EU delivering on rare diseases for patients and families

February 2025

Did you know?

Between **27** and **36** million people in the EU live with a rare disease.

Between **6 000** and **8 000** distinct rare diseases are estimated to exist today in the EU.

Whilst one rare disease may affect only a handful of patients, another may touch as many as **245 000.**

Around **80%** of rare diseases are of genetic origin and **70%** already start in childhood.

What is the Commission focusing on?

Strengthened European action on rare diseases

The EU's action on rare diseases strives to improve access to knowledge, diagnosis, and treatment of patients with rare diseases through funding research on rare diseases and newborn screening, making available dedicated tools, such as the telemedicine tool, pooling of resources and cooperation.

24 European Reference Networks (ERNs) for rare and complex diseases are in place and fully operational.

This includes **1 613 specialised centres** and **380 hospitals** in 27 EU Member States and Norway.

Since their creation in 2017, the ERNs have received EU funding and recently a direct grant of more than **EUR 77 million**, covering their activities for 2023-2027.

What were the main achievements in 2024?

Better availability of and access to medicines and medical devices for EU patients with rare diseases

By the end of 2024, **260 orphan medicines were authorised**. From around 3 000 products that have received an orphan designation, more than 2 000 are still under development. A programme launched in 2024 under EU4Health supports the development of orphan medical devices.

In 2024, **17 orphan medicines have been authorised** and around **140 products** received an **orphan designation**.

EU level guidance, published in June 2024, set for the first time criteria for medical devices intended for small patient populations (e.g. affected by a rare disease) to qualify as an 'orphan device'. Those devices may benefit from free advice from expert panels.

The first evaluation of ERNs showed positive results

The ERN Evaluation report concluded that the objectives of the ERN system are correctly pursued and **the ERNs deliver highly specialist work for rare disease patients**. This includes consultations for diagnosis and therapies, specialised training actions, work on registries and clinical guidelines. However, the ERNs need to be better integrated into national healthcare systems to ensure their sustainability.

24 ERNs and 836 healthcare professionals participated in the evaluation. 100% of ERNs and almost 88% of their members achieved satisfactory results and 3.7% of members had their membership terminated. In comparison, 72 members (8,6%) of ERNs with unsatisfactory results have submitted an improvement plan and will be re-evaluated in 2025.

The CPMS 2.0 - Clinical Patient Management System has been upgraded

A new, dedicated and secure IT platform has been developed to:

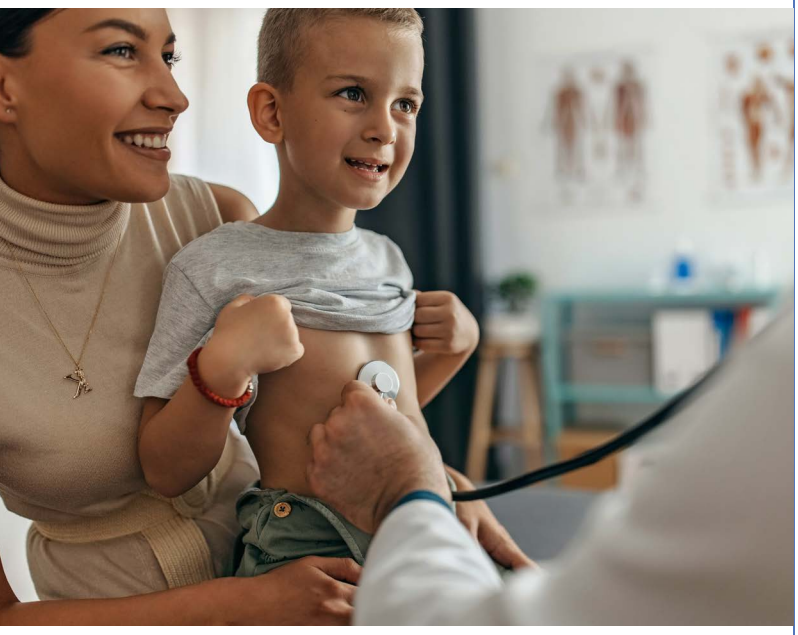
- facilitate discussions between healthcare professionals across the EU and Norway
- support the diagnosis and treatment of rare, low-prevalence and complex diseases.

CPMS 2.0 is simpler, **easier to use**, **more secure**, fully compliant with the Data Protection Regulation (GDPR), f**acilitating collaboration between ERNs**. In the future, it may be used by other health networks at national or European level as it is planned to be distributed under an open-source license.

The Joint Action (JARDIN) has started

The Joint Action on Integration of ERNs into national health systems (JARDIN) was launched in March 2024 to ensure the sustainability of the ERNs and **improve their accessibility**.

27 Members States and Norway received funding totalling **€ 18.75 million** to develop together national rare disease governance models, **improve national plans for rare diseases**, national care pathways, referral systems and improving data management in Member States.



Continued improvement of recognition, visibility, and coding of rare diseases

The **EU continued to support the European Platform on Rare Disease Registration** (EU RD Platform), which copes with the fragmentation of rare disease patients' data in more than 600 registries across Europe.

The Platform makes data of rare diseases registries searchable and findable, thus increasing visibility for each registry, maximising the value of each registry's information, and enabling extended use and re-use of data.

The EU continued to support Orphanet, the European portal on rare diseases and orphan medicines. Among its activities, **Orphanet is developing and maintaining a rare disease nomenclature** (ORPHAcode), which can make it easier to identify and diagnose a patient's disease and to improve the visibility of rare diseases in the health and research information system.

Going local and raising awareness

The **Commission has co-organised 8** workshops with EU countries at national level to **raise awareness** of patients' rights in cross-border healthcare and on the existence of the ERNs and their activities. Citizens, healthcare professionals, healthcare providers, payers, administrations, and patients' organisations participated in the events involving Malta, Italy, Ireland, Greece and Cyprus, Poland, Latvia, Estonia, Finland Belgium and the Netherlands.

Moreover, the Commission has revamped its website on rare diseases and ERNs. A new booklet about the ERNs and their **success stories has been released for the 2025** Rare Disease Day. The booklet provides an updated overview of the ERNs and some examples of activities undertaken by the ERNs, **showing the added value for rare disease patients**.

What's next?

Reform of the EU pharmaceutical legislation and promotion of orphan drugs

The EU is proposing the revision of the pharmaceutical legislation and **2025 will see major achievements**. The new legislation aims to improve **access to safe and more effective medicines**, promote orphan medicines for underserved rare diseases and target incentives and regulatory support to reward exceptional therapeutic advancement.

Promoting the use of data on rare diseases

Currently, there are **more than 90 000 patients registered in all ERNs registries**. The European Health Data Space will open new opportunities for research on rare diseases, to further develop EU-wide rare disease registries to speed up research, improve healthcare planning and patient care.

The European Partnership on Rare Diseases

ERDERA (2025-2031), the European Partnership on Rare Diseases co-funded by the European Commission and the EU Member States, Associated Countries and beyond, was kicked off in September 2024 under the Horizon Europe Programme. It includes a total budget of **€380 million** and up to **€150 million** of EU contribution, involving more than **170 organisations from 37 countries**. It will shape the research landscape for rare diseases until 2030.

Educating a new generation of doctors on rare diseases

Work is currently underway to use the Erasmus+ Programme to offer students in medical faculties opportunities for acquiring specialised knowledge in rare diseases.

Additionally, there are ongoing efforts to develop curricula for academic or specialisation courses on rare diseases and mapping the situation in the Member States as regards their availability.

Communicate more and better

An EU-level event will be organised in 2025 to draw conclusions on the lessons learnt from the different national workshops on patients' rights and ERNs. New communication material related to these topics will be released in all EU languages.

© European Union, 2025

Reuse of this document is allowed, provided appropriate credit is given and any changes are indicated (Creative Commons Attribution 4.0 International license). For any use or reproduction of elements that are not owned by the EU, permission may need to be sought directly from the respective right holders. All images © European Union, unless otherwise stated.
© Gettyimages

Luxembourg: Publications Office of the European Union, 2025

PDF ISBN 978-92-68-24331-2 doi:10.2875/2895621 EW-01-25-013-EN-N
Print ISBN 978-92-68-24332-9 doi:10.2875/9306756 EW-01-25-013-EN-C