European Commission

EU ACTION ON RARE DISEASES

Improving patient access to knowledge, diagnosis and care

FEBRUARY 2023

«Rare diseases, numerous patients»

Up to **36 million people** in the EU live with a rare disease.
There are more than **6000** distinct rare diseases in the EU so whilst one rare disease may affect only a handful of patients, another may touch as many as **250,000**.
Around **80% of rare diseases are of genetic origin** and of those **70% already start in childhood.**
EU action on rare diseases aims to improve the diagnosis, care and treatment of patients with rare diseases through the pooling of resources and cooperation.

Strengthened European cooperation and coordination to improve access to knowledge, diagnosis and treatment of rare diseases

More than 1600 specialised centres and almost 400 hospitals in 28 countries, connected by **24 European Reference Networks (ERNs).**

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

More than **200 orphan medicines** have been authorised and over **2700 orphan medicinal products** designated.

Improving recognition, visibility and coding

Support to the **European Platform on Rare Diseases Registration.**

Support to **Orphanet,** the European portal on rare diseases and orphan medicines, also developing and maintaining the coding system for rare diseases (**Orphacodes**).

Support for training, development and research

Support **ERN Exchange of Professionals Programme** to share knowledge and stimulate collaboration between healthcare professionals.

ERN Clinical Practice Guidelines programme.

EU-funded research projects.

Support national policies in EU Member States

Several **Joint Actions** to develop national information and data systems, national rare diseases plans, as well as advance work on rare cancers.

Support international collaboration

Participating in **International Classification of Diseases**, led by WHO.

Towards a stronger EU approach on rare diseases

Revision of the legislation on medicines for rare diseases

To foster the development of medicines addressing unmet medical needs, and more timely and affordable access for all.

Improved and increased funding for greater impact

€ 77 million as direct grants to ERNs.

Promote the use of big data on rare diseases

Set up EU-wide registries to facilitate clinical research, improve healthcare planning and patient care, in line with the European Health Data Space.

Launch of European Partnership for research activities on rare diseases in 2024

Over €2.4 billion has been allocated to support national, local and European research and innovation programmes.

Increased support to Member States in 2023-2025

Launch of the Joint Action on integration of ERNs into national health systems.

Enhanced European Reference Networks

Creation of Virtual ERN Academy in 2022.

An improved version of the IT platform to support the cross-border discussion of clinical cases is being developed and will be available in 2024.

First periodic evaluation of ERNs and their members after 5 years of existence.

Support for the creation of a Ukrainian Hub for rare and complex diseases

Since the beginning of its activity in 2022, over 600 cases of patients have been referred to the hub.

Funding under **EU4Health** will continue to support research on rare diseases and the development and improvement of the ERNs ecosystem.