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EDITORIAL

Rare diseases: a priority in public health and research

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Since the 1990s at both European Union (EU) and country level political concepts and initiatives concerning rare diseases have emerged. At European level, there are currently three key policy documents establishing a political framework for action in the field of rare diseases and orphan medicinal products at European level: **(1) The Orphan Medicinal Product Regulation (Regulation (EC) No 141/2000 of the European Parliament and of the Council of 16 December 1999 on orphan medicinal products)** was proposed to set up the criteria for orphan designation in the EU and describes the incentives (e.g. 10-year market exclusivity, protocol assistance, access to the Centralised Procedure for Marketing Authorisation) to encourage the research, development and marketing of medicines to treat, prevent or diagnose rare diseases. (2) **The Commission Communication on Rare Diseases: Europe's challenge** 7, adopted on 11 November 2008, set out an overall Community strategy to support countries in diagnosing, treating and caring for the 36 million EU citizens with rare diseases. This Communication focuses on three main areas: 1) improving recognition and visibility of rare diseases, 2) supporting policies on rare diseases in MS for a coherent overall strategy, and 3) developing cooperation, coordination and regulation for rare diseases at EU level. (3) **The Council Recommendation on an action in the field of rare diseases** was adopted on 8 June 2009. The Recommendation engages the responsibility of Member States and concentrates on supporting and strengthening the adoption before the end of 2013 of national plans and strategies for responding to rare diseases, on improving recognition and visibility of rare diseases, on encouraging more research into rare diseases and forging links between centres of expertise and professionals in different countries through the creation of European reference networks in order to share knowledge and expertise and, where necessary, to identify where patients should go when such expertise cannot be made available to them. The role of patients' organisations is also highlighted as particularly important. To aid the European Commission with the implementation of Community activities in the field of rare diseases, **The European Union Committee of Experts on Rare Diseases (EUCERD)** was formally established in 2009. At the same time, national strategies and policies in the field of rare disease (RD) are rapidly evolving. Up till now, several countries have taken action to adapt their health care system to meet the needs of the RD patient community or plan to do so. The first action point is to spot the expertise and organise healthcare pathways to reduce time to diagnosis and ensure appropriate care. A few European countries have already established centres of expertise for RD. The other ones have agreed to work on it according to the recommendations of the EUCERD. This will allow for health care users to access optimal care. Some experimental European reference networks of centres of expertise have been established. They are recognised as a desirable form of cooperation in the directive on cross-border healthcare. Maximising scarce resources and coordinating efforts are also key elements for success in the field of research. Worldwide sharing of information, data and samples to boost research is currently hampered by the absence of an exhaustive rare disease classification, standard terms of reference and common ontologies, as well as harmonised regulatory requirements. The **International Rare Disease Research Consortium (IRDiRC)** was launched in April 2011 to foster international collaboration in rare diseases research. The European Commission and the US National Institutes of Health initiated the discussions, and other stakeholders, including other funding agencies, were also invited to join the consortium. In October 2011 there was a meeting to identify principal research topics and regulatory challenges in this international context, as well as to establish the governance of the consortium. IRDiRC will team up researchers and funding agencies in order to achieve two main objectives by the year 2020, namely to deliver 200 new therapies for rare diseases and diagnostic tools for most rare diseases.

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