

Meeting Report

11th Meeting of the European Commission Rare Disease Task Force Luxembourg, 30 April 2009

On 30 April 2009, the eleventh meeting of the Rare Disease Task Force (RDTF) included the attendance of:

RDTF Members and Observers:

J. Adamski	N. Kerlero de Rosbo	J. Sandor
V. Anastasiadou	O. Kremp-Roussey	R. Stefanov
S. Aymé	Y. Le Cam	F. Suzan
H. Dolk	J. Llenares-Garcia	S. Tanner
L. Fregonese	S. Lynn	D. Taruscio
G. Gatta	M. Macek Jr	J. Torrent Farnell
J. Geissler	C. Nourissier	M. Vihinen
M. Guillou	B. Pizzera Piantanida	J. Vives Corrons
H. Cansen	A. Ramirez Vanegas	S. Webb
A. Kent	C. Rodwell	K. Westermark
B. Wojtyniak	G. Zambruno	C. Zeidler

DG SANCO: A. Montserrat, N. Fahy

Welcome and Discussion of Agenda:

Ségolène Aymé

Leader of the Rare Disease Task Force

Director of Orphanet

S. Aymé opened the meeting by welcoming the new participants:

- Bogdan WOJTNIAK – National Institute of Hygiene, Poland – Department of Medical Statistics
- Yann GEISLER – observer
- Marie GUILLOU – representing L. Dubertret
- Charlotte RODWELL – assistant of the RDTF scientific secretariat

No changes were made to the agenda.

Antoni Montserrat apologised for the last minute change of plans due to emergency H1N1 (Novel Flu) meetings. He highlighted the effect of this pandemic on the rare disease community, especially the effects of the infection (and flu vaccines) on rare disease patients.

A. Montserrat proposed to contact Nick Fahy on behalf of the RDTF to draw the attention of the European Commission and of the Member States about the case of the populations not suitable to receive medication based on Neuraminidase Inhibitors (which includes Tamiflu and Relenza). Therapeutical alternatives for these populations should be part of the national protocols to prevent possible pandemics of Novel Flu.

1. State of play of the Council Recommendation on a European Action in the field of Rare Diseases

Antoni Montserrat

DG Sanco

A. Montserrat told the RDTF members that the latest revision of the text of the Council Recommendation on a European Action in the field of Rare Diseases should have been discussed on 29 April 2009. However, this meeting was cancelled due to emergency meetings concerning Novel Flu. The discussion by the Council of Ministers, for the adoption of this Recommendation, is scheduled for 9 June 2009 in Luxembourg: this date could possibly change, due to the proximity of the elections on 7 June, and the possible evolution of the pandemic. A. Montserrat remarked that the European Commission is pleased with the evolution of the discussion of the text and highlighted three significant points:

1. That a deadline would be established to fix the date at which Member States should have implemented their national plan for Rare Diseases: the document suggests a deadline of 2010 – 2011, but this is more likely to be revised to 2013 considering the evolution of different projects linked to the development of national rare disease plans. The UK and Czech Republic have already started efforts to establish a national plan (as reported in OrphaNews Europe), a meeting will be held in June in Spain to the same effect, and Cyprus equally intends to adopt a national strategy for RD. Estonia is the only Member State opposed to the plan, and other countries such as Slovakia predict difficulties with meeting the deadline. On a side note, it was mentioned that certain Member States opposed the use of the word ‘plan’ for historic reasons, and preferred ‘actions’ or ‘strategies’: the revised text currently reads “national plans, strategies or actions” to allow for preferences in different Member States.

It was proposed that the next Task Force meeting should discuss the development of these proposals for national plans.

2. Reference networks also posed problems on the discussion of the text: the concept of reference networks was not opposed, but rather the relation between the Cross Border Healthcare Directive and Member State’s national plans for rare diseases. A. Montserrat stated that the European Commission has always maintained that the mobility of knowledge is its main priority in the rare disease context. He also stated that the directive creates the concept of European Reference Networks, concretised so as to become physical, which is positive as it empowers patient organisations.

3. The Committee of Regions has endorsed the text without amendment. The report of the European Parliament and the European Social and Economic Committee both raised the same point: the need to allocate common funding for rare disease actions. The Council decided that an implementation report on the recommendation be implemented in 2012 (the next health programme starts in 2013-2014). The advantage of an implementation report in 2012 would be to assess what is necessary to make advances in the field of rare diseases. A. Montserrat concluded that many elements are in place to arrive at the next health programme in a good

position. The Council Recommendation will push Member States to be more active in rare diseases. A. Montserrat congratulated the Czech presidency, and in particular Milan Macek Jr for his dedicated work in coordinating the revision of the Recommendation.

Milan Macek Jr

M. Macek Jr presented an overview of the four revised versions of the text. He highlighted that all concrete attempts to reintroduce an explicit promise for funding were rejected. However, major changes were accepted by the Council. Firstly the working definition of prevalence was kept in the text. In Article 5, the text now states that very rare diseases (less than one case in 100'000) should receive very special attention, unlike Cystic Fibrosis (knowledge less disparate, and services very active). In Article 9, the text now highlights the need for cooperation between research networks (not only national and local incentives). The Council also accepted the need for a better classification of rare diseases in the ICD-11, the necessity to support Orphanet (which is currently available in only 5 languages, and ideally abstracts should be available in all European languages for patients) and the need for more sustained funding for rare diseases in the EU27 Member States. The Council equally supported the role of independent patient groups and the development of recommendations and guidelines to be proposed by the Europlan project. The Recommendation also states that information be freely accessible, financed publicly and updated regularly (mentioning Orphanet). Issues concerning intellectual property rights were raised in relation to the mention of "sharing of best practices and medical/social care in the field of RD". The difficulty of finding a common denominator amongst MS was exposed in relation to sharing assessment methods and benchmarking certain terms in the area of medical and social care. One very positive theme of the text is the empowerment of patients: the recommendation will urge MS to consult patients and patient organisations on policy and to give them free access to expert-validated, updated information.

Questions concerning the text revolved around the definition of 'third countries' in relation to research on RD which M. Macek Jr clarified to mean all non-EU countries, in the perspective of liaison between EU and non-EU research projects. S. Aymé highlighted an inaccuracy in Section 1: the WHO has not appointed the RDTF to be the TAG for the revision of ICD10; S. Aymé has been appointed as chair and the rest of the TAG are not task force members. Also highlighted, in reference to centres of expertise, was the difficulty of including social care in the text. Another subject of discussion was the absence of primary prevention (even environmental primary prevention) in text due to the sensitivity of the issue. Discussion also revolved around the part of the text about training concerning RD of medical professions in different specialities

A. Montserrat stressed that one reason for creating an EU Committee of Experts on Rare Diseases is to have a representative of each Member State to consult rapidly on a given topic (such as the revision of this text): Member States are not currently obliged to be part of the RDTF.

2. Implementation Plan of the Council Recommendation and the Commission Communication

Antoni Montserrat

A. Montserrat briefly presented the draft road map which will be communicated to RDTF members for consultation and revision for the implementation of the Communication and Council Recommendation. The document indicates the timing, type and instrument of each action. It was proposed that the document also be discussed and finalised at the first meeting of the new advisory committee of the EU RD committee.

3. State of play of the Commission Decision setting up a European Union Advisory Committee on Rare Diseases

Antoni Montserrat

The decision to set up an EU Committee of Experts on Rare Diseases is explicitly stated in the Communication. For legal reasons, this Committee (which will replace the RDTF) will be called the European Committee of Experts on Rare Diseases instead of Advisory Committee and will have 54 members: 1 representative from each member state, 4 representatives of patient organisations (chosen by a panel composed of representatives of the DG Sanco, DG Enterprise, DG Research), 4 representatives of the pharmaceutical industry, 6 representatives of past RD projects, 2 representatives from the COMP, 1 representative of the EMEA and 1 representative from ECDC. Each representative will have a 3 year mandate and also a deputy. The secretariat will be provided by the commission. The Committee will be able to make recommendations to MS (i.e. recommendations concerning national RD plans), the EU Parliament, EMEA, COMP and the UN.

Questions on this subject revolved around the duration of the presidency mandate and the structure of governance (to be defined at the 1st meeting of the new committee): it was also confirmed that the first meeting would only take place when all members have been selected. A. Montserrat also confirmed that the representatives of each MS would be freely selected by each MS: ideally this should be a person with political influence in the MS in the field of RD. It was also confirmed that meetings would take place in Luxembourg, but working group meetings (as for the Joint Action) could very well take place in other cities. The first meeting of the new Committee could take place in October 2009 if all representatives are duly selected, if not the next meeting of the RDTF will be maintained.

4. Outcomes of the European Rare Diseases Day in 2009

Yann Le Cam

Eurordis

Y. Le Cam presented a report on the 2nd Rare Disease Day, initiated and coordinated by Eurordis, which focussed on patient centred care (“Patient Care, a Public Affair!”). This year 19 national alliances helped organise the day at local level and 600 patient groups participated in Europe: in total 30 countries were involved in RDD. The target audiences of the day were policy makers (health authorities, national parliamentarians, European parliamentarians and candidates) as well as the general public, media, health professionals, academics and researchers. Y. Le Cam briefly presented the RDD website and the use of platforms such as YouTube and networks such as Facebook to promote RDD. In total, 21.000 visits were made to www.rarediseaseday.com this year. Media coverage of the day was strong with over 1500

media articles in total. The 2nd Rare Disease Day also spread beyond Europe with the participation of the US (organised by NORD – the National Organisation for Rare Disorders), Canada, Argentina, Colombia, Australia, Taiwan and China.

The event received the patronage of EU Commissioner for Health Androulla Vassiliou, who attended the Rare Disease Day book launch of *The Voice of 12.000 Patients* at the European Commission in Brussels. A dinner debate, hosted by MEP Antonios Trakatellis (Rapporteur of the Parliament's Opinion on the Council Recommendation on Rare Diseases), was held at the European Parliament and was attended by policy-makers at the European Commission, patient advocates, parliamentarians and the representatives of the biopharmaceutical industry. Rare Disease Day 2009 also saw lobbying action at national level (Spain, UK, US, China), and also provided the momentum in the push for national plans and strategies (Belgium, Spain, Portugal, Bulgaria, Ireland, Czech Republic), as well as creating momentum in the push for Centre of Expertise (i.e. visit of Centre for Rare Disorders by MEP in Denmark), and in the construction of emerging national alliances (Switzerland, Australia). Rare Disease Day also played an important role in raising awareness amongst the general public (this was helped through VIP and celebrity patronage this year) to inform, educate and involve the public in the issues surrounding rare diseases.

Y. Le Cam suggested that the new European Committee of Experts on Rare Diseases to recommend that Rare Disease Day becomes an international event. It was also highlighted that Rare Disease Day provides a focus for fund raising – this year's Telefon in Catalonia was dedicated to rare diseases.

Y. Le Cam also distributed brochures on the Polka project and gave the dates of the 5th European Conference on Rare Diseases to be held in Krakow, 13-15 May 2010.

5. State of play of the Joint Action on support to the Rare Disease Task Force and perspectives for the future

Ségolène Aymé

S. Aymé presented an overview of the objectives of this action: 1) to survey initiatives and incentives at MS and EU level, and to liaise between stakeholders through an electronic newsletter; 2) to define indicators to monitor the public health impact of RD and the effect of policies; and 3) to improve the codification and classification of RD.

The methodology for objective 1) was outlined and improvements concerning access information (i.e. through RSS flux and the publication of OrphaNews content on the Orphanet website) were proposed. The first workshop to discuss the content and the format of the report on initiatives and incentives will be held in Paris on 22 October 2009.

Objective 2) is a continuation of work already carried out by the RDTF expert group on indicators: in Year 1 of the project, the list of tentative indicators will be examined with a first workshop in Leiden, 10 November 2009. In Years 2 & 3 data will be collected to populate selected indicators and to publish reports.

Objective 3), concerning the improvement of codification and classification of RD, has two sub-objectives: a) to submit a proposal to WHO for ICD11 to make RD visible and codable in the next edition (2014), and b) to cross reference coding systems (Orpha nomenclature, ICD10, MeSH, SnoMed-CT, MedDRA).

S. Aymé presented an overview of the ICD-10 revision process and the work of the TAG for Rare Diseases which she chairs. The preparatory work carried out at Orphanet was then presented in detail: from collecting published classifications to building a clinical one, to analysing ICD-10 and identifying problems linked to the existing classification, to the final step of contributing to ICD-10+ and ICD-11 by proposing a new classification.

The methodology proposed to carry out the cross-referencing of the second sub-objective was described: a partner kick-off meeting of the coding and classification workgroup was held in Paris on 11 March 2009 during which several intellectual property rights issues were identified. The next meeting of the coding and classification working group will be held in Manchester, 27 January 2010.

RDTF members will be invited to participate in the revision of ICD10 as experts in their specialty, and also as experts for the planned workshops on 'initiatives and incentives', on 'indicators' and on 'coding'. The previous RDTF working groups have been discontinued. S. Aymé highlighted that overlap should be avoided with Europlan working groups.

Questions concerning the Joint Action clarified aspects of the ICD revision process at Orphanet and WHO, and the involvement of experts who are invited to read the revised chapters concerning their specialities. The classification of complex, multi-system rare diseases was discussed: S. Aymé explained that at the last meeting of the ICD revision steering committee in Geneva a proposal for a multi-system chapter was accepted (but not endorsed).

6. Call for Tenders and direct agreements in the framework of the Second Health Programme in 2009

Antoni Montserrat

A Call for Tender was announced for Newborn screening (June 2009): three deliverables will be demanded: to describe existing practices; to provide an expert opinion on the feasibility of an EU recommendation on newborn screening; and to create a network of experts in the field of neo-natal screening. A consortium should be created for the call, and a preannouncement will be made at the International Conference of New Born Screening.

Another Call for Tender was intended to support repertory activities (and in particular Orphanet): this Call will not be launched in 2009 but in all probability in 2010 as a Joint Action. The reason behind this decision was explained. By 2010 the Council Recommendation (which explicitly mentions Orphanet) will be in force and will oblige MS to actively engage in Orphanet's activities.

Finally a direct agreement was announced with WHO for the classification and codification of rare diseases: this agreement would address the problem of there being no existing funding for meetings to discuss chapters. It was also announced that the next session of the European Committee of Health Information in 7-8 July 2009 will be devoted to the revision of the ICD.

7. State of play of the Europlan project – European Project for Rare Diseases National Plans Development

Domenica Taruscio

Europlan

D. Taruscio presented a report on the Europlan project which began in April 2008: the main goal of the project is to elaborate recommendations on how to define a national plan/strategy/action for rare diseases. Focusing on the previous experience of MS, Europlan plans to share information, models and data concerning effective RD strategies. 26 of the 27 EU MS are partners of the project; the proposed "Council recommendation on a European action in the field of rare diseases" invites MS to define plans/strategies/actions around ideas

set out in the Communication. The recommendations developed by Europlan will provide practical guidance on how this can be done on the basis of existing best practices: an efficient and effective action for RD will depend on a coherent overall strategy based on a common approach and integrated into a common European effort.

Europlan has several specific objectives: to collect information on EU MS initiatives on RD; to identify successful experiences and lessons learned; to elaborate indicators for monitoring the implementation and evaluating the impact of national plans for RD; to elaborate recommendations; to discuss recommendations with local stakeholders; and to disseminate Europlan recommendations. The methodology behind the collection of information on MS RD initiatives was exposed, as were the areas to be investigated: national/regional RD strategies; definition, codification and inventorying of RD; research on RD; centres of expertise; empowerment of patients organisations; sustainability of RD activities (funds dedicated to RD); orphan drugs development and reimbursement; early diagnosis; specialised social services. The limitations of this methodology were exposed as well as predictions of findings. To compensate for these limitations, a survey will be carried out to collect additional information. Another planned step is to develop indicators to monitor implantation and evaluate the impact of national plans for RD (the first draft of these indicators is under revision by Europlan partners (due by 28/04/09), with a workshop planned in Madrid, 18-19 June 2009 to which RDTF WG indicators members will be invited. A first draft of elaborated recommendations is equally under revision by partners, with a workshop planned at The Hague, 17 September 2009. These recommendations will be discussed with local stakeholders through a Call for Conference (managed by Eurordis and launched on 19 December 2008) addressed to National Alliances. The final selection of proposals was carried out in April 2009.

In addition to this, on 18 November 2008 the document “National Strategies and Plans for Rare Diseases in Europe – State of the art and sharing experiences: toward EU Recommendations” was disseminated under the auspices of Europlan, the French Presidency of the EU, Eurordis and the European Commission.

Discussion revolved around the possible cross-over of the Europlan project and the Joint Action concerning initiatives and indicators. S Aymé drew attention to the fact that the focus of Europlan is to monitor the progress of EU recommendations and so indicators should be devised to monitor political progress and not everything generally concerning RD: this point was contested as all these indicators are linked to the reflection on the recommendation. Other questions concerned the methodology applied to the collection of information on initiatives, carried out in English, with a survey sent to all MS who will collect information in their country in their language. The surveys will be sent to partners of the project, national alliances and members of the RDTF.

8. Next meeting

The next meeting of the Rare Disease Task Force will take place on 23 October 2009 if the new European Committee of Experts on Rare Diseases has not at that moment been put in place.