

## **Minutes of 5th meeting of the Rare Diseases Task Force Luxembourg, 8 June 06**

### **Changes to the agenda:**

#### **Minutes from the previous meeting**

Minutes from the previous meeting were not received in advance and therefore could not be approved. Members were asked to comment on the minutes and send their remarks to the secretary who will then distribute the revised version.

**News update from the commission** (added to the agenda).

#### **Report from Antoni Montserrat:**

Brief update on the new programme for Public health:

The Commission is finalising the approval of a new programme for public health for 2007-2013. This programme was initially submitted as a joint programme on health and consumer protection but was rejected by the council and the parliament. The new programme has been split, allowing the programme for public health to be integrated into the new financial perspectives of the EU. In terms of content, the initial programme contained a strand specifically labelled 'diseases'. Unfortunately, this proposal was rejected and the new programme contains three strands (Health Security, Prosperity and Solidarity, and Generation of Knowledge in the EU), with no specific strand for diseases. New programme will be approved by the council and parliament by the end of the year.

There are two major new priorities of relevance to the RDTF: Children's health and health inequalities. Rare diseases are recognised in the new programme. **A. Montserrat** concluded that the representation of rare diseases in the new programme is satisfactory. However, it is important to note that no specific disease is mentioned in the new text and all diseases are considered at the same level.

In terms of funding, globally the new programme will receive more money than in the past but the three new agencies created in the past years in the field of public health will be supported and maintained by money from the programme. In practical terms, this means that the money available in the new programmes is reduced. For *e.g.* In 2005-2006 the money available for the public information strand of the old programme was around 15 million per year, for the new programme this will be 10 million or less. Existing projects will not be affected by these new funding decisions but their continuation will.

In addition, the new executive agency for the implementation of the public health programme will now be involved in the implementation and evaluation of new projects. The commission also announced in the text of the new programme that it will use increasing the method of 'call for tender' rather than 'call for proposals' for new projects. There will be three types of call for tender:

Free competition between institutions on a contract in a given field

Restricted to a small number of institutions for leaner operations (<125 000 euros)

Restricted to one institution termed the 'monopoly of knowledge'. This type of call for tender is not really applicable for rare diseases but may be used for precise actions should as a conference.

In terms of new projects, the number of submitted projects up in the field of rare diseases has increased from 4-5 in previous years, to 14 this year. Selection will be completed by December 2006.

**H. Sundseth** commented that it was also pointed out that from the cancer patient perspective that while the new programme may benefit rare diseases it is a disaster for major chronic diseases to delete the 'disease' strand.

**C. Zeidler:** Is there any decision on money sharing between applicant institutions and the EC?

**Answer:** Original proposal of 95% was rejected. 80% is the current figure. Criteria should be more flexible.

**N. Ruperto:** Will there be a 2-step procedure involved in the evaluation process (letter of intent prior to full application) ?

**Answer:** Whilst unfamiliar with the FP7 process, it was agreed that this may be an interesting possibility to provide a step between a restricted call for tender and call for proposals.

### **Presentation of the new EU Health Portal:**

The new EU Health Portal was presented at the recent conference (10-11 May) in Malaga. The portal contains a guide to websites and other resources from all member states and international organisations. The portal address is as follows: [http://ec.europa.eu/health-eu/index\\_en.htm](http://ec.europa.eu/health-eu/index_en.htm)). Under the theme health problems, there is a page on rare diseases. There is also a link towards the DG SANCO website, from where links to all DG SANCO projects can be found. Resources in the portal will incorporate national resources from the member states and will be available first in 21 languages and then eventually 27. At present the site is currently only available in English. While specific questions concerning rare diseases should still be sent to **A. Montserrat**, comments concerning the portal should be sent to **Daniel Mann** ([daniel.mann@ec.europa.eu](mailto:daniel.mann@ec.europa.eu)).

### **DG research: report by Catherine Berens**

The figure for the FP7 has now been set at a little more than €50 billion (there was no doubling of the budget as originally proposed by the European Commission). However, the health theme of the research programme is less affected by budget cuts than other areas: €8.3 billion was requested, just less than €6 billion will be received. However, the details of this allocation and the text of the framework is still under discussion. The first reading will hopefully be adopted in June by the European Parliament and, once a consensus is reached and any amendments incorporated, the second reading should take place after the summer. Following this schedule, the process should be finished by the end of 2006, ready for launch of the first call for proposals at the beginning of 2007.

With regard to the budget it is important to note that the general plan for the programme is a progressive effort.

Regarding rare diseases, there is no conclusion as yet ; the text is still being discussed. Rare diseases come under the second pillar of the Health theme in collaborative research: translating research for human health. Several bullet points cover various diseases, including a bullet point on natural history studies, patho-physiology and development of preventive, diagnostic and therapeutic approaches. However there is some crossover between other bullet points included under the health theme.

Overall, the impact of FP7's adopted budget will probably mainly relates to alterations in the number of rare diseases projects funded rather than on the orientation of the calls themselves. A clear picture of FP7 should be available at the next meeting in December.

FP6: evaluation of the fourth call for proposals is now complete and negotiations are now underway on the projects that have been selected. For rare diseases there are three new projects accepted under the bullet point "Cardiovascular, diabetes and rare diseases" of the "major diseases" section.

Figures are not final but for projects on rare diseases under the bullet point "cardiovascular diseases, diabetes and rare diseases", we have around €46 million. If taken together, the budget available for projects relevant to rare diseases in the health priority, amounts to more than €200 million (compared to €64 million in FP5). Programmes relating to the coordination of rare disease issues at EU level, such as the Orphanplatform project, have also received attention in FP6. This will continue in FP7. In contrast, funding of clinical trials is not well covered and this needs to be taken into consideration for the next programme.

**N. Ruperto:** What about children's health?

**C. Berens:** Children's health issues will be treated as a horizontal priority throughout the FP7 Health theme. The aim is to identify topics that are particularly relevant to children's health. Studies will be conducted under the framework programme for research dedicated to developing new medicines for children, in particular medicines that are no longer under patent (off-patent medicines). This is to be discussed with EMEA.

**N. Ruperto :** Who is on the advisory board?

**C. Berens:** Renowned scientists, Heads of research institutes, industrial expertise and one patient representative

**S. Aymé** expressed concern that the FP6 calls for proposals are too specific and restrict the applicants. How were these topics selected and by whom?

Here is an outline of the plan: Scientific community were first asked to send in ideas for proposals / Evaluation with independent experts / Priorities selected / Other ideas stemming for various sources and agencies / Discussions with the advisory groups to define final topics. However, it has been recognised that these topics were perhaps too narrow and prevented applications. For FP7 a balance needs to be found between broadening the subject areas to allow more interested parties to apply and receiving a number of proposals that is appropriate for the restricted budget (balance between more open topics and reasonable success rate).

**H. Dolk:** What is the average size and range of the budgets in FP6?

**C. Berens:** €10-€12 million for big projects (duration 4-5 years) and €2-€3 million for small projects (over 3 years).

### **EMEA – Jordi Linares-Garcia (Report of activity over the past 6 months)**

**Inventory of national initiatives:** Data collection was completed in 2005 by the secretariat of the Rare Diseases task Force. The validation process by DG Enterprise is on its way. It should be published soon on the commission's website.

**Guidelines in preparation for article 8A2 of the Orphan drug designation:** these will have a major impact on Orphan drug authorisation and may lead to withdrawal of exclusivity for a given drug if this drug is deemed to be sufficiently profitable. However there is concern that this will be a dis-incentive for drug companies. The commission is currently reconsidering the guidelines and a decision will be reached in June.

**Impact of the regulation:** The regulation on orphan drugs has very positive effects: more than 100 applications this year and 29 market authorisation of an Orphan drug so far.

**New regulation:** Any drug wishing to be authorised or wishing for an extension will have to consult with the EMEA for a potential application in children. If there is a potential use in children, an agreement must be reached between the EMEA and the sponsor as a way of scrutinizing all drugs for potential use in children.

**C. Zeidler:** What about drugs that haven't been introduced as Orphan drugs but are used as such and are very expensive? Is there any way to get these drugs into the Orphan drug register to make them less expensive?

**Answer:** No.

**N. Ruperto:** What about already patented drugs with no paediatric authorisation:?

**Answer:** For these drugs, the EMEA will force revision for use in children when any application for an alteration or extension of the authorisation is requested. Authorisation will be blocked if the paediatric potential of these drugs is not developed.

**H. Sundseth:** What is the composition of the paediatric committee that will be set up at the EMEA? It was noted that a balance has been established: companies are obliged to search for potential use in the paediatric population but they get a 6 months patent extension in exchange.

**Answer:** Not confirmed but is expected to contain 1 member per member state plus co-opted members and representative of the CHMP programme for paediatrics.

**C. Nourissier:** said that Eurordis has been actively advocating for the vote of paediatric drug regulation since Dec 2000, because rare diseases affect many children.

### **Reports on recent National initiatives**

**Hungary (J. Sandor)** There is a project to assign the mission of surveying rare diseases in Hungary to the epidemiology centre in charge of monitoring congenital malformations. Decision will be taken soon.

**Italy (D. Taruscio):** Italian drug agency (AIFA) is funding academic trials on orphan drugs and has just completed the evaluation phase.

Launched a new programme to fund research projects on rare diseases with main emphasis on diseases without a diagnosis with the aim of reaching a consensus for diagnosis of these diseases. There is also an epidemiological theme, starting and collaborating with the national registry to fund projects on risk factors.

In Italy there is a division of duties regarding pre-clinical and clinical trials, with the Italian drug agency funding projects concerning clinical studies.

**N. Ruperto** reported on the 2 stage application system used by the Italian Drug Agency: 130 short applications were received and one third of these were then followed-up with a longer full application being requested. Around 10% of the original projects were selected for funding. This allows the evaluation to concentrate on the most interesting applications.

**S. Aymé** noted that the funding in this case comes from private industry (5% of all marketing expenses go to funding of academic clinical research and trials). She also commented on the fact that the topics selected for funding were very well thought out, using academic trials to evaluate protocols and thus allowing comparisons between treatments.

**Spain (M. Posada de la Paz):** Setting up of new commissions to study all aspects of rare diseases in Spain. New initiatives are being put in place for basic research New decree for regulation of rare diseases

**Bulgaria (R. Stefanov):** Very successful lobbying for rare diseases Current discussion for the establishment of a national plan for rare diseases

**Germany (C. Zeidler):** Announced that rare diseases networks in Germany will be funded for another two years but no new projects have been announced.

**France (S. Aymé):** France is currently expanding labelling of centres of reference (already 67 but the aim is 100 this year). Analysis of the situation shows that now most of the rare diseases are covered by an existing centre. Diseases treated by surgery pose a specific problem as surgeons tend to be less willing to organise the long-term follow-up of patients with rare diseases. It may be difficult to find CRs for these diseases.

**Denmark (M. Jespersen):** At present, Denmark is undergoing social institutional reforms that will lead to redistribution of care services; these reforms will most likely have an impact on management of rare diseases.

**Luxemburg (Y. Wagner):**

Luxembourg has been very active in the field of rare diseases over the past year. It has set up a multidisciplinary committee working with the medical association in the fields of screening for newborns and establishing patient directories. There is also a national plan for a rare diseases forum that will provide information and guidance for all people with questions concerning rare diseases. There has also been progress towards setting up of a national registry and a national inquiry is underway for rare diseases.

## **Activities of the RDTF (S. Aymé) :**

### **Classification of rare diseases.**

Two international collaborations have been established. The first was with the WHO regarding the revision of the international classification of diseases. At the previous meeting on the 21<sup>st</sup> of Dec 2005, it was agreed that the preliminary data collected by Orphanet was useful for this revision and it was agreed that there should be collaboration to establish a common agenda for classification but the revision should be open to world experts with any suggestions collected and made visible. **S Aymé** is concerned that the WHO representatives show too much interest and expectation on a classification based on genetics. The meeting planned for May was cancelled but will be rescheduled.

The second was with the Office of Rare Diseases in the US with a general agreement for increased communication. The plan is to have a common meeting in Madrid in October at the EPPOSI partnering workshop on Orphan drugs. The ICORD (International Conference on Rare Diseases) which aims to establish an international society for rare diseases will also provide an opportunity for collaboration and the chance to establish a common agenda.

### **OrphaNews Europe:**

The newsletter has 6700 registered readers but also a significant number of non-registered readers receiving the newsletter through inter-establishment distribution.

**S. Aymé** reminded the participants that the newsletter covers both scientific and political issues and that members should send in relevant information including that concerning upcoming events and workshops.

To assess the satisfaction of readers, an on-line survey was performed. 500 responses were received. The majority of readers have a strong research interest followed by academic health. Most of the comments came from individuals working for small biotech companies, noting that OrphaNews provided an essential link to events and information on rare diseases. Readers came from 26 countries. Most of the readers are satisfied - only 2.4% stated that the newsletter did not fulfil their needs, but no further comments were made. Most readers (3/4) reported that the newsletter was well adapted to their level of knowledge. Most interesting topics covered were research issues (as a result the research aspects will now receive more attention) followed by issues concerning Orphan Drugs and the national and international news section. **Main comments:** 1) Congratulations!!!! 2) Include a section on primary care and general practice in the field of rare diseases. **S. Aymé** proposed that the results of a systematic search for publications in this area could be included in the newsletter. 3) Include a section on new diagnostic tools. **S. Aymé** proposed that the manufacturers association could be contacted to provide information concerning new products relevant to rare diseases. Eurogentest could also be contacted. 4) Section on reimbursement for diagnostics. **S. Aymé** thinks it will be too difficult to gather information on this topic. 5) Section on changes concerning pharmaceutical and biotech companies (**S. Aymé** thinks this is a good idea as there are frequent movements in the setting up of new companies). 6) Job opportunities in the field of rare diseases. 7) More space for patient organisations. 8) General requests included addition of a search tool, translation of the newsletter into other languages, creation of a summary section of the main info at the top of the newsletter.

The majority of readers discovered the newsletter through Orphanet or Google (only 3% discovered it through Cordis).

### **Discussion of Newsletter:**

**H. Dolk: Archiving of the newsletter in sections and search engine tool available to search this archives.** S. Aymé proposes to extract information from the newsletter section by section and put it on the website of the Rare Diseases Task Force. Then introduce a search engine tool into the website rather than the newsletter.

**C. Zeidler:** Commented that the size of the newsletter is good; additional entries may make the newsletter unreadable on a monthly basis. Most of this additional information could be managed by adding a list of links to appropriate websites.

**A. Micheli** pointed out that there is a need for a link between research and policy as research knowledge is not always strongly represented in health policy and society.

**D. Taruscio:** pointed out that Eurordis is also doing a good job and therefore a link should be constructed between the patient communities and professional organisations. Secondly, concerning the job opportunities section, **D. Taruscio** was in favour of introducing a site on job opportunities as this promotes exchange between companies involved in research and doctors. With regard to translation into other languages, she appreciates that this is a lot of work but it may encourage patient groups and patient organisations to read the newsletter.

**C. Nourissier:** Pointed out that readership of the Eurordis newsletter increased 3-fold after translation into 6 languages. S. Aymé: A link should be introduced in the Eurordis newsletter towards Orphanews Europe. **H. Sundseth** also commented that patient groups would appreciate news in their own language and that this would increase readership and suggested that her cancer association website should also include a link to Eurordis.

**S. Aymé:** Translation is very difficult and many mistakes are introduced. RDTF has no funding for this effort. She asked the representatives of the commission if there is a possibility for them getting involved in this project.

With regard to the request to have a summary at the top of the newsletter – **S. Aymé** is not convinced that this is feasible or important, in particular as the importance of the topic depends on the reader – many readers only read sections that are of interest to them. However, she agrees that the content of the newsletter should be extracted and classified with a search engine introduced. Job opportunity section will be tested and the feasibility of the diagnostic tools section will be investigated. For the patient organisations, each newsletter reports documents published by PO's but as there are thousands of Pos it is not possible to follow the activities of each organisation. PO's are welcome to send information for publication in the newsletter.

For job opportunities, nothing is centralised, so this could be a very good idea.

**S. Aymé** reminded all members to encourage their colleagues and other administration bodies to sign up for the newsletter.

**The RDTF website:** the website will be redesigned, listing sections on topics covered in the newsletter and containing a section on national incentives and actions. Compared to the newsletter, the RDTF website is not sufficiently developed. The design will be re-thought over the summer and proposals will be transmitted to the RDTF members electronically.

**Global report on health in the EU.** Project to analyse all data collected from member states from projects funded by DG Sanco. A small section of this report will cover the subject of rare diseases, covering aspects such as prevalence and incidence. This report will be prepared this summer by **S. Aymé** and the first draft is expected for November this year. After receiving comments from external readers, the final version is due in Feb 2007.

**Other meetings:** The RDTF has been invited to several meetings. Recently **S. Aymé** went to the Malaga conference on e-Health to present Orphanet as an example of a successful e-

Health tool and discussed activities of the RDTF. Other invitations have been received to discuss the activities of the RDTF.

**OJRD:** **S. Aymé** explained that OJRD is an e-journal with open access (publishing fee paid by scientists) published by BMC. The journal was launched only a few months ago but all of the publications have been accessed over 500 times. Two weeks after the launch of the journal, the papers published were indexed in PubMed. The journal is expected to obtain an impact factor within the next 2 years. The main objectives are to publish regularly, provide a place for publication for difficult subjects such as clinical trials with negative results, and establish rare diseases as a field.

#### **Questions/comments:**

**C. Berens:** Although only 3% of readers hear of the RDTF newsletter through Cordis, this is not a negative result as it amounts to 200 readers. **C. Berens** also made an important point for the next newsletter: she requested that members are given a little more time to read the newsletter before it is published (the previous deadline of 2 days was difficult to reach). A deadline of at least 3 to 4 days was requested. **S. Aymé** apologised for the short deadline and agreed that 3 to 4 days is an acceptable delay.

**New members and participants of the RDTF:** **S. Aymé** noted that there is no EU Commission scientific officer for rare diseases; A. Montserrat does his best but contact with his secretary is limited. As a result, there is no list of participants for this meeting. Query to DG Sanco: would it be easier for the RDTF to manage RDTF meetings?

New participants: a national expert from Norway was present and working as a structural trainee for DG Sanco.

#### **Activities of the Working Groups (WG).**

**Coding and classification:** This Working Group has not yet met but will be formed in collaboration with the WHO with representatives being invited to Working Groups. Yolanda Huizer working on Orphan drugs in the Netherlands also asked to attend and will visit Orphanet in two weeks. Offer was made to other RDTF members to attend. The next meeting will be held in Luxemburg on 11 October 06.

#### **Matters for discussion:**

- MeSH terms for rare diseases
- ICD 10 (the timescale is 6-7 years)
- Classification of rare diseases: collection of all published classifications has already begun at Orphanet with all existing classifications available by the end of the year. The idea is that Orphanet does not choose a given classification but makes available all existing information. However, in the end a 'super' classification must be defined so that all rare diseases can be assigned to a medical field. In addition, this classification will have to interact with existing coding systems.

#### **Comments/Questions**

**H. Dolk:** Different countries have different updates of IC10; this could create problems in trying to construct a European database. The subject of 'what to do with national initiatives on ICD's should be addressed by the WG as some of the coding modifications are contradictory to the original ICD, leading to different uses of the codes.

**Manuel Posada:** Classification is an enormous task with many different goals and objectives. What is the main aim of this WG? Is ICD the best classification to use?

**S. Aymé:** The WG should define the goals and decide on the best method to obtain them. It should also identify the problems.

**H. Dolk:** Should we not include OMIM among the coding systems with some guidance on how and when to use it?

**S. Aymé:** OMIM is not a classification and the link with rare diseases is already made.

**H. Dolk:** Is there a way to find a link between OMIM and classification codes?

**S. Aymé:** This is a very good idea and should be used to inform people about what OMIM is and what it is not.

The name of **Charles Taylor** was proposed as he has made an interesting study of childhood cancer classification. This expert will be invited to attend the WG.

**Working Group on Prevalence:** A survey of all published prevalence data has been started and the first report was published last June. This work will continue with the help of a grant awarded to the Alliance Maladies Rares from the French PSG football club! This survey will continue to collect data from publications, books and websites. A new updated report will be published in 3 months as part of the Orphanet report series with diseases listed both in order of prevalence and as an alphabetical list of diseases with prevalence data.

This Working Group met for the first time in January 06: the main point of discussion was prevalence but other topics included selection of diseases with a strong need for surveillance, indicators relevant to rare diseases and the use of mortality data to estimate life expectancy for major rare diseases in Europe.

**J. Donadieu** reported on the major findings of January 06 Working Group meeting in Jan in Paris. The group should meet again in January 07 but the date has not yet been set. Members were invited to join this Working Group.

#### **Questions for J. Donadieu:**

**M. Posada de la Paz:** Is there any possibility of funding from the Commission for projects recommended by the WGs?

**S. Aymé:** The aim of the WGs is to stimulate projects. These proposals then compete with other projects for funding.

**H. Dolk:** What is on the agenda for the next meeting? Will how to contribute to the survey and will the issue of confidentiality and consent be on the agenda?

**S. Aymé and J. Donadieu:** These meetings are not just about indicators but also cover topics related to the epidemiology of rare diseases. Both the topics mentioned will be discussed in the WG.

**Working Group on Health care:** Centres of Reference (CORs) are not the only topic covered by this WG but at present there hasn't been much time for anything else. Aim was to promote CORs and networks and consider requirements for medical and health care funding. These aims have been achieved. This project was carried out at the request of the High Level Committee which asked the WG to review existing concepts in Europe, discuss added value of centres, map existing centres and provide guidance on how to proceed.

Conclusions of expert group that met twice last year were published. These proposals should be adopted. The major messages were: although the concept of CORs is interesting, it is not understood in the same way by all actors. The term associated with CORs need to be defined, in particular with reference to the main role of these centres. In some countries, CORs are

diagnosis treatment centres, whereas in others they are expert centres associated with advice and clinical research, etc. Issues such as whether to opt for national or regional centres and whether centres should cover research, teaching and care aspects were also discussed. In addition, CORs already exist in some countries but are not recognised as such.

The general opinion of **S. Aymé** is that the Commission is moving too quickly in this area. The member states have to agree on a definition for the concept of a COR before continuing. The aim is now to continue identifying CORs, identify networks nationally or at the European level, make the expert centres visible with information about whether or not they belong to a network, with the goal of covering all European countries.

In some countries, health care is organised at the regional rather than national level; is it appropriate to have national centres in these countries? Should people be pushed to go to specific centres?. The role of the Task Force is to analyse the reality of these centres in the few counties where they are established and to identify the problems. The negative effects of moving too fast need to be considered. A new report should be presented to the High Level Group in September 06 covering the national scenarios via reports provided by each country on the difficulties encountered if CORs were imposed.

The next meeting of the working group will take place in Paris on 1 September.

#### **C. Nourissier reported on the Rapsody project:**

Eurordis is going to organise 10 workshop in 10 different countries to discuss the concept of ECR in the framework of the national health system. A final European workshop will be held in Prague to reach a consensus to be presented at the European Conference on Rare Diseases in Lisbon.

#### **Comments on CR/Questions for C. Nourissier:**

**S. Aymé** disagreed with the comments of the patient groups with regard to the need for flexibility in the definition of a COR. **S. Aymé** reinforced her view that the definition has to be rigid to clarify the role of the centres (centres of knowledge, centres for treatment etc.).

**M. Posada de la Paz** commented that it will be very difficult for CORs to respond to the expectations of the patient groups – how can any one centre respond fully to the call for multidisciplinary care and clinical research?

**J Donadieu** commented that the definition of a COR in France is very refined and may be restrictive, requiring that patients are treated at the COR.

**S. Aymé:** There is a need for both types of centre: centres for clinical research and guidelines and training as well as centres where patients go to obtain advice and care. These two types of centres could be termed differently to avoid confusion: In France the ministry has taken the decision to have expert centres (centres for clinical research with a leading role at the national or international level) and a network of regional centres (termed competent centres, attached to the expert centres) to ensure that patients find their way to these centres through the health system. The CR has to be the leading centre working at the world class level.

**C. Zeidler:** Agrees with **S. Aymé** that a definition is needed and that both types of centres are needed. The definition should be precise, using different wording reflecting the different methodology behind the two types of centre.

**M. Jesperen:** Agrees that the high level committee is jumping to conclusions. We have to identify and map expertise first. This approach may lead to a clear definition for CORs. More attention needs to be paid to differences between countries during this mapping procedure. The criteria should be defined once the mapping is complete and no conclusion should be made until this study is complete.

**A. Montserrat** (in response to the criticism that the Commission was too quick regarding the work plan for the CORs): The problem is with the concept of the COR in that at the moment that the work plan for 2006 was being established, the Rhapsody project was on the reserve list and not endorsed. At the time when new projects were proposed, the status of Rhapsody was undecided and there is a risk that some of the new projects will be a duplication of Rhapsody. The main purpose at the moment is development of the concept, and care should be taken that new projects do not overlap with Rhapsody. Globally, DG Sanco is in favour of CORs, therefore the problem of a flexible or rigid definition is not the major issue. Flexibility is important but there is a political obligation (to the High Level Committee) to reach a consensus definition, taking into account inter-country differences in health systems, funding opportunities for rare diseases, *etc.*

**C. Nourissier** said that Eurordis is very aware of inter-country differences. This is why the annual membership conference was organised this year in Berlin. Rhapsody proposes a bottom-up approach, creating a dialogue within national health systems.

**N. Ruperto** commented that **A. Montserrat's** concerns about the duplication of work may have a positive aspect: it may provide the opportunity for enrichment if the two projects have the same goal achieved by different methods. Even two definitions may be positive as it allows the High Level Committee to choose and avoids presentation of a monopoly view.

**S. Aymé** agreed with **N. Ruperto's** point. For example, the Rhapsody project is driven by the views of the patient organisations rather than professionals. Patients need to express their views, but these views, like those of the professionals, will be biased. The views of all stakeholders need to be considered. **S. Aymé** disagreed with **C. Nourissier** with respect to her comment that Rhapsody covers the views of all stakeholders but **C. Nourissier** pointed out that the workshops will include all stakeholders: patient representatives, health care professionals and public health authorities, including representatives of the Rare Diseases Task Force and of the EU High Level Group.

**A. Montserrat:** When the committee chooses a project to develop a concept, they try to select projects that will obtain input from all stakeholders – this was the impression the committee gained from Rhapsody. However, he did not agree with **N. Ruperto** that arriving at the High Level Committee with two proposals was a plus. One consensus must be reached. Ministers of health should take into account that reports like Rhapsody may not have input from all stakeholders.

**M. Posada de la Paz** commented that the status of the report on the CORs is not clear: Why was the RDTF asked to evaluate CORs if the question is still open? What was the purpose of the final report?

**S. Aymé:** When the report of the RDTF was presented in September it received a positive reception by the French Ministry of Health which was holding the presidency of the Working Group. Once the TF reported that CORs were a good idea this committee pushed for the identification and funding of CORs. A request was then made to fund a pilot project to determine whether this system is working or not. Thus, the High Level Group requested that the committee include CORs in the last call for proposals in order to provide funding for a network of centres and assess whether this improved care for patients. Then the decision can be taken whether or not this process can be more widely applied. Now a new report is expected to fine-tune the concept and define the next steps.

**A. Montserrat** agreed with **S. Aymé** that the aim of the new report is to decide on the future direction of the project. However, the High Level Committee commented that the concept of CORs needs to be discussed more thoroughly. It is the role of Rhapsody to involve a wider range of stakeholders. The High Level Committee should now look towards ending discussions and wait for the new conclusions of the RDTF and the Rhapsody project. However, it is clear that there is some confusion as some responses to the call for proposals

are individual hospitals or research centres applying for the label of COR. The national workshops taking place as part of the Rhapsody project could help resolve this misunderstanding if they are well done. Agreed with **S. Aymé**'s concept of competence and expert centres

**D. Taruscio** explained how the centres are labelled in Italy, with centres defined as centres for care, diagnosis, etc in regional and national networks. Patients are obliged to use these centres if they want free expert treatment for rare diseases.

**A. Montserrat**: This process should be finished at the time of the second conference on rare diseases in Portugal in December 2007. This will allow this process to be finished in a way that has political impact.

**H. Dolk**: Agrees with the comment of **M. Jesperen** that we shouldn't get too involved in definitions and labelling. All patients want access to expertise and expert care. How this operates depends on the size of the nation's population, the rareness of the disease, when expertise is required (e.g. diagnosis or continued treatment) etc. Is Orphanet / Rhapsody going to produce a typology of the different models of reference centre that are currently in operation?

**S. Aymé**: This is exactly what was done (by Orphanet) in the first report in September.

**C. Zeidler** commented that the confusion about the process stems from the fact that the WG made its report and gave guidelines to the High Level Group making it look like the process of identification was already done. A clear statement should be made on the present status of the project. Countries without any COR may consider that 2007 is a long wait when they see that labelling has already begun in other countries. It needs to be made clear that this is an ongoing process and provide a date to indicate when other countries can join the process.

**N. Ruperto**: Why consider new applications for COR proposals of the deadline for a decision in 2007?

**A. Montserrat**: The deadline of Dec 2007 is for the announcement of strategy but does not mean that the work cannot be finished before this date. There are three aspects: 1) a document on the concept of CORs, 2) High Level Group requested a broader discussion in the member states (Rhapsody project) and 3) Testing the feasibility of CORs with pilot trials (projects have only just been received and have not been evaluated but there is some confusion). There will be funding for proposals aimed at setting up national workshops.

### **What are the new priorities for the September workshop/report?**

**S. Aymé**: Production of a better report, with a more mature concept. More annexes should be added, including data from countries with more experience in the field and a deeper analysis of what can be learned from these experiences. Typology of centres and criteria according to the size of the population etc. This will enable the RDTF to provide the High Level Group with a more comprehensive report and the concepts outlined in this document may be used by magistrates for national systems: these concepts must be adopted at national level as health care is funded at national level, without funding these concepts are of no use. This is work to be carried out at the national level. At the EU level, guidance can be provided on the criteria and selection of centres. Even countries that are opposed to CORs are now organising workshops using the RDTF definitions. Overall aim is an improvement of the previous recommendations. **S. Aymé** strongly encourages members to participate.

**C. Nourissier**: Process of national workshops has not yet started. Request that members participate in these workshops and use these events to present the conclusions of the WG at the national level.

**S. Aymé**: Agrees that the document should be used in the national workshops. Also recommended that a specific list of topics should be discussed at each workshop. This list should be the same at each workshop.

**H. Dolk:** The question of accessibility of the CORs is one of the quality criteria that needs to be addressed.

**FUTURE MEETINGS:**

Next RDTF workshop on CORs: 1 September 06 in Paris

Next RDTF workshop on coding and classification : 11 October 06 in Luxembourg

Next RDTF meeting: 14 December 06 in Luxembourg.

Other WG meetings are still to be decided.

Members were reminded to read the minutes of the last meeting and send any comments quickly. If no comments are made in the following week the minutes will be adopted.