

# **Meeting Report**

## **9th Meeting of the European Commission “Rare Disease Task Force” Luxembourg, 28 February 2008**

On 28 February 2008, the ninth meeting of the Rare Diseases Task Force (RDTF) included the attendance of:

### **RDTF Members and Observers:**

V. Anastasiadou	E. Jessop	B. Pizzera Piantanida
S. Aymé	A. Kent	M. Posada de la Paz
R. Capocaccia	N. Kerlero de Rosbo	J. Sándor
H. Dolk	A. Kole	A. Schieppati
A. Fourcade	O. Kremp	S. Rumenov
L. Fregonese	J. Llinares-Garcia	J. Torrent Farneli
G. Gatta	S. Lynn	A. Vegnente
M. Gattorno	C. Nourissier	G. Zambruno

**DG SANCO:** A. Montserrat

**DG Recherche:** C. Berens

**PHEA:** G. Dargent

### **Welcome and Discussion of Agenda**

**Ségolène Aymé**

**Leader of the Rare Disease Task Force**

**Director of Orphanet**

S. Aymé opened the meeting by stressing the importance of the day's agenda as it most importantly includes a discussion on the public consultations of the Communication.

The minutes of the 8<sup>th</sup> RDTF meeting have not yet been distributed as they have not been approved by the Commission. They will be distributed shortly.

S. Aymé welcomed new participants:

- Stephen Lynn of the TREAT-NMD Network
- A. Vegnente, member of the EuroWilson project
- M. Datorno, member of the PRINTO network.

It was asked of the Commission if the leaders of newly funded RD projects have been invited to join the RDTF.

A. Montserrat replied that they will be officially invited for the next RDTF meeting.

No requests were made to change the agenda.

## **Public Consultation Regarding European Action in the Field of Rare Diseases**

**Antoni Montserrat  
DG SANCO**

### **The Communication Process**

A. Montserrat prefaced his presentation of the public consultation asking members to remember that we are in a transitional stage of the process in which the content of any documents may change very quickly. As such, no electronic copies of the documents under discussion were distributed. Nor were any slides used.

In 2008 the European Commission (EC) accepted a Communication in the field of RD addressed to the Council, the Parliament, and the Committee of Regions. The Committee of the Regions has explicitly asked to see the document. Although this is not an obligatory step in the process, it will give good political weight to the initiative and thus a positive aspect.

We have now completed the first step of the Communication process – the public consultation. The public has consulted on a document finalized by the following voluntary RDTF members for whom we thank for their hard work: Rumen Stefanov, Jordi Llinares Garcia, Domenica Taruscio, Laura Fregonese, Catherine Berens, Manuel Posada, Segolene Ayme and Christel Nourissier. They produced a document for consultation making as acceptable as possible for the EC, for the Parliament, and for the Member States (MS) – not an easy task. The most complicated task will follow with the incorporation of the responses of the public.

The Communication for public consultation was first presented at the European Conference of Rare Diseases (ECRD). The document was presented in 22 languages with the deadline of consultation on February 14, 2008. The authorities of several MS such as the United Kingdom, Germany, Spain, Denmark, and France have requested additional time. The EC will of course accept these any other late coming responses to the Communication, but will firmly close the invitation once all the contributions are published on the EC website (to be expected on Monday, March 3, 2008).

Over 600 contributions were sent to the EC regarding this communication. Many were simply confirmations of the document or congratulations on the work. Of these, 488 had significant content and will be posted on the EC website. This quantity of contributions the most successful in the area of health in the EC and is an enormous success in relation to other public consultations. For example, 200 contributions were sent for the consultation on a Communication regarding cross-border care and 60 contributions were received for a Communication on bio-terrorism.

Of the 488 public contributions:

- 25 came from the pharmaceutical industry (a very large and useful contribution)
- 46 came from international patient organisations
- several international orgs (who, etc)
- 26 from authorities of MS (this does not mean responses from 26 MS as some MS sent responses from several authorities)
- national orgs of patients
- 52 from patients themselves
- 12 came from reference networks
- 16 came from local authorities
- 4 came from research centres
- 40 came from universities
- 47 came from other parties

The next step entailed the analysis of contributions to arrive at the **Staff Working Document accompanying the White Paper on Rare Diseases** received today. In the Commission procedures a Staff Working Document is an annex to a Communication. Its presence is not obligatory, but it permits an extension of the (15 page) Communication another maximum of 40 pages (39 in this case). The boxes in this text summarize the actions that can be done. It becomes a sort of intermediate document between the Communication and the Impact Assessment. RDTF members have not received it electronically as it is only a draft, and will change according to the discussion.

After the completion of this annex the Impact Assessment will be drafted and presented to a tribunal where the added-value of the actions is assessed. This is currently scheduled for April 16. If the impact assessment is judged as contributing a significant added-value the document is presented to the Inter-Service Group on Health. After this passage the documents will be presented to the EC Inter-Service Group on Rare Diseases.

The impact assessment will be presented to the Commissioners sometime between July and September 2008. At this moment the Communication becomes a White Paper and will be presented to the Parliament if the rapporteur agrees. It is known that several people are fighting for position as rapporteur – which is a ‘good’ sign.

The following step is a process in the Council which includes a discussion with the Member States. This is a very frustrating experience as all must agree. The presence of the French presidency and the favourable attitudes of the Health Commissioner and the President of the Council are all good signs.

## **Discussion**

S. Aymé asked a question about timing of this process: if the Parliament only receives the White Paper in September can they have enough time to review it?

A. Montserrat responded that although there is no specific health group in the Parliament and time must be shared with all other social issues presented, it is possible to complete the review in 2 months.

A. Fourcade confirmed that the French Presidency supporting all actions and that they will do their best for it to be done by the end of 2008.

H. Dolk suggested that a list of those who responded to the Public Consultation favourably (even if without content) be created and added to the publication of responses. C. Nourissier agreed that it is very important to acknowledge all contributions even if they are not unique.

A. Montserrat presented a rough presentation of the list of contributions as a table which included the person or institution responding, the language, and a link to the response. From the 488 responses, 300 were in English, 150 in French, 70 in Polish, and the fourth largest groups in German and Spanish.

S. Aymé mentioned that it was unfortunate that there is not more time to review the responses in more depth as they are very valuable. She suggests that the RDTF publish a report of this analysis in the future. C. Nourissier stated that Eurordis will also be doing a more in depth analysis. A. Montserrat agreed that this sort of exercise is relevant material for many EU groups such as the COMP, Eurordis, and Orphanet.

H. Dolk expressed concern about contributions in foreign languages that were not translated. She suggested asking contributors to be made aware that if they would like their contribution to be distributed widely, to translate into English.

A. Montserrat responded that the Commission is not allowed to request for contributions in English as MS have the right to contribute in their native language. Unfortunately, the Commission does not have the resources to translate every contribution.

### **The Content of the Staff Working Document**

A. Montserrat introduced the Staff Working Document as having a similar structure to the Communication in that each section is concluded with a summary of defined actions. In Section 1 very few modifications are made in relation to the Communication. There are, however, a few changes in the RD provided as examples. Section 2 is an explanation of the political context of the Communication.

Section 5 is where differences between the content of the Communication and comments become more significant. The previously presented aims are proposed to be reached by 11 objectives and actions: 11 objectives detailed one by one.

1. To improve information, identification and knowledge on rare diseases
2. To improve prevention, diagnosis and care of patients with Rare Diseases

3. To develop national/regional centres of reference and establish EU reference networks
4. To ensure equal access to all EU patients to orphan drugs and compassionate use
5. To develop specialised and adapted social services for rare diseases patients
6. To gather at European level the limited and scattered expertise on rare diseases.
7. To accelerate research and developments in the field of Rare Diseases and Orphan Drugs
8. To empower patients with Rare Diseases at individual and collective level
9. To support implementation of National Plans for Rare Diseases
10. To develop the international cooperation on rare diseases
11. To coordinate the policies and initiatives at EU level

Participants of the meeting agreed on the following conclusions for each objective:

### **Objective 1 – To improve information, identification and knowledge on RD**

#### **Actions:**

- The EU definition of rare disease based on a prevalence of less than 5 per 10,000 is maintained (Commission, EMEA)
- An EU or an international project exploring an incidence based definition of rare diseases will be launched (Commission)
- The EU will contribute to the ongoing process of revision of the ICD (International Classification of Diseases) in order to ensure appropriate codification and classification of rare diseases in the future ICD-11. A working group will be supported for all the period of this revision (Commission, WHO)
- The EU will establish since 2009 an Inventory of Rare Diseases to be periodically updated (Commission)
- The database Orphanet will be supported using appropriate financial instruments from the Health Programme or the FP7 (Commission)
- The support to the disease information networks through the Health Programme and the FP7 should be pursued (Commission)

The fact that there is no intention to change or discuss the threshold of a RD as it applies to the Orphan Drug regulation should be emphasized. It should rather be stated that both indicators – prevalence and incidence- have different values and should be used in different contexts. Using the EMEA text on the use of these two indicators should be used to find correct language. An EU or international project should collect incidence data as well and a purpose-oriented approach should be employed when using these indicators. If RD are classified by prevalence as well, the classification of rare and ultra-rare will be implied.

## **Objective 2 – To improve prevention, diagnosis and care of patients with RD**

### **Actions:**

- Development of e-Health in the field of RD using on-line and electronic tools (Commission)
- To create a help line unique EU-wide number for information and social services on rare diseases a 116 number (Commission)
- An evaluation of population screening (including neonatal screening and Pre-implantation Genetic Diagnosis) strategies for Rare Diseases in the Member States will be launched (Commission, Member States)
- To launch a European series Patient Leaflets on some rare diseases in all the EU languages (Commission, Member States)

It must be clarified that the 116 help-line involves the establishment of national help lines that will all use the same number.

Currently, an evaluation of population screening strategies in the field of RD should be launched at the EU level as there is a need for a combined European effort. It is premature to give recommendations on how MS should implement policies on population screening, but it must be emphasized that the decision is ultimately up to the MS. Screening and diagnosis should be separated as concepts and although the discussion of Pre-implantation Genetic Diagnosis should remain in the preceding text (as it was frequently expressed in Communication responses), it should not be included in the action points.

E-Health is mentioned as a highly supported development at the European Commission. The field of RD can greatly benefit from online and electronic tools created at the European level. It is perhaps one of the fields in health that can most greatly benefit from such tools.

## **Objective 3 – To develop national/regional CE and establish ERN**

### **Actions**

- To repertory in an EU list the existing Centres of Expertise identified throughout the Member States by the end of 2010 (Commission)
- To establish by Commission Decision a procedure for designation and accreditation methodology of EU Reference Networks for Rare Diseases (Commission)

- To provide adequate, long-term public funding to Centres of Expertise in order to ensure their sustainability and continuity of care for patients (Commission, Member States);
- To recommend inclusion in the National Plan for Rare Diseases provisions on the creation of Centres of Reference and their participation in European Reference Networks (Commission, Member States)

Orphanet currently maintains a list of existing Centres of Expertise (CE) throughout Europe. It must be decided how this effort will be supported and sustained by the EC.

The accreditation of CE should only be at the national level. It is questionable how and why ERN should be evaluated at the EU level, though it is clear that such a surveillance could ensure that such networks benefit countries which do not have adequate resource to serve RD patients and support RD research on their own. The next RDTF A procedure for the creation, selection, evaluation, etc. of European Reference Network (ERN) must be established. This will be the centre of the discussion during the next RDTF Standards of Care Working Group meeting.

The definition and responsibilities of a CE and a ERN must be more correctly and precisely defined. A significant number of hospital representatives responded that they do not have adequate resources to support CE (in most cases hospitals) activities as described in the Communication.

In the third action point 'Centres of Expertise' should be replaced by 'European Reference Networks'. Despite this clarification in vocabulary, the difficulty in guaranteeing long-term funding for ERN remains. Currently participating institutions of any networks in the field of RD are funded by DG research. DG Research funding competitive and renewable by nature (and is unknown after the end of and thus, long-term funding is problematic. In addition, it is unknown what will happen at the end of the Programme of Community Action in Public Health in 2013.

Several RDTF member expressed reservations in the official designation of ERN as doing so would mean the exclusion of networks that although are not official are equally if not more effective. The scope of these networks also remains unclear. Nevertheless, RDTF members expressed the opinion that ERN should focus on actions not manageable at the MS level (e.g. databases and registries, outreach to MS in need).

#### **Objective 4 – To ensure equal access to OD and compassionate use**

##### **Actions**

- To explore additional incentives at national or European level to strengthen research into rare diseases and development of orphan medicinal products, and Member State familiarity with these products (Commission, EMEA)

- To establish of a European Committee to evaluate the appropriateness of compassionate use (Commission, EMEA)
- A European guideline should be developed clarifying responsibilities in a Compassionate Use situation (Commission, EMEA)
- A European Guideline should be developed clarifying requirements for import licenses and labelling (Commission, EMEA)
- The Commission should present, a report to the Council and the Parliament identifying bottlenecks on orphan drugs access (delays, marketing, access, reimbursement, prices, etc.) every two years, proposing the necessary legislative modifications in order to guarantee equal access to orphan drugs throughout the EU on the basis of a COMP/EUACRD European collaborative scientific assessment (Commission, EMEA)
- An EMEA Committee for the assessment of the Therapeutic Added Value of Orphan Drugs should perform a common scientific assessment of the TAV for each Orphan Drug and deliver an opinion document (Commission, EMEA)
- A feasibility study should be launched to explore incentives for industry in the field of medical devices and diagnostics for RD (Commission)

Significant contributions were received concerning this objective, namely from the COMP, Eurordis, and the pharmaceutical industry. One simple observation in the contributions is that big pharmaceutical companies were largely in favour of the OD section of the Communication whereas smaller companies were not.

Activities described in Action point 1 currently exist for drugs in general; the specification for these activities for OD may be unrealistic. It was suggested that 'medical devices' be added to Action point 1.

Many contributions reflected the opinion that RD patients are 'used as guinea pigs' in the compassionate use program. It was agreed that it should simply be stated that compassionate use is an issue, without specifying the proposal of a committee dedicated to the topic as compassionate use should be regarded as part of a national health plan.

Guidelines clarifying responsibilities in a Compassionate Use situation are homogenous in many MS and even non-existent in others. As such it could be beneficial to provide such guidelines at the EU level.

The discussion of reimbursement and pricing posed difficulty as reimbursement is done at the MS level and pricing is determined by pharmaceutical companies with an attempt to keep them the same across Europe. RDTF members agreed that the pharmaceutical industry should be more transparent in how they arrive at a price.



The therapeutic added-value of orphan drugs is linked to reimbursement and pricing. As some MS would welcome this assessment at the European level, others reject it. It was suggested that Action point 6 should be made more general.

A European approach assessing the use medical devices is not agreed upon by all stakeholders. Therefore, at this time, the action point should either be removed or softened.

### **Objective 5 – To develop specialised and adapted social services for RD patients**

#### **Actions**

- The European Commission should recommended the adoption of national initiatives in the National Plans for Rare Diseases on specialised social services (Member States)
- Provide financial support for this activity through European Networks of Therapeutic Recreation Programmes, using the Health Programme and the Disability Action Plans (Commission, Member States)

The development of social services is a national competence. The Commission can only recommend that the development of such services be included in national initiatives and provide financial support through the support of networks in general and not specific activities such as recreational programs.

### **Objective 6 – To gather limited and scattered expertise on RD at the EU level**

#### **Actions:**

- The Health Programme and the FP7 will continue to support, in a coordinated way, registries, databases and biobanks on rare diseases with appropriate financial tools for a sustainable funding (Commission)
- The Commission will establish, by Commission Decision, a publicly accessible EU Register of Rare Diseases patient registers databases and biobanks defining criteria for register accreditation and qualification and the access to samples. EU registering will become mandatory for publicly funded or co-funded repositories under the Health Programme and the FP7 (Commission)
- Specific support to further research into biomarkers should also be given to encourage long-term follow-up, and the acquisition of robust evidence on clinical effectiveness (Commission)

The second action point is included to control the inappropriate nomination of a EU registry, database, etc. This will simply include the documentation (not accreditation) of patient registries, databases, biobanks, etc.

C. Berens suggested moving the third action point under Objective 7 as it relates more to RD research. She strongly urges finding an appropriate solution for long-term funding of such registries because it will not come from DG Research.

### **Objective 7 – To accelerate research and developments in the field of RD and OD**

#### **Actions**

- The EU Advisory Committee on RD and the Committee for Orphan Medicinal Products (COMP) in the EMEA (European Medicines Agency) will address to the FP7 biannual recommendation on research priorities on RD (Commission, EMEA)
- To create an adequate EU mechanism through the FP7 to specifically support the clinical research development of designated orphan medicinal products up to the end of phase II and the specific Genetic and molecular characterisation for the more than 4 000 diseases for which it remains to be done (Commission)
- To launch the creation of a public-private foundation for RD, the European Research Foundation for Rare Diseases (Commission, Member States, private sector)

Though research priorities cannot be established in this document, a procedure of how to communicate biannual recommendations on research priorities in RD can be established.

The second action point was proposed by several pharmaceutical companies in their response to the Communication as a development that can benefit both the industry and patients.

The role of the proposed public-private foundation for RD should be better defined. A feasibility study on its function was proposed.

### **Objective 8 - To empower RD patients at individual and collective level**

#### **Actions**

- The Health Programme will continue to integrate the support to the patient's organisations as a priority for action (Commission, Member States)

As the goal of support for patient organizations is to create equity, patient organization representatives should receive support to make sure that they are not disadvantaged by their voluntary participation in RD work (meetings, committees, etc.)

### **Objective 9 – To support implementation of National Plans for Rare Diseases**

**Actions**

- The Member States are invited to establish national or regional action plans for RD in order to implement the actions suggested in the Commission Communication and the Council Recommendation before 2010 (Member States)
- The Commission will provide European guidelines for the elaboration of these action plans for RD. Appropriate international conferences will be organised (Commission)

Instead of enforcing a precise deadline (which introduces a risk of MS not meeting the deadline) MS should be required to submit yearly reports on their progress towards establishing a national plan on RD.

This objective should be introduced earlier in the document.

**Objective 10 – To develop international cooperation on RD****Actions**

- An international cooperation framework on rare diseases with other countries (e.g. US, Canada, Japan, Singapore, Australia,...) will be adopted (Commission)
- A proposal of resolution on an international action in the field of rare diseases will be submitted by the European Commission to the World Health Assembly (Commission)

This objective was missing from the Communication, but appeared in many contributions and was welcomed by RDTF members. It was suggested that this could be an appropriate Objective under which the cooperation with the WHO on the Coding and Classification of RD can be reaffirmed.

**Objective 11 – To coordinate the policies and initiatives at EU level****Actions**

- An EU Advisory Committee on Rare Disease (EUACRD) will be created, by Commission Decision, in order to advice the European Commission. A specific budget for this Committee will be allocated in a sustainable basis in the EU budget (Commission)
- The European Conferences on Rare Diseases will be organised every two or three years. Funds will be provided by the Health Programme (Commission).
- A Rare Diseases Fund will be included in the Financial Perspectives (2014-2020) (Commission).

- A feasibility study for the creation of a European Agency on Rare Diseases will be launched in 2009 by the Health Programme (Commission).
- Every two years the Commission should produce an Implementation report on the Commission Communication and Council Recommendation addressed to the Council, the Parliament, the Social and Economic Committee and the Committee of the Regions (Commission)

The EU Advisory Committee on Rare Disease (EUACRD) is not a new title for the RDTF, but the creation of a new structure. The members of this committee will include a representative from each of the 27 MS (more if it is decided to include candidate or neighbouring countries). This framework of an advisory committee will result in a loss of intimacy but a gain in legitimacy in relation to the current structure of the RDTF. Current RDTF members expressed concern regarding the expertise of each designated MS representative, the flexibility in the agenda, and the possibility of inviting other experts as needed. The EUACRD will be supported until 2013 after which its support will have to be reassessed.

Among contributions to the public consultation of the Communication most patient organisations were in favour of the creation of a European Agency on RD, most pharmaceutical companies were not, and MS health authorities were divided. It is proposed that a study should be launched to investigate the relevance and feasibility of such a structure.

## **European Conference on Rare Diseases 2007**

**Christel Nourissier**

**Eurordis**

The 2007 European Conference on Rare Diseases was organized in Lisbon, Portugal under the Portuguese presidency and its partners. It was attended by participants from 35 countries by a good balance of stakeholders. The conference received good media coverage which was encouraging as one goal of the conference was to raise awareness of RD in Portugal. This was a crucial moment and Portugal is currently working on a National Plan of Rare Diseases.

The main outcome of the conference was the successful launch of the Communication as we see from the large amount of responses received. This meeting also served as a platform for the first presentation of the outcomes of the RAPSODY project.

The next ECRD is scheduled in Poland under the DG SANCO funded POLKA project. This conference will be much larger with more satellite meetings, more patient representatives and more of an opportunity for communication with neighbouring countries. The conference is proposed to take place in Poland in May 2009 though this has not yet been confirmed.

S. Aymé congratulated Eurordis on their previous ECRD meetings. She stated that as the RDTF was not very involved in the previous meetings, it would be nice to work more together on future conferences.

## **Workshops of the Task Force**

**Ségolène Aymé**

**Leader of the Rare Disease Task Force**

**Director of Orphanet**

Four workshops have been planned this spring. The first – Workshop on Coding and Classification – took place on February 6, 2008. Outcomes of this workshop will be presented soon. The Coding and Classification Working Group has already met several times. It has agreed on several principles of action:

- 1) Rare Diseases should be traceable in mortality and morbidity information systems
- 2) There are two categories of RD:
  - The recurrent RD (~1,500 to 2,000) should have a specific code in ICD-11
  - The ultra-rare (~4,000) should be coded as “other specific RD” within relevant subcategory but indexed nevertheless

The Working Group has also agreed on the following criteria for assigning RD with a specific code in ICD-11:

- Any disease coded in a registry of patients or an information system
- Any disease covered by a support group
- Any disease with a clinical test

Using these criteria, Orphanet has identified almost 900 diseases of which 400 do not have a specific code in the ICD-10 and must assigned a code. In the next three years these suggestions will reviewed by experts and presented for comments using the WHO’s technology platform for the ICD revision process.

The WHO is fully open in its organisation of RD in ICD-11 is fully open in its organization. For RD a clinical approach based on medical specialties is most appropriate where some classifications are by aetiology and others by anatomy. All RDTF members will have an opportunity to comment on the organisation via WHO technological platform.

The Workshop on Assessing the Added-Value of Centres of Expertise (CE) and European Reference Networks (ERN) in RD will take place on March 11, 2008 in Paris. Participants will be provided with a workshop working document which will serve as a working document. Following discussion with workshop participants, and consultation with other experts, a new draft of the document will be published as an RDTF Scoping Paper on the topic by June 2008.

The Workshop on Health Indicators (HI) will take place on March 12, 2008 in Paris. The working document for this workshop is prepared by L. Fregonese and the RDTF Secretariat and will include the following topics of discussion:

- Discussion of HI in the Communication
- Definition, objectives, and legal basis for HI
- Past and ongoing projects on HI
- Potential Sources of Data
- Criteria of Selection of appropriate HI for RD

The ultimate goal is to create a list of feasible HI for RD.

The last workshop on Health Registries and Databases will take place on March 13, 2008 in Paris. The aim of this workshop is to produce recommendations and guidelines for:

- requirements in maintaining and maximising the use of registries
- obtaining funding to support tools and resources shared by rare disease registries on a European level;
- creating a repository of data in the event of a termination of registry funding.

In the next coming years the RDTF will have to identify new working groups addressing new pressing issues. Suggestions are welcome.

### **New projects selected for funding in 2007**

**Gemma Gatta**

**RARECARE**

**Fondazione IRCCS Istituto Nazionale dei Tumori**

The project began in April 2007 and is funded for three years. The aims of the project are:

- To provide an operational definition of “rare cancers”, and a list of cancers that meet this definition
- To estimate the burden of rare cancers in Europe
- To improve the quality of data on rare cancers
- To develop strategies and mechanisms for the diffusion of information among all the key players involved in Europe-wide surveillance on and treatment of rare cancers

The proposed actions of the project include:

- Estimation of incidence, survival, prevalence and mortality for all rare cancers
- Analysis of data quality for a subset of cancers, by confirming the diagnostic data and, if possible, analysis additional data on stage and treatment
- A web-site on rare cancers will be designed to disseminate the results of the project, and in particular, to inform clinicians, patients and health planners

The major aim of year one is to agree on an international level on a list of rare cancers. To attain this goal, periodic meetings of clinicians, advocacy groups, epidemiologists, etc. have been organized. During the most recent meetings, the topography and morphology of approximately 400 entities was presented and incidence and prevalence was calculated. Using the threshold of 3 per 100,000, a list of rare cancers was defined. Four additional meetings of this nature are planned.

J. Llinares-Garcia asked why the threshold of 3 per 100,000 was chosen. He also requested if the EMEA could be provided with this data as a large portion of applications concern rare cancers.

G. Gatta replied that the threshold of 3 per 100,000 was chosen because with this threshold 55% of all patients are included. She also responded that an EMEA representative was invited to the meeting, but responded that they could not participate because the RARECARE project measured incidence and not prevalence.

S. Aymé stated that she did not agree with some of the approaches. She felt that the threshold is not relevant, and that it should not matter the percent of patients included as long as it reflects reality. She stated that in her opinion experts who specialized in one area (such as cancer) lose perspective on what cases are rare because they see them relatively often. Oftentimes this is not the reality and their work loses credibility if a solid foundation for choosing a threshold is not established.

A. Montserrat concluded this presentation by stating that purpose of any project is that is proves useful for the European Commission. Consequently, the results should have utility, continuity, etc.

## **New projects selected in 2007 for future funding**

**Antoni Montserrat**

**DG SANCO**

- European Haemophilia Safety Surveillance System (EUHASS)
- Patients' Consensus on Preferred Policy Scenarios for Rare Diseases (POLKA)
- European network of paediatric Hodgkin's lymphoma – European-wide organisation of quality controlled treatment (Paediatric Hodgkin Network)
- The PRES Network for Autoinflammatory Diseases in childhood (EuroFever)
- European Network of Reference for Rare Paediatric Neurological Diseases (nEUroped)
- European Project for Rare Diseases National Plans Development (EUROPLAN)
- A reference network for Langerhans cell histiocytosis and associated syndrome in EU (EURO-HISTIO-NET 2008)

## **Next call for proposals**

**Antoni Montserrat  
DG SANCO**

The next Call for Proposals is being launched tomorrow. The deadline for this call is **23 May 2008**.

In the call several new important themes can be observed:

- Co-financing of projects intended to achieve a Programme objective (call for project proposals)
- Co-financing of the operating costs of non-governmental organisations or specialised networks (operating grants) – intended for projects requiring stability such as registries.
- Co-financing of conferences intended to achieve a Programme objective (call for conference proposals) – intended for small conferences and small working groups
- Joint actions by the Community and Member States as well as other (third) countries participating in the Programme – A joint action exist between (at least five) MS and the Commission. This instrument will allow for faster turn-around. This instrument will be used to fund the activities of the RDTF.
- Tendering of actions to achieve a Programme objective – This instrument will not affect RD.

On March 12, an Info day on calls for proposals is scheduled but unfortunately it is already full. A few additional more information days in other MS are being organized as the Luxembourg Info Day is full.

## **Future Actions of the Rare Disease Task Force**

**Ségolène Aymé  
Leader of the Rare Disease Task Force  
Director of Orphanet**

S. Aymé emphasized that input of RDTF member is very important as we will have to apply for funding together under the new instruments of funding. She herself has listed several possible topics and welcomes any others: indicators, population screening for RD, principles guiding compassionate use.

N. Kerlero de Rosbo proposed a workshop on ethics in the context of trans-European RD projects, networks, data sharing, etc.

A. Montserrat responded that a subgroup of the Network of Competent Authorities is already working on this but that a discussion specifically about RD could be useful.

Cornelia suggested work on prenatal diagnosis.  
The group agreed that this was not a topic for the RDTF.



L. Fregonese suggested a workshop on the sharing of health indicators. So many projects are funded to collect health indicator data and the results could be consolidated to try and make the most of the results of past and ongoing data collection projects in the field of RD.

Several RDTF members agreed and A. Montserrat agreed to propose it to the PHEA.

**Presentation of the 3<sup>rd</sup> Eastern European Conference on Rare Diseases and Orphan Drugs**  
**Ruman Stefanov**  
**ICRORD**

This conference takes place in Plovdiv, Bulgaria beginning on March 1<sup>st</sup>, 2008. The Bulgarian vice Minister of Health will open the conference and 200 participants are anticipated. Twenty-five participants will attend a satellite workshop on the Marketing of Orphanet drugs. The results of this conference will be presented to RDTF members.

S. Stefanov added that he just received news of the resignation of Commissioner Markos Kyprianou and asked how this will affect the work on RD.

A. Montserrat responded that indeed Commissioner Markos Kyprianou has resigned and that he will most likely be replaced by Commissioner Maglena Kuneva to allow for a smooth transition.

**Rare Disease Meetings of the French Presidency**  
**Ségolène Aymé**  
**Leader of the Rare Disease Task Force**  
**Director of Orphanet**

On November 18, 2008 a conference on National Health Plans will take place in Paris. Please note the change in date. On October 13 or 14 (to be confirmed) another a session on RD will take place as part of a larger conference at the Ministry of Health.

Though not an official event sponsored by the French presidency, the next EPPOSI meeting (European Platform for Patients' Organisations, Science and Industry) will also take place in Paris (date to be decided).

**Next Meeting of the RDTF**

July 3<sup>rd</sup>, 2008