



5th Meeting of the European Union Committee of Experts on Rare Diseases

20-21 June 2012

Executive Summary

The fifth meeting of the European Union Committee of Experts on Rare Diseases (EUCERD) took place on 20-21 June 2012 in Luxembourg.

- **CLINICAL ADDED VALUE OF ORPHAN MEDICINAL PRODUCTS (CAVOMP)**

The EUCERD reviewed the final draft of the « EUCERD Recommendations to the European Commission and Member States on improving informed decisions based on the clinical added value of orphan medicinal products information flow ». The modifications made to this draft during an extended drafting meeting on 19 June 2012, which brought together the key actors in the proposed information flow, were explained in detail and responses were given to the questions and suggestions made by the EUCERD during the 4 week consultation period prior to the meeting. The EUCERD Members were in agreement with the changes made and the recommendations were provisionally adopted with the adoption to be confirmed via written procedure. The next step will be to organise a meeting of the Bureau as well as a representative of Industry and Member States with Mrs Paola Testori Coggi to present these recommendations and the discuss the implementation process.

- **EUROPEAN REFERENCE NETWORKS**

Another key topic at the meeting was the issue of the European Reference Networks (ERNs) for Rare Diseases in the context of the Directive on Cross-Border Healthcare. The European Commission gave an update on the recent steps in the process of implementing this Directive. In addition, the Commission requested the opinion of the EUCERD for October 2012 on a number of questions concerning the implementation of the Directive 2011/24/EU on the application of patients' rights in cross-border healthcare with regard to RD patients. The Bureau will prepare a first draft of the reply and will send this for the EUCERD's consultation and contributions with the aim of obtaining a consensus by October 2012 as requested. The EUCERD also discussed a first draft of the EUCERD Recommendations on ERNs for rare diseases, which are to be discussed and elaborated further during a workshop planned in the context of the EUCERD Joint Action on 25-26 September 2012 in Newcastle. These recommendations are to consider elements such as the scope and governance, capacity building and tools and resources to support ERNs, and quality assurance for ERNs.

These recommendations will be proposed for adoption at the next meeting of the EUCERD in November 2012.

- **EUCERD JOINT ACTION: WORKING FOR RARE DISEASES**

The coordinator of the EUCERD Joint Action, which started on 1 March 2012 for a duration of 42 months, gave an overview of the planned work and workshops in the coming months. The Kick-Off meeting of the associated partners was held on 12-13 March 2012 in Paris. There are various activity areas:

- An update on the elaboration of the 2012 EUCERD Report on the State of the Art of Rare Disease Activities in Europe was presented: the report is due to be published in July 2012.
- The new website of the EUCERD, developed in the context of the Joint Action, was demonstrated, along with the new EUCERD logo. The website will have information concerning the EUCERD activities as well as the activities of the Joint Action.
- The EUCERD Joint Action will provide support to the process of elaboration of national plans/strategies for rare diseases and to this end a workshop will be held in Rome on 10-11 September 2012 to bring together the persons elaborating national plans or strategies for rare diseases at national level to exchange on their experiences and difficulties. The plans for the second round of 'Europlan' conferences were also detailed as were the countries interested in hosting one of these conferences.
- Recent developments concerning the work financed by the EUCERD Joint Action on the revision of the International Classification of Diseases were presented. Experts were advised to give their feedback on the ICD-11 beta version which is now online in three months time in order to signal the remaining issues concerning rare diseases to the WHO. Members were encouraged to promote the use of the Orphacode as an alternative until ICD11 is implemented in the planning of their national plans/strategies for rare diseases. An expert workshop will be held in the context of the EUCERD Joint Action on the cross-referencing of terminologies on 27-28 September 2012 in Paris.
- A first workshop on the work package on specialised social services is being planned for 6-7 December 2012 in Zalau, Romania at the NoRo Centre. Members were encouraged to help identify appropriate participants, especially in the relevant authorities.
- A workshop on registries in the scope of the integration workpackage is scheduled for 13 November 2012 in Luxembourg ahead of the next meeting of the EUCERD and will bring together members from all relevant initiatives and in particular Epirare and the Parent Joint Action to work on a EUCERD recommendation on rare disease registries.

- **NEW BORN SCREENING**

A proposal for further action in the field of newborn screening prepared by the EUCERD Scientific Secretariat was discussed by the Members: this follows a request from the Commission to consider the results of the Tender on New Born Screening financed by the Health Programme and to issue proposals for next steps. A workshop will be planned for the

first trimester of 2013 to discuss possible areas where collaboration and information-sharing at EU level could take place in order to improve newborn screening practices in Member States and elements that could be shared at EU level. A document will be prepared by the Bureau ahead of a workshop of the EUCERD in the first trimester of 2013 to identify future steps and to inform the Commission of the direction the EUCERD is advising to take in this area.

- **COLLABORATION WITH EUROPEAN PARTNERSHIP ACTION AGAINST CANCER**

A presentation was given of the potential areas of collaboration between the EUCERD Joint Action and the European Partnership Action Against Cancer. This discussion is at the preliminary stages based on fact-finding with the objective of making sure that work is not duplicated in these areas. A number of areas for collaboration will be identified and the outcomes of collaboration monitored.

- **JOINT RESEARCH CENTRE WORKSHOP ON THE GENETIC TESTING OFFER IN EUROPE**

Details of the workshop being organised by the Joint Research Centre in collaboration with Eurogentest and the EUCERD on 19-20 November 2012 in Ispra were presented to the EUCERD. This workshop will deal with a number of priority topics with the aim of producing recommendations for consideration by the EUCERD. These topics include the accreditation, certification and External Quality Assessment of genetic laboratories and tests, as well as ways to rationalise testing services. The workshop will also discuss the future of genetic testing services as regards the latest advances in technology.

- **FRAMEWORK PROGRAMME 7/HORIZON 2020 & THE INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM (IRDiRC)**

An overview of the 2012 work programme of the Health Theme of FP7 and the content of the future call for proposals, expected to be opened in July 2012, was presented to the EUCERD. There will be no specific rare disease area in this call, but areas applicable to rare diseases were highlighted. More details were given concerning the likely structure of the Horizon 2020 programme, due for publication in November 2012. An update was also given on the next steps for the IRDiRC: the strategy and communication plans of the Consortium will be presented at the next meeting of the EUCERD. The EUCERD was presented a short promotional video concerning the initiative.

- **UPDATE ON THE ADVANCEMENT OF NATIONAL PLANS/STRATEGIES FOR RARE DISEASES**

The EUCERD Member State representatives were invited to give an update to the Committee on the latest steps forward in the elaboration/implementation of their national plans/strategies for rare diseases. On the whole, much progress has been made in 2012, although the implementation of these plans may be difficult due to the current economic climate.

- **HEALTH AND GROWTH PROGRAMME 2014-2020**

The Commission proposal for the future health programme, entitled provisionally 'health and growth' was outlined. Rare diseases and European Reference Networks are explicitly

mentioned in the proposal. The timeframe and procedure for the adoption of the programme was explained to the EUCERD.

- **EUROPEAN CONFERENCE ON RARE DISEASES AND ORPHAN PRODUCTS**

The outcomes of the European Conference on Rare Diseases and Orphan Products, held on 23-25 May 2012 in Brussels, were presented to the EUCERD. The conference, organised by EURORDIS in collaboration with the DIA (Drug Information Association) was a great success with many EUCERD members participating, with a greater proportion of researchers and policy makers participating at this year's edition than in previous conferences. The next conference will be held in Berlin in 2014. The EURORDIS Membership Meeting 2013 will be taking place end of May in Dubrovnik, Croatia, with capacity building workshops on national plans and strategies.

- **OTHER BUSINESS**

Members were presented at the meeting with translations into their language of the "EUCERD Recommendations on Quality Criteria for Centres of Expertise for Rare Diseases in Member States".

The EUCERD will be asked via written procedure if it wishes to maintain the current Bureau for one more year.

- **NEXT MEETING**

The tentative dates for the sixth meeting of the EUCERD are 14-15 November 2012 in Luxembourg.