

Meeting Report

7th Meeting of the European Commission “Rare Disease Task Force”

Luxembourg, 20 June 2007

On 20 June 2007, the seventh meeting of the Rare Diseases Task Force (RDTF) included the attendance of:

Rare Disease Task Force:

S. Aymé	A. Kole	A. Trama
E. Daina	J. Llinares Garcia	J. Vasquez
H. Dolk	C. Nourissier	J.L.Vives Corrons
J. Donadieu	M. Posada de le Paz	Y. Wagener
L. Fregonese	A. Ramirez Vanegas	S. Webb
G. Gatta	J. Sándor	
HK Hartle	R. Stefanov	

Observers:

R. Capocaccia	O. Kremp
A. Federico	Y. Le Cam
G. Filocamo	A. Phinikaridou
A. Fourcade	B. Piantanida Pizzera
S. Giampaoli	A. Ramirez Vanegas
	M. Rätsep

European Commission:

K. Freese
A. Montserrat

A. Welcome and Approval of Agenda

No changes.

B. Presentation on the Commission Communication on a European Action in the Area of Rare Diseases (Including Genetic Diseases)

Antoni Montserrat
European Commission
DG SANCO Health Information Unit

The concept of “rare disease” (RD) emerged in 1978 with the publication of an article (Holzman NA. Rare diseases, common problems: recognition and management. Pediatrics, 1978; 62(6): 1056-1060) stating that rare diseases, though diverse, have common problems of being recognised by physicians and problems of being effectively managed as knowledge about each is very limited and little clinical research in the field exists. In the early 1980’s, rare diseases gained recognition as a political issue with the fight of patient advocacy groups to obtain a set of incentives for the development of therapeutic products in the USA. In the early 1990’s the debate cantered around research challenges 1990s when rare diseases, which are mostly genetic, became instrumental for mapping human genes. More recently, the public health dimension of rare diseases has been recognised by the European Commission and by several individual countries and consequently specific action plans were developed.

A Community action programme on RD, including genetic diseases, was adopted for the period of 1 January 1999 to 31 December 2003 with the aim of ensuring a high level of health

protection in relation to RD. As the first EU effort in this area, specific attention was given to improving knowledge and facilitating access to information about these diseases.

Rare diseases are now one of the priorities in the EU Public Health Programme 2003-2008. According to the DG SANCO Work Plans for the implementation of the Public Health Programme, the two main lines of action are the exchange of information via existing European information networks on rare diseases, and the development of strategies and mechanisms for information exchange and co-ordination at EU level to encourage continuity of work and trans-national co-operation.

For the period 2008-2013 a new Public Health Programme will replace the existing Public Health Programme (if approved by the Council and the Parliament). It will include a new strand 'Generation of knowledge' and no 'Diseases' strand.

In 2007, the Commission plans to adopt a new Health Strategy, which includes the need of a Commission Communication in the field of RD. During 2007 the Impact Assessment and Strategy White Paper will be developed and adoption of this strategy is expected mid to late 2007 at which point the European Council will discuss the strategy.

During 2007/2008, DG SANCO will elaborate a proposal for a Commission Communication on the European Health Information and Knowledge System:

- Summarising the principles basing the EU health information and knowledge system,
- The responsibilities of the different actors in this field, the role of DG SANCO,
- The national and EU responsibilities on the mechanisms for collecting data (health surveys, hospital information, etc.),
- The interoperability of different systems of health indicators, the cooperation with other actors (Eurostat, ECDC, OCDE),
- The role of the consultative structures
- A code of good practices on health information
- Obligations that the European Commission should assume respect to the Member States in the field of health information.

The proposal should be discussed in all the existing SANCO advisory structures. A Consultation regarding Community action on health services is in place. Before the Commission brings forward proposals for Community action on health services, all stakeholders involved in the health services sector are consulted, on the basis of a specific consultation document.

There is probably no other area in public health in which 27 national approaches could be considered to be as inefficient and ineffective as with rare diseases. The reduced number of patients for these diseases and the need to mobilise resources could be only efficient if done in a coordinated European way.

This initiative is one proposal in the Annual Management Plan 2007. Article 152 provides for the adoption by qualified majority by the Council of Recommendations, on the basis of Commission proposals, for the purposes set out in that article. These Recommendations are the only legislative tool provided for in Article 152 on public health except for the few areas where measures or incentive measures may be adopted (see Article 152.4). Recommendations

are without legal force but are negotiated and voted on according to the appropriate procedure. Recommendations differ from regulations, directives and decisions, in that they are not binding for MS. Though without legal force, they do have a political weight. The Recommendation is an instrument of indirect action aiming at preparation of legislation in Member States, differing from the Directive only by the absence of obligatory power. In order to coordinate the position of the Member States in respect of this important field, it is considered that a Recommendation is the appropriate legal instrument. In the case of the proposed Communication on rare diseases, it is considered necessary to accompany that Communication with a Proposal for a Council Recommendation on rare diseases covering the areas of:

- Common definition of rare diseases in the EU.
- Necessity of national plans for rare diseases in the EU Member States
- European guidelines for the elaboration of the national plans for rare diseases
- Common databases and medical protocol for the identification of genetic rare diseases
- Common approach for a better codification and classification of rare diseases in the process of revision of the International Classification of Diseases
- Creation of the EU Forum on Rare Diseases
- The European Conference on Rare Diseases organised by the EU Forum on Rare Diseases with the specific budget be fixed in the EU Budget during the coming years for the activities of the EU Forum.
- Common approach to the support of patient's organisations
- Creation of the EU Rare Diseases Portal as a part of the EU Health Portal and as common tool for rare diseases identification
- Using e-health facilities for information and treatment
- Better integration of the EU rare diseases public health action with other rare diseases policies (research, orphan drugs, advanced therapies, etc)

- The participation in the COMP of all the EC DG's involved in the field of rare diseases as well as the most relevant NGO's
- A procedure for the creation and recognition of EU networks of reference for rare diseases. The EC will prioritise cooperation in sharing knowledge as the most efficient approach.
- EU identification and certification of laboratories worldwide that perform gene tests for rare genetic diseases, the methodology employed, and whether the tests they provide are in the investigational stage, or are being used for clinical diagnosis and decision making.
- Networking Bio Banks in the EU
- Data protection
- Training of rare diseases researchers and professionals
- Intensifying Therapeutic Research, toward a Public–Private Partnership
- A systematic report on the situation of rare diseases in the EU: The Commission should produce every three years a report on the situation of rare diseases in the EU. An Atlas of the epidemiology on rare diseases should be also produced on a five-year basis.
- A monitoring for the future: On the basis of the work of DG SANCO and the advice from the European Forum on Rare Diseases, the creation of a European Office on Rare Diseases could be considered as an appropriate way of action in the framework of the future EU Programme of Public Health (2014-2020).

A. Montserrat stated that the CR WG of the High Level Group, Eurordis, EMEA-COMP, and Orphanet are the preferable bodies consulted for the Communication. It is important to

remember that after the finalisation of the Communication it is not known how it will be received by the Council.

Next Meeting of the Drafting Group

The next meeting of the Drafting Group will take place in Luxembourg on 18 July and will include S. Aymé, D. Taruscio, L. Fregonese, J. Llinares Garcia, A. Montserrat, M. Posada de le Paz, R. Stefanov, and C. Nourissier.

C. Commission Communication for a Community Action in the Area of Health Services and Activities Related to Rare Disease Reference Networks

Alexandra Fourcade

French Ministry for Health

Working Group on Reference Networks of the High Level Group on Health Services

The High Level Group (HLG) established in 2004 in response to the decision to address the issue of RD patient mobility. This HLG is made up of many Working Groups (WG), one of which is the WG on European Networks of Centres of Reference (ENCR). This WG aims to encourage a close link between research and health professionals and patients for very RD in which healthcare professionals are rare as well. This WG seeks input from the RDTF on the definition of networks, from the HOPE project for a patient perspective, and from EURORDIS to help with the consensus of how to define a Centre of Reference at the European level. Eventually, it was agreed by the WG members that ENCR should comply with the following criteria:

- appropriate capacity to diagnose, to follow-up and manage patients with evidence of good outcomes when applicable
- sufficient capacity to provide expert advice, diagnosis or confirmation of diagnosis, to produce and adhere to good practice guidelines and to implement outcome measures and quality control
- demonstration of a multi-disciplinary approach
- high level of expertise and experience documented through publications, grants or honorary positions, teaching and training activities
- strong contribution to research
- involvement in epidemiological surveillance, such as registries
- close links and collaboration with other expert centres at the national and international levels and a capacity to network
- close links and collaboration with patients associations where they exist.
- Although an ENCR should fulfil most of the above criteria, the comparative relevance of these various criteria will depend on the particular disease or group of diseases covered.

This definition is rather vague. As such, in 2004 a questionnaire was sent to many centres of expertise to assess the reality of such centres. The results indicated that the centres were very diverse in their characteristics. The first report from the RDTF assessing CR presented the status of centres at the country level. The second report included recommendations to the HLG.

The WG on ENCR began its discussions around the criteria required to label a centre a CR. As the work of the WG progressed the discussion moved towards that of networks of CR. It was agreed by the WG that

- Hierarchy between national or regional networks of centres of reference should be avoided.
- Networking of expert centres should be favoured, rather than isolated CR.
- In principle, expertise should travel rather than patients themselves. However, it should be possible for patients to travel to centres when necessary.

Heavy emphasis is placed on the links of a CR with patient groups. After the establishment of such suggested criteria, 5 pilot projects were launched to test the criteria at the EU level. It has been clear from the activities of these 5 CR that there is much diversity among the way these criteria are interpreted. Another concern included the sustainability of these centres; what happens after their funding runs out?

The WG on ENCR of the HLG include is eager to be consulted on this topic for the Commission Communication. The outcome of the European Workshop on Centres of Reference for Rare Diseases in Prague sponsored by EURORDIS will also serve as very valuable feedback for the WG.

Discussion

One RDTF member was particularly impressed by the emphasis placed on collaboration between public health agencies, research professionals, and patients. Previously, research was very much on one “track” while the public health and patient agendas were on another.

It was agreed that the outcomes of the Prague workshop will be important to assess appropriation and evaluation of the WG. When asked how flexible the WG was on its CR criteria as it is already clear from the national workshops (preparatory workshops for Prague) that the outcomes of the Prague workshop will require the CR criteria to be refined, A. Fourcade responded that indeed the WG must be very flexible in its CR criteria as even with only 5 pilot projects there is great diversity. It is certain that certain criteria will remain, such as epidemiological surveillance, research development, access to diagnosis and links with patient groups. The details of these criteria, however, must certainly be flexible and ideally not too exhaustive. Most importantly the criteria that are agreed upon must be agreed upon at the EU level.

It was suggested that when presenting the idea of ENCR one must be clear in specifying that funding will not come from a common “pot” but will remain up to CR at the MS level.

A. Fourcade continued by explaining that the very idea of pilot projects is to highlight unexpected findings. As such, it will be important for the WG on ENCR to follow up on them in a few years to see what kind of concrete impact they have had (ex. has diagnosis improved as a result?).

A. Montserrat added that indeed the experience of all five projects has been very different. For each project, the idea of ENCR was interpreted in different ways. They differed, for example, in the transportation of patients or in the use of e-health.

One member disagreed with the approach and suggested that the five pilot projects should be seen more as case studies. As the CR were built on very vague criteria it is no surprise that

they function so differently depending on the disease area. It would be better to look at each project's achievements and then apply the lessons to an *a priori* set of criteria.

It was also expressed that there has been much confusion during this and previous discussions between CR and ENCR. One must be very careful that when assessing ENCR to limit the discussion to the added value of such linkages. Only when assessing individual CR can the previously described criteria be applied. It is imperative to separate the two.

Finally, RDTF members were reminded that underlying this work is the goal of improving access for patients though it is not explicitly stated anywhere. It seems necessary to demonstrate that patients, the end-users, are benefiting from these networks.

D. Progress Report on the Rhapsody Project

Yann Le Cam
EURORDIS

Y. Le Cam presented progress on the Rare Disease Patient Solidarity project funded by DG SANCO including patient surveys providing information on patient satisfaction with the provision of healthcare for rare diseases and national workshops organised to initiate the debate on centres of reference for rare diseases at the national level.

Surveys were sent across Europe to patient organisations covering 16 rare diseases representing a diversity of prevalence, age of onset, clinical manifestations, handicap generated, and availability of treatments. Surveys have been completed in France, Hungary, and Spain and are in progress in the remaining 23 countries. These surveys have proved to be a complicated process as many questions must be catered to each disease and the surveys have been translated into 16 languages.

The number of surveys sent to each patient organisation varies depending on the prevalence of the disease but also the number of members of an organisation. A preliminary analysis of the responses (focus on questions common to all surveys) is underway.

On 11 June 2007, the Advisory Group met to discuss the results of the 11 completed national workshops (Luxembourg has not yet turned in its report). The synthesis of the 10 completed national reports will be structured around the three following Questions:

- Q1: Needs and expectations for national Centres of Reference/Expertise
- Q2: Proposals for the evaluation of national Centres of Reference in the country
- Q3: Cooperation with other countries and recommendations for European Reference Networks

The European Workshop in Prague will indeed give good insight on the expectations of the patients. It is important to emphasize that these are opinions of patients in each Member State and not the official expectations of the MS. It is already clear from national reports that the added value of European Reference Networks is real and impressive. The national workshop participants include patients as well as other stakeholders that are consulted.

Y. Le Cam continued by presenting the Draft Agenda of the European Workshop in Prague.

European Workshop in Prague – Day 1

	First day: starts at 11.00 am
	Introduction 11.00 to 12.00 am <i>Co-chairs: Birthe Holm (Rare Disorders Denmark), Martin Benes (SUKL director)</i>
5 min	Welcome speech
5 min	<i>Czech patient representative</i>
	Workshop objectives
20 min	<i>François Houyez, EURORDIS, EU</i>
20 min	Presentation of the proposals of the High Level Group on Health Services and Medical Care
	<i>Aude Marlier Sutter, Ministry of Health, France</i>
10 min	Presentation of the DG Sanco Rare Disease Task Force report
	<i>Dr Ségolène Aymé, DG SanCo Rare Disease Task Force, EU</i>
	Discussion
10 min	Presentation of selected European networks 12.00 – 1.00 pm <i>Co-chairs: Birthe Holm (Rare Disorders Denmark) Martin Benes (SUKL director)</i>
10 min	- Establishing the six first European Reference Networks: practical experience
10 min	<i>Prol Lara Fragonese, Rare Bleeding Disorders Network, Netherlands</i>
30 min	- Cystic fibrosis, a European Reference Network supported by DG SanCo
	<i>Prof Thomas Wagner, ECORN-CF, Germany</i>
	- EuroAtaxia, a European Network of Centre of Expertise supported by DG. Research
	<i>Prof Olaf Riess, EuroAtaxia, Germany</i>
	Discussion
90 min	Lunch 1.00 pm to 2.30 pm
	EurordisCare Survey results, 2.30 – 4.30 pm <i>Co-chairs: Terkel Andersen +/- Panos Kanavos LSE</i>
	Survey on patient experience and expectations in access to health services (EurordisCare3)
	<i>Pierre Chauvin, INSERM, Unit Public Health</i>
	Panel with representatives of the selected diseases

European Workshop in Prague - Day 2

	Second day
	Synthesis of national workshops 9.30 am – 1.00 pm <i>Chair person: Terkel Andersen (EURORDIS), Maryna Krenkova (SUKL, Czech Republic)</i>
10 min	Overall presentation of the national workshops
10 min	
30 min	Synthesis of responses to question 1 by <i>Christel Nourissier, EURORDIS, EU</i>
10 min	Discussion
30 min	Responses to question 2 by <i>Simona Bellagambi, UNIAMO, Italy</i>
11.00 am	Discussion

10 min	Coffee break 11.00 am to 11.20 am
30 min	Responses to question 3 by <i>Birthe Holm, Rare Disorders Denmark</i>
12.00 am	Discussion
	Proposed priority of criteria and their content for European Reference Networks
30 min	<i>Yann Le Cam, EURORDIS, EU</i>
	Discussion
	Lunch 1.00 pm to 2.30 pm
	Methodology to assess the outcomes of European Reference Networks, 2.30 to 4.00 pm
20 min	<i>Chair person: Prof Birgitta Strandvik, Sweden and Prof Olaf Riess, Germany</i>
	Discussion based on DG SanCo RDTF meeting June 2007
20 min	<i>Dr Edmund Jessop, Ministry of Health, UK</i>
	Description of the evaluation plan as proposed by the French policy on centres of expertise
30 min	<i>HAS representative (France)</i>
	Discussion
15 min	Closing remarks 3.45 to 4.00 pm
	The way forward from the Commission's perspective
	<i>Toni Montserrat, DG SanCo, EU (tbc)</i>
	Ends at 4.00 pm

A report of the outcomes of the European Workshop will be distributed to all national and European Workshop participants along with a CD including all presentations and national workshop reports.

E. Presentation on RDTF WG on Coding and Classification of RD

Ségolène Aymé
Orphanet
INSERM-SC11

On 2 May 2007 a meeting of the WG on Coding and Classification occurred in Paris just after WHO's launch the ICD revision.

Orphanet has recently added a lot of relevant information to its database. Currently, MESH terms, MIM codes, proteins (via SwissProt collaboration), and genes are linked to 2,000 diseases in the database. The prevalence, age of onset and other text is also associated with almost all of the diseases in the database.

The goal of the WG on Coding and Classification is to make RD traceable in terms of morbidity and mortality by defining one group of the "main" rare diseases which must have a specific ICD code and organising the remaining "ultra-rare" diseases under "other RD" subcategories of the more general codes.

The steps in this process include:

1. making a list of the diseases deserving a specific code
2. Analysing the current ICD coding system and identifying problems by cross-referencing many coding lists (with the cooperation of other coding bodies)

3. Contributing to ICD-10plus, a tool provided by the WHO to communicate suggestions to the current ICD version

These steps will require a significant amount of technical work and thus we will apply for support from DG SANCO.

The revision process will be led by a Revision Steering Committee under which several Topical Advisory Groups (TAG) will represent a particular field. The rare disease TAG represents Europe and several other countries have been approached for a truly international contribution. Steve Groft of the Office of Rare Diseases at the National Institute of Health in and Roberta Pagon of GeneTests have joined the effort from the US. Colleagues from South America are currently being approached.

Currently there are 384 specific codes for RD so a lot of work remains. Simple coding mistakes in the ICD-10 will be easy to correct, however, many codes will be the source of difficult debate and will require face to face discussions. As such, the next WG on Coding and Classification workshop is scheduled 13 November in Luxembourg.

S. Aymé continued by sharing Robert Jakob's (of the WHO) presentation on the organisation of this revision process. The next meeting of the TAG will be in Trieste this year. The next meeting of the WG on Coding and Classification will be in Washington DC.

A. Montserrat stated that this activity of the RDTF is very important. In 2008 DG SANCO can help increase the number of meetings for the activity. As for further funding for the work DG SANCO is investigating the possibilities. DG SANCO has approached the WHO to establish an agreement for funding but the WHO has not responded with any interest yet. A. Montserrat clarified that funding must be sent to an identifiable body (i.e. institute, laboratory, etc.). The identification of the body responsible for revision of RD alone is not obvious. The use of the EMEA database of experts in this endeavour was fully supported.

Though the ICD-11 revision will truly be an ambitious process reviewing RD codes "from scratch", the TAG is aware that drastic changes are not seen as favourable and the role of the Revision Steering Committee is to make sure that changes are only dramatic when necessary.

It was suggested that it may be important to include an explanation of why such a revision process is necessary and helpful in the Communication.

DG SANCO agreed to participate in the revision process by sending a representative to workshop meetings. J. Vasquez stated he would also like to contribute to the process.

F. Presentation on Other Activities of the Rare Disease Task Force

Ségolène Aymé
Orphanet
INSERM-SC11

Communication

RDTF Secretariat has continued the publication of the RDTF electronic newsletter, OrphaNews Europe. Currently distributed to more than 8,500 registered readers from 32

countries, subscribers are free to opt in or out of the service at any time. Since its creation in June 2005, 16 issues have been completed. In May 2006, a satisfaction survey reflected a high level of readership satisfaction and suggestions to expand topics covered in OrphanNews Europe to include more relevant issues on the political level and in research findings. To decrease the quantity of information in each newsletter, it has been decided to publish a newsletter every two weeks. All stakeholders including RDTF members are encouraged to send their contributions to orphanews@orpha.net.

S. Aymé added that she and several other RDTF members presented at the first Canadian Conference for Rare Disorders and Orphan Products Policy. She is also participating in the 3rd International Conference on Rare Diseases and Orphan Drugs (ICORD). The conference is organised by the NIH in the US and the Karolinska Institute in Sweden. S. Aymé also visited the Minister of Health in Hungary who said she will consider proposals in RD very favourably.

It was suggested that the RDTF Secretariat prepare a basic RDTF presentation for members to be able to use in presenting the RDTF activities to others.

Workshop on Orphan Drugs

On 30 May 2007, in Paris, a workshop was organised for a subgroup of the WG on Standards of Care on the subject of the future of orphan drugs. This topic was selected because the discussion of the proponents of Orphan Drug Acts as being victims of their own success is becoming more and more common. Many fear that innovation will lead to an orphan drug (OD) for each rare disease and no healthcare system will be able to support it. At the moment, no one has data to show otherwise. The workshop in Paris facilitated a discussion on identifying the number of treatable RD and forecast the number of OD expected in the next ten years using the US experience and a model constructed by François Faurisson of EURORDIS. Using this information it looks as the ~200 new OD will appear on the market in the next 10 years.

The communication of this message will be difficult. It is a sensitive subject as on the one hand patients may be very discouraged if a number of OD is defined and does not include treatment for their disease. Several participants were wary of providing any exact figures during the communication of this discussion.

It should be kept in mind that off-label drugs are used very often and truly help the quality of life of patients. It is clear that industry is not ready to coordinate clinical trials to show this but it could be helpful to mention in communicating the forecast of OD without discouraging patients. This information is available on Orphanet. It is known that ~500 RD are treated by drugs with other indications. Not only the ~200 OD but also the number of drugs in clinical trial could be reported in this forecast.

There continues to be a great need for better collaboration between industry and Public Health professionals as there is always a critical phase of drug development after the basic research phase and before clinical trials. The success rate of development is approximately 15%.

It was agreed during the Paris workshop that one must always present this dilemma in terms of solidarity and equity (as opposed to cost effectiveness).

S. Aymé stated that she felt it was a very productive meeting and that she hopes the RDTF will be able to have more such meetings, perhaps in conjunction with EMEA.

Workshop on the Future Needs of the Rare Disease Community

S. Aymé announced her participation in this expert workshop along with several other RDTF members. The Workshop included presentations of RD activities supported by DG Research and DG SANCO, and a discussion of research needs of the rare disease community. The next step in the discussion of RD research needs is the 'Rare Diseases Research: Building on Success' conference on 13 September 2007 in Brussels. Members are encouraged to get involved in this conference which provides the rare disease community with the opportunity to express their needs in terms of research. The conclusions from this conference will be presented at the European Conference on Rare Diseases in Lisbon in November 2007 ([ECRD Lisbon 2007](#)).

G. Presentation on the European Conference on Rare Diseases (Lisbon, 27-28 November 2007)

Yann Le Cam
EURORDIS

Y. Le Cam presented the European Conference on Rare Diseases sponsored by EURORDIS in Lisbon. Registration and all relevant information is available on the website www.rare-disease.eu which will progressively be updated. The conference is organised by a program committee of 14 members with two chairs, T. Andersen, President of EURORDIS and Professor Josep Torrent i Farnell, former chairman of the COMP. The local organising committee is the Directorate General of Health in Portugal and 10 patient organisations. It is very important that this conference takes place in conjunction with the new presidency as it will place RD on the national agenda.

Several pre-meeting workshops will take place the day before the official start of the conference including:

- Workshop on help-lines for rare diseases
- Thalassaemia International Federation and PBSA annual meeting
- Orphanet network meeting
- Council of National Alliances meeting
- Drug Information, Transparency and Access Task Force meeting

The official meeting will include 11 sessions spanning two days. Participants will be able to choose among several coinciding sessions on day 2. A fee reduction is available for those who register before the end of July and there is room for only 400 participants so early registration is encouraged. The deadline for submission of abstracts has also been extended to the end of July. Organisations that would like to pass out fliers during the conference should contact the Secretariat of the conference.

H. RDTF Future Initiatives

Ségolène Aymé

European Networks of Centres of Reference

The third technical and scientific report on ENCR will be published as promised. This report will include a discussion on the added value of ENCR and more specifically on the methods used to assess this added value. A workshop, in collaboration with Eurordis and the High Level Group on Health Services and Medical Care, will be organised to produce this report. If desired by RDTF members, this can be the topic of the second semester RDTF meeting.

Emergency Guidelines/Emergency Cards

An initiative to provide patients and health professionals with concise information on emergency guidelines for rare diseases has begun in France. A pilot study, producing such guidelines for several rare diseases is already underway, thanks to the funding of the French National Plan of Rare Diseases. This type of effort could be valuable at the EU level. The question remains at what point guidelines should be managed at the national level due to the diversity of traditions in disease management and existing laws.

Essential Tests

Based on the concept of “Essential Medicines” designated by the WHO, the rare disease community could profit from a list of “essential diagnostic tests”. It was mentioned that the Council of Europe has a WG on this topic that could be consulted.

National Plans on RD

National Plans on RD were suggested as a future topic for the RDTF including the creation of best practice guidelines that can be implemented at the MS level. As such, there would be some guidance or steering of the evolution of National Plans on Rare Diseases.

All additional suggestion for future RDTF initiatives are welcome and should be emailed to the RDTF Secretariat.

Next Meetings

The drafting group for the Communication will meet July 18th. The following members voiced their interest in participating: S. Aymé, D. Taruscio, L. Fregonese, J. Llinares Garcia, A. Montserrat, M. Posada de le Paz, R. Stefanov, and C. Nourissier.

8th RDTF Meeting will be held 23 October 2007 in Luxembourg.

The next WG on Coding and Classification will take place on 13 November in Luxembourg.