Rare Diseases Task Force

Report of the RDTF meeting
Luxembourg
13 November 2008

1- Commission Communication and Council Recommendation on a European Action in the field of Rare Diseases

Antoni Montserrat (DG SANCO) announced the adoption, last 11th November, of the Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions on Rare Diseases. He explained the process which led to the current text, from the public consultation (November 2007 – February 2008), to the impact assessment (March 2008 – June 2008) and to the inter-service consultation (July 2008 – October 2008) which involved, between others, DG Enterprise, DG Research, DG Budget, DG Employment, DG Relex, DG Internal Market and the Legal Service of the European Commission. The discussion with DG Enterprise regarding the section of the text dedicated to measures for orphan drugs was particularly challenging. The text was finally adopted, by an oral procedure, by the college of Commissioners, on the 11th of November, together with the proposal for a Council Recommendation on a European action in the field of rare diseases.

The two documents are now available in the 23 EU languages on the Commission website. They have already been officially transmitted to the Parliament, the Council, the European Economic and Social Committee and the Committee of Regions. The first discussion at a Council working party will take place on 27 November. The discussion by the Council of Ministers, for adoption of the documents, is scheduled under the Czech Presidency, on the 9 June 2009, providing that the Parliament has already expressed its opinion. This opinion is not mandatory but desirable. There is a possibility that the Parliament will not deliver on time due to the fact that, with the upcoming elections taking place in 7th June, most parliament members will be busy campaigning. The name of the Rapporteur is not yet known. If the documents are not adopted in June 2009 they will have to be submitted to the Council under the Swedish Presidency, probably in November 2009.

Antoni Montserrat presented the project of the establishment of an EU Advisory Committee on Rare Diseases to advice on the implementation of this Communication. He indicated that this committee (which will replace the RDTF) will be composed of representatives of all MS to be nominated by their Ministry of Health, representatives of the main DGs of the European Commission involved in the field (DG Public Health, DG Research, DG Enterprises, DG Information and Society; Eurostat); and of EC agencies (EMEA, ECDC) as well as experts to be selected from among the coordinators of relevant EC projects in the field of rare diseases.

All the nominations will be nominative (plus an alternate). For the experts and patient representatives, an expression of interest must be sent to DG Sanco.

Antoni Montserrat thanked the RDTF members for their contribution to the elaboration of this Communication, in particular its chair person, Ségoléne Aymé, and also thanked Eurordis for its active advocacy to put the project on the right track under the French Presidency. He concluded by saying this is only the first step. The next steps will not necessarily be easy.

The ensuing discussion got underway with questions geared toward understanding why some key elements of the initial text were removed from the final version. Some RDTF members expressed their regret and concern that (a) the proposal for a public-private partnership in the field of R&D for rare diseases was removed; (b) they regret deeply that the proposal for a scientific assessment, at EMEA level, of the medical added-value of each orphan drug was deleted at the request of DG Enterprise. The explanation given by the representative of DG Enterprise, Narti Ulla, was unconvincing as it revealed that the decision was based on a misunderstanding on the part of DG Enterprise, contradictory to the conclusions reached by the EU Pharma Forum Working Group on pricing on the same subject, chaired by Mrs Lalis;; (3) They also regret the removal of the whole chapter on patient empowerment from the Communication.

The discussion then covered possible action at the level of Parliament to ensure a timely examination of the documents, before the next election. It was concluded that RDTF members should contact their national MEPs who are sitting on the Committee that will be in charge (ENVI). Similarly, RDTF members were encouraged to establish contact with the two key persons responsible for the Communication adoption at the Council level: the national representative for health in their permanent representation and the individual in charge of commenting on the document at the national ministry of health level.

The RDTF members made a request to Antoni Montserrat, asking to be consulted regarding the text establishing the EU Advisory Committe. A. Montserrat stated that a first draft will be available before Christmas, and will be sent to RDTF members. In response to a question regarding the resources to be allocated to this Committee to fulfil its mandate, he explained that the only resource at DG Sanco level will be himself and that the funding for the necessary working groups will come from already funded projects such as EuroPlan and the RDTF joint action, both of which will act as back office to the Committee. This committee is expected to meet at least four times a year.

2- Outcomes of Conferences organised under the French Presidency of the European Council

Four conferences dedicated to Rare Diseases were organised under the French Presidency of the EU. The three past ones were real successes. Summary reports for each have already been published in *OrphaNews Europe*. The fourth conference will take place on 18 November at the French Ministry of Health, in conjunction with the EuroPlan project and Eurordis.

3- European Reference Networks for Rare Diseases

In the absence of Alexandra Fourcade, chairperson of the working group on European Reference Networks of the High Level Group on Health Services and Medical Care, the conclusions of this working group could not be presented.

Ségolène Aymé summarised the conclusions of the RDTF working group, which held a workshop in March 2008, followed by the publication of a Report in July 2008. This report is annexed. She concluded that many aspects of a possible policy to establish European Reference Networks require further clarification. It is too soon to decide before a systematic analysis of the preliminary outcomes of the ten current pilot networks is available. The RDTF scientific secretariat will contribute to this analysis for further discussion at the next RDTF meeting. To a question relative to the continuation of funding of the first set of networks, A. Montserrat answered that these pilot networks could apply for an operational grant for one additional year. Until the future EU Directive on cross-border health care legal and maybe a financial instrument could be provided. It was concluded that there is still a lot to do before going from a self initiated process to a direct designation of European Reference Networks. It was also underlined that the mandate of potential ERN should be proportionate to a potential budget.

4- New services developed by Orphanet

Ségolène Aymé presented the new services developed by Orphanet in 2008, to better serve the RD Community. The main achievements are:

- The launch, in March 2008, of a new portal fully accessible to disabled users and more user-friendly than the previous version of the website
- The extension in June 2008 of the information provided for each rare disease to include prevalence data, mode of inheritance, age of onset and causative genes. Orphanet also became cross-referenced with other gene databases such as HGNC, SwissProt, OMIM and Genatlas.
- The extension of the information on expert clinics in Europe allowing to sort by various criteria: type of clinic, paediatric or adult, centre of reference or not, disease management or genetic counselling.
- The extension of the information on medical laboratories to encompass information on the quality management of laboratories, in order to promote quality services. This was achieved from a collaborative effort with EuroGentest, an FP6 network of excellence.
- The classification of all RD following a medical logic, permitting for the access of all information through any general query, along with the visualisation of these classifications directly on the website, as of October 2008.

RDTF members congratulated the Orphanet team for these new services, which are highly welcomed.

5- Agenda of the RDTF in the field of Indicators

Laura Fregonese presented the conclusion of the RDTF working group on indicators in the field of RD. This WG held a meeting in March 2008 and published a report in September 2008 which is annexed. She presented the work plan for the three years to come, as defined by

the Joint Action selected for funding this fall and which will officially start in January 2009. A discussion clarified the concepts of Indicators versus Information. The relative merits of aggregated indicators versus disease-specific indicators were also discussed. Currently no decision has been taken regarding indicators to be documented. This will be discussed at the next meeting of the working group and presented later at the RDTF meeting.

6- Agenda of the RDTF in the field of coding and classification

Ségolène Aymé presented the conclusion of the RDTF working group on coding and classification in the field of RD. This WG held a meeting in February 2008 and again on 14 November 2008. She presented the work done by Orphanet in this field, in line with the previous decisions of the WG. Currently, all published classifications in the field of RD have been collected and introduced in the Orphanet database. In addition Orphanet has established a general classification by medical speciality and sub-specialty. All the data have been transferred to WHO headquarters to be used as a pilot set of data for the alpha version of the next International Classification of Diseases. S. Aymé emphasised the fact that Orphanet has now established a unique nomenclature of RD which is available to third parties on request.

The work plan for the three years to come, as defined by the Joint Action, includes a cross-referencing of Orphanet diseases with ICD10, OMIM, SnoMed-CT and MedDRA. It also includes proposing codes for all recurrent RD in the next edition of the ICD. The proposal will be submitted to WHO by the end of 2009.
Antoni Montserrat asked for clarification regarding the relative role of the EC and of the US NIH in the revision process of the ICD. S. Aymé reminded him that the work in the field of RD is the responsibility of the WHO Topic Advisory Group which she chairs. EC funding is instrumental in providing the resources to generate all the proposals to WHO. The US NIH, as with other equivalent bodies in different regions of the world, will be involved in commenting on the proposals and making final decisions.

7- Outcome of the 2008 call for proposals in the field of Public Health

Giorgios Margetidis briefly presented the six projects which were selected for funding in 2008. Not all the negotiation rounds are completed yet. The six projects are:

- The funding for the Conference on national plans for RD to be held in Paris on 18 November
- An operating grant for Eurordis for 1 year covering the year 2009
- A three year grant for a Joint Action in the Field of RD to provide resources for some RDTF activities: in the field of indicators, for coding and classification, and for publication of *OrphaNews Europe*.
- Three more pilot networks on chronic hyperventilation syndrome, on rare anaemias, on cerebral palsy and related disorders.

8- Next meeting

The next RDTF meeting will take in Luxembourg on Thursday 30 April 2009