type of current living arrangement.

Current living arrangement	Percentage of participants with disabilities in each type of current living arrangement (n=8351)	Percentage of participants with SEVERE disabilities (WGSS) in each type of current living arrangement (n=1398)
With family (n=7614)	87% (6603/7614)	15% (1140/7614)
Alone (n=1624)	88% (1422/1624)	9% (146/1624)
Apartments with support services (n=63)	97% (61/63)	44% (28/63)
Institutions (n=143)	94% (135/143)	38% (54/143)
Other (n=147)	88% (130/147)	20% (30/147)

Source*: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

4. SOCIAL PARTICIPATION

Social participation is defined as a person's involvement in activities providing interactions with others in community life and important shared spaces, based on the societal context and what individuals want and is meaningful to them[16].

Areas of participation are diverse and include work, school, access to healthcare, as well as political, physical, and cultural activities. Our survey focuses on participation at work, school and in the community.

4.1. PEOPLE WITH RARE DISEASES FACE DISCRIMINATION

More than **1 out of 2 participants reported having already faced discrimination** in various contexts because of their disease or disabilities. This includes discrimination in healthcare (25%), in public places such as hotels, transport and museums (24%), in employment (20%) and in housing (6%) (**Table 8**).

Perceived discrimination was different depending on European subregions:

- It was significantly higher for healthcare in Northern Europe (33%) and in Eastern and Central Europe (29%)
- It was higher for education in Western Asia (33%), in Eastern & Central Europe (25%) and in Southern Europe (23%).

Table 8. Have you ever experienced discrimination related to the rare disease or disability?

	In healthcare	In other public accommodations (hotels, restaurants, transport, museums, etc.)	In education	In employment	In housing	Other, specify	In social environment (family, friends)	Never	Total
Western Europe	22% (754)	22% (747)	17% (592)	21% (725)	5% (166)	5% (185)	1% (35)	45% (1558)	100% 3428
Southern Europe	22% (526)	24% (581)	23% (558)	21% (495)	5% (129)	4% (107)	1% (27)	40% (964)	100% 2405
Northern Europe	33% (526)	25% (407)	16% (254)	21% (335)	7% (113)	7% (111)	1% (11)	41% (656)	100% 1597
Eastern & Central Europe	29% (292)	25% (246)	25% (250)	16% (164)	4% (43)	3% (25)	0% (2)	40% (399)	100% 996
Western Asia	18% (62)	22% (78)	33% (115)	11% (40)	8% (29)	4% (15)	1% (2)	42% (146)	100% 349
Total	**25% (2160)**	**23% (2059)**	**20% (1769)**	**20% (1759)**	**5% (480)**	**5% (443)**	**1% (77)**	**42% (3723)**	**100% 8775**

All participants who declared their country of residence, 'Don't know' excluded (n=8775). The relationship is very significant: p-value≤0.01; Chi2= 253.8; dof= 28. Under-represented elements; over-represented elements. ***Source****: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

4.2. WORK PARTICIPATION IS LIMITED AMONG PEOPLE WITH RARE DISEASES

4.2.1. EMPLOYMENT RATE IS LOWER AMONG PEOPLE WITH RARE DISEASES

Among working-aged participants (16-64 years old), **48% were employed, 23% were unemployed, and 13% were retired (Table 9).** In 2023, the EU employment rate was 75.3 %, while the unemployment rate was 6.1%.

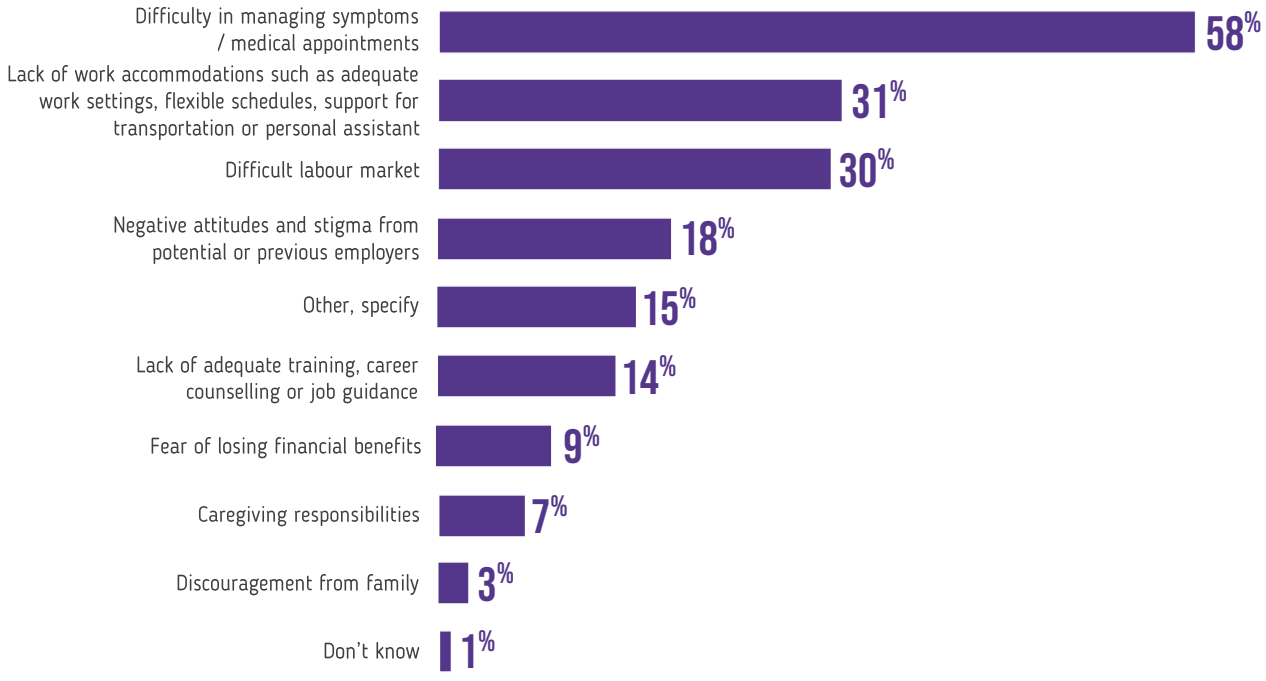
Figure 7 shows that although participants recognised the role of external factors as reasons for being unemployed, such as a difficult labour market (30%) or the lack of adequate training, career counselling or job guidance (14%). However, the main reasons they cited for being unemployed were related to their difficulties in managing symptoms and medical appointments (58%) and lack of work accommodations (31%). In addition, 18% referred to negative attitudes and stigma from potential or previous employers, while 3% were discouraged from working by family members (26/916). It is also important to note that 9% cited fear of losing financial benefits as a reason for being unemployed (75/916).

Table 9. Work-related situation

What is your current situation?	% (n)
Employed	**48%** (2567)
'Employed (or partially employed)'	44% (2329)
'Self-employed'	4% (238)
Unemployed	**23%** (1216)
'Unemployed'	13% (716)
'Cannot work because of the disease'	9% (500)
'Retired'	**13%** (714)
'Student/pupil'	**9%** (500)
Stay-at-home	**3%** (177)
'Not of school age yet.'	**0%** (4)
'Other'	**3%** (154)

Working-aged participants, 'Don't know' excluded (n=5332). ***Source****: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

Figure 7. Reasons for being currently unemployed (several answers possible).



Unemployed participants (n=707) ***Source****: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

4.2.2. LIVING WITH DISABILITIES IS A PREDICTOR OF UNEMPLOYMENT AMONG PEOPLE WITH RARE DISEASES

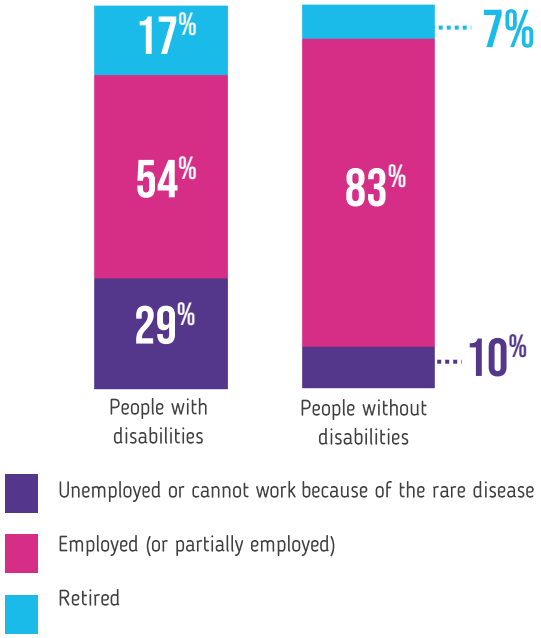
To better understand which characteristics predicted employment and unemployment the most, we conducted bivariate analyses (**Figure 8**) and multivariate analyses (**Table 10**) only among the active population, *i.e.* working-aged participants (16-64 years old) who were employed, unemployed, or retired: 57% (2567/4497) of them were employed, 27% (1216/4497) were unemployed, and 16% (714/4497) were retired.

Figure 8 shows a significant difference in employment rate depending on the disability status (p-value ≤ 0.01, chi2=159.8.):

- The employment rate was 54% among active participants living with disabilities and 83% among active participants without disability.
- **The unemployment rate was nearly 3 times higher among working-age active participants with disability** (29%) than among active participants without disability (10%), highlighting potential inequalities driven by disability status within the rare disease community.

Among participants living with severe disabilities, the employment rate was even lower: 24%. For this subsample, the unemployment rate (47%) and retirement rates (29%) were higher than the employment rate.

Figure 8. Employment status among people with and without disability.



Working-aged (16-64 y.o.) participants who were employed, unemployed or retired (n=4497). ***Source****: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

The average retirement age is 62.7 in our survey and 61.3 years in the EU: in our survey, 14% of active participants below 62 were retired, and 97% had a disability.

The logistic regression we conducted (**Table 10**) confirms that **disability status was one of the main determinants of employment for people with rare diseases**, along with age, geographic area, having or needing a personal assistant, having access to social benefits, having a strong social environment, and facing discrimination:

- Participants who reported a strong social environment (close ones or people to count on when needed) were more likely than participants with a poor social environment to be employed.
- Living in Eastern or Central Europe, in Northern Europe, or in Southern Europe also increased the likelihood of employment compared to living in Western Europe.
- People with disabilities, people who received social benefits (such as pensions, work and education accommodations, mobility aids, adapted housing or housing support), people who had or needed personal assistance, and people who were exposed to discrimination were less likely to be employed.

These findings might indicate that:

- Participants who had or who needed a personal assistant faced more barriers to employment than participants who did not need a personal assistant.
- Those barriers were not mitigated enough by their current social benefits and support. Besides, social benefits, such as certain pensions, are often attributed only to people who are not employed.

The education level, which is a usual predictor of employment, is not statistically significant in this study, confirming that the health determinants of people with more common diseases differ from those of people with rare diseases.[17]

Table 10. Logistic regression: determinants of employment.

Characteristics	Odd ratios (Standard Deviation)	Characteristics	Odd ratios (Standard Deviation)
European subregions (United Nations Geoscheme)		**Age**	**0.988***** (0.004)
Western Europe	ref	**Age when left school (education level)**	
Eastern and Central Europe	**3.739***** (0.792)	≤ 15 y.o.	Ref
Northern Europe	**1.284**** (0.148)	16 - 19 y.o.	0.747 (0.147)
Southern Europe	**1.247**** (0.130)	20 - 23 y.o.	1.070 (0.209)
Western Asia	1.506 (0.719)	≥ 24 y.o. or more	1.266 (0.248)
Disability (WGSS)		**Gender**	
No Disability	ref	Male	ref
Disability	**0.557***** (0.092)	Female	0.917 (0.098)
Personal assistance		**Social environment**	
No, but it is/was not needed	ref	Poor support	ref
Yes	**0.414***(0.071)	Intermediate	**1.405***** (0.153)
No, but it is/was needed	**0.414***(0.046)	Strong support	**1.783***** (4.200)
Social benefits		**Discrimination**	
No	Ref	Never	Ref
Yes	**0.275***** (0.025)	Yes	**0.751***(0.071)
Constant cut	12.742 (0.329)		
Observations	3,299		
R^2	0.15		

*Working-aged (16-64 y.o.) participants who were employed or unemployed (n=3299). Constant Cut or "cut point" values are defined by the ratio of cases below the cut point to cases above the cut point. OR Odd Ratios, R^2 Coefficient of determination, *** p<0.01, ** p<0.05, * p<0.1; Ref = Reference group.* ***Social environment****: 'How would you qualify your overall perceived social support (close people to count on, concern shown by other people, practical help from neighbours in case of need)?'.* ***Social benefits****: "Do you currently benefit from…"* ***Discrimination****: "Have you ever experienced discrimination related to your rare disease or disability?"* ***Personal assistant****: "Do you have a personal assistant to help you live independently?"* ***Source****: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

4.2.3. PEOPLE WITH RARE DISEASES FACE DISPARITIES IN THE LABOUR MARKET DUE TO GENDER OR DISABILITY STATUS

Most employed participants, excluding those who were self-employed, had a full-time contract (67%), while 28% had a part-time contract.

The relationship between gender and the type of contract is very significant (chi2= 47.2, Pr=0.000): women were more likely to have a part-time contract (32% vs 17%), while men were most likely to have a full-time contract (79% vs 63%).

In general, women are more likely to have precarious employment throughout their lives, when compared to men, regardless of their level of education.[18] In 2023, in the general EU population, the share of women between 20 and 64 years old with part-time employment was 27.9 against 7.7 for men. [19]

Disability status also had a significant impact on the type of contract (chi2(2) = 24.0, Pr=0.000), as fewer participants with disabilities had full-time contracts (65%) compared to participants without disabilities (76%).

Likewise, **participants with disabilities had more part-time contracts (30%) than people without disabilities (20%)**.

However, those disparities in gender or disability status might also be the expression of preferences, since among survey participants who were unemployed and were looking for employment, 70% (431/616) would have preferred part-time employment rather than full-time employment.

As an example, in 2023, the general EU population reported the following reasons conditioning their part time working arrangements:[19]

- Care of adults with disabilities or children (30% of female part-timers against 8% of male part-timers).
- No full-time job found (18% for women against 28% for men).
- Their own illness or disability (6% of women and 11% of men).

4.2.4. PEOPLE WITH RARE DISEASES HAVE LIMITED PARTICIPATION AT WORK, ESPECIALLY WHEN THEY HAVE DISABILITIES

64% (1942/3054) of the participants who were employed or self-employed declared fully participating at work, 29% (892/3054) declared that their work participation was somewhat limited, and 7% (220/3054) said that it was very limited.

Participation at work was worse for participants with disabilities: **only 59% of employed people with rare diseases and disabilities declared fully participating at work**, against 87% among participants living without disabilities (p-value ≤ 0.01, chi2=146.5).

4.2.5. PEOPLE WITH RARE DISEASES NEED MORE WORKPLACE ACCOMMODATIONS

73% (2195/3002) of employed participants had regular employment without support or workplace accommodations, **24% (732/3002) benefitted from support or accommodations in their workplace**, and only **2% (73/3002) were in sheltered employment** (organisations mostly employing people with disabilities).

The barriers to employment faced by people with rare diseases could be mitigated by workplace accommodations: **82% of participants (2460/3002) considered that work accommodations would improve their participation at work.**

Table 11 also shows that participants have a preference towards organisations that offer work accommodations over those providing sheltered employment or that do not provide support. **39% (1175/3017) of employed participants would prefer to work in an organisation that provides accommodation**. Only 24% of them currently benefit from it.

Sheltered employment seems to be the least attractive type of workplace setting: participants working in sheltered organisations were mostly living with disability (92%), **34% of those working in sheltered employment would have preferred regular employment**, 20% had no preference, and 46% preferred to stay in sheltered employment.

Some actionable measures were highlighted by participants when they were asked about factors that could improve their employment participation:

- **Work accommodations** including adequate work settings, flexible schedules, and remote work: 80% of employed participants.
- **Inclusion and positive attitudes** from colleagues and employers: 39% of employed participants.
- **Adequate training or career counselling**: 15% of employed participants.

Table 11. Current types of working organisations and preferences.

Work organisation	Current situation		Preferred situation		Difference	
	N	%	N	%	N	%
In regular employment without support or workplace accommodations	2195	73%	990	33%	1205	40%
In regular employment with support or workplace accommodations	732	24%	1175	39%	-443	-15%
In sheltered employment (employing mostly people with disabilities)	75	2%	120	4%	-45	-2%
No preference	-		732	24%		
TOTAL	**3002**	**100%**	**3017**	**100%**		

Employed and self-employed participants, 'Don't know' excluded (n=3080). ***Source****: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

4.3. MOST STUDENTS WITH RARE DISEASES DO NOT FULLY PARTICIPATE IN EDUCATION

School participation was measured through the corresponding module of the Child and Adolescents Scale of Participation (CASP). Questions were addressed directly to the 167 participants attending school or college (self-report) and the 1381 family members of people with rare diseases attending school or college (proxy-report).

The CASP module for school participation includes five items: academic activities, using educational materials, moving around at school, communicating with other students and adults, social play and recreational activities. For each item, participants were asked if, compared to people the same age, they (or the person living with a rare disease) had *full participation*, were *somewhat limited*, *very limited*, or *unable* to do the activities listed.

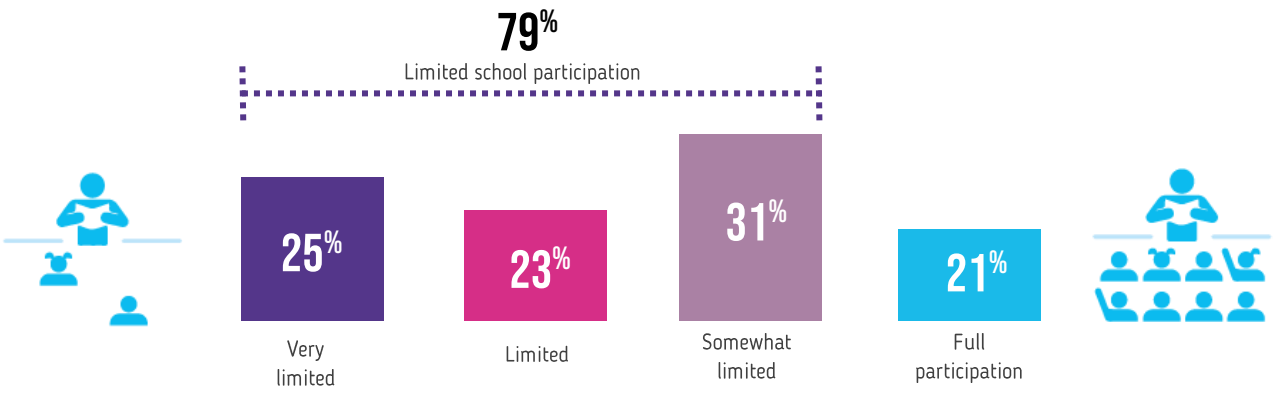
Individual scores were summed up for each item and divided by the maximum possible score, based on the number of items rated (5 scores in the school module). They were then multiplied by 100, ranging from 0 to 100.

The higher the score, the closer the participants were to age-expected full participation:

- **Full participation** (CASP score 100–97.5): pupils' or students' participation is equal to or greater than their peers, with or without assistive devices or equipment.
- **Somewhat limited participation** (CASP score 97.5–81.0): pupils and students participate in fewer activities than their peers and may need occasional supervision or assistance.
- **Limited participation** (CASP score 81.0–68.5): pupils and students participate in fewer activities than their peers and may need supervision or assistance.
- **Very limited participation** (CASP score 68.5 or less): pupils and students participate in activities much less than peers, and they may need a lot of supervision or assistance.

Figure 9 shows the percentage distribution of school participation: **only 21% of pupils and students with rare diseases reported full participation in school or college.** The rest reported participation restrictions, among which 25% reported severe participation limitations.

Figure 9. School participation as per the Child and Adolescent Scale of Participation (CASP).



Pupil and students (n=1548). ***CASP****: 'Compared to other people the same age, what is your current level of participation in: educational activities; recreational activities; mobility; use of educational material & equipment; communication?'.* ***CASP scores****: Full participation = 100-97.5; Somewhat limited=97.5-81.0; Limited= 81.0-68.5; Very limited=<68.5.* ***Source****: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

We also compared average school participation scores reported by students and pupils with rare diseases (self-report) and those reported by family members (proxy-report). The average and median scores of school participation are reported in **Table 12**.

Table 12. CASP scores of school participation among pupils and students.

People living with a rare disease (self-report) n=167	Median (IQR)	90 (80-95)
	Mean (SD)	85.6 (13.9)
Family members of people with rare diseases (proxy-report) n=1381	Median (IQR)	85 (65-95)
	Mean (SD)	78.7 (19.2)
Total n=1548	Median (IQR)	85 (70-95)
	Mean (SD)	79.4 (18.9)

Pupils and students (n=1548). Median = median value; IQR = Inter Quartile Range; Mean = average value; SD= standard deviation. ***Source****: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

Table 13. Scores of school participation per level of disability severity of pupils and students.

		School participation scores	
		Median (IQR)	Mean (SD)
Disability severity (WGSS)	No disability (n=271)	100 (95-100)	96.3 (6.8)
	Milder disabilities (n=465)	90 (80-95)	88 (12.3)
	Moderate disabilities (n=536)	75 (65-85)	74.9 (14.6)
	Most severe disabilities (n=276)	55 (45-70)	57 (18.7)

Pupils and students (n=1548). Median = median value; IQR = Inter Quartile Range; Mean = average value; SD= standard deviation. ***Source****: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

On average, students with a rare disease reported somewhat limited school participation (85.6), while family members reported limited school participation (78.7). The median self-reported score for school participation was 90 (IQR=80-95), and the median proxy-reported score was 85 (IQR=65.0-95.0).

School participation was worse when students with rare diseases lived with a disability. Pearson correlation (assuming normality of participation and severity scores) shows a moderately strong negative relationship between school participation and disability severity, with a coefficient of -0.6, meaning that participation scores vary significantly depending on disability severity.

Table 13 presents average and median participation scores by disability severity. The participation scores vary across levels of severity: the more severe the disability, the more important the participation restrictions. Personal, social, environmental, and policy factors influence participation at school. A strong support system, including not just family but also peers, is another facilitator, with family - notably parents' and carers' involvement - being a fundamental determinant of children's participation.[20]

Therefore, disability status is not the only determinant of participation at school, as illustrated in the linear regression reported in **Table 14** which shows the role of social environment, geographic area, age and access to personal assistance in school participation:

- The R-squared value of 0.35 indicates that all the independent variables (age, disability, personal assistance, disability, subregions, and social environment) together in the model explain 35% of the variation in school participation. This is statistically significant (Prob > F; 0.000).
- The regression coefficient for age indicates that as students and pupils age of one year, their average school participation increases by 0.17 (95% CI: 0.045-0.308; p<0.01), regardless of the remaining factors.
- The regression coefficient for disability status is -14.34 (95% CI:-17.306-11.886; p= 0.000), which means that the average participation score of participants with a disability was 14.34 less than that of participants without disability, regardless of the remaining factors.
- The positive regression coefficient of participants with a strong social environment means that the average participation score of participants with a strong social environment was 7.573 higher than those with a poor social environment. School participation of those with a fair social environment was higher than those with a poor social environment, but to a lesser extent (2.839).

Table 14. Linear regression: determinants of school participation (CASP - Child and Adolescent Scale of Participation).

Variables	School participation (CASP)	Variables [continued]	School participation (CASP) [continued]
Age	**0.17**** (0.06)	**EU subregions**	
Personal assistance		Western Europe	ref
No, but it is/was NOT needed	ref	Eastern and Central Europe	**0.330** (1.543)
YES	**-12.97***** (1.399)	Northern Europe	**-5.784**** (1.400)
No, but it is/was needed	**-15.02***** (1.143)	Southern Europe	-2.7601. (1.192)
		Western Asia	1.375 (2.183)
Disability (WGSS)		**Discrimination**	
Without disability	ref	Never	ref
With disability	**-14.34***** (1.402)	Yes	-0.918 (1.096)
Social environment			
Poor support	ref	**Constant**	79.441*** (2.454)
Intermediate	**2.839*** (1.30)	**Observations**	1,078
Strong support	**7.573***** (1.375)	**R-squared**	0.35

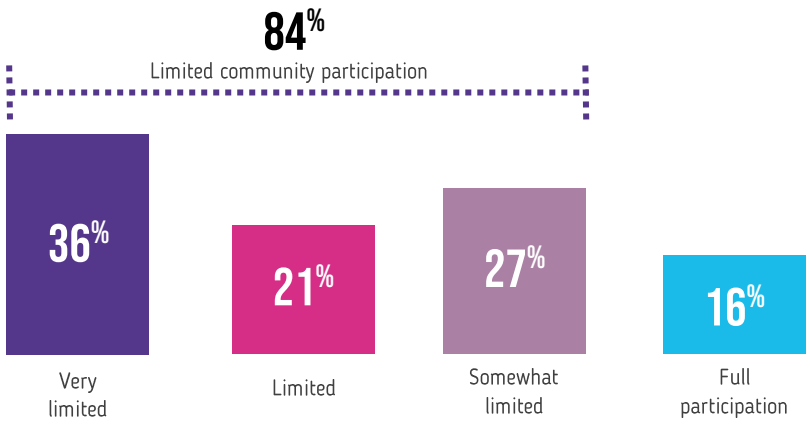
*Standard errors in parentheses; *** p<0.01, ** p<0.05, * p<0.1; Ref (-) Reference group.* ***CASP****: 'Compared to other people the same age, what is your current level of participation in: educational activities; recreational activities; mobility; use of educational material & equipment; communication?'* ***Social environment****: 'How would you qualify your overall perceived social support (close people to count on, concern shown by other people, practical help from neighbours in case of need)'.* ***Discrimination****: "Have you ever experienced discrimination related to your rare disease or disability?"* ***Personal assistant****: "Do you have a personal assistant to help you live independently?"* ***Source****: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

4.4. COMMUNITY PARTICIPATION IS LIMITED, ESPECIALLY FOR THOSE WITH DISABILITIES

The community module of the CASP evaluates children's and adolescents' involvement in the community. It includes four items: social/leisure (friends), structured activities, mobility, and communication.

The scoring strategy is similar to the one for school participation, presented on page 27. **84% of participants have very limited, limited or somewhat limited participation in the community** (**Figure 10**).

Figure 10. Community participation as per the community module of the CASP.



All participants, 'Don't know' and 'non applicable' not included (n=8692). ***Community participation module of the CASP:*** *'Compared to other people the same age, what is your current level of participation in: neighbourhood and community activities; social, play or leisure activities; structured events and activities; moving around; communicating with others?'. CASP scores: Full participation = 100-97.5; Somewhat limited=97.5-81.0; Limited= 81.0-68.5; Very limited=<68.5. Pupils and students (n=1548).* ***Source****: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

4.4.1. COMMUNITY PARTICIPATION IS LOWER AMONG ADULTS

Community participation was assessed for all participants, but we conducted a separate analysis for students and pupils (young people) to allow for comparison with other populations (see **Box 2**). The mean difference test showed a significant difference between community participation among young people and adults.

Young participants' average community participation score is 5 points higher than that of adults (for self-report). Similarly, proxy participants tend to declare a higher participation score for young people than adults (**Table 15**).

Table 15. Community participation scores among students and pupils (including homeschooled) and adults.

	Pupils and students		Adults	
	Median (IQR)	Mean (SD)	Median (IQR)	Mean (SD)
People living with a rare disease (self-report)	81.3 (68.8-93.8)	79.6 (16.7)	75 (62.5-87.5)	74.6 (18.8)
Family members of people with rare diseases (proxy-report)	75 (56.3-93.8)	72.3 (22.3)	62.5 (43.8-81.3)	62.1 (22.9)

Students and pupils (n=1675); adults (n=6838); "not applicable" excluded. Median = median value; IQR = Inter Quartile Range; Mean = average value; SD= standard deviation. ***Source****: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

Box 2. The CASP and participation restrictions of children, adolescents and young adults with acquired brain injury and developmental disorders.

Gary Bedell originally developed the **CASP for children and adolescents** with acquired brain injury (ABI). In a recent cohort study[22], Bedell et al. measured school and community participation among people with ABI in The Netherlands. They found:

- For school participation, self-report from pupils was on average 78.9, while proxy-report from parents was on average 88.4 (against 85.6 and 78.7 respectively in our survey).
- For community participation, self-report from pupils was on average 80, while proxy-report from parents was on average 71.1 (against 79.6 and 72.3 respectively in our survey).

Both studies show higher school participation than community participation (for both self-reports and proxy-reports). However, while proxy scores were higher than self-reported scores in school and community participation in the Bedell study, our study showed lower proxy scores than self-reported scores for school and community participation.

It has been reported that situations where parents overestimate participation are usual with the CASP, which authors attributed to the fact that activities usually occur outside of the home environment, especially for adolescents and young adults (school, concerts, etc.). It is worth noting that in Bedell's study, participation was reported by people with ABI and parents at the same time, allowing for accurate comparison between restrictions using both points of view. However, in our survey, participations are reported either by the person with a rare disease or by the family member and family members underestimated participation at school and in the community compared to those who self-reported. Therefore, the discrepancies between the two groups can be attributable to other factors, such as proxies answering for people with more severe conditions and, therefore, more participation restrictions.

Another study conducted in Spain showed a significant difference between children with and without developmental disorders (NDD) across all domains of participation (ranging from 1.7 to 5.5 difference points)[23]. Regarding school and community participation, people with rare diseases have on average slightly better participation (73.1 for community participation and 79.4 for school participation) than children with NDD (68.1 for community participation and 77.5 for school participation) but worse participation than children without NDD (96.3 for community participation and 97.8 for school participation).

4.4.2. A STRONG SOCIAL ENVIRONMENT INCREASES COMMUNITY PARTICIPATION

Table 16 shows that the main determinants of community participation were disability status, needing personal assistance, having a strong social environment, accessing social benefits, facing discrimination, and geographical area of residence. The R-squared value of 0.34 indicates that all the independent variables (age, disability, personal assistance, disability, subregions, and social environment) explain 34% of the variation in school participation, which is statistically significant (Prob > F; 0.000).

A strong social environment positively correlated with community participation. People with intermediate and strong levels of strong social enviroment scored respectively 9.3 and 5.8 points more that people with poor social enviroment. Social environment refers to close people to count on, concern shown by people, and practical help from neighbours in case of need.

The average community participation score of participants with disabilities was 12.182 less than that of participants without disabilities. This can be influenced by accessibility barriers, including in built environment, transportation, information and communication.

The average community participation score of participants who did not have personal assistance but who needed it was 12.393 points less than the average score of participants who did not need personal assistance. Likewise, the average community participation score of participants with personal assistance was 11.152 less than that of participants who didn't need personal assistance. This could be linked to the disability status, as 96% of participants with personal assistants had disabilities.

However, our results show that **although people who need personal assistance have lower community participation, those with a personal assistant are slightly better off than those do not have one.** Having a personal assistant might, therefore, reduce the gap in community participation when the number of hours is adequate and the relations with the difficulties to manage the personal assistant are minimised. In our survey, 22% of participants with personal assistance expressed dissatisfaction with the number of hours, and 55% had difficulties managing the personal assistant, which can also explain the low impact of having a personal assistant on community participation.

Average community participation was also lower for participants:

- With social benefits, such as pensions, work and education accommodations, mobility aids, adapted housing or housing support (-7.95); 95% of them were living with disabilities.
- Who experienced discrimination related to a rare disease or disability (-4.136).

Table 16. Linear regression: determinants of community participation (CASP).

Variables	Community participation	Variables	Community participation
Disability (WGSS)		**Social benefits**	
Without disability	ref	No benefits	ref
With disability	**-12.182*** (0.715)**	Benefits	**-7.95*** (0.480)**
Personal assistance		**Discrimination**	
No, but it is/was not needed	ref	Never faced discrimination	ref
Yes	**-11.151***(0.828)**	Faced discrimination	**-4.136*** (0.480)**
No, but it is/was needed	**-12.393*** (0.618)**		
Social environment		**European Subregions**	
Poor support	ref	Western Europe	ref
Intermediate	**5.843*** (0.594)**	Southern Europe	0.108 (0.543)
Strong support	**9.309*** (0.616)**	Eastern and Central Europe	-0.327 (0.882)
		Western Asia	-1.026 (2.336)
Age	**-0.101***(0.014)**	Northern Europe	**-3.193***(0.588)**
Constant	93.625 (1.300)		
Observations	**5,213**		
R-squared	**0.34**		

*Standard errors in parentheses; ***p<0.01, **p<0.05, *p<0.1; Ref = Reference group.* ***CASP****: 'Compared to other people the same age, what is your current level of participation in: neighbourhood and community activities; social, play or leisure activities; structured events and activities; moving around; communicating with others?'* ***Personal assistant****: "Do you have a personal assistant to help you live independently?"* ***Social environment****: 'How would you qualify your overall perceived social support (close people to count on, concern shown by other people, practical help from neighbours in case of need)?'* ***Social benefits****: "Do you currently benefit from…"* ***Discrimination****: "Have you ever experienced discrimination related to your rare disease or disability?"* ***Source****: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

Table 17. Average community participation score per European subregions.

European subregions (UN geoscheme)	Community participation
Western Europe	75.1
Southern Europe	72.9
Eastern and Central Europe	71.0
Northern Europe	69.7
Western Asia	61.8

p-value≤0.01; Fisher=45.4 Source: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.

Table 17 shows that **community participation was significantly lower in Northern Europe than in other European subregions** (-3.192 compared to Western Europe), even if the raw average score for Western Asia was lower than for Northern Europe. While we expected disparities due to differences in healthcare infrastructure, socioeconomic factors, school inclusion, accessibility, and cultural attitudes toward disabilities, better scores in Northern Europe could have been assumed given the influence of the Nordic model, characterised by a comprehensive welfare system. Lower scores in Northern Europe could be due to cultural reasons or higher expectations for full participation. This would lead people with rare diseases to score lower in community participation, especially since the CASP assesses participation relative to other people of the same age.

4.5. MOST PEOPLE WITH RARE DISEASES HAVE A POSITIVE OPINION OF THEIR SOCIAL ENVIRONMENT

Only 23% (2080/9227) of participants felt that they had a poor social environment based on the presence of people they could count on, concern shown by other people, or practical help from neighbours in case of need.

As shown in **Figure 11,** the level of perceived social environment was different depending on disability status (Error! Reference source not found.p-value≤ 0,01; Chi2= 112.9; dof= 2). However, there is no significant relationship between disability and involvement in voluntary work (p-value=0.8, Chi2=0.1).

Figure 11. Level of perceived support from the social environment by disability status.

*All participants (n=9227. **Question**: How would you qualify the social support you receive from people (close people to count on, concern shown by other people, practical help from neighbours in case of need)?. Answers "don't know" are excluded. **Source**: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

Box 3. The GALI and Social Participation: voluntary work and overall social support

In the general EU population, the level of social participation is assessed in the SILC survey through voluntary work and social support. In 2019, 31.9% of people with disabilities (as measured by the GALI) in the EU perceived a strong level of social support, 48.1 % perceived an intermediate level of support, and 20% perceived a poor level of support. As for people without disabilities, 40% declared receiving a strong level, 48.5% an intermediate level and 11.5% a poor level of social support. [24]

The question on voluntary work has been altered in our survey: while the SILC distinguished between formal and informal voluntary work and active citizenship, our question referred to "unpaid, not compulsory work or informal activities". Nonetheless, 12.3% to 14.3% of the EU population were involved in formal or informal voluntary activities in 2022, and these percentages were slightly lower for those with disabilities in the EU (10.3% to 13.3%). In our survey, 31% (2364/7681) of people with disabilities were volunteering.[25] The higher involvement in voluntary activities in our survey might be inherent to the rare disease community's context and the fact that questionnaires were disseminated through and by networks of civil society organisations representing people with rare diseases. It should also be noted that 23% of our participants identified as patient representatives, being involved in in voluntary and/or policy activities to support the cause of rare diseases.

5. METHOD

5.1. QUESTIONNAIRE

5.1.1. DESIGN

The survey was based on a self-administered online questionnaire (using Sphinx software) elaborated based on a literature review and consultations with:

- Fourteen members of a Topic Expert Committee, who have contributed to clarifying the objectives of the survey and the topics to include in the questionnaire.
- EURORDIS national alliances, representing people with a wide range of rare diseases in each country, and EURORDIS European Federations, representing specific rare diseases in several countries, who gave their opinions and feedback on the questionnaire.

The questionnaire was written in English and translated by professional translators specialised in health-related issues into the following 25 languages: Bulgarian, Croatian, Czech, Danish, Dutch, Finnish, French, German, Greek, Hungarian, Italian, Latvian, Lithuanian, Norwegian, Polish, Portuguese, Romanian, Russian, Slovak, Slovenian, Spanish, Swedish, Ukrainian, and Turkish. Seventeen translations were reviewed: French, German, Spanish, Czech, Danish, Finnish, Greek, Hungarian, Italian, Dutch, Norwegian, Romanian, Sweden, Slovak, Slovenian, Ukrainian and Turkish. Native speakers with expertise in rare diseases, disability and/or disability policies reviewed the translations to check their cultural validity and consistency with the original English version.

MEMBERS OF THE TOPIC EXPERT COMMITTEE

Valentina Bottarelli, Head of Policy and Public Affairs, EURORDIS-Rare Diseases Europe. **Petra Bruegmann,** President, European MEN Alliance; member of EURORDIS' Social Policy Action Group (SPAG). **Dorica Dan,** President, Romanian National Alliance for Rare Diseases; President, Romanian Prader-Willi Association; member of SPAG. **Jakub Gietka,** President, Aiming for the Future Foundation; member of SMA Europe's Adult Committee; member of SPAG. **Stavros Goulidis,** Civil Servant, Ministry of Labour and Social Affairs. **Haydn Hammersley,** Social Policy Coordinator, European Disability Forum. **Kirsty Hoyle,** CEO, Metabolic Support UK; member of SPAG. **Gavin McDonough,** Disability Project Manager, Orphanet. **Maria Montefusco,** Investigator, Swedish Agency for Participation. **Pauline McCormack,** Medical Sociologist, Newcastle University. **Adéla Odrihocká,** Board member, Rare Diseases Czech Republic; member of SPAG. **Petra Rantamaki,** Board Member, European Association of Service Providers for Persons with Disabilities. **Sara Rocha,** Vice-President, European Council of Autistic People; President, Associação Portuguesa Voz do Autista; member of Women's Committee of the European Disability Forum. **Ariane Weinman,** Public Affairs Senior Manager, EURORDIS-Rare Diseases Europe.

5.1.2. DISTRIBUTION

The survey was conducted online from July 10 to September 8, 2024. The study population included people with rare diseases and their close family members worldwide. 10478 responses were received worldwide, with 9591 being from Europe. 31% of participants were directly contacted through the Rare Barometer panel, and 69% were reached through social media posts, organisations representing people living with rare diseases, and EURORDIS networks.

5.2. DATA MANAGEMENT AND ANALYSIS

In this report, we conduct exploratory analyses of the association between our variables of interest. The data has been tested using bivariate analysis, such as chi-squared tests (to determine if two variables are related) and multivariate analysis (linear and logistical regressions) to consider relationships between multiple variables.

5.3. SURVEY SAMPLE

5.3.1. PROFILE OF PARTICIPANTS

Figure 12. Number of participants per country in Europe.

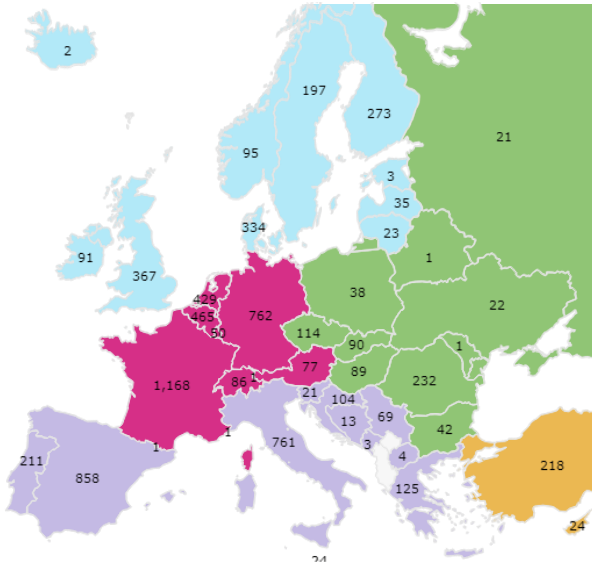


Table 18. Composition of the survey sample for Europe.
All participants (n=9591).

Type	Percentage (n)
Participants' status	
People living with a rare disease	65% (6260)
Family members of people living with a rare disease	35% (3331)
Patient representatives Participants involved in voluntary and/or policy activities to support the cause of rare diseases	23% (2210)
Gender of participants	
Female	75% (5807)
Male	25% (1864)
Age of participants	
<24	2% (169)
24-29	5% (333)
30-39	16% (1114)
40-49	26% (1833)
50-59	26% (1847)
60 and over	25% (1733)

Figure 12 presents the country distribution of participants, which is impacted by population size, the level of involvement of patients' organisations in disseminating the survey and other cultural factors. Countries were grouped in subregions as defined in the United Nations geographic regions for Europe (Figure 12 and **Annex**):

- Western Europe (in pink): 39% of the participants.
- Southern Europe (in purple): 27% of the participants.
- Northern Europe (in blue): 18% of the participants.
- Eastern and Central Europe (in green): 11% of the participants.
- Our sample also comprises 4% of participants from Western Asia (in yellow) as the United Nations geoscheme classifies some transcontinental countries as Western Asia, such as Cyprus and Türkiye.

Given the relatively low number of participants in some countries, only countries with more than 30 participants and with significant results (p-value<0.05) will be considered in per-country analyses.

Table 18 shows the sample's characteristics: 65% were people living with rare diseases, and 35% were family members of people with rare diseases, consistent with the participants' distribution in previous Rare Barometer surveys. 23% of participants declared that they are involved in voluntary or policy activities to support the cause of rare diseases.

The proportion of female participants (75%) was higher than the European Union general population (51%)[1] but is consistent with other surveys carried out among the rare disease community[21], reflecting women's over-representation in caregiving roles.

93% of participants (people with rare disease or their family members) were over 30.

[1] https://ec.europa.eu/eurostat/statistics-explained/index.php?title=Gender_statistics

5.3.2. RARE DISEASES

95% of the people with rare diseases in the survey had received a confirmed or an initial diagnosis. In comparison, 5% only had a partial diagnosis or knew their disease was rare but remained undiagnosed (**Table 19**). Again, those numbers are consistent with previous Rare Barometer surveys[1,15,26-30].

The population of people with rare diseases is diverse: over 6,000 distinct rare diseases and various disease groups exist. Our survey sample represents this diversity and comprises 1,754 individual rare diseases. Error! Reference source not found. presents the repartition of participants' disease in one or several therapeutic areas, based on the classification developed by Orphanet and available on orphadata.org. The conditions that are most represented were also listed in the **Annex**.

The name of the disease allows an estimation of the point prevalence, which is the proportion of a particular population living with a given disease at a specific time. It was calculated using Orphanet epidemiological data based on the name of the rare disease and participants' country of residence. In our survey (**Table 19**), the point prevalence is known for 53% (5053/9,591) of participants, of which 25% (1244/5053) were living with a very rare disease (less than 1 case for in 100,000 people), and 75% (3796/5053) were living with a more common rare disease (from 5 cases for in 10,000 people to 1 case for in 100,000 people). Among the 47% (4,538/9591) of participants for which point prevalence is unknown, 50% (2263/4538) did not declare their rare disease, and 50% (2275/4538) were living with a rare disease for which epidemiological data is not yet available.

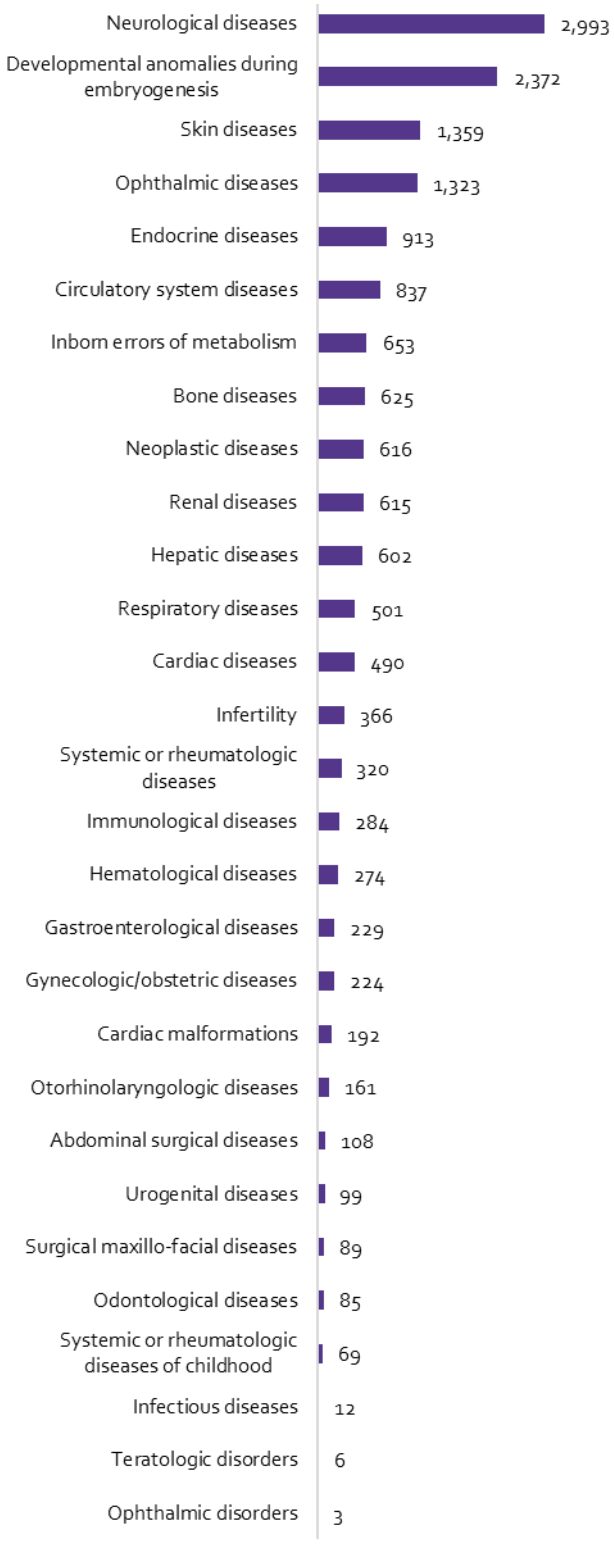
Table 19. Characteristics of the rare diseases represented.

All participants (n=9591).

Type	Percentage (n)
Diagnosis status	
Diagnosed	95% (9080)
Undiagnosed	5% (510)
Point prevalence of the rare disease (Orphanet)	
Between 5/10,000 and 1/100,000	75% (3796)
<1/100,000	25% (1244)

Figure 13. Number of people with rare diseases per therapeutic area.

All participants (n=9591). Grouping based on the Orphacode of the disease entered by participants and on the Orphanet classification of rare diseases. One rare disease can be classified in several therapeutic areas.



5.3.3. PROFILE OF PEOPLE LIVING WITH RARE DISEASES

Table 20. Sociodemographic characteristics of the people living with a rare disease.
(n=6260)

Type	Percentage (n)
Patient representatives	
Participants involved in voluntary and/or policy activities to support the cause of rare diseases	20% (1255)
Gender of the person living with a rare disease	
Female	78% (3992)
Male	21% (1086)
Age of the person living with a rare disease	
<18	21% (1612)
18-24	7% (516)
25-30	6% (456)
31-40	12% (942)
41-59	34% (2646)
60 and over	20% (1550)

We also asked about the characteristics of people with rare diseases represented in the survey, *i.e.* those who have answered the survey about their situation and those whose family members responded on their behalf (**Table 20**). Most of them were females (78%), and 34% were under 30. 20% of people living with rare diseases were involved in voluntary and/or policy activities to support the cause of rare diseases.

While 93% of participants who answered regarding their situation were over 30 years old, most proxy participants (family members) responded on behalf of people with rare diseases aged 30 or less.

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ACRONYMS AND ABBREVIATIONS

CASP: Child and Adolescent Scale of Participation.

EHIS: European Health Interview Survey.

ERN: European Reference Network.

EU: European Union.

EURORDIS-Rare Diseases Europe: European Organisation for Rare Diseases.

Eurostat: Statistical Office of the European Union.

EU-SILC: European Union Statistics on Income and Living Conditions.

GALI: Global Activity Limitation Indicator.

Orphacode: Code assigned to rare diseases in the Orphanet database.

Orphanet: Portal dedicated to providing information on rare diseases and orphan drugs.

PA: Personal assistant.

SPAG: EURORDIS' Social Policy Action Group

UN: United Nations.

UNCRPD: United Nations Convention on the Rights of Persons with Disabilities.

WGSS: Washington Group Short Set on Functioning.

WHO: World Health Organization.

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APPENDIX

Participation per country, grouped by United Nation geosheme, and comparison with population distribution.

	Rare Barometer survey		Population	
	%	N	%	N
Western Europe	**32%**	**3572**	**23%**	**195,381,649**
Austria	1%	77		
Belgium	5%	465		
France	12%	1,168		
Germany	8%	762		
Liechtenstein	0%	1		
Luxembourg	1%	50		
Monaco	0%	1		
Netherlands	4%	429		
Switzerland	1%	87		
Unknown country and questionnaire answered in Dutch, French or German	6%	532		
Southern Europe	**26%**	**2473**	**18%**	**152,130,606**
Andorra	0%	1		
Bosnia and Herzegovina	0%	13		
Croatia	1%	104		
Greece	2%	150		
Italy	10%	946		
Malta	0%	24		
Montenegro	0%	3		
Macedonia	0%	4		
Portugal	3%	274		
Serbia	1%	69		
Slovenia	0%	27		
Spain	9%	858		
Northern Europe	**17%**	**1657**	**13%**	**106,197,357**
Denmark	4%	427		
Estonia	0%	3		
Finland	4%	338		
Iceland	0%	2		
Ireland	1%	91		
Latvia	1%	46		
Lithuania	0%	32		
Norway	1%	123		
Sweden	3%	228		
United Kingdom	4%	367		

Participation per country, grouped by United Nation geoscheme, and comparison with population distribution [continued].

	Rare Barometer survey		Population	
	%	N	%	N
Eastern and Central Europe	**11%**	**1046**	**35%**	**291,464,162**
Belarus	0%	1		
Bulgaria	1%	71		
Czech Republic	2%	154		
Hungary	1%	114		
Moldova	0%	1		
Poland	1%	44		
Romania	4%	368		
Russia	0%	21		
Slovakia	1%	127		
Ukraine	0%	33		
Unknown country and questionnaire answered in Bulgarian, Croatian, Czech, Hungarian, Latvian, Lithuanian, Polish, Romanian, Russian, Slovak, Slovenian or Ukrainian	1%	112		
Western Asia	**4%**	**392**	**11%**	**88,810,566**
Cyprus	0%	24		
Türkiye	4%	368		
TOTAL	**95%**	**9140**	**100%**	**833,984,340**
Unknown country but questionnaire answered in a language mostly used in English or in Spanish	5%	451		
TOTAL	**100%**	**9591**		

***Source**: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

Participation per rare disease (individual orphacodes): rare diseases with at least 30 participants.

Rare condition (individual orphacode)	Number of participants
Hypermobile Ehlers-Danlos syndrome	243
Hereditary hemorrhagic telangiectasia	241
Addison disease	127
Myasthenia gravis	112
Sarcoidosis	107
Neurofibromatosis type 1	106
Primary sclerosing cholangitis	96
22q11.2 deletion syndrome	68
Autoimmune hepatitis	67
Cystic fibrosis	67
Phenylketonuria	65
Williams syndrome	60
Systemic sclerosis	52
Marfan syndrome	51
Friedreich ataxia	50
Tuberous sclerosis complex	50
Classical Ehlers-Danlos syndrome	49
Common variable immunodeficiency	48
Hereditary spastic paraplegia	48
Facioscapulohumeral dystrophy	46
BehÃ§et disease	45
Primary lymphedema	45
Alpha-1-antitrypsin deficiency	44
Wilson disease	43
Arnold-Chiari malformation type I	41
Diffuse cutaneous systemic sclerosis	40
Primary biliary cholangitis	39
Duchenne muscular dystrophy	38
Idiopathic achalasia	38
Monosomy 5p	38
Amyotrophic lateral sclerosis	37
Chronic primary adrenal insufficiency	36
Fabry disease	35
Idiopathic pulmonary arterial hypertension	35
Pulmonary arterial hypertension	35
Autoimmune polyendocrinopathy type 1	34
Familial Mediterranean fever	33
Chronic inflammatory demyelinating polyneuropathy	32
Granulomatosis with polyangiitis	32
Angelman syndrome	31

***Source**: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

Participation per EURORDIS European Federation (at least 25 participants), defined based on Orphacodes and disease lists validated by each European Federation.

European Federation	Number of participants
Ehlers-Danlos Society	364
HHT Europe (Hereditary Haemorrhagic Telangiectasia)	241
NF Patients United (Neurofibromatosis)	135
Federation of European Scleroderma Associations	133
European Myasthenia Gravis Association	126
PHA Europe (Pulmonary Arterial Hypertension)	123
Albi France (Inflammatory Biliary Diseases)	114
Sarcoidosis	113
Lupus Europe	97
Sclerosing Cholangitis	96
Marfan Europe Network	75
22Q11 Europe (22q11 Deletion syndrome)	74
European Society for Phenylketonuria	74
OIFE - Osteogenesis Imperfecta Federation Europe	68
CF Europe (Cystic Fibrosis)	67
European Federation of Williams Syndrome	60
European Federation for Hereditary Spastic Paraplegia	54
European Tuberous Sclerosis Complex Association	50
SMA Europe (Spinal Muscular Atrophy)	49
FSHD Europe (Facioscapulohumeral Muscular Dystrophy)	46
Duchenne Muscular Dystrophy	38
European Huntington Association	38
Gaucher	37
MPS Europe (Mucopolysaccharidosis)	37
Perineural cyst	30
European Haemophilia Consortium	29
Naevus Global	27
Rett Syndrome Europe	27

***Source**: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

Participation per European Reference Network (ERN), n fdefined based on Orphacodes and disease lists validated by each ERN.

European Reference Network (ERN)	Number of participants
EYE *(rare eye diseases)*	1,779
ERKnet *(rare kidney diseases)*	990
MetabERN *(inherited metabolic diseases)*	727
VASCERN *(rare multisystemic vascular diseases)*	531
RITA *(rare immunological disorders)*	434
NMD *(rare neuromuscular diseases)*	373
EpiCARE *(rare and complex epilepsies)*	335
ITHACA *(rare malformation syndromes and rare intellectual and neurodevelopmental disorders)*	324
Lung *(rare respiratory diseases)*	321
Liver *(rare liver diseases)*	301
Endo *(rare endocrine conditions)*	157
eUROGEN *(rare uro-recto-genital diseases and complex conditions)*	147
GENTURIS *(genetic tumour risk syndromes)*	133
ReCONNET (rare connective tissue and musculo-skeletal diseases)	131
BOND (rare bone diseases)	124
GUARD-Heart (rare complex heart diseases)	114
ERNICA *(rare inherited and congenital digestive and gastrointestinal anomalies)*	95
RND *(rare neurological diseases)*	89
EuroBloodNet *(rare hematological diseases)*	85
Skin *(rare and complex skin diseases)*	73
CRANIO *(rare and complex craniofacial anomalies, and ear, nose and throat disorders)*	15

***Source**: Rare Barometer survey conducted July-Sept. 2024. 'The impact of living with a rare disease: barriers and enablers of independent living and social participation'.*

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