Rare Diseases Task Force

Meeting Report

12th Meeting of the European Commission Rare Disease Task Force
Luxembourg, 23 October 2009

1. State of play of the Commission Decision setting up a European Committee of Experts on Rare Diseases

Toni Montserrat
DG Sanco C2

The draft of the Commission Decision setting up a European Committee of Experts on Rare Diseases was submitted to an inter-service consultation at the European Commission, and the document was given the green light from most Directorates General except from DG Enterprise and Industry who gave an 'avis négatif' on the draft. This, amongst other reasons, has blocked the development of the committee during 5 months. The main point of contention with DG Enterprise, as they wished the representatives of the EMEA and COMP not to be full members of the Committee: they have accepted the current formulation which places representatives of the EMEA and COMP at the same level as DG Sanco. The 'avis négatif' was transformed into 'avis positif' in early October. The text is will be translated into the 22 official EU languages. It will then be published in the official journal, and the official adoption should be made by end-November 2009, followed by the process of nomination of members.

The committee will have 51 members, instead of the 54 members evoked in the first draft. After publication of the adopted text in the official journal, DG Sanco will address a letter to permanent representations of the EU27 in Brussels asking for the nomination of one representative for each of the MS: DG Sanco will advise that nominees should have responsibilities, or experiences, in the ministries or agencies dealing with Rare Diseases in the MS. The other members will be designated on the basis of a call for expression of interest: this call will be published on the DG Sanco website and official journal in English (the date will be communicated to relevant bodies shortly). A call of expression of interest will be published for:

- 4 members and 4 alternates of patient organisations;
- 4 members and 4 alternates from the pharmaceutical industry;
- 9 representatives (and 9 alternates) of ongoing and/or past Community projects in the field of RD financed by the programmes of Community action in the field of health (including 3 representatives of existing pilot networks of European Reference Networks);
- 6 representatives (and 6 alternates) of the ongoing and/or past rare diseases projects financed by the Community Framework Programmes for Research and Technological Development.

Internally, a group will be constituted to make the selection of representatives of DG Sanco and DG Research.
One representative of the European Centre for Disease Prevention and Control (ECDC) will also be nominated. The Commission will be represented by representatives of DG Sanco, DG Research, DG Enterprise and Eurostat. Representatives from the EMEA, as well as the Chair or Vice-Chair of the

COMP may attend. EFTA states party to the Agreement of on the European Economic Area can also request representation. There is also possible to grant representatives of international or professional organisations acting in the field of rare diseases making duly substantiated requests to the Committee observer status.

The functions of the scientific secretariat will be revised as the Task Force will no longer exist: it is most probable that the Joint Action will provide scientific support to the new Committee: this is an administrative detail that will be clarified between the EAHC and the partners of the Joint Action to ensure the continuity of the Joint Action.

The first meeting of the new Committee will be held exceptionally in Brussels (meetings will usually be held in Luxembourg) on the 26 February 2010 to mark the 3rd World Rare Disease Day, providing that all the members of the Committee have been selected and their names published in the official journal by this date.

The draft Commission Decision provoked many comments from Task Force members and requests for clarification concerning the powers of the new Committee. Task Force members asked for the minutes show that it is within the scope of the Committee to address ***access*** to orphan medicinal products. Task Force members were disappointed that the EMEA and COMP – who have made such active contributions to the Task Force – would no longer be full members. The question of whether alternates would be able to attend meetings was approached, it was suggested that for certain agenda items this could be possible. It was also clarified that the likely number of Committee meetings per year would be 3, held in Luxembourg. Concern was expressed that a quorum of two thirds of the Committee is required to adopt opinions, reports or recommendations; A. Montserrat explained that alternates are possible to ensure full attendance and will be asked for by name before each meeting; he acknowledged that it is a difficult task to ensure the attendance of each MS. Concerning the possible collaboration between the Committee and the High Level Group on European Networks of Reference, it was suggested that the possibility of the Committee playing the role of pre-implementing European Reference Networks for rare diseases be explored when the Cross-border Directive is approved. It was also decided that internal rules of the Committee are drafted by Sanco C2 before the first meeting of the new Committee to curtail lengthy discussion, notably on nomination procedures.

2. Implementation of the Second Health Programme
Antoni Montserrat

The Commission will anticipate the publication of the results of the 2009 call so as to proceed immediately with the implementation of the call 2010. The call for the work plan 2010 will be published in November, with a deadline for proposals in February 2010 (3 month delay).

Three proposals were retained for funding from the call for 2009 concerning RD: RD Portal 2 for the development of Orphanet (main partner INSERM); “Care NMD - Dissemination and Implementation of the Standards of Care for Duchene muscular Dystrophy in Europe (including Eastern countries)” (main partner University of Frankfurt); and “BURQOL–RD - Social economic burden and health-related quality of life in patients with rare diseases in Europe” (main partner Canary Islands Foundation for Research and Health”).

Two conferences on rare diseases are funded from the 2009 call: the 2nd Pan-European Conference on Haemoglobinopathies (Thalassaemia International Federation); and the European Autism Action 2012: Working Conference on a European Strategic Plan for Autism, Dublin 2009 (Irish Autism Action).

An Operating Grant was awarded to Eurordis. Operating grants are awarded to NGOs to support the continuity of their activities and on going projects. The European Commission privileges Eurordis for operating grants (as Eurordis is seen as representing the European RD patient community); however there are more and more requests for operating grants from Eurordis' members. A. Montserrat suggested that Eurordis coordinates these demands to avoid frustration. It was decided to schedule a meeting between Eurordis, DG Sanco and the Executive Agency in order to discuss these issues.

The result of the call for tender launched in August 2009 for newborn screening for some rare disorders has not yet been announced. The Commission intends to use the results of this project to draft a Council Recommendation on new born screening in 2011-2012, so the project has a political dimension.

An overview was presented of the Work Plan for 2010 to be published in November 2009. This year's work plan is explicit in its intentions and objectives. Firstly, the intention of the Commission to establish two joint actions in 2010 (a joint action is the common work of the Commission in Member States for a certain purpose) for rare diseases, one for Orphanet and one for Eurocat - actions specifically mentioned in the Council Recommendation. Two preparatory meetings will be held. Secondly, explicit support will be given to certain registers and database on the basis on proposals, to avoid problems with evaluators who do not understand the importance of certain projects (i.e Ercusyn, Euromyasthenia). Thirdly, there will be a call for tender for a project examining the creation of a working group on the clinical added value of orphan medicinal products. The deadline for these calls will be in February 2010.

It was stated that the areas to be supported should have been discussed by the RDTF, especially the criteria for selection and prioritisation of registries: the new Committee to be involved in the process prior to the drafting of the work programme.

3. European Reference Networks on Rare Diseases

a) Situation of the process discussion on the proposal for a Directive on Cross-Border Healthcare Antoni Montserrat

A. Montserrat gave an update on the proposal for a Directive on Cross-Border Healthcare; the modified text was presented under the Swedish presidency to the Parliament, the MS and the Commission: the parliament has no problem with the text, however, resistance at MS-level blocked the dossier from advancing: however, the concept of ERN is virtually accepted by all, although there is a problem as some states wish to exclude private institutions from ERN, which is a problem as for rare diseases many centres that could make up these ERN as private. The draft Directive will probably go on to the Spanish presidency in 2010. Then the question of how to proceed for the designation of future ERN for RD will be addressed. The Cross-border Directive will be a framework for all types of diseases, but the Commission has taken RD as an example to prove the utility and validity of European action in the form of ERN. The task of the new Committee will be to create criteria for the designation of future ERNs; this discussion will take place probably in the second semester of 2010. The Directive doesn't provide financing, but does provide reinforcement for proposals for the third programme. Advocacy from stakeholders is still helpful at this point, and the position of the RDTF and Commission is that the mobility of expertise should be favoured above the mobility of patients, and e-Health solutions should be explored.

b) List of ongoing pilot projects

Ségolène Aymé

A spreadsheet prepared by the RDTF scientific secretariat showing country participation in the 9 existing pilot European Reference Networks (ERNs) for RD (see Annex) was presented to participants. The countries that participate in the most pilot ERNs for RD are Belgium, Germany and United Kingdom with 9 ERNs. The only EU MS that does not participate in an ERN for RD is Luxembourg. France is the main partner for 3 pilot ERNs for RD, followed by Germany with 2. Dyscerne is the ERN with the most participating countries (21 EU MS).

The process by which these pilot ERNs for RD were selected and funded was not the process which was defined by the HLG on ERN, so many of these networks were already in place before the concept of ERN (networks of registries etc) and so they sometimes do not cover the criteria one would usually expect for expert centres. An analysis of services provided by these pilot ERNs is needed and a report should be published on this matter, to aid the HLG in deciding on the next steps to take. This work is not covered by a contract, but the scientific secretariat would be willing to do the preparatory work. Access to the activity reports of these pilot ERNs for RD can be obtained from Sanco C2 to work from. DG Sanco will help with contacting these networks for completion of the questionnaire, an analysis will then be made of these replies, and a draft report submitted to the RDTF for comment and validation. This report will then be available to the HLG on ERN.

c) POLKA Project
Yann Le Cam
Eurordis

Eurordis started working on the concept of Centres of Expertise for RD from a patient perspective in 2006. This work continues with the production of 2 fact sheets for advocacy use, on Centres of Expertise and ERNs, and the POLKA (WP4) project's Patient's Consensus on Preferred Policy Scenarii for RD uses a Delphi-like method to discuss Centres of Expertise. WP5 of the POLKA project aims to instigate collaboration between Patient Representatives and ERNs for RD, and develop recommendations towards good practice of collaboration: however the preliminary investigation into the 'real life' experience of pilot ERNs (feedback from patient representatives involved in ERNs, feedback from leaders of pilot ERNs) has been disappointing, possible due to a misunderstanding of the need for and nature of this collaboration. WP5 has thus been adapted: the Declaration of Common Principles on Centres of Expertise and ERN will be disseminated further and possibly revised following feedback. Guidelines for good practice of collaboration between patient organisations and Centres of Expertise at national level and at EU level will be created. A method for the evaluation of ERNs/Centres of Expertise by patients *or* regarding patient outcomes could be a possible task for the new Committee of Experts. One-to-one interviews with the project leaders and the federations concerned are underway, instead of a general meeting.

Comments included the difficulty of identifying ways of collaborating to obtain relevant information, and the need to support patient organisation involvement in ERNs.

4. Classification and Codification of Rare Diseases
Progress report of the working group and planned EU/WHO workshop on Classification and Codification on Rare Diseases (Luxembourg May-June 2010)
Ségolène Aymé

The objectives of the working group on Coding and Classification are: 1) to submit a proposal to WHO for ICD11; 2) to cross-reference coding systems to improve coding of RD. The structure and timeline of the ICD-10 revision process and the role of the Topic Advisory Group for Rare Diseases was outlined. The revision process and peer-review process was explained. The electronic version of the ICD will reflect the poly-hierarchy classification proposed by the RD TAG. A linearization will be needed for the book form, which will be used in countries without easy access to PCs. The Alpha Draft for ICD-11 should be published on the WHO website by May 2011 and a tool will be provided that allows for comment by disease.

To date 5 ICD-11 chapter proposals have been drafted: Haematology, Immunology, Endocrinology, Nutrition and Metabolic Diseases; the Neurology, Malformation and Internal medicine chapters should be sent out soon (these are transversal chapters where the most major changes have been made), possibly along with the Ophthalmology and Dermatology chapters. So far helpful contributions have been made by the Italian revision group, by ENERCA (haematology chapter) and by Eurocat whose suggestions for the malformations chapter will be integrated into the chapter proposal by the TAG RD.

Cross-referencing using automated methods and completed manually is underway at the University of Manchester and at Rouen: Orphanet entries have been converted into an ontology and matches with Snomed CT and MeSH are on the way. A meeting of the workgroup is planned for the 27th January 2010 in Manchester.

Discussion revolved around the different uses of classifications for RD (clinical versus policy-making) and the classifications and cross-referencing available to both groups of users during the revision process to make RD visible. It was explained that during the wait for ICD-11, basic changes can be integrated into ICD-10 using the annual revision process, and alongside this the Orphanet classification makes the RD visible in its classification and is stable despite evolutions and should be well matched with ICD-11.

A coding and classification of rare diseases conference, co-organised by DG Sanco and WHO, is planned for June 2010 to which around 100 experts will be invited. A draft agenda will soon be proposed for this 2 day event which will include plenary sessions and workshops. Topics for discussion include: present state of art of ICD revision process in the field of RD; present state of art of cross-referencing; present state of art of ontologies and of web semantics; the coding of rare cancers; different coding systems in different countries; and present implications for Health Information Systems. S. Aymé asked for volunteers for the organisation committee to contact her.

5. State of Play of the Europlan Project
a) State of implementation
Domenica Taruscio
Europlan

The status of collection of information on 27 EU MS initiatives and incentives on RD was presented: a questionnaire has been sent to all Europlan partners, and representatives of Ministries of Health, and follow-up telephone calls are being made. This information will be organised using the structure of the Council Recommendation. A workshop on recommendations for the development of RD

national plans an strategies was held in September 2009 following the first draft of these recommendations. Comments were received on this draft and a revised version is planned for October 2009. A next step will be the development of indicators for monitoring the implementation and evaluating the impact of national plans and strategies: a first draft version of these indicators was released in April 2009 to partners, and in June and September workshops of RDTF members and other experts were held to discuss these proposed indicators. A draft recommendation will be circulated to the RDTF and EU27 MS health authorities in spring 2010 with a follow-up workshop for with these authorities back to back with the ECRD conference in Krakow. This will be followed by the national conferences organised by Eurordis.

Questions were asked concerning the proposed indicators for monitoring national plans. The difference between the work of the RDTF WG on Public Health Indicators and Europlan indicators was explained. It was highlighted that countries' progress in implementing RD plans or strategies should not be evaluated in the same way: some countries have already implemented a plan, others are in preparation, and some countries have not taken any steps as of yet

A. Montserrat underlined that in 2013 DG Sanco will have to produce the implementation report of the Council Recommendation addressed to the Council and to the Parliament, and these indicators will be needed as an objective tool to demonstrate the impact of the Recommendation and determine whether the MS have implemented the Recommendation, which will be important for the next health programme.

b) Planned actions for the coming months
Christel Nourissier
Eurordis

C. Nourissier presented the state of play of WP 8 of the Europlan project which aims to organise national conferences to present and discuss the transferability at local level of the EC Council Recommendation on RD and the Europlan recommendation. These national conferences aim to gather all relevant stakeholders keen to advance the development of the National Plan. So far, 6 advisors have been chosen to establish an advisory group. A common layout with a plenary session and working groups on topics specific to each country has been established and dates are being fixed. The national alliances are responsible for cooperation with national authorities and stakeholders, obtaining funds and drafting a final report. Themes of the working groups could include: definition and inventory of RD, centres of expertise, gathering of expertise at EU level, research, sustainability, patient empowerment, specialised services, and orphan drugs.

6. Cooperation between six member states
Guenther Leiner and Mathias Schuppe
European Health Forum Gastein

This project was initiated by the Austrian Ministry of Health and the EHFG. Small and medium sized countries face specific challenges to tackle RD, with only a few numbers of potential Centres of Expertise per country. In Central Europe most countries are small or medium-sized and these countries have cultural, historic and political links. An initiative was introduced in August 2008 to establish mechanisms for e-health governance at EU level, and a consensus was reached on the importance of wider regional cooperation for RD, with a recommendation to include cross-border care and ERN and Centres of Expertise in future Forum activities, and a decision that EHFG act as a platform for networking. A follow-up meeting in August 2009 led to a consensus on the cooperation and activities at regional level (between Austria, Czech Republic, Germany, Hungary, Italy,

Slovenia) as well as a draft position paper (adopting the criteria set out by the RDTF to define a Centre of Expertise, suggesting that represented countries should create a national RD office, and that these offices start the networking process to enhance Central European cooperation in this field with larger countries becoming involved on a regional level) and an initiative to be open to other countries and stakeholders. Strategies proposed are the identification of potential Centres of Expertise, the networking of these centres and supporting activities to address RD on a political level. A project meeting is planned for March 2010 and next steps include the definition of a detailed work plan and funding requirements.

Different opinions were voiced, concerning the regional versus European approach and how to incorporate this initiative into existing EU level frameworks. Another question was the validity of including larger countries in this initiative and which countries should be considered.

7. Preparation of the European Conference on Rare Diseases, (Krakow May 2010)
Yann Le Cam

The 5th European Conference on Rare Diseases will be held in over three days on the 13-14-15 May 2010 at the Jagiellonian University, Krakow, Poland. This biennial conference provides the state-of-the-art of the rare disease environment, monitoring and benchmarking initiatives and will bring together all stakeholders in the field of rare diseases, and topics will cover research, new treatments, health care, social care, information, and public health at national and regional levels. Four to six hundred participants are expected, and the conference will be interpreted in 5 languages. There will be a call for abstracts for a poster session on the theme "Services to patients, families and carers". A report will be produced in the 3 months following the conference.

On 13 May, satellite meetings will be held, including: a workshop of the Council of National Alliances, a meeting of the Orphanet Network, the Eurordis General Assembly, etc.

Registration will be opened in November 2009.

8. Rare Disease Day 2010
Yann Le Cam

The next Rare Disease Day will be on 28 February 2010 and will focus on research and bridging the gap between the scientist and the patient. The website for RDD 2010 will soon be launched along with an information pack. Eurordis and E-Rare are organising on 1 March 2010 a one day workshop in Brussels for 100-120 participants with the support of DG Research. Orphanet, RD Platform and Europlan will also be involved in the workshop.

9. Upcoming RDTF workshop on Indicators – 10th November, Paris
Laura Fregonese
RDTF WG PHI

A workshop on Indicators has been organised for the 10th November in Paris following the publication of the 2008 RDTF report on the State of Art of Public health Indicators (available to access on the RDTF website (http://www.orpha.net/testor/doc/RDTFPHISept08Final.pdf).

It has been decided to focus on the data collected by networks and registries and data collection methods: it is impossible to ask Member States to collect data on rare diseases when this is highly expensive and specialised and when data collection methods are already in place. The aim of the working group is to look at the reliability of the data collected by networks, and the

characteristics of this data in order to decide what type of indicators can be produced with this data, and a possible choice of pilot RD for these indicators. A report will be prepared after this workshop on the decisions made for future action.

10. Next meeting of the RDTF

This is the last meeting of the RDTF. The first meeting of the new Committee of Experts on Rare Diseases will be held exceptionally in Brussels (meetings will usually be held in Luxembourg) on the 26 February 2010 to mark the 3rd Rare Disease Day, on condition that all the members are nominated by that time. Ségolène Aymé as chair of the former RDTF thanked the RDTF members for their commitment and decisive input, especially the EMEA and COMP representatives. She concluded that the RDTF can be proud of the achievements since 2004, which were made possible thanks to the efficient support of the Sanco Unit C2 staff.