European Parliament

# World Rare Diseases Day: Advances in research give patients new hope

Rare diseases, though seldom recognised and frequently misdiagnosed, affect approximately 36 million people in the EU. Despite their significant collective prevalence, patients with rare diseases are often overlooked by healthcare systems, facing challenges in diagnosis, treatment, and access to advances in research. Researchers are increasingly using networks to share their findings and make better progress, giving new hope to patients.

## World Rare Diseases Day

World Rare Diseases Day, observed every year on 28 February (or 29 in leap years) – the rarest day of the year – was set up in 2008 by EURORDIS (a non-profit alliance of over 1 000 rare disease patient organisations from 74 countries) and partner organisations to raise awareness and generate change for the 300 million people worldwide living with a rare disease, and for their families and carers.

## Background

In the EU, a rare disease is defined as life threatening, chronically debilitating and of low prevalence (fewer than five cases per 10 000 people according to Decision No 1295/1999/EC of the European Parliament and of the Council of 29 April 1999). Rare diseases can be of genetic origin (such as Duchenne muscular dystrophy, Huntington's disease, amyotrophic lateral sclerosis, Creutzfeldt-Jakob disease, and metachromatic leukodystrophy, to name just a few), but the group also includes metabolic diseases, infectious diseases, autoimmune diseases and rare cancers. Common symptoms can sometimes mask underlying rare diseases, resulting in misdiagnosis and delayed treatment. Moreover, symptoms differ not only between diseases but also among patients suffering from the same disease. Rare diseases are often disabling, significantly affecting a person's quality of life due to their chronic, progressive, degenerative, and often life-threatening nature; loss or lack of autonomy further exacerbates these challenges. Parents of a child with a rare disease often struggle to manage the rising costs of care and to find schools equipped to support their child's needs. Additionally, stigma and a lack of public awareness can deepen feelings of isolation for both the person living with a rare disease and their families.

The June 2024 study on tackling rare diseases, offered by the European Parliament's Policy Department for Economic, Scientific and Quality of Life Policies, details the most salient challenges for rare diseases:

- a lack of data, information, and research;
- problems around diagnosis, and the codification of individual rare diseases in and across EU Member States;
- market failure in developing treatments;
- problems in establishing market or patient access to treatments;
- high prices for treatments.

## Facts and figures

Approximately 6 000 to 8 000 rare diseases have been identified worldwide, though the exact number fluctuates as new conditions continue to be discovered and classified. Medical and scientific knowledge about rare diseases is lacking: fewer than 1 000 diseases benefit from even minimal amounts of scientific knowledge. Around 80 % of rare diseases have a genetic cause, almost 70 % of which are present in childhood. Despite numerous scientific advances, about 95 % of the known rare diseases still lack approved treatment. About 30 % of children with a rare disease die before the age of five.

In the EU, it is estimated that around 36 million people are living with a rare disease. Diagnoses, if made at all, are often very slow: the average time for an accurate diagnosis in the EU is between four and five years.


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## EU action

EU Member States are responsible for their own healthcare policies. However, according to Article 168 of the Treaty on the Functioning of the European Union, the EU can complement and add value to national actions. In 2009, the Council adopted a recommendation on an action in the field of rare diseases, encouraging EU Member States to develop national plans to ensure high-quality healthcare and research, and improve patient access to treatments. Many patient organisations have repeatedly called for further action at European level. In its June 2024 conclusions on the future of the European health union, the Council invited the Member States and the European Commission to strengthen the Healthier Together – EU Non-Communicable Diseases Initiative by adopting a comprehensive EU-level approach to rare diseases, including a European action plan on rare diseases. The European Economic and Social Committee also called for such an action plan in an exploratory opinion in October 2024.

The European Medicines Agency plays a central role in facilitating the development and authorisation of medicines for rare diseases (known as 'orphan medicinal products'). In 2023, the EU launched a revision of its pharmaceutical legislation, including the 'EU Orphan Regulation', which is still ongoing.

The EU supports the definition, codification and inventory of rare diseases. To this end, it established the Orphanet portal, which has expanded into a consortium of 40 countries within the EU and worldwide. The EU also maintains the 'Orphanet rare disease nomenclature' (ORPHAcode), an international classification system. In addition, the European Platform on Rare Disease Registration provides a unified access point to rare disease registries.

Expanding knowledge of rare diseases and developing new therapies and diagnostic tools through research have been EU priorities for more than two decades. These priorities have been pursued through successive programmes for research and innovation, with the participation of universities, research organisations, healthcare providers, industry, and patient organisations from across Europe and beyond.

On 28 October 2024, a European partnership on rare diseases was launched ('European Rare Diseases Research Alliance' ERDERA). Co-funded by the EU Member States and the Commission under the 2021-2027 Horizon Europe research programme, it will drive research in prevention, diagnosis, and treatment of rare diseases, with an estimated overall budget of €380 million until 2031, and brings together over 170 organisations from the public and private sectors.

The EU also contributes to research coordination through cross-border networks, the European reference networks (ERNs), which have been bringing together European hospital centres of expertise since 2017 to tackle rare, low-prevalence and complex diseases and conditions requiring highly specialised healthcare. There are currently 24 ERNs (which are evaluated regularly), including 1 619 specialised centres located in 382 hospitals. In March 2024, the Commission launched a joint action on the integration of ERNs into national healthcare systems (JARDIN) to create a bridge between ERNS and national health systems, facilitating the accessibility of the ERNs for patients across the EU. For the 2024-2027 period, the action will receive €15 million from the EU4Health programme and €3.75 million from the EU Member States. On 4 December 2024, the Commission launched a new IT platform to offer better support to the ERNs (the Clinical Patients Management System 2.0). The platform enables free of charge, remote online discussions between healthcare professionals from different Member States, reducing the need for patient travel.

## European Parliament position

Parliament has consistently highlighted the need for research into rare diseases and the development of medicines. In a 2021 resolution on the EU's pharmaceutical strategy, it stressed the importance of advancing research for unmet medical needs. Previously, in a 2020 resolution, it called for the creation of an EU action plan for rare and neglected diseases as part of a post-pandemic public health strategy.

Since the start of the current legislature, Parliament has submitted several written questions to the Commission on issues related to rare diseases. They include questions on paediatric transplants, on orphan drugs and on the shortage of critical medicines in the European market.

The new standing Committee on Public Health (SANT) – the successor to the SANT subcommittee – will continue to monitor the shortage of critical medicines in the EU, which impacts, notably, patients suffering from rare diseases.