SEVENTH MEETING OF THE EUROPEAN UNION COMMITTEE OF EXPERTS ON RARE DISEASES

31 January – 1 February 2013, Luxembourg

Executive Summary

The seventh meeting of the European Union Committee of Experts on Rare Diseases (EUCERD) took place on 31 January – 1 February 2013 in Luxembourg: the plenary session was preceded by breakout sessions by stakeholder group to discuss the draft EUCERD Recommendations on Common Principles and Consensus on patient registries and data collection for rare diseases.

- ***European Reference Networks***

The EUCERD adopted unanimously the Recommendations on European Reference Networks for Rare Diseases[1] elaborated during a previous workshop and meetings of the EUCERD. The adopted recommendations will be shared with the Expert Group on Cross-Border Healthcare so as to be used, in terms of the specificities of rare diseases, in their future work in elaborating criteria for the ERNs in the context of the implementation of the Cross-Border Healthcare Directive. Following the EUCERD meeting, the Recommendations were presented to the Expert Group on 6 February 2012. The EUCERD will keep in close contact with the Expert Group concerning next steps.

- ***Registries***

The discussions of the stakeholder breakout sessions concerning the first draft of the EUCERD Recommendations on Common Principles and Consensus on Patient Registries and Data Collection for rare diseases were reported on during the plenary session. It was decided to revise the drafted recommendations according to discussions and to send the new draft to Members for their input during one month by the end of February 2013. An EPIRARE/EUCERD Joint Action workshop will be held on 22-23 April 2013 in Brussels to further discuss the new draft, which could be then proposed for further discussion at the next EUCERD meeting before adoption. The EPIRARE/ EUCERD Joint Action workshop will also be aimed at working on a technical document of possible policy scenarios for rare disease patient registration concerning the direction the proposed European registry platform to be based at the EC's Joint Research Centre in Ispra could take to meet the needs of the various actors in the field.

Some members of the EUCERD expressed their concern that the proposed General Data Protection Regulation threatens data collection for rare diseases. The position being formulated by EURORDIS

[1] http://www.eucerd.eu/?post_type=document&p=2207

on this subject will be shared with the EUCERD so that they may contact their MEPs with their concerns.

- ***Coding and classification of rare diseases***

Members were provided with an update on the revision process concerning rare diseases for the update of the World Health Organization's International Classification for Diseases (ICD) which is due to be adopted at the World Health Assembly in 2015. The revision process for rare diseases is coordinated through the EUCERD Joint Action, which also supports the ongoing efforts to cross-reference ICD, SNOMED-CT, OMIM, MeSH, MedDRA and HPO with the Orpha codes: the Orphanet classification is the only classification specific to rare diseases. A EUCERD Joint Action/Eurogentest workshop in September 2012 brought together experts from each of these nomenclatures to establish a number of recommendations and actions for the future[2] as well as to set up the International Phenotype Terminology Consortium. Member States were encouraged to consider the use of the Orphacode in addition to ICD/SNOMED in their information systems. The EUCERD will explore liaison possibilities with EUROSTAT in this area and the EC agreed to approach the WHO concerning a long-term collaboration for the update of rare diseases in ICD.

- ***Genetic testing in Europe***

The outcomes of an expert workshop, organised by the EC's Joint Research Centre in Ispra with the support of Eurogentest and EUCERD in November, to discuss a number of areas where European cooperation could give added-value in terms of the quality of genetic services and the organisation of genetic services, were presented. The topics discussed included the organisation of quality assessment schemes, the challenge presented by next generation sequencing techniques and direct-to-consumer testing. Areas for harmonisation at European level could be envisaged, and the outcomes of the workshop will be considered by the EC. The EUCERD will discuss the possibility of elaborating recommendations in this area based on the outcomes of this workshop at the next meeting.

- ***New Born Screening***

At the previous meeting of the EUCERD, the Members decided to review the prioritisation of topics that could be dealt with in this field at European level in terms of the quality of screening and the efficiency of newborn screening processes. A document was prepared proposing a ranking of 10 points for action, and MS were asked to prioritise the topics in order to assess whether there was a consensus concerning the priority areas. During the discussion, it was clear that there was insufficient consensus concerning the priority areas to be tackled in this field to further work on a recommendation of the EUCERD on this topic, it was decided to draft a report of the work carried out at the EUCERD meetings and discussions to reflect the opinion of EUCERD Member State representatives, particularly that this is a primarily national matter and that the exchange of information be promoted so effort is shared at the level of Health Technology Assessment agencies.

- ***National plans and strategies for rare diseases***

The EUCERD heard from each Member State representatives on the elaboration and/or implementation of their national plans or strategies for rare diseases which the Council

[2] http://www.eucerd.eu/?post_type=document&p=1728

Recommendation on an Action in the Field of Rare Diseases encourages Member States to do before the end of 2013. Member States were asked in particular about their approaches to rare disease registries.

- ***EUCERD Joint Action (EJA)***

Members were given an update on the progress to date with the EUCERD Joint Action which had not yet been covered in the meeting and the scheduled activities for the year ahead:

- The annual meeting of the partners of the EUCERD Joint Action was announced for 19-20 February 2013.
- Work has started on a EUCERD Recommendation for Indicators for National Plans/Strategies for Rare Diseases, to be further discussed at the EJA Europlan meeting on 25 March 2013 in Rome. The recommendations will be sent to the EUCERD for input at the end of April 2013 before finalisation by the Bureau with view to possible adoption at the EUCERD meeting in June 2013.
- The elaboration of the annual EUCERD Report on the State of the Art of Rare Diseases Activities in Europe[3] is underway. Member States representatives received on 6 February 2013 the updated information concerning their country's activities in 2012 for further elaboration by stakeholders at national level before 31 March 2013. The final version for publication will be sent for MS approval in May 2013 to ensure publication in early July 2013.
- It was suggested for the EJA to work on the topic of rare disease identification cards.

- ***International Rare Diseases Research Consortium (IRDiRC)***

Members were informed of the activities of the newly established Scientific Secretariat of the IRDiRC which is based at the Rare Disease Platform in Paris to support the Scientific Committees and Working Groups of the Consortium. A new website www.irdirc.org has been created to provide information concerning the IRDiRC's activities. The IRDiRC's first conference will be held in Dublin on 16-17 April 2013 and registration is now open for the remaining places: http://jk-events.com/IRDiRC2013/.

- ***Rare Disease Day 2013***

The plans for Rare Disease Day (28 February 2013) were presented: the theme will be International Solidarity with the slogan 'Rare Disorders Without Borders'. EUCERD members were encouraged to participate in a number of ways:

- By engaging with and supporting their National Alliances coordinating the RDD campaign at national level as well as attending/endorsing/co-organising national advocacy events.
- By participating at the public hearing on 26 February 2013 at the European Parliament in Brussels and to encourage their MEPs to attend.
- By becoming friends of RDD through the website, placing the RDD logo and web address on their website, and by raising and joining hands on the day and submitting an official photo of their institution.

[3] http://www.eucerd.eu/?page_id=163#StateArt

- ***Other business***

The EUROCAT recommendations on policies to be considered for the primary prevention of congenital anomalies in national plans and strategies on rare diseases were presented to the Members. This document will be made accessible via the EUCERD website and other ways in which this information could be disseminated will be envisaged.

The question of long term financing solutions for Orphanet was raised by Members : the EC foresees this issue to be discussed within the future Health Programme. Member State representatives were encouraged to examine national solutions in the short-term with their Orphanet country coordinator.

- ***Next meetings***

The next meeting of the EUCERD is planned for 5-6 June 2013, with the third of the year scheduled for 24-25 October 2013.