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

14 & 15 November 2012

Executive Summary

The sixth meeting of the European Union Committee of Experts on Rare Diseases (EUCERD) took place on 14 -15 November 2012 in Luxembourg: the plenary session was preceded by breakout sessions by stakeholder group to discuss the draft EUCERD Recommendations on European Reference Networks for Rare Diseases.

- ***European Reference Networks***

The outcomes of the EUCERD Joint Action workshop to elaborate the draft EUCERD Recommendations on European Reference Networks (ERNs) for Rare Diseases (RDs) were presented, the comments heard at the breakout sessions on the draft sent for discussion prior to the meeting were reported to the plenary. This draft has now been revised taking into account this comment and has been sent to the Committee for final comments before the document is finalised for adoption at the next meeting of the EUCERD in January 2013.

EUCERD members also heard from the European Commission on the development of the implementation of the Directive 2011/24/EU on the application of patients' rights in cross-border healthcare led by the Committee on Cross-Border Healthcare and the Cross-Border Healthcare Expert Group, in terms of the preparation of the Delegated and Implementing acts. Notably, the EUCERD members were informed of the public consultation[1] on the implementation of European Reference Networks open until 24 January 2013. The EUCERD and the Expert Group will keep each other informed of their work in the field of ERNs and a representative of the EUCERD will attend the next meeting of this Group to ensure effective cross-talk, as the work of the two groups is taking place in parallel.

- ***Registries***

The EUCERD heard from the European Commission on the proposed European Union framework for action in the field of rare diseases registration. The European Commission has financed around 50 registries for rare diseases, as well as a number of initiatives such as EPIRARE (to build consensus and synergies to address regulatory, ethical and technical issues associated with the registration of RD patients and to elaborate possible policy scenarios as well as to evaluate the experience of the registries in place and the most appropriate criterion to use EU funding) in addition to the PARENT (Cross-Border Patient Registries Initiative) and RD-Connect (focused on registry initiatives for

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[1] http://ec.europa.eu/health/cross_border_care/consultations/cons_implementation_ern_en.htm

research purposes) initiatives. The European Commission now intends to create an EU platform for RD registration to further integrate registry activities: this platform would be established via an administrative agreement with the Joint Research Centre in Ispra, Italy, and €2 million has been foreseen the Work Plan 2013 of the Second Health Programme for this ‘support to rare diseases registries and networks in view of their sustainability’. The platform would provide a range of services to registries and to aggregate data for re-use from different registries using a set of standardised Common Data Elements.

The outcomes of the EUCERD Joint Action Workshop on Rare Diseases Registries, held prior to the EUCERD on 13 November 2012 were presented. This workshop heard from the different initiatives in the area, and discussed the Commission proposal for a platform in the light of the past and current work in this area at EU level. The workshop concluded that the EUCERD would be the appropriate advisory body for an EU platform for rare disease registration and that the EUCERD Joint Action and EPIRARE projects should have the flexibility to work together to produce recommendations for the development of this platform.

During the plenary, Members welcomed the willingness of the Commission to create a platform, albeit a little too soon seeing as the projects aimed at examining this possibility (EPIRARE and RD-Connect), have yet to come to term. It was proposed and accepted to produce two EUCERD documents in the field of registries:

1. A **policy document on key principles and consensus** based on the work of existing projects in the field to guide discussions on data collection and registries at ERN and national plan/strategy levels. This would be a EUCERD Recommendation;
2. A **technical document of policy scenarios** concerning the direction such a platform could take to meet the needs of the various actors in the field

These documents will be elaborated through two different workshops in the framework of the EUCERD Joint Action and the EPIRARE project to be scheduled for the first semester of 2013, and will be scheduled for possible adoption at the June 2013 EUCERD meeting.

- ***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. It was encouraging to hear that progress is being made in all countries even if there may not be a budget for all the planned actions.

The outcomes of the EUCERD Joint Action Europlan Inception Workshop held in Rome in September 2012 were presented: this workshop was aimed at constituting a network of people involved in the planning and implementation of national plans/strategies in order to hear the needs of different Member States in terms of European level technical support and to further refine the indicators developed during the Europlan project for use in monitoring and evaluating these plans/strategies.

The plans for the organisation of the 20 national plans and strategies foreseen in the EUCERD Joint Action Europlan workpackage were presented. These conferences will take place in 2013 and will be organised by the National Alliances of patient organisations with the support of EURORDIS. They will aim to discuss with a wide range of stakeholders the themes presented in the Council Recommendation, to examine the transferability to the national context of the Commission Communication and Council Recommendation as well as the EUCERD recommendations, to present the EUROPLAN recommendations and a selection of indicators, and to discuss the national situation and next steps. The conferences will be outcome-orientated and will have sufficient flexibility to reply to the needs of each country.

Members of the EUCERD were keen for summaries of the existing national plans/strategies for rare diseases to be made available so as to facilitate the exchange of experience between countries.

- ***EUCERD Joint Action***

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:

- A number of workshops are planned in the scope of the EUCERD Joint Action for 2013, including: the second Europlan workshop, several national conferences on rare diseases, follow-up workshops on registry policy scenarios in cooperation with EPIRARE, a workshop on centres of expertise once initial work has been completed, and training workshops for social service providers.
- The coding and classification workpackage held a workshop[2] in September 2012 bringing together representatives of all major classification systems from across the world. Outcomes included the decision that it is not reasonable to have a unique terminology to code phenome elements due to the multitude of needs of the different communities, however a core set of 2000 terms should be agreed on at international level and recommended for use as well as proposed to SNOMED CT and ICD. The participants also decided to establish the International Consortium of Phenotype Terminologies so as to be in the position to update the proposed core terms and cross-referencing of terminologies.
- The Specialised Social Services workpackage is progressing with the identification and mapping of Therapeutic Recreational Programmes and Respite Care Services and two country visits have taken place and interview guides for these visits have been produced. A paper on the needs for Specialised Social Services and integration of these services into Social Policy is being drafted to be distributed to the EUCERD in 2013.
- The 'integration' workpackage, in addition to the work on ERNs and registries, continues to liaise with the European Partnership Action Against Cancer and EUNetHTA to promote effective cross-talk in mutual areas of interest. Liaison with PARENT JA, RD-Connect the IRDiRC is also planned.
- In terms of communication, the new EUCERD website is now online and the 2012 EUCERD Report on the State of the Art of Rare Disease Activities has been published, with the 2013 edition to be elaborated over the coming months in collaboration with the EUCERD.

- ***New Born Screening***

Members discussed the potential areas for further action at European level proposed by the Scientific Secretariat. It was decided to streamline the proposed areas of discussion in the document and ask MS representatives to decide on the prioritisation of the topics for discussion. There will be time to discuss this prioritisation whilst the Bureau determines if funding for a workshop on these topics is possible.

- ***Clinical Added Value of Orphan Medicinal Products (CAVOMP)***

The EUCERD Recommendations concerning the CAVOMP information flow have been transmitted to the European Commission for their consideration and a decision on the next steps as to the information flow's implementation. As with all the other recommendations of the EUCERD, dissemination of this recommendation is key. Members were encouraged to participate in strategic and adhoc dissemination of the recommendations. The Scientific Secretariat will produce a short Executive Summmaries and powerpoint presentations for re-use by Members to help them with this dissemination effort, including the core elements to be conveyed.

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

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

The Members were given an update on the Scientific Secretariat activities which will be supported by the FP7 Support-IRDiRC action. The action will provide support to the members of the IRDiRC's Scientific Committees and their activities, will collect and disseminate pertinent information and results concerning ongoing research projects, will enable and implement cooperation with other stakeholders, and will monitor progress towards goals. Updates will be provided via the OrphaNews Europe newsletter, reports and the IRDiRC website. Members were encouraged to participate in the IRDiRC conference next year in Dublin (16-17 April 2013).

- ***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.

- ***Other business***

Members were encouraged to widely disseminate the 2012 EUCERD Report on the State of the Art of Rare Disease Activities in Europe[3].

Member States representatives were encouraged to take note of the revision of the data protection directive, as this will have an impact in the field of registries, and to keep in touch with those involved in the process at national level.

- ***Next meeting***

The 7th Meeting of the EUCERD will be held on 31 January & 1 February 2013, with a workshop on 30 January 2013 probably on registries.

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