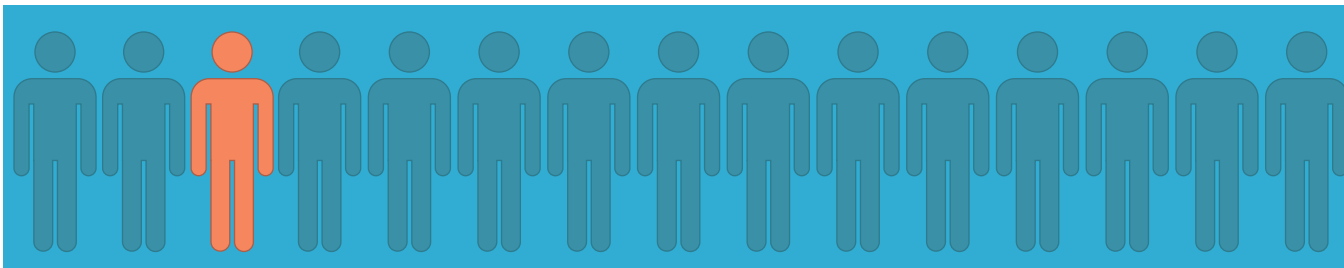




European  
Commission



## EUROPEAN RARE DISEASES DAY: TOP FACTS ON EU ACTION

A disease or disorder is defined as rare in the EU when it affects less than five in every 10 000 citizens. Yet, because there are so many different rare diseases – between 6 000 and 8 000 – taken together they affect a significant share of the population. Between 30 and 40 million people in the EU, many of whom are children, suffer from rare diseases. Most rare diseases have genetic origins while others are the result of infections, allergies and environmental causes. They are usually chronically debilitating or even life-threatening. The impact of such diseases on sufferers, their

families and carers is substantial and patients often go undiagnosed due to lack of scientific and medical knowledge or to difficulty in accessing expertise. The fragmentation of knowledge about rare diseases and the small numbers of patients affected by a single disease makes it indispensable to work across borders. At the European Commission we take an integrated approach to further improve the access and equity to prevention, diagnosis and treatment for patients suffering from a rare disease throughout the European Union. We do this by:

### ... SUPPORTING ACTIONS FOR AN EARLY DIAGNOSIS

One of the greatest challenges for sufferers of rare diseases and their families is getting a timely and correct diagnosis. This is an essential first step before treatment options can be explored. Commission action helps in several ways.

Firstly, the Commission has supported the ORPHANET project and Joint Action through its Health Programme. This has resulted in the [ORPHANET database](#) which, to date, lists the descriptions of almost 6000 rare diseases, and has grown into the number one online source of information on rare diseases worldwide. This tool is an invaluable resource for clinicians, health professionals and patients seeking a diagnosis.

The Commission is also well advanced in the establishment of European Reference Networks (ERNs), foreseen in the [Directive on Patients' Rights in Cross-border Healthcare](#). These ERNs will facilitate cooperation between Member States in the development of diagnosis and treatment capacity to provide highly specialised healthcare for rare or low prevalence complex diseases or conditions. As a result, patients will have easier access to expertise on rare diseases beyond their national border. The Directive also sets up national contact points where patients can receive information on where to find the most appropriate treatment for their disease, anywhere across the EU.

## ... SUPPORTING EUROPEAN RESEARCH FOR BETTER UNDERSTANDING AND TREATMENT OF RARE DISEASES

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With regard to collaborative research, the European Commission will continue the strong commitment to funding excellent research in rare diseases, established through previous framework programmes. The Horizon 2020 Work Programme 2015 for “Health, demographic change and wellbeing” includes an earmarked budget of € 62 million euro for developing new therapies for rare diseases. The Commission is also co-funding the ERA-NET project [E-RARE-3](#) which will strengthen the collaboration between participating EU countries in funding rare disease research.

Rare diseases are too big a challenge for any country or world region to master alone. This is why the European Commission, together with European and international partners, initiated the [International Rare Diseases](#)

[Research Consortium](#). Launched in 2011, it is the biggest collective rare diseases research effort worldwide. Its key objective is to deliver, by 2020, 200 new therapies for rare diseases and the means to diagnose most of them.

IRDiRC has currently over 40 member organisations from four continents committed to working together towards the initiative’s goals. Members are composed of funding bodies investing a minimum of \$US 10 million over five years in research projects/programmes contributing towards IRDiRC objectives, and invited advocacy groups. International partners include organisations from Australia, Canada, China, South Korea, Georgia and the USA.

## ... INCENTIVISING PHARMACEUTICAL COMPANIES

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Patients suffering from rare conditions should be entitled to the same quality of treatment as other patients.

In response to this public health concern and in order to stimulate the research and development of orphan medicines, in 2000 the EU introduced new legislation with the aim to provide incentives for the development of orphan and other medicinal products for rare disorders. The [EU Regulation on Orphan Medicines \(EC/141/2000\)](#) establishes a centralised procedure for the designation of orphan medicinal products and puts in place incentives for the research, marketing and development of medicines for rare diseases.

Today, companies with an orphan designation for a medicinal product benefit from incentives such as fee waivers, a 10 year market exclusivity period post authorisation for designated products, scientific assistance for marketing authorisations, and the possibility of a Community marketing authorisation. Thanks to these incentives, 112 orphan medicines have been authorised by the European Commission and 1132 medicines under research & development have been designated as orphan medicines since the Regulation entered into force.

## ... ESTABLISHING A EUROPEAN PLATFORM ON RARE DISEASES REGISTRATION

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The Joint Research Centre (JRC), the European Commission’s in-house science service, in collaboration with the Directorate-General for Health and Food Safety will establish a European Platform on Rare Diseases Registration. The platform is conceived to provide a central access point for all registry data on rare diseases, to act as a ‘hub’ improving access to patient registries, as well as to promote interoperability between registries. A further important goal is to improve data comparability, reliability and harmonisation among rare diseases registries throughout Europe.

The creation and maintenance of the platform is be part

of the EU strategy for sustainable research in this area. The JRC will work in close cooperation with the Member States and stakeholders, which include national, regional, local registry holders, research institutes, hospitals, patients’ organisations, and pharmaceutical companies. The aim is to ensure full integration of data to further epidemiological research in rare diseases, as well as to guide clinical trials for specific patient groups and to steer health policy in the field. The final outcome will be to improve the quality of health-care and the quality of life for rare disease patients.

## ... HELPING THE ORGANISATIONS THAT SUPPORT PATIENTS

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Through its Health Programme, the Commission provides an operating grant to the European Organisation for Rare Diseases (**EURORDIS**), a non-governmental patient-driven alliance of patient organisations. EURORDIS represents 614 rare disease patient organisations in 58 countries, and is the voice of 30 million people affected by rare diseases throughout Europe.

EU funding helps EURORDIS with its mission to improve the quality of life of people living with rare diseases in Europe through advocacy at the European level, support for research and medicines development, facilitating networking amongst patient groups, raising awareness, and many other actions designed to reduce the impact of rare diseases on the lives of patients and their families.

## ... ASSISTANCE AND SUPPORT FOR MEMBER STATES' EFFORTS

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In 2008, the **Commission adopted a Communication on Rare Diseases**, followed in 2009 by a Council Recommendation. Both seek to improve recognition and visibility of rare diseases and develop European cooperation, coordination and regulation for rare diseases.

The Recommendation called upon Member States to adopt national plans or strategies for rare diseases by the end of 2013 in order to ensure universal access to

high quality care for all patients. Technical assistance and training tools to help EU countries create these national plans has been developed thanks to the **EUROPLAN Project and Joint Action** co-financed by the Commission's Health Programme.

To date, 20 Member States have adopted national plans and several others are nearing adoption. A **report** on the progress made by Member States and the Commission was published in September 2014.

## ... GATHERING THE BEST EXPERT ADVICE

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Since the end of 2009, the Commission has sought the advice of experts to guide EU policy on rare diseases. These expert groups comprise representatives of European level patients' organisations, professional associations, scientific societies and associations producing products or providing services in the field of rare diseases. The Commission may consult the expert group on any matters relating to rare diseases, for example:

- ▶ Help with drawing up legal instruments and policy documents, including guidelines and recommendations
- ▶ Advice on the implementation of EU actions and suggestions for improvement
- ▶ Advice on the monitoring, evaluation and dissemination of results of measures taken both at EU and national level
- ▶ Assistance with fostering exchange of experiences and practices between EU countries and the various parties involved.

## ... SUPPORTING PROJECTS

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Under the Second Programme of Community Action in the Field of Health (Health Programme 2008–13) more than 30 projects on rare diseases have been funded, covering different types of actions and stakeholder groups. This has led to a number of EU-wide resources to pool scarce expertise and provide patients and

health professionals with improved access to medical information, treatment centres, patient support groups and epidemiological/research data.

The third Health Programme (2014-2020) will continue to support projects on rare diseases.

### FOR MORE INFORMATION:

[http://ec.europa.eu/health/rare\\_diseases/policy/index\\_en.htm](http://ec.europa.eu/health/rare_diseases/policy/index_en.htm)

<http://ec.europa.eu/programmes/horizon2020/en/h2020-section/health-demographic-change-and-wellbeing>

[http://ec.europa.eu/research/health/pdf/rare-diseases-how-europe-meeting-challenges\\_en.pdf](http://ec.europa.eu/research/health/pdf/rare-diseases-how-europe-meeting-challenges_en.pdf)