



**SECOND MEETING OF THE EUROPEAN UNION
COMMITTEE OF EXPERTS ON RARE DISEASES**

SUMMARY REPORT

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DECISIONS ON MEMBERSHIP OF THE EUCERD AND ON THE RULES OF PROCEDURE

Members requested that the rules of procedure be followed to the letter and that documents should be sent ahead of meetings at least 2 weeks prior to the meeting, and preferably 4 weeks prior to the meeting to allow, in particular, Member States representatives to consult their hierarchy.

Many countries outside of the EU are interested in developments at European level in the field of rare diseases: the Bureau proposed to include representatives of countries outside the EU (especially those preparing national plans/strategies for rare diseases) to attend meetings of the Committee as observers to participate in discussions and to automatically receive the documents circulated to members. As observers they will not have the power to vote nor will their approval of documents be sought.

UPDATE ON THE ACTIVITIES OF THE SCIENTIFIC SECRETARIAT OF THE EUCERD

EUCERD WORKSHOPS 2011

- ***Workshop on centres of expertise for rare diseases and networking of centres of expertise (21-22 March 2011)***

A workshop on national centres of expertise for rare diseases and networking of national centres of expertise was held prior to the meeting on 21-22 March 2011. The work shop was discussed topics such as the models for expert care (health care pathways, coordinating centres and expert centres), the scope of national expert centres (including disease coverage), European Reference Networks and networking between expert centres: the work shop participants also revisited the criteria for national centres of expertise established by the RDTF/High Level Group on Health Services for Medical Care.

It was proposed that the discussions on the quality criteria for national centres of expertise would be used to fine tune these criteria to be proposed as a EUCERD recommendation for adoption at the next meeting of the EUCERD, which will help MS in their reflections or policy developments concerning national plans and strategies for rare diseases. This recommendation will be revised in around 2 years by the EUCERD based on the experiences of EU MS in the field. A synthesis will be worked on to form a EUCERD strategic vision on health care pathways and networks based on the Commission Communication and Council Recommendation, taking into account ERN and health care organization at local, national and EU levels, to be presented at an upcoming meeting of the EUCERD for further consultation. The EUCERD will develop and adopt guidelines on ERN to provide a strategic vision and quality criteria for implementation in the field of rare diseases.

Member states representatives expressed the need for work to be done in the area of national centres of expertise which should be seen as priority area.

- **Workshop on disease-focused versus drug-focused registries (date to be confirmed)**

A workshop on this topic in collaboration with the EMA will be organised in London. The workshop will focus on the current situation in the EU in this field, will consider the usefulness of data for drug development, registration and post-marketing surveillances and also discuss steps towards moving to from the current situation to publicly-owned, multi-purpose databases.

- **Workshop on rare diseases in health information systems (4 November 2011)**

This workshop will be held in the context of the *Rare2011* rare diseases conference organised by EuroBioMed in France which will be held over 2-4 November 2011 in Montpellier, France.

PROPOSITIONS FOR ADDITIONAL WORKSHOPS IN 2011

Two extra workshops on topics the EUCERD sees as important could be organised in 2011. Suggestions included:

- An additional workshop on the technical and qualitative criteria for the designation of national centres of expertise for RD and ERN for RD.
- A technical workshop on the classification of genetic diseases in ICD with expert groups.

UPDATE ON THE ELABORATION OF THE EUCERD REPORT ON THE STATE OF THE ART OF RARE DISEASES ACTIVITIES IN EUROPE

The Scientific Secretariat is finalising the draft of information on each Member State and additional countries from the previous report¹ published in July 2010. During April 2011 the country sections will be sent to stakeholders at country level for further elaboration (including EUCERD MS representatives and alternates, contributors to the previous reports, national COMP representatives, National RD patient alliances, Orphanet country coordinators and other identified experts). The editorial process for further editions of the report will be presented at the next EUCERD meeting.

REPORT ON RARE DISEASE DAY – 28 FEBRUARY 2011

Rare Disease Day (RDD) is an annual, awareness-raising campaign coordinated by EURORDIS at the international level in collaboration with National Alliances of Patient Organisations at the national level. Individuals, patients, patient organisations, health professionals, researchers, drug developers, public health authorities can all take part in the event as “Friends of Rare Disease Day”. The theme of this year’s event was “Rare Diseases & Health Inequalities” with the slogan “Rare but Equal”. 55 countries from across the globe participated in 2011 and the www.rare-disease.org site saw an increased traffic on last year with nearly 50,000 visits (from 1 January to 3 March 2011) from 150 countries with 10,000 alone on the 28 February. RDD also used social networks to its advantage with over 14 000 Facebook fans, 1,446 Twitter followers, 432 photos on Flickr and 100 videos on YouTube. Announcements towards national plans came from Ireland, Croatia, Luxembourg, Hungary and Spain. Highlights from across the globe were given, including the designation by the US Senate of 28th

¹ <http://www.orpha.net/nestasso/EUCERD/upload/file/Reports/2009ReportInitiativesIncentives.pdf>

February 2011 as RDD, and the Pope giving a personal message to RD patients and families in St Peter's Square. In addition a European Symposium entitled "Rare but Equal - Addressing Health Inequalities for Rare Disease Patients in Europe" was organised by EURORDIS with the support of DG Sanco with 86 participants: 380 people tuned into the live streaming. Next year the event will be held on 29th February 2011. The next step is to make this an internationally recognised day.

The EUCERD endorsed Rare Disease Day and will add the weight of the Committee to these actions. RDD continues to provide excellent opportunities to launch initiatives and gain publicity and may be the best date to launch EUCERD reports such as that on the State of the Art of Rare Diseases Activities in Europe.

PRESENTATION OF THE RESULTS OF THE EUROBAROMETER ON AWARENESS OF EUROPEANS ON RARE DISEASES

The results of the Eurobarometer on the awareness of Europeans on RD were presented to the EUCERD (72.3²). This survey was carried out through interviews of 27 000 people in the EU (for 500 to 1000 people in each EU MS) between 27 November and 15 December 2010. This questionnaire was established in June 2010 with the collaboration of Orphanet and EURORDIS and aimed to investigate the awareness of Europeans of RD. At national level only France has carried out this sort of survey before. The overall results are available in English, French and German. This Eurobarometer shows that we do something that is supported and appreciated.

FINAL REPORT AND CONCLUSIONS OF THE EUROPLAN PROJECT

This three-year project will end at the end of March 2011 and there is planned continuation of the work of the project in the context of the 2011 Joint Action on Rare Diseases. The project has resulted in a joint report with the Scientific Secretariat of the EUCERD on initiatives and incentives in the field of RD at national level, a guidance document containing the EUROPLAN recommendations for the elaboration of the national plans or strategies for rare diseases, a report on indicators for monitoring the implementation and evaluating the impact of national plan or strategy for rare diseases, the organization of 15 EUROPLAN National Conferences and a report on the results.

KEY CONCLUSIONS FROM THE EUROPLAN NATIONAL CONFERENCES

15 national Europlan conferences were organised in 2010/2011 to present the Commission Communication and Council Recommendations and to discuss their implementation at national level.

² http://ec.europa.eu/health/rare_diseases/docs/ebs_361_en.pdf

The key results, facts and figures of the national conferences are available in the final reports of the conferences³. Over 2200 participants were present overall. The national conferences have effectively helped to:

- present the two new EU key policy texts on RD (Commission Communication and Council Recommendation on Rare Diseases) by reaching broader national audiences;
- make RD communities more aware that national efforts are “embedded” into a more comprehensive and long-term approach integrating EU and national levels;
- specific recommended actions and national measures to be integrated into the debate and the language of stakeholders participating to the conferences;
- endorse EUROPLAN recommendations and in many cases national audiences have amended

The conferences had a real awareness raising impact, although the impact was different in countries already with a plan as it was ultimately confusing for those attending in these countries as they saw these conferences as assessing the strategy: this approach should be rethought for the next steps.

ROAD MAP FOR THE IMPLEMENTATION OF THE EU POLICY ON RARE DISEASES 2010-2013: DISCUSSION OF THE THIRD DRAFT COMMISSION PROPOSAL

The third draft of a Commission Proposal of the Road Map for the implementation of the EU policy on rare diseases 2010-2013 was presented.

The domains of the Road Map follow the main points of the Commission Communication and Council Recommendation, and a number of simple actions with instrument and suggested timing, as well as risk level, have been identified. These areas are not described in great detail in order for the document to be easily readable and flexible.

It was proposed that the Members of the EUCERD be given 4 weeks to transmit their views on the Road Map to the Commission including their comments on which actions should be prioritised. These comments will be used to produce a fourth draft for endorsement by the EUCERD at their next meeting. The final draft will align the content of the Road Map with the definitive content of the Joint Action 2011.

EU PUBLIC HEALTH – WORK PROGRAMME 2011 AND CALL FOR PROPOSALS

An overview was given of the EU Public Health work programme 2011⁴ which falls under the second programme 2008-2013. An amount of 5 million € has been dedicated to RD this year. The work programme was adopted on 22 February 2011 and a call for proposals was launched soon after with a deadline for the proposals of 27 May 2011.

³ <http://www.eurordis.org/content/europlan-guidance-national-plans-and-conferences#EUROPLAN%20%20National%20Conference%20Final%20Reports>

⁴ http://ec.europa.eu/eahc/documents/health/calls/2011/WP2011_en.pdf

There are two distinct subject areas in the call for proposals in the field of rare diseases: a Joint Action on rare diseases with 60% EC co-funding (for a tentative 3 million €) and calls for projects on rare disease information networks (this definition is intentionally ambiguous to allow for proposals from ERNs, registries etc) with a total EC co-financing of 1.5 million €, with 60% EC co-funding. Leading and associated partners in these Joint Actions or projects cannot receive more than 20% financing from industry. 4 million € is available for operating grants (which can be used to finance the overall work of an NGO, network or project) this year and these beneficiaries must receive no more than 20% of industry financing: this is calculated retroactively for the duration of the grant.

11. JOINT ACTION 2011 ON RARE DISEASES

A Joint Action to support the implementation of the Council Recommendation and the Commission Communication on Rare Diseases during the period 2012-2014 is in preparation. The maximum EU co-funding is 3 000 000€ with 60 % co-funding over three years (January 2012 – December 2014). The deadline for submission is 27 May 2011.

The Joint Action will continue the work started by Europlan and the Joint Action to support the RDTF (including the revision of the ICD for RD) and it will be an innovative action that gives continuity and a new technical and political framework to the projects on European Reference Networks for rare diseases supported by EU funding between 2006 and 2009.

The following workpackages are planned: “Implementation of plans or strategies at MS level (continuation of EUROPLAN)”; “Contribute to the standardisation of nomenclatures at international level to ensure the visibility of rare diseases in health information systems”; “Provision of specialised social services and integration of rare diseases into social policies”; “European networking for improving the quality of care in the field of rare diseases”; “Scientific Support to the EUCERD and integration with other projects in the field of rare diseases to plan for sustainability of tools and resources”. There are 3 transversal WPs: coordination of the project, dissemination and evaluation of the project.

Members were reminded that only countries who have expressed their interest in the Joint Action before 27 May 2011 via their government to their permanent representation who will then inform the EC can participate as partners: for the moment 16 countries have done so including France, UK, Belgium, Portugal, Slovakia, Lithuania, Latvia, Spain, Bulgaria, Austria, Italy, Hungary, Poland, Romania, Greece and Malta.

12. UPDATE ON FP7 AND DG RTD ACTIVITIES IN THE FIELD OF RESEARCH ON RARE DISEASES

An update on international cooperation activities in the field of research on rare diseases was presented. During an initial bilateral EC/NIH workshop on 27-28 October 2010 in Reykjavik⁵, the

⁵ Report of the meeting : http://ec.europa.eu/research/health/medical-research/rare-diseases/events-03_en.html

creation of an international consortium on rare diseases research (IRDiRC) has been suggested and an interim steering committee has been set up to prepare a draft policy document describing research objectives, develop policy goals and establish a governance. A second workshop will be held in Washington DC 6-8 April 2011 to discuss and finalise a first draft of the policy document, to officially launch the initiative and to propose working groups on important policy goals. The general goals of the consortium are to establishing diagnostic tests for all RD and to develop 200 new therapies by 2020. Now it should be determined how to best cooperate on international level in order to reach these goals. Grand challenges to be tackled include: establishing and providing access to harmonised data/samples, molecular and clinical characterisation of RD, translational/preclinical research and clinical research.

The consortium will not launch calls or joint calls, but national calls and FP calls will feed into the consortium and projects will be gathered together in working groups where discussion will take place on how to best cooperate. The DG Research FP will put funding towards the consortium's goals. Interest from Canada, Australia, Japan, Spain, Italy, Germany and the UK has been voiced and it is hoped that more will join. The initiative will not just deal with genetic diseases, but with all RD. It was suggested that after the next workshop the policy document should be made available for comments to the EUCERD.

13. EUROPEAN REFERENCE NETWORKS ON RARE DISEASES: SITUATION OF THE PROCESS DISCUSSION ON THE PROPOSAL FOR A DIRECTIVE ON CROSS-BORDER HEALTH CARE

The Cross Border Health Care Directive was, after two years of discussion, accepted by the European Parliament. Article 13 explicitly mentions rare diseases, with Orphanet cited as the source of information for appreciating whether or not a disease is rare. The Directive will be published in the Official Journal during the Hungarian presidency, and following publication MS will have 30 months to implement it. A Joint Action will start in 2012 (TBC) to implement the directive.

The EUCERD should contribute as of now to the reflection on the quality criteria for centres of expertise and ERN and the designation of the ERNs. Ideally the EUCERD should recommend criteria for centres of expertise before the Joint Action to implement the Directive comes into force. Reflection needs to be carried out on whether ERNs should just be networks of centres of expertise at national level where these exist, and if these centres do not exist, whether another approach will need to be considered. Disease coverage will also need to be considered, as will different types of network (i.e. networks across Europe and networks between neighbouring countries). Orphanet will also be an important tool for repertorying centres of expertise and ERNs for RD. It needs to be considered as soon as possible which Committee will accredit and designate ERN for RD within the Joint Action to implement the Directive: will this be the EUCERD or another Committee?

14. NEXT MEETINGS AND OTHER BUSINESS

The tentative dates for the next meeting of the EUCERD are 24-25 October 2011.

It was also proposed that smaller groups be organised before the plenary session to enable Members to discuss agenda points in order for discussions to be more animated.

It was suggested that the EUCERD Bureau address the permanent delegations of the next three European presidency countries in order to explore the possibility of a one of these countries hosting a meeting of the EUCERD. This could help raise the profile of RD in these countries. Members, however, wish to stress that the priority is to efficiently set dates and agendas.

15. CONCLUSIONS

- The Commission has opened a consultation on whether countries outside of the EU/EFTA will be invited to participate at EUCERD meetings as observers: if no objections are voiced in the coming 2 weeks, this proposal will be adopted.
- The Members of the EUCERD be given 4 weeks to transmit their views on the Road Map to the Commission including their comments on which actions should be prioritised. These comments will be used to produce a fourth draft for endorsement by the EUCERD at their next meeting. The final draft will align the content of the Road Map with the definitive content of the Joint Action 2011.
- MS wishing to express their interest in the 2011 Joint Action for Rare Diseases should do so before 27 May 2011 via their permanent delegations to the European Commission services.
- The Bureau will, in the next two weeks, amend the draft letter concerning the elaboration and adoption of national plans/strategies for RD and the Council Recommendation to be sent to MS permanent representatives to refer to the final outcomes of the Europlan project and the 2011 JA for Rare Diseases once this is better defined. The draft will be sent to the EUCERD for an open consultation period of 2 weeks: if no objections are received during this period the letter will be sent to the permanent delegations.
- The Bureau will draft and send a letter to each of the 3 future presidencies of the Council to organise one of the next meetings of the EUCERD in one of these countries.
- The final draft '*EUCERD Report: Preliminary analysis of the outcomes and experiences of pilot European Reference Networks for Rare Diseases*' will be sent following this meeting to participants of the December 2010 and March 2011 to the workshops' participants and members of the EUCERD for their comments/validation during a period of 4 weeks.
- The work process and Editorial Committee for future editions of the Report on the State of the Art of Rare Disease Activities in Europe will be further defined for approval by the EUCERD.
- The Bureau will set the dates for the EUCERD meetings in 2012 in time for the next meeting.