



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

SUMMARY REPORT

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Establishment of the Committee and of its Bureau

The European Union Committee of Experts on Rare Diseases (EUCERD) met for the first time on 9-10 December 2010 in Luxembourg. The EUCERD is an expert committee of the European Union, established through the Commission Decision of 30 November 2009¹. Following the publication of the Commission Decision, Member States' permanent delegations in Brussels were asked to each nominate a representative. An expression of interest was launched for interest groups' representatives: these representatives were then selected by an internal selection committee and the results published in the Commission Decision of 27 July 2010 on the appointment of the members of the European Union Committee of Experts on Rare Diseases set up by Decision 2009/872/EC².

The EUCERD elected the Bureau of the Committee for a two year term: Ségolène Aymé (Orphanet) was elected Chair, and Yann Le Cam (Eurordis), Kate Bushby (Treat-NMD) and Helena Kääriäinen (the Finnish representative from the National Institute for Health and Welfare Helsinki, Finland) were elected as Vice-Chairs. The Bureau will be in charge of the agenda of meetings and of assuring the good functioning of the Committee.

Internal rules of the EUCERD

The Council Decision of 30 November 2009 stipulates that the EUCERD adopt rules of procedure. The proposed rules of procedure are the standard rules for an expert committee, adapted to the needs of the Committee, but these rules should not step outside the bounds of the legal tool (the Commission Decision establishing the EUCERD). The only adaption to the standard rules proposed is that the Bureau is chaired by others than the Commission. The rules also focus on the scope of the Committee in comparison to other committees (i.e. the Committee on Orphan Medicinal Products): the committee cannot make decisions on matters for other committees to decide (see Article 2, Point 3). However, members of other committees may be invited to meetings and working groups of the EUCERD when specific points of the agenda or topics of the working group require their presence.

The drafting of the agenda will be coordinated by the chair in consultation of the Bureau. Members may suggest topics for the agenda by contacting the Bureau. Alternates may participate at the meetings, but they will not be reimbursed.

EUCERD documents are eligible to be made public and available and therefore the minutes must comply with Article 9 and document the following: the decisions reached and the elements of the

¹ <http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:L:2009:315:0018:0021:EN:PDF>

² <http://www.eucerd.eu//upload/file/Docs/CouncilDecision270710.pdf>

discussion on each point of the agenda. No individual attribution of a position will be recorded unless a member wishes for a certain statement to be recorded.

The Commission services provide support to the EUCERD, but delegates this service to the Scientific Secretariat which is currently supported to 31 December 2011 by Joint Action N° 2008 22 91. In 2011 the EC will propose a new Joint Action for the scientific support of the EUCERD. This new Joint Action will be elaborated in collaboration with the Bureau of the EUCERD and in consultation with the members of the EUCERD. Member States are invited to participate in the elaboration and support of the proposal.

Road Map for the Implementation of the EU Policy on Rare Diseases 2010-2013

A draft road map for the implementation of the EU Policy on Rare Diseases was discussed. The actions cover the following areas: the definition of rare diseases; the classification and codification of rare diseases; the dissemination of knowledge and information on rare diseases; improving universal access to high-quality healthcare for rare diseases in particular through the development of national/regional centres of expertise and establishing EU reference networks; access to specialised social services; access to orphan drugs; e-Health; screening practices; quality management of diagnostic laboratories; primary prevention; registries, cohorts and databases; research and development; empowerment of patients organisations; and other actions not explicitly included in the Commission Communication – i.e. creating a framework of cooperation between the pharmaceutical industry and EC in the area of RD, public/private partnership on research and action on RD, future priorities on RD in the EU 3rd Health Programme (2014-2020), future priorities on RD in the 8th EU Framework Programme (2014-2020), future priorities on RD Structural Funds Regulations (2014-2019), workshop on using Joint Assistance to Support Projects in European Regions funding in new MS.

The Road Map will be revised according to Members' comments and will be discussed at the next meeting of the EUCERD.

Past activities of the Rare Diseases Task Force

The past activities of the Rare Diseases Task Force were presented to the Committee.

Coding and classification working group

The activities of the coding and classification working group were presented, starting with the ICD Revision Process at the WHO. The work of the Topic Advisory Group (TAG) for rare diseases was presented, including the life-cycle of a proposal, the major problems encountered with ICD-10 and the proposed organisation of chapters and basis for a revision in the field of rare diseases. An overview was given on the process at Orphanet to revise the classification, including the process behind the chapter by chapter comparisons between ICD-10, Orphanet classification systems and published classifications when available and the elaboration of proposals for ICD-11 for each chapter. The current and provisional schedule for the chapter by chapter revision was presented, along with the experts and groups invited to review the proposed chapters. The TAG RD will be one of the very

few TAGs able to propose an alpha draft as scheduled by March 2011, she also expressed concerns about the structure, as most TAGs are not sufficiently staffed for in depth work, and also evoked the problems posed by conservative forces who are against the polyhierarchy system and changes to the structure, as well as the lack of strategy to ensure consistency and updates in the future. It was suggested that an alternative for RD should be envisaged from now so as to ensure the traceability of RD in health information systems. It was decided that the Bureau will draft a letter to the WHO highlighting the importance of the visibility of RD in the new classification and the importance of the polyhierarchy approach.

Indicators working group

The indicators working group decided to concentrate on data from existing RD patient registries and indicators that could be derived from this data. Five registries accepted to participate in a pilot study (ERCUSYN, ENRAH, EURO CARE CF, Marfan cohort and registry, EUROFEVER) to see which indicators they could propose, concentrating on available data. Following this it was seen that interesting indicators can be derived from registry data over time and there are possibilities to evaluate outcomes from different countries. It was decided that the EUCERD write to the EMA for collaboration on the issue of disease registries versus product registries.

Tools for the EUCERD

The two available tools for the EUCERD were presented: *OrphaNews Europe*, the newsletter of the EUCERD, and www.eucerd.eu, the website of the EUCERD.

The planned evolutions in 2011 of the newsletter were presented: this includes personalised subscription to articles of most interest to the reader, an RSS feed, a function to search OrphaNews Europe's archives (full text search option) and the further implication of EUCERD members.

Concerning the website, it was requested that more documents at national level be added to the list of documents provided on the website and that the EUCERD site be rethought to make it more user friendly so that documents by country can be easily found.

Report on Initiatives and Incentives in the Field of Rare Diseases in 2009

A report entitled *2009 Report on Initiatives and Incentives in the Field of Rare Diseases of the EUCERD* was published in 2010: members were given an overview of the methodology, validation process and content of this report which covers activities in the field of rare diseases and orphan drugs at EU and MS level before 2009 and during 2009. A report covering activities in 2010 will be elaborated in 2011, and suggestions were sought from Members on how to improve the report, how they wished to contribute to the report, their opinions on the validation of the report and the dissemination process.

The feedback on the report was highly positive, and it was suggested that the document should be made widely available, possibly via a press conference.

A glossary of acronyms and terms should be added to the next report for easier reading for the 'uninitiated'. It was decided that the title of the report should be changed to '*State of the art of rare disease activities in Europe*'.

State of Play of the Europlan Project

The Recommendations for the Development of National Plans or Strategies for Rare Diseases Guidance Document elaborated by the project was presented as was the work to date on process indicators to monitor the implementation and evaluate the impact of national plans/strategies for RD.

An overview was then given of the state of play in each of the EU MS concerning the elaboration of a national plan/strategy for RD, along with an outline of future plans, i.e. to support countries without a national plan strategy (with tools, workshops, discussion), and to monitor the implementation of actions recommended by the Council in those countries with a national plan/strategy. A final Europlan Conference will be held on 25th February 2011 in Rome to present the outcomes of the project.

It was decided that the EUCERD draft and adopt a two page recommendation in 2011 to the Commission to be sent to MS asking them to support the creation of a national plan/strategy with financing: 2011 is the appropriate timing as it will be mid-way between the adoption of the Council Recommendation and December 2013 deadline, right after the creation of the EUCERD.

Creation of the Network of Experts on Newborn Screening for Some Rare Disorders in View of a Council Recommendation in 2012

A European Union Network of Experts on Newborn Screening (EUNENBS) was established in 2010; an overview was given of the steps made to date. Discussions after this presentation revolved around the importance of the topic and its sensitivity and it was highlighted that enough time and resources must be dedicated to the discussion of the conclusions of the project. It was suggested that a report on the outcomes should be given to the EUCERD at the meeting following the publication of the final document of the project.

Update on FP7 and DG Research Activities in the Field of Research on Rare Diseases, and Report of a Workshop Organised by the National Institute of Health USA and the European Commission

The activities in the field of research on rare diseases in the past DG Research framework programme were presented before an overview was given of the Health theme of the 7th Framework Programme which includes the topic of translational research in the field of rare diseases. This topic focuses on Europe-wide studies of natural history, pathophysiology and the development of preventive, diagnostic and therapeutic interventions in order to shed light on the course and/or mechanisms of rare diseases and to test diagnostic, preventive and/or therapeutic approaches to alleviate the negative impact of the disease on the quality of life of the patients and their families.

In 2012-13 the next two FP7 calls will focus on the gaps in the research portfolio identified from the results of previous calls, the contribution to EU policy objectives and the priorities discussed with the Health Theme Advisory Group and the Health Theme Programme Committee.

An overview was then given of the ongoing cooperative efforts between the EU and the USA. There is a recognised need for more international cooperation in research on rare diseases: to this effect a joint EU/USA workshop with the participation of stakeholders was held in Reykjavik, Iceland on 27-28 October 2010 to identify areas that would most benefit from trans-Atlantic and international cooperation and to reflect on potential strategies and contributors for implementation.

Areas for cooperation that have been identified are: access to harmonised data/samples, molecular and clinical characterisation of RD, translational/preclinical research and clinical research, nomenclatures and ontologies.

Another workshop in Washington DC, USA is planned in early spring 2011 in order to organise an international consortium on rare diseases, and will define priorities and the organisation for research.

European Reference Networks on Rare Diseases: Situation of the Process Discussion on the Proposal for a Directive on Cross-Border Healthcare

An update on the current situation was presented and discussed.

Next Meeting

The second meeting of the EUCERD will be held on the 22-23 March 2011 in Luxembourg, and will be preceded by a workshop on Centres of Expertise and European collaboration between Centres of Expertise for Rare Diseases.

Final Conclusions

The EUCERD decided during the meeting:

- That the Commission services will revise the Rules of Procedure as discussed during the meeting and send a final version to members.
- To revise the Road Map for the implementation of the Commission Communication and the Council Decision presented at the meeting so that it is easier to follow. This revised version will then be sent to the EUCERD Members for their input.
- To start in January 2011 to prepare the framework of the next Joint Action for the Scientific Support of the EUCERD: the Bureau will start to draft this and to consult members on what should be included. On 19-20 January 2011 a meeting will be held in Luxembourg on the Joint Actions in general.

- To send a letter to WHO expressing the expectations of the Committee of the revision process regarding rare diseases.
- To send a letter to MS reminding them of the Recommendation of the Council to elaborate or adopt a national plan or strategy by the end of 2013 and referring to the key documents.
- For 2011 to organise the working groups of the EUCERD within the constraints of the current Joint Action (N° 2008 22 91). The proposed workshops for 2011 will cover:
 - Centres of Expertise and European Reference Networks for RD (scheduled for 21 - 22 March 2011 in Luxembourg);
 - Registries for RD (disease specific versus product registries) to encourage dialogue between stakeholders (with the collaboration of the EMA in London);
 - Coding and classification of RD: this will be a public conference on 4 November 2011 in Montpellier in conjunction with the EuroBioMed RD conference.
- To improve the website with a section for EUCERD members including a list of members and contact details as well as working documents and presentations from the meetings, an improved list of national websites and documents and a list of acronyms.
- To make modifications to the type of data included in the Initiatives and Incentives report covering 2010: i.e. include marginal indicators of the evolution of policy and science in the field so as to show how the field of RD is driving innovation. The next report will be named '*State of the art of rare diseases activities in Europe*'. It should be easier to see the 'bigger picture'. An editorial board of those who wish to volunteer in the elaboration of the next report will be established.
- To call for collaboration for the newsletter: EUCERD members were encouraged to send news for the newsletter and to comment on the documents they receive (this is also applicable for the input to the report on Initiatives and Incentives).