

Meeting Report

8th Meeting of the European Commission “Rare Disease Task Force”

Luxembourg, 23 October 2007

On 23 October 2007, the eighth meeting of the Rare Diseases Task Force (RDTF) included the attendance of:

Rare Disease Task Force:

S. Aymé	C. Nourissier	R. Stefanov
H. Dolk	M. Posada de le Paz	D. Taruscio
G. Gatta	A. Ramirez Vanegas	S. Tanner
A. Kole	J. Sándor	J. Torrent Farneli
J. Llinares Garcia	A. Schieppati	A. Trama
M. Jespersen	H. Segura	G. Zambruno
E. Jessop	D. Sheppard	

Observers:

R. Capocaccia
H. Jensen
O. Kremp
M. Rätsep

DG SANCO:

K. Freese
A. Montserrat

Public Health Executive Agency:

G. Margetidis

A. Welcome and Approval of Agenda

S. Aymé welcomed new members and suggested that the composition and organisation of the RDTF be presented to these new members.

A. Montserrat listed the composition of the group to include representatives of Member States (MS) chosen by MS health authorities, project leaders of on-going or past Rare Disease (RD) projects, NGO and patient organisation representatives, leaders from FP7 RD projects funded by DG Research, representatives of relevant bodies and institutions such as the EMEA and the High Level Group on Health Services and Medical Care, and other observers invited for their expertise in the field. The RDTF is assisted by a Scientific Secretariat supporting communication and administration of RDTF meetings and tasks.

S. Aymé asked to add the discussion of RD in the Programme of Community Action in the Field of Health 2008-2013.

A. Montserrat stated that the explicit mention of RD in the Health Strategy and the Public Health Program shows the Commission commitment to RD as a priority.

B. Public Consultation on the Commission Communication on a European Action in the Area of Rare Diseases

Antoni Montserrat

1. Producing the Communication Document

A Communication Drafting Group was composed to create the 1st Draft of the Public Consultation of the Communication on European Action in the Area of Rare Diseases. A. Montserrat clarified that before any Communication or Recommendation to the council a public consultation is required to allow all EU citizens to have input on the document. The Public Consultation will eventually be translated in to 21 languages. Although the document will be available in some languages earlier than other, all translations of the document will be allotted a minimum of 8 weeks for public consultation. The English version of the text will be presented and distributed during the 2007 European Conference on Rare Disease (ECRD) in Lisbon, Portugal on November 27-28.

Today the 3rd Draft of the document will be approved and it will be the last opportunity to input on its content before its presentation for public consultation. Eventually, this document will be accompanied by an Impact Assessment which includes statistics, background documents, *etc.* supporting the content in the Communication. These documents are then presented to the Impact Assessment Committee after which they are passed to the Council and the Parliament.

It was asked who makes changes to the document after public consultation. A. Montserrat replied that in February 2008, the Drafting Group, perhaps an enlarged version, will propose how to incorporate the comments from the public.

A. Montserrat went on to explain that the Drafting Group made the 1st draft. This draft was sent to the following specialised bodies: Rare Disease Task Force, the Higher Level Group, EMEA Committee for Orphan Medicinal Products, and Eurordis. On 15 October the Drafting Group met to incorporate the contributions of these bodies and finalised a 2nd draft. It was explained that although there are no limitations in length of the document, we must remember that it is best to keep it as short as possible with respect to translation into 21 languages. The accompanying Health Impact Assessment is certainly unlimited in length.

2. Section by Section Discussion of the Communication Document

A. Montserrat explained that the word “Action” was specifically included in the title as the role of the Council is to approve actions. He stated it was important to retain the definition of RD to show that although they are very diverse, they can still be defined. The term ‘ultra-rare diseases’ was removed at the request of the EMEA-COMP as it introduces an unnecessary additional category. RDTF members agreed on the following modifications to the text (Draft dating from October 23, 2007 Communication from the Commission: Public Consultation regarding European Action in the Field of Rare Diseases).

Section 1

- It should be stressed that RD are complex as this is a major argument for why collaboration at the EU level is imperative.
- The definition of RD prevalence does not apply to rare cancers. As such incidence, which is more appropriate, should be included in the Health Impact Assessment.

Section 2

- The objective of this section is to explain in reference to RD, an EU level approach will be efficient where Member States will be less efficient.
- Text describing a RD's patients right to equal treatment should be adopted from the text of the Orphan Drug Regulation.
- The descriptions of "very" rare diseases and "small" countries should be removed.
- Using the phrase "biological test" may not be clear and it should be monitored to see if this misunderstanding comes out during the public consultation.

Section 3

- Rearrange main lines of DG SANCO action so that DG SANCO's priority of networks of RD is mentioned first
- Directive of the European Parliament and of the Council on Health Services should be approved by the end of November 2008.

Section 4

- In every public consultation, the inclusion of questions is obligatory.

4.1

- Keep the common definition of RD in the EU as less than 5 per 10,000 and perhaps include an excerpt of the Orphan Drug Regulation.
- Include other disease classification systems
- Add why it is so important to sustain financial support of coding and classification activities and emphasize that the Commission has already supported so much activity in this area it would be a waste to discontinue it.

4.2

- Change the title of the section to include prevention
- Remove redundant text at the end of Question 2.
- Change wording to "online tools and electronic records" in Question 3
- "Biological tests" should be changed to "diagnostic tests" in Question 4
- Change wording about laboratories.
- Wording about population screening should be changed to emphasize that it should be done consistently across EU even if it is not for the same diseases in each MS.
- Population screening seen as a controversy. Notion of 'recommendations' should be changed to 'assessment'. (Question 5)
- Sentence referring to improved clinical training in MS where it is not sufficient should be re-included as it was removed from a previous draft. (Question 6)
- Cost of registration should be removed and comments from EPPOSI discussion should be included: OD administered in hospitals should be administrated at a level higher than the local hospital.
- Omit sentence concerning the question whether RD patients "deserve" payment of drugs. (Question 8)
- Article 83 includes a provision of compassionate use and should be included as a footnote. (Question 8)
- No reference to home-care and should be included. (Question 8)
- Health Economic Evaluation should be changed to Health Technology Assessment

4.3

- Clarify that EMEA does not “approve” the use of biomarkers as efficacy endpoints in clinical trials.
- Text referring to registries should be moved to the previous section.
- Remove “Therapeutic” from bullet title Intensifying Therapeutic Research
- Mention that the Clinical Research Directive has been shown to deter clinical trials occurring in Europe because of cost.
- Add that clinical trials should be supported at national level using example of Italy, France, Spain.

4.4

Agreement

Questions for Consultation will be changed to the following:

All agreed that S. Aymé and A. Montserrat will incorporate all suggestions. A. Montserrat will finalise the test tomorrow and shortly disseminate the timeline for translations. All responses to the public consultation will be visible. The Drafting Committee will meet around the end of January and the next RDTF meeting will follow (a bit earlier than usual in terms of interval). All members of the RDTF will receive the 3rd Draft

C. Calls for Proposals

G. Margetidis

Public Health Executive Agency

By the beginning of December the results of the evaluation will be made public. Previously there were 8-9 RD proposals most related to disease-specific networks (building up of reference networks), but what it now more important will be an emphasis on a more horizontal, transversal approach.

The RDTF Secretariat has not been supported in this call justified by the opinion that its activities were redundant. The Commission responds with the following proposals:

1. To give the task of Orphanews Europe and the Coding and Classification WG to another project. (This proposal was subsequently rejected by the “other project”)
2. Call for tender: different non-competitive process
3. Joint project: submit another proposal limited in scope.

Both the Commission and the Public Health Executive Agency ensured that a solution will be identified to prevent a gap in the funding of the newsletter and activity related to coding and classification.

The Public Health Programme for Action next year includes a budget of ~40 million/year and has just been officially adopted. The first two years of the 2008-2014 funding period will include the implementation of financial tools per activity/project. Pilot Reference Networks will not be included as priorities.

The results of the European Health Info Survey will be included in the 1st quarter of the 2008 period.

It was asked what happened to the role of the RDTF in contributing to the Work Plan? G. Margetidis said the RDTF would be solicited for a written consultation.

D. Presentation on RDTF Upcoming Initiatives

Ségolène Aymé

Leader of Rare Disease Task Force

Director of Orphanet

Achievements of the Coding and Classification Working Group

The previous meetings were held on 11 October, 2006 and 2 May, 2007. The WG on Coding and Classification is involved in tackling the following issues:

- Comparing and correcting state-of-the-art coding systems
- Contributing to the WHO ICD-revision process
- Establishing a database of expert classifications of RD

Part of Orphanet mission is already to collect this information including MESH terms, MIM codes, proteins (via SwissProt collaboration), and genes which are linked to 2,000 diseases in the database. The prevalence, age of onset and other text is also associated with almost all of the diseases in the database. It is for this reason that S. Aymé has been selected as leader of the Rare Disease Topical Advisory Group.

RD can be made more traceable in mortality and morbidity information systems by defining one group of the “main” rare diseases which must have a specific ICD code and organising the remaining “ultra-rare” diseases under “other RD” subcategories of the more general codes.

The steps in this process include:

1. making a list of the diseases deserving a specific code
2. Analysing the current ICD coding system and identifying problems by cross-referencing many coding lists (with the cooperation of other coding bodies) which will be discussed at the next WG meeting as ~1200 disease have been crossed thus far.
3. Contributing to ICD-10plus, a tool provided by the WHO to communicate suggestions to the current ICD version

Cross-referencing data sets will allow the identification of mistakes in respective data sets, improve coding sets, and identify problems in ICD10 such as need for category rearrangement and need for more specific categories which reflect homogeneous groups of RD.

Consequent activities include:

- Continued matching of lists of codes
- Release of the new version of Orphanet with the classifications: Dec 2007
- Organisation of Coding and Classification WG meeting
 - Priority list
 - mismatches
- Apply for funding

Additional RDTF Initiatives in 2008

The proposed topic for the next Standards of Care WG workshop is the evaluation of the added value of European networks of Centres of Expertise. During this discussion the current networks scheme, the evaluation process used in France, and a proposal for a framework guiding an evaluation of the European added value will be analysed.

The proposed topics for a Health Indicators WG workshop is the indicators used to monitor RD and RD policy in Europe. More specifically, existing indicators, implementation of new indicators, and the areas of demography, epidemiology, health status, socioeconomic factors, health services, R&D outcomes, and policies will be reviewed.

A second workshop of the Health Indicators WG is proposed regarding RD registries. During this workshop existing registries, optimisation/exploitation of data, and the production of recommendations on ways to establish a registry, rules for accessing data, and prioritisation can be discussed.

The outcomes of these workshops will include a report on the methods used to assess the European added value of Networks of Centres of Expertise, a report on the indicators used to monitor the impact of the implementation of the Communication, as well as guidelines created to establish and exploit RD registries.

The activity of the Scientific Secretariat of the Rare Disease Task Force will unfortunately end on May 30, 2008.

Next Meetings

The drafting group for the Communication will meet February 13th in Brussels.

The 9th RDTF Meeting will be held 28 February, 2008 in Luxembourg.

The following WG workshops will be taking place in Paris:

Coding and Classification: February 6, 2008

European Networks of Centres of Expertise: March 11, 2008

Health Indicators: March 12, 2008

Health Registries: March 13, 2008