European Commission

Rare diseases

Sharing expertise to tackle rare disease

Rare diseases affect millions of Europeans but can be difficult to diagnose, treat and study because the patient population and disease experts are dispersed across Europe. Through the second EU health programme 2008-2013, Europe is supporting projects that share knowledge and expertise on rare diseases.

EU action for enhancing protection

In EU countries, any disease affecting fewer than five people in every 10 000 is considered rare. These conditions, including rare cancers, metabolic conditions and other genetic disorders, affect between 27 and 36 million people in the EU. Patients with rare diseases can find it difficult to get an accurate diagnosis and to access appropriate treatments. Rare diseases are also difficult to tackle because the patient population is small and scattered.

The EU can make a significant impact in this area by bringing together patients and experts to improve research, diagnosis, treatment and care. Encouraging cooperation between EU countries, pooling knowledge and establishing networks of experts can deliver important advances in scientific understanding.

By deepening health professionals' understanding of the natural history of the diseases and encouraging medical innovation in the development of new treatments, European policies have the potential to improve quality of life for patients.

Consumers, Health and Food Executive Agency


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Adding value through collaboration

Several measures have been taken at EU level to improve the treatment of rare diseases and the lives of those suffering from them. The European Commission Communication on Rare Diseases: Europe's challenge, adopted in 2008, set out an overall strategy to support EU countries in diagnosing, treating and caring for EU citizens with rare diseases. It focuses on: improving the recognition and visibility of rare diseases; supporting national policies on rare diseases as part of a coherent overall strategy; and developing cooperation, coordination and regulation for rare diseases at EU level.

In addition, EU governments agreed a series of recommendations for action in the field of rare diseases in a Council Recommendation in 2009 which confirmed the need for national strategies to be put in place. It focused on the definition, codification and inventory of rare diseases, research, European reference networks, gathering expertise at EU level, empowerment of patient organisations, and sustainability.

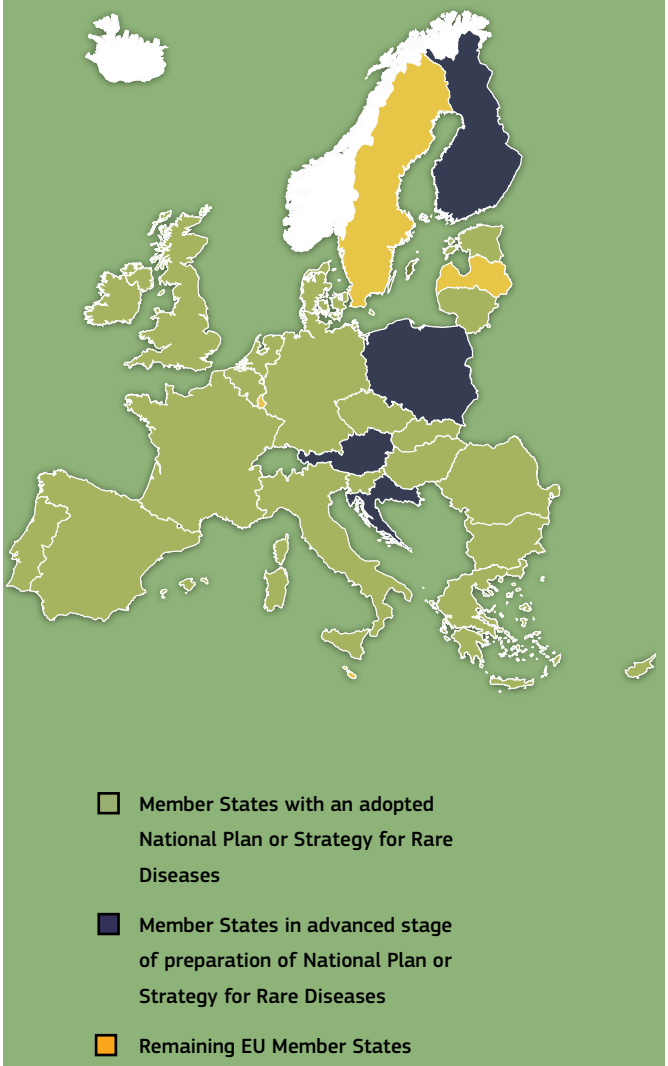
The International Rare Diseases Research Consortium (IRDiRC) was also launched in 2011. This international collaboration, which includes the European Commission, the US National Institutes of Health and several other funding agencies, brings together researchers from across the globe in a bid to deepen understanding of rare diseases.

The 2011 EU Directive on patients' rights in cross-border healthcare encourages measures to raise awareness among health professionals of diagnostic tools for rare diseases, and clarifies the possibility for referral of patients to other EU countries.

To accelerate the approval of new orphan medicines, the EU medicines regulatory system offers incentives for products designed to treat, prevent or diagnose rare diseases.

The second EU health programme 2008-2013, implemented by the Consumers, Health and Food Executive Agency (Chafea), finances a range of projects and actions designed to enhance the sharing of knowledge and expertise in this area.

Stages of national plans or strategies for rare diseases in the EU countries as of the first quarter of 2014.



Source: European Commission implementation report on the Commission Communication on Rare Diseases: Europe's challenges, September 2014 (Italy and Ireland adopted the plans after the publication of the report).

Building networks of knowledge

Project name: Joint Action: working for rare diseases (**EUCERD**)

Number of partners: 9 from 7 countries: DE, ES, FR, IT, PT, FI, UK. **EU funding:** €2 994 023. **Duration:** 2010-2013.

The EU Committee of Experts on Rare Diseases (EUCERD), which was recently replaced by the Commission expert group on rare diseases, supported the European Community in formulating and implementing policy on rare diseases. The Committee fosters exchanges of relevant experience, policies and practices between EU countries and stakeholders.

The joint action enhances the visibility of rare diseases and dissemination of knowledge, and contributes to improvements in access to services. It has supported the implementation of rare diseases plans at national level and the application of coding and classification of rare diseases in international nomenclatures, and proposed a model for sustainable action in the area of rare diseases.

Visit: www.eucerd.eu/

The 'go to' hub for rare diseases

Project name: Development of the European portal of rare diseases and orphan drugs (**JA-Orphanet Europe**)

Number of partners: 25 from 21 countries: BE, CZ, DE, EE, EL, ES, FR, IT, CY, LV, LT, HU, NL, AT, PL, PT, RO, SK, FI, SE, UK. **EU funding:** €3 295 857. **Duration:** 2011-2014.

Orphanet is an established website which provides an inventory of rare diseases, an encyclopaedia in multiple languages and a directory of expert services. It is a reference source for all citizens, including health professionals, healthcare managers and policymakers.

This joint action aimed to improve the encyclopaedia and the directory of services, and to put in place a process to ensure swift management of translations, timely collection of data and validation by national health authorities prior to publication. It also made the website more user-friendly.

Visit: www.orpha.net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Establishing the natural history of rare diseases

Project name: An EU Rare Diseases Registry for Wolfram Syndrome, Alstrom Syndrome and Bardet Biedl Syndrome (**EURO-WABB**)

Number of partners: 10 from 6 countries: EE, ES, FR, IT, PL, UK. **EU funding:** €900 000. **Duration:** 2011-2014.

This project aimed to implement an EU registry for rare diabetes syndromes (RDS) containing genetic, clinical and outcome data. The registry established the natural history of these rare diseases, assessed clinical effectiveness of their management, and provided an inventory of patients for recruitment in intervention studies.

The registry has the potential to deliver a step change in the volume and quality of clinical research in RDS diseases. It will provide data for assessing the clinical effectiveness of standard care and new interventions in a real-world setting.

Visit: www.euro-wabb.org


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Improving diagnosis and care

Project name: European registry and network for Intoxication type Metabolic Diseases (**E-IMD**)

Number of partners: 13 from 10 countries: DK, DE, ES, FR, HR, IT, NL, PL, PT, UK. **EU funding:** €779 746. **Duration:** 2011-2014.

E-IMD aimed to provide evidence-based and consensus-agreed diagnostic and management protocols for individuals affected with rare organic acidurias (OADs) or urea cycle defects (UCDs). The overall goal was to improve access to rapid diagnosis and care for patients.

The project evaluated current management strategies and outcomes in 15 European countries to better understand the natural history of these rare diseases. It established a European patient registry describing the disease course, epidemiology, diagnostic and therapeutic strategies and provided information to national and EU healthcare authorities.

Visit: www.e-imd.org/en/index.phtml


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United against rare cancers

Project name: Information network on rare cancers (**RARECARENet**)

Number of partners: 10 from 8 countries: BG, IE, FR, IT, NL, SI, FI, UK. **EU funding:** €1 000 631. **Duration:** 2012-2015.

RARECARENet builds on the work of a rare cancers information network consisting of oncologists, general practitioners, researchers, health authorities and patients. It strives to improve the timeliness and accuracy of diagnosis, to facilitate access to high quality treatment for patients, to identify centres of expertise for rare cancers in Europe, and to standardise practices across EU countries.

The network also promotes international collaboration on very rare cancers and seeks to empower patients.

Visit: www.rarecarenet.eu

Harnessing e-Health for rare anaemias

Project name: New e-Health Services for the European Reference Network on Rare Anaemias (**e-ENERCA**)

Number of partners: 13 from 7 countries: BE, IE, ES, FR, IT, NL, UK. **EU funding:** €1 163 275. **Duration:** 2013-2016.

The e-ENERCA project builds upon the work of the European Network for Rare and Congenital Anaemias which was established in 2002. It aims to provide health professionals and patients with e-Health tools and to harmonise levels of access to services throughout Europe for people with rare anaemias.

By implementing three platforms on the ENERCA website, the group aims to: enhance the creation and use of European inventories; support centres of expertise; facilitate continuing medical education and best practices; and create a telemedicine platform for the provision of healthcare and medical advice.

Visit: www.enerca.org


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Find out more

Directorate-General for Health and Consumers
of the European Commission
http://ec.europa.eu/health/rare_diseases/policy/index_en.htm

Consumers, Health and Food Executive Agency (Chafea)
ec.europa.eu/chafea/index.html

Database of actions co-funded under the EU health programmes
ec.europa.eu/chafea/projects/database.html

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