Rare diseases: European action

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The Commission adopted a report on the implementation report on the Commission Communication on Rare Diseases: Europe's challenges and Council Recommendation on an action in the field of rare diseases.

Rare diseases affect between **27 and 36 million people in the European Union** and are a key health policy priority for the EU.

With a view to enhancing the European dimension and the co-operation between Member States in this area, to achieve this goal in 2008, the Commission adopted a Communication on Rare Diseases, setting out an overall strategy to support Member States in diagnosing, treating and caring for EU citizens with rare diseases. Alongside the Communication, a Council Recommendation on action in the field of rare diseases was adopted a few months later, calling on Member States to put national strategies in place.

Directive 2011/24/EUon the application of patients' rights in cross-border healthcare also addresses rare diseases as does the Orphan Medicinal Products Regulation (Regulation (EC) No 141/2000. The **Rare Diseases Task Force** was also set up within the Commission.

Member States committed to use for the purposes of Community-level policy work a common definition of a **rare disease** as a **disease affecting no more than 5 per 10 000 persons**.

This report presents an **overview of the implementation** of the rare diseases strategy to date and takes stock of achievements and lessons learnt. It seeks to draw conclusions on the extent to which the measures foreseen in the Commission Communication and the Council Recommendation have been put in place and the need for further action to improve the lives of

patients affected by rare diseases and their families.

Plans and strategies: in order to support Members States in the process of developing national plans and strategies, the European Commission co-financed the **EUROPLAN** project from the EU Health Programme.

The project, running from April 2008 to March 2011, involved representatives of national health authorities of **21 Member States**, and brought together 57 associated and collaborating partners from 34 countries. One of the deliverables was a report on **indicators for monitoring** the implementation and evaluating the impact of a National Plan or Strategy for rare diseases.

This has supported a significant number of Member States to put in place dedicated plans to address rare diseases: 16 Member States now have rare diseases plans (as compared to only 4 in 2008) and seven further countries are well advanced in the development of their plans/strategies.

The EU has funded close to **120 collaborative research projects** relevant to rare diseases through its Seventh Framework Programme for Innovation and Technological Development (FP7). With a total budget of over EUR 620 million, these projects span across several disease areas such as neurology, immunology, cancer, pneumology, and dermatology.

In collaboration with its national and international partners, the European Commission spearheaded the launch of the **International Rare Diseases Research Consortium (IRDiRC)** in early 2011. Its key objective is to deliver, by 2020, 200 new therapies for rare diseases and the means to diagnose most of

them by stimulating, better coordinating, and maximising output of rare disease research on a global level. As of January 2014 there were 588 rare diseases registries distributed as follows: 62 European, 35 global, 423 national, 65 regional and 3 undefined. Most of the registries are established in public and academic institutions.

It should be noted that the European Commission's Joint Research Centre is currently developing a **European Platform on Rare Diseases Registration.** The main objectives for this platform are to provide a central access point for information on **rare diseases patients' registries.**

Future proposals: despite such encouraging progress, there is **still a long way to go**. There are still Member States who do not yet have a national plan or strategy. This is why action on rare diseases features prominently in the new Health Programme and the new EU Research and Innovation Programme Horizon 2020.

The following actions are envisaged to continue supporting Member States:

- maintain the EU's coordinative role in the development of the EU policy on rare diseases and to support Member States in their activities on the national level;
- continue to support the development of high quality National Rare Diseases Plans/Strategies in the European Union;
- provide continued support for the International Rare Disease Research Consortium and initiatives developed under its umbrella;
- continue to ensure proper codification of rare diseases;
- work further to decrease inequalities between patients with rare diseases and patients suffering from more common disorders and to support initiatives promoting equal access to diagnosis and treatment;
- continue to promote patients empowerment in all aspects of rare diseases policy development;
- continue activities increasing public awareness about rare diseases and EU activity in this field;
- support the development of the tools facilitating cooperation and interoperability of the European Reference Networks for rare diseases;
- stimulate development and use of eHealth solutions in the area of rare diseases;
- implement and continue support for the European Platform on rare diseases registration;
- continue playing a global role in the rare diseases initiative and collaborating with important international stakeholders.