8th Meeting of the European Union Committee of Experts on Rare Diseases

5-6 June 2013, Luxembourg

Executive Summary

The 8th Meeting of the European Union Committee of Experts on Rare Diseases (EUCERD) was held on the 5-6 June 2013 in Luxembourg and marked the last meeting of the current mandate. The EUCERD's Bureau and Members bowed out having received the thanks of the European Commission from the Director for Public Health, Mr. John F. Ryan, with the adoption of five recommendations as proof of the previous two and half years of hard work and consensus building.

- **Update on the future of the EUCERD activities**

The EUCERD's current mandate ends in Summer 2013. The new Commission Decision concerning the new group of experts on rare diseases is intended to be published soon and will be followed by the call for expression of interest. It is hoped that a meeting of the new group will be possible in late autumn 2013. The new group will adopt at their first meeting rules of procedure. In the transition phase, the EC will interact with the current Bureau.

- **Adoption of the draft EUCERD Recommendations on Patient Registries and Data Collection for Rare Diseases**

The recommendation was adopted by full consensus by the EUCERD. Rare disease registries are valuable instruments for increasing knowledge on rare diseases, and for supporting fundamental, clinical and epidemiological research, as well as for post-marketing surveillance of orphan medicinal products and medicines used off-label. This data is also crucial for the planning of healthcare services. The recommendation calls for the international operability of registries and databases and use of appropriate coding systems to enable the necessary pooling of data for public health and research purposes, gives advice concerning the establishment of registries and collection of data, highlights the various uses of patient data and how to best share this information, underlines the importance of adherence to good practice guidelines in the field, stresses the need for registries to be adaptable to meet future needs, and emphasises the importance of sustainability for the time span of the registry's utility.

- **Possible policy scenarios for rare disease patient registration in view to the establishment of a European platform for registries**

The EUCERD were presented with the outcomes of the EUCERD Joint Action/EPIRARE workshop which was organised on 22-23 April 2013 in Paris to gather the input of different stakeholders concerning the proposed European platform for rare diseases registries to be created at the EC's Joint Research Centre (JRC) in Ispra, Italy. The breakout sessions prior to the EUCERD plenary were

also the occasion to collect the views of the Committee concerning the needs of different stakeholders in the field and priorities in the context of a number of possible policy scenarios. Four main missions for the platform were identified by the EUCERD: support for new registries, promotion of the interoperability of existing stand-alone registries, provision of a hub for access to all data collection in the field of rare diseases, and provision of IT tools to maintain already existing data collection. The discussions of the workshop and EUCERD meeting is to be considered by the EC and integrated into the concept paper being developed internally by the EC, which should be presented at a future meeting of the new committee. The Members were also informed that the administrative agreement between the EC and the JRC in view to the establishment of this platform is being finalised at the moment.

- **EUCERD Recommendation on Core Indicators for National Plans/Strategies for Rare Diseases**

The second set of recommendations adopted at the meeting was the EUCERD's Recommendations on Core Indicators for National Plans/Strategies for Rare Diseases. This recommendation provides a list of 21 core indicators which are intended to capture relevant data and information on the process of planning and implementing of these plans and strategies on a regular basis at national level. These indicators will provide information notably to the European Commission on the implementation of the Council Recommendation on an Action in the field of Rare Diseases which encourages Member States to establish a national plan or strategy in the field by the end of 2013. They will also serve as a basis for the elaboration of indicators at national level tailored to the specific actions foreseen in the plans/strategies. This set of recommendations will be revised in the future to take into account the experiences of the Member States.

- **EUCERD Opinion on areas of potential European collaboration in the field of New Born Screening**

The EUCERD Members reviewed the proposed final draft of the EUCERD Opinion on areas of potential European collaboration in the field of New Born Screening, which includes 11 areas which would potentially welcome European collaboration in the field. This document is being adopted by written procedure and will be made available mid-July 2013. The opinion is destined for the EC, Member States and interested third-parties. The EC will also receive a full report of the various discussions on this subject over the previous meetings in order to facilitate policy-making decisions in this area.

- **Possible topics for EUCERD recommendations on areas of action at European level in the field of genetic testing**

Following the expert workshop organised in November 2012 by the JRC in Ispra in collaboration with Eurogentest and the EUCERD, a set of possible topics for EUCERD recommendations on areas of action at European level in the field of genetic testing. These possible recommendations are directly drawn from the report of the workshop report[1] and identify a number of areas and action points covering the organisation of genetic services, quality assurance, next generation sequencing and direct-to-consumer testing. It was decided to revisit this proposal, which will be considered as a pre-draft, at the next meeting of the new Committee for a full discussion and decision on next steps.

[1] http://ihcp.jrc.ec.europa.eu/our_activities/public-health/jrc-publishes-a-report-entitled-genetic-testing-offer-in-europe/at_multi_download/file?name=finalreportWSgenetic%20testing.pdf

- **ERIC on rare diseases information for research purposes**

The EUCERD was given more information concerning an idea to establish a European Research Infrastructure Consortium (ERIC) on Rare Diseases information for research purposes. An ERIC would provide a legal status for a structure which could regroup a number of existing, or foreseen, networking activities in the field of rare diseases. To initiate the discussion on this ERIC, an invitation has been addressed to the Permanent Representation of Member States, with copy to the EUCERD MS representatives, inviting them to a preparatory meeting on 17 June 2013 in Ispra to find out more concerning the role of MS, to gauge interest, and to give information on the process for application to the Commission. The EC is interested in facilitating this process up to the stage of application for an ERIC to the Commission, but the content will be determined by the MS. The EUCERD Members agreed that the real added value of an ERIC must be determined before it is decided to apply for this status.

- **Commission Report on the Implementation of the Commission Communication and Council Recommendation**

A questionnaire has been addressed to MS by the EC via the permanent representations to help the EC elaborate the implementation report concerning the Commission Communication and Council Recommendation on an action in the area of rare diseases which is due by the end of 2013.

- **Outcomes of the working group on coordinated access to orphan medicinal products (MoCA)**

The EUCERD was presented with the outcomes[2] of the working group on coordinated access to orphan medicinal products (MoCA). This working group was situated within the context of a platform within DG Enterprise's process on Corporate Responsibility in the Field of Pharmaceuticals which aimed to find common, non-regulatory approaches to ensure better, timely and equitable access to medicines after their marketing authorisation. The working group has brought a number of Member States and stakeholders together to establish a dialogue on a range of subjects (assessing OMPs, organising structural access, and organising individual access (bearing in mind issues of sustainability). One of the achievements was the establishment of a European Transparent Value Framework, to provide a common basis for further discussions in this area. In addition the group proposed the establishment of an *ad hoc* taskforce with the voluntary collaboration of EU/EEA countries and stakeholders to continue these discussions, and to monitor which steps initiatives have been taken/are envisaged, as well as to test the proposed framework through concrete pilots.

- **Update on the elaboration/implementation of national plans/strategies for rare diseases**

Member States representatives gave short updates on the process at national level to either elaborate or implement national plans/strategies for rare diseases as encouraged by the Council Recommendation. Although most countries will have adopted a plan by the end of 2013, the issue of adequate financial support for the implementation remains problematic due to the economic climate. The EUCERD also heard more from the project at the German Institute of Medical Documentation and Information (DIMDI) to include the Orpha codes for rare diseases in their coding

2 http://ec.europa.eu/enterprise/sectors/healthcare/competitiveness/process_on_corporate_responsibility/platform_access/index_en.htm

system. An information leaflet on the Orpha code[3] has been produced to provide more information on the use and benefits of the codes.

- **Update on the EUCERD Joint Action**

Members were updated on the activities of the EUCERD Joint Action which is at its 16th month. Two workshops are planned for this year: the Workshop on Training for Specialised Social Service Providers, 10-11 October 2013 in Copenhagen, and the Workshop to present the preliminary results of the study and interviews with centres of expertise organised by work package 7, on 11-12 December 2013 in Madrid. The State of the Art of Rare Diseases Activities in Europe report for 2013 (covering up to the end of 2012) is in the process of being finalised and should be published early this summer and will be conveyed to members and announced in OrphaNews Europe. Members were warmly thanked for their contributions and asked to disseminate the report at national level, especially the country sections, when they become available.

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

The IRDiRC Policies and Guidelines were presented, which were recently adopted by the Executive Committee. This document is aimed at researchers and funding bodies[4]. Next steps include the elaboration of the IRDiRC road map using proposals from the 12 working groups in June-July 2013, so as to submit a draft to the Executive Committee at the end of September. A survey of trends and gaps is expected to analyse the projects funded by IRDiRC members and identify possible areas of collaboration and resources/tools/data to be promoted. An overview of the outcomes of the IRDiRC's first conference on 16-17 April 2013 was also presented.

- **Preparation of the European Conference on Rare Diseases and Orphan Products 2014**

The EUCERD heard about the plans for the European Conference on Rare Diseases and Orphan Products which will be held in Berlin and organised by EURORDIS with the DIA on 9-10 May 2014. EUCERD members are present in the Programme Committee, and will be encouraged to reply to the call for posters, to disseminate information concerning the conference, and to attend the conference and networking reception. More information on the conference will soon be available via the official website www.rare-diseases.eu.

- **Progress with the 116 number for rare diseases**

Members were updated concerning the process to reserve a unique European phone number for access to rare disease help lines. EURORDIS is in the process of preparing an information pack on this subject to share with the EUCERD in order to further discuss this topic at the next plenary. In the meanwhile MS can help identify the relevant contact point in the appropriate Ministry, usually that of Industry.

[3] http://www.eucerd.eu/wp-content/uploads/2013/06/OrphaCode_Leaflet.pdf
[4] http://www.irdirc.org/wp-content/uploads/2013/06/IRDiRC_policies_24MayApr2013.pdf

- **Proposal for a European Year of Rare Diseases in 2019**

EURORDIS presented a proposal and timeline for requesting a European Year of Rare Diseases in 2019. A document with more information will be presented at the next EUCERD meeting to be adopted so as to have a show of support from the Committee for this initiative.