Rare diseases: European action

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Rare diseases are considered as such in the EU when they affect not more than 5 per 10 000 persons. This nevertheless means than between 5 000 and 8 000 different rare diseases affect or will affect an estimated 29 million people in the EU. The lack of specific health policies for rare diseases and the scarcity of the expertise, translate into delayed diagnosis and difficult access to care. This results in additional physical, psychological and intellectual impairments, inadequate or even harmful treatments and loss of confidence in the health care system, despite the fact that some rare diseases are compatible with a normal life if diagnosed on time and properly managed. Misdiagnosis and non-diagnosis are the main hurdles to improving life-quality for thousands of rare disease patients.

The national healthcare services for diagnosis, treatment and rehabilitation of people with rare diseases differ significantly depending on their availability and quality.

Depending on the Member State and/or region where they live, EU citizens have unequal access to expert services and available care options.

Objectives: the Communication sets out an overall Community strategy for support to Member States in ensuring effective and efficient recognition, prevention, diagnosis, treatment, care, and research for rare diseases in Europe. The Communication orients the operational actions in three main fields of work:

- improving Recognition and Visibility on Rare Diseases: the key to improving overall strategies for rare diseases is to ensure that they are recognised, so that all the other linked actions can follow appropriately. The existing definition of rare diseases in the EU was adopted by the Community action programme on rare diseases 1999-2003 as those diseases presenting a prevalence not more than 5 per 10 000 persons in the EU. The same definition is set out in Regulation (EC) 141/2000 and, accordingly used by the European Commission for the designation of orphan drugs. The EU will maintain the current definition. The Commission aims to put in place a thorough coding and classification system at European level, which will provide the framework for better sharing knowledge and understanding rare diseases as a scientific and public health issue across the EU;

- supporting policies on rare diseases in Member States: the Commission proposes that Member States base themselves on common approach for addressing rare diseases, based on existing best practice, through the adoption of a Council Recommendation. (For the Commission's proposal for

a Council Recommendation, please see summary of the same date.)

- developing European cooperation on rare diseases: Community action will help Member States to achieve efficiency in bringing together and organise the scarce resources in the area of rare diseases, and can help patients and professionals to collaborate across Member States in order to share and coordinate expertise and information. The establishment of an EU dynamic inventory of rare diseases will contribute to tackle some of the main causes of neglecting the issue of rare diseases including the ignorance of which diseases are rare. The Commission will ensure that this information continues to be available at European level, building in particular on the Orphanetdatabase;

- disease information networks: priorities for action regarding the existing (or future) specific disease information networks are, inter alia, to guarantee the exchange of information via existing European information networks; to develop strategies and mechanisms for exchanging information between stakeholders; and to develop comparable epidemiological data at EU level.

Operational actions to develop European cooperation and improve access to high quality health care for rare diseases: the Communication proposes that this will be done, in particular through development of national/regional centres of expertise and establishing EU reference networks. It also discusses access to specialised social services, and access to Orphan Drugs. With regard to the latter, the Commission will set up a working party to exchange knowledge between Member States and European authorities on the scientific assessment of the clinical added value of orphan medicines. The paper goes on to describe proposed compassionate use programmes, incentives for Orphan Drug development, e-Health, the evaluation of current population screening (including neonatal screening) strategies for rare diseases and of potential new ones, quality management of diagnostic laboratories, and primary prevention. It discusses registries and databases, noting that a key issue will also be to ensure the long-term sustainability of data collection systems. With regard to research and development, the communication notes that the development of therapies faces three hurdles: the lack of understanding of underlying patho-physiological mechanisms, the lack of support of early phases of clinical development and the lack of opportunity/cost perception from the pharmaceutical industry. The paper outlines proposed action in this area.