

# Rare Diseases



- Consultation period: 28 February to 31 March 2025
- Methodology: online survey
- 4,108 responses: 2,679 individuals, 383 patient support groups, 899 healthcare professionals, 102 stakeholder interest groups, 45 international organisations/governments/public bodies

In the EU, between **27 and 36 million people** live with a rare disease. Rare diseases are conditions that affect fewer than 5 in 10,000 people, with more than 6,500 distinct disorders identified.

## Barriers in care-seeking and obtaining support for rare disease patients

**Obtaining a rare disease diagnosis, having access to existing treatments, and the availability of specialists** were identified as major challenges to patients by all respondents.



**70.0%**

of patient support groups reported difficulty in obtaining an accurate diagnosis



**58.0%**

of patient support groups reported difficulties accessing care



**39.8%**

of individuals reported lack of information about available support services



**47.6%**

of individuals reported no treatment is available for their rare disease



**48.3%**

of individuals reported a lack of specialists

### The personal and emotional impact of a delayed diagnosis for those living with rare diseases:

*"It took me over 15 years to get diagnosed with a Primary Immunodeficiency. That's my entire young adult life. It has cost me so much. I'm scared for my future. I spent all my money on endless amount of doctors and their useless treatments because no one thought of a Primary Immunodeficiency."* (Individual)



### Limited awareness and knowledge of rare diseases within the medical community:

*"They don't know about my illness, if what they know is out of date, if they don't recognise many of the symptoms as being related to the illness, how can they help? If they don't bother to listen, how can they help?"* (Individual)



### Disparities in the availability and reimbursement of treatment between countries:

*"As a rare disease patient, I took part in a clinical trial. I was put on new medication for 3 months, it was life changing, the improvement was immediate & they've kept me out of hospital. Once the trial finished so did the supply of medication. I now travel to another EU country to buy the medication (at great expense). They should be available in my country."* (Individual)



### Individuals living with rare diseases often endure significant discrimination and isolation, both within healthcare systems and society at large:

*"I cannot go out in public without getting stared at, get questions from strangers, kids laughing, teens and young adults being mean like pointing and laughing."* (Individual)



*"I cannot take out loans because banks do not want people like us. I cannot work. I earn an income like a worker, but I am not entitled to the same benefits. It is not fair."* (Individual)



## Researching and managing rare diseases

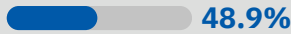
According to healthcare professionals, **lack of funding and investments, lack of adequate treatments, and a lack of disease awareness** are the main challenges in researching and managing rare diseases.

## Gaps in the healthcare system for rare disease patients

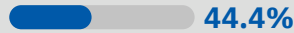
According to the international organisation and public authority responder group, the following issues were identified to be the most important gaps in the healthcare system for rare disease patients requiring urgent policy action:



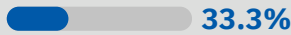
Data sharing issues



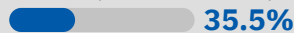
Low level of public awareness about rare diseases



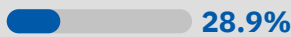
Limited healthcare system budget in a country



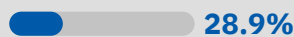
Limited competences of healthcare professionals



High cost of care



Insufficient access to medicines and treatment



## Recommendations:



### Diagnosis

Shorten diagnosis time through **equal access to genetic testing and early screening** for all treatable rare diseases across European countries.



### Treatment

Make treatments more accessible by implementing a **faster approval procedure of new treatments** and **improving access to existing treatments abroad**.



### Research

**Boost research investment and coordination**, aiming for faster diagnosis and more therapies through EU-supported initiatives like IRDiRC and EJP-RD. Ensure **patient involvement** in policy and research.



### Impact of rare diseases in daily life

Promote social inclusion by implementing **supportive measures at schools and in the workplace** to accommodate people with rare diseases.

Implement an **EU-wide harmonisation of disability recognition**, with automatic recognition for all rare diseases, to ensure equitable access to support across Member States.



### Cross-border healthcare

Facilitate cross-border healthcare by providing **better patient information and awareness**.



### EU actions and policies

Adopt a **new European Rare Disease Plan** to align and renew national strategies, reduce inequalities, and **integrate European Reference Networks (ERNs) into national healthcare systems**.

Support existing and the development of new **interoperable EU-wide registries** to improve research and care, leveraging the **European Health Data Space**.

Increase **awareness of EU initiatives and funding opportunities** among professionals and stakeholders.

This infographic was prepared by Leila JOUINI from the Policy Department for Transformation, Innovation and Health of The Directorate-General for Economy, Transformation and Industry (DG ECTI) and is based on the analysis produced by Sciensano, the Belgian institute for health, at the request of the European Parliament's Committee on Public Health (SANT).

**For more details, scan the QR code:**

