

EU ACTION ON RARE DISEASES

Improving patient access to knowledge, diagnosis and care

FEBRUARY 2024

«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

1619 specialised centres and 382 hospitals, located in 27 EU Member States and Norway, connected by **24 European Reference Networks (ERNs)**.



Integration of European Reference Networks into national health systems

Joint Action JARDIN, covering a period of three years, has received a funding of €18.75 million to better integrate ERNs in national healthcare systems and develop patient pathways. It involves all EU Member States, Norway, and Ukraine.



Evaluation of European Reference Networks, including 836 members

The ERN ecosystem is functioning well, meaning they deliver on highly specialist work for rare disease patients, such as consultations for diagnosis and therapies, production of clinical guidelines and specialised trainings. 100% of the ERNs and 88% of their healthcare providers obtained satisfactory results, with termination of 4% of their healthcare providers.



Continued support for the European Reference Networks

New grants covering the next four years (2023-2027), with a total budget of €77.4 million for 24 ERNs, will enable the ERNs to continue their work and measure their impact through 24 impact indicators.



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

Around **250 orphan medicines** have been authorised and around **2000 products** received an orphan designation and are under development. New programme to support the development of orphan medical devices, in particular for children, under the EU4Health Programme.



Improving recognition, visibility, and coding

Further development of the **European Platform on Rare Disease Registration**.

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

Towards a stronger EU approach on rare diseases



Reform of the EU pharmaceutical legislation

Further **promotion of orphan medicines** for underserved rare disease areas and **targeted incentives and regulatory support** to reward exceptional therapeutic advancement.



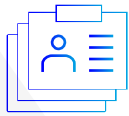
Improved and increased funding for greater impact

€77 million provided to ERNs as **direct grants**, covering the 2023-2027 period.



Development of the Clinical Patient Management System (CPMS)

New IT platform to support **cross-border medical discussions** of rare clinical cases: the CPMS 2.0 is simpler, more user friendly, more secure, fully GDPR compliant and distributed under an open-source license. This facilitates collaboration between ERNs, and can be used by health networks created at national level.



Promote the use of data on rare diseases

Further develop EU-wide rare disease registries to speed up research, improve healthcare planning, and patient care. The European Health Data Space will open up **new opportunities for research on rare diseases**.



Launch of a European Partnership on rare diseases

European Partnership on Rare Diseases co-funded by Member States and the Commission under the Research & Innovation Programme Horizon Europe (EU funding up to €150 million).



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