



Rare Disease Task Force Workshop on Initiatives and Incentives Summary Report

9 November 2009, Paris - France

1. General Introduction

The Scientific Secretariat of the European Commission's Rare Disease Task Force is supported over a three-year period (starting in January 2009 and ending in December 2011) by a DG Public Health funded Joint Action (Contract N°2008 22 91).

One of the principal objectives of this project is to organise the surveillance of initiatives and incentives in the field of rare diseases put into place at member state (MS) and EU levels. This work is based on the systematic surveillance of international literature and on a systematic query of key stakeholders in the EU27 MS. These are the members of the RDTF, EU27 country coordinators of Orphanet, and the 110 members of the Orphanet scientific advisory board. Queries are sent electronically, twice a month, by the RDTF scientific office. The data is published as news in *OrphaNews Europe*, the newsletter of the RDTF, which currently has over 11,700¹ registered readers. This data has been analysed to produce a report providing an overview of progress made towards finding a better approach to meeting the needs of the RD patient community, in order to use this information to propose recommendations for future action at the MS and EU levels.

It was initially planned to produce the report via a three-step approach:

- 1) draft report produced by INSERM (sent to workshop participants 10 days before the workshop);
- 2) workshop (09/11/09) of 20+ experts to discuss the proposed analysis, to suggest improvements and methods of dissemination, and to agree on recommendations;
- 3) dissemination of the revised document to a much wider audience composed of stakeholders who were consulted on a regular basis for the data collection to obtain their comments and criticisms, and consultation of the wider community for a duration of 2 months by putting the report on the RDTF website for public discussion.

The yearly reports will be published electronically during months 12, 24, 36 and will be made available on the DG Sanco and RDTF websites. In addition to this, 500 paper reports will be printed and mailed to key stakeholders.

2. Workshop agenda and introduction

A workshop was held on the 9th November 2009 in Paris: 30 experts attended this session. Firstly, Charlotte Rodwell presented the Joint Action and the work package concerned before presenting the sources, methodology and structure of the proposed draft report.

The main sources of data for this report was the data collected by the systematic surveillance of international literature and the systematic query of key stakeholders carried out in order to produce the *OrphaNews Europe* newsletter, in addition to past reports published by the European Commission, including past reports of the working groups of the Rare Disease Task Force. From this data and this documentation a retrospective of actions at EU-level and the state of affairs in the field in each EU Member State was constituted (i.e. pre-2009), and an inventory of Initiatives and Incentives for 2009 at EU and MS level was constituted.

¹ Number of registered readers in October 2009.

Orphanet Country Coordinators and information scientists were contacted once a draft version of the section concerning their country was drawn up using the data collected from the aforementioned sources. They were asked to validate and complete the data collected on the state of affairs pre-2009, and the initiatives and incentives in 2009

The report is structured into two main parts: the first main section concerns Initiatives and Incentives at EU level, and the second main section concerns Initiatives and Incentives at EU Member State level, with a secondary section concerning four other European countries where information was available. In annex to this report is a bibliography outlining the sources used to produce this report, which includes a list of the European Commission documents referred to in the report and a list of web addresses where National Plans for Rare Diseases in place in various EU Member States can be accessed.

The main section concerning the Initiatives and Incentives at EU level is split into 4 sections: the first 3 sections deal with each European Commission Directorate General having a major influence on rare disease and orphan drug policy in turn: DG Enterprise and Industry (including the work of the EMEA), DG Public Health (SANCO), and DG Research. The last section gives details of other RD meetings held at a European level in 2009.

The sections dealing with the three Directorates General of the European Commission are split then into two main sub-sections: a retrospective of actions pre-2009, and an account of initiatives, incentives and developments in 2009. The section concerning the actions of DG Sanco in 2009 is divided into two separate areas: direct initiatives and indirect initiatives.

The main section concerning the Initiatives and Incentives in each of the 27 Member States (plus Croatia and Turkey as candidates for EU membership, and Norway and Switzerland as Orphanet country teams from whom we regularly receive information on rare disease initiatives and incentives) is organised in alphabetical order by country. The information on each country is clearly divided into two sections: the first describing the state of affairs in the field of rare diseases before 2009, and the second outlining the developments made in the field of rare diseases at Member State level so far in 2009 (this includes advances made in the development of national plans and strategies for rare diseases).

The first section describing the state of affairs in each Member State pre-2009 covers the following categories:

- Definition of prevalence of RD
- National plan for rare diseases
- Centres of expertise
- Registries
- National alliances of patient organisations
- Sources of information on rare diseases and national help lines
- Rare disease events
- E-Rare partnership and research activities
- Orphan drug committee and incentives
- Orphan drug availability

The second section on initiatives and incentives and developments in 2009 in each Member State covers information collected on the following topics:

- new decisions regarding health care in the field of rare diseases (i.e. national plans)
- new initiatives to support patient organisations
- new information services on rare diseases and orphan drugs
- congresses and conferences concerning the field of rare diseases and orphan drugs/ events
- Rare Disease Day 2009
- new initiatives in the field of education and training
- new production of best practice guidelines both clinical and for laboratory testing
- new proposals for funding research into rare diseases and orphan drugs
- new incentives for the pharmaceutical industry in the field of orphan drugs
- new initiatives to facilitate access to orphan drugs and reimbursement
- any recent negative developments concerning initiatives and incentives for rare diseases and orphan drugs

The categories for which information is provided depends wholly on the information available for 2009 following data collection from the described sources and contact with Orphanet country coordinators.

The report is followed by its Annexes as aforementioned and described.

3. The Europlan Initiatives report

L. Vittozzi, RDTF Joint Action and Europlan partner, explained the scope of the Europlan Initiatives report (WP4), a one-shot report (rather than annual, as the RDTF report) which aims to create an inventory of initiatives in the field of rare diseases in order to develop recommendations for national plans and strategies for RD. The information collected is structured in the same way as the Council Recommendation. Questionnaires have been sent to Europlan partners, and replies have been followed up with telephone calls for clarification. The content is the same sort as the RDTF Initiatives report for the initiatives sections, and the same problems are encountered i.e. completeness, the institutional reliability of sources etc. The question was raised of whether collaboration between these two projects on a common report was possible, and in which ways and to what extent this collaboration could be pursued. It was agreed that a joint report would be more efficient and would ensure consistency between the two documents. Permission from DG Sanco will be asked in order to finalise this project.

4. Sources

The question of which are the most appropriate sources for this report was discussed amongst workshop participants. For the country section, it was agreed that a multi-source approach was the most appropriate, and that a coordinator for each country should suggest the most appropriate sources of information in their country: this could be undertaken by the Orphanet country coordinator who was previously contacted for their help in preparing the section on their country prior to the compilation of the draft report. Information should be factual and objective: any conflicting information should be reconciled where possible.

Other sources of information were suggested, such as:

- the presidents of national human genetics societies,
- COMP members,
- websites of patient organisations,
- Annual national rare disease plan implementation reports (when available).

The question of referencing sources was also addressed: it was decided that the names and institutions of those who had contributed to the revision of the document be noted for each country section

It was suggested that only reliable and objective information be kept in the final version of the report, and sources be explicitly cited for each country section.

5. Content

Another issue discussed was the possible validity and usefulness of describing initiatives and incentives at regional level for countries such as Spain and Germany which are highly regionalised. Also, the issue of whether similar weight be given to institutional initiatives and incentives and initiatives of patient organisations (as in some countries such as Romania, most RD initiatives come from patient organisations).

Section A – Europe and European Commission:

This section was felt to be globally complete and satisfactory in its structure. It was suggested that a separate section concerning the E-Rare project be included in the DG Research sub-section, rather than dividing this information (incorrectly) between the 6th and 7th framework programme sections.

Section B – Countries:

The content of the draft report was discussed, specifically concerning the topics/sub-titles proposed for the country sections. Certain topics were addressed:

- National Plans: it was noted that progress in this area is difficult to follow as there is no single model or approach. European data could be of use in this area.
- Centres of Expertise: should only official centres of expertise be inventoried, or should non-official centres of expertise be also included. Should we highlight the fact a policy for designation is in place as this also suggests financial backing? Should European Reference Networks also be inventoried (i.e. participation of the country in European Reference Networks)? What are the criteria for inclusion of a centre of expertise in the report?
- Research: this section was the weakest for most countries. It was suggested that additional information be collected (when possible) on how many projects are funded at national level, what is the budget allocated to RD funding, which funding agencies and patient organisations fund RD research, etc.

Additional sections were suggested for inclusion:

- New born screening programmes
- Best practice guidelines for clinical and laboratory testing
- Actions in the field of rare cancers (i.e. are separate initiatives taken?)
- Reference laboratories
- Participation of countries in international networks (not just European ones)
- Other therapies for RD (i.e. reimbursement of ocular implants).
- Transparency of clinical trials at national level.

It was also suggested that the section on “negative developments in the field of RD and OD” be removed.

Concerning the choice of countries covered outside the EU27 MS, it was decided that Switzerland be included as an interesting model, and Norway be included as member of the European Economic Area along with Lichtenstein and Iceland. As an extension of this logic all EU candidate countries should be included in the report (Macedonia, Croatia and Turkey).

Annexes:

Concerning the annexes, it was suggested that the sources of information from patient organisation websites be expanded, and that a possible annex be added detailing the RD research projects funded by DG Research’s 5th, 6th and 7th framework programmes with the amount of funding mentioned explicitly.

It was decided that an annex listing all general meetings and conferences linked to RD and OD in Europe was not necessary seeing as these are inventoried by country in the main text.

6. Review process

A strategy for revising and validating the content of the draft report was discussed.

a) For the section on the European Commission’s initiatives and incentives, validation from Toni Montserrat (DG Sanco), Catherine Berens (DG Research) and DG Enterprise and their hierarchies will be solicited.

b) It was decided that for each country, a list of key stakeholders should be proposed. These should be people in charge of taking initiatives and incentives at national level and they should be solicited to check the information for their country.

c) Specific experts will also be solicited for their opinion on sections concerning their scope of expertise, i.e:

- National drug agencies

- Representatives of the EMEA COMP
- Ministries of health/ Rare diseases offices at ministries of health
- Research funding agencies
- Patient alliances
- RDTF members

It was decided that at country level, Orphanet country coordinators should be in charge of contacting these people and sending revisions to the RDTF scientific secretariat. Following the decision to ask permission from DG Sanco to produce a joint Europlan/RDTF report, the timetable for validation and finalisation of the report needs to be reviewed (the final report was originally scheduled for publication in December 2009) and will probably take place in April 2010.

7. Conclusions

It was discussed whether conclusions should be drawn from the data or an analysis made. It was decided that the report as it stands is objective, and that for the report to be of greater use for policy making, that a table be included as a conclusion which recapitulates for each country the areas in which initiatives and incentives are present, i.e. national plans, calls for proposals for RD research funding. This information could also be produced as maps. This would allow readers to draw their own conclusions: however this could invite readers to compare what cannot be compared. A sort of disclaimer should be drafted and added to the introduction of this report, explaining that not all countries have set off from the same starting blocks (i.e. different health care systems, economical differences, political differences) and thus are not easily comparable: approaches are too different across countries to compare. This report should not convey incorrect messages and evaluation cannot be undertaken without indicators. It was suggested that the report should include in the introduction a comment on how the various annual revisions of this report will enable the progress at a political level to be monitored, and that the report itself could serve as an incentive for initiatives in the field of RD as stakeholders will see what other countries are able to put in place.

8. Dissemination

Participants agreed that the title "Report from the Rare Disease Task Force on Initiatives and Incentives in the field of Rare Diseases" was the most preferred. It was decided that publication on the RDTF website would not disseminate this document widely enough to all stakeholders and the general public. One suggested possibility was to transform the report into a paper for the Orphanet Journal of Rare Diseases, which would cost €1,000 and could be financed by the joint action. ORJD is an electronic, free-access journal and would thus have a bigger impact. Further questions concerning this suggestion of journal publication revolved around the need for expert/peer review and possible copy right and reproduction issues. Permission would also be asked from the Commission to publish this information. The type of article ("state of the art", "review paper", "special report") was also discussed and whether this should be a summary of the whole report, or the whole report.

500 printed copies will also be produced: 20 copies will be allocated for each country to be sent to relevant stakeholders. The importance of producing paper copies was highlighted, as this encourages stakeholders to read the document. It was suggested that a greater volume of flyers could be produced as an advert for the report, with a summary and a link to this report to encourage people to download the report. This text should summarise the content and methodology, and the flyers should be sufficiently visual to attract attention (i.e. graphic content).

Another issue was the importance of translating the report and summary. If the leaflet is translated into local language then the relevant sections of the report (at least the relevant country section) should also be translated. There is no budget for this translation, so it has to be done on a voluntary basis. It is important for this information to be accessed by stakeholders in each country in order to generate support in the Member States for DG Sanco's health programme.

9. Next steps

These next steps for the draft report were determined:

- Define timeline for publication
- Redefine validation process
- Add additional sections to Section B: European countries
- Investigate possibility for publication in OJRD
- Draft a flyer for disseminating the report

The next meeting of the RDTF Initiatives and Incentives Working Group will be held in late 2010.

Annex – Workshop Participants

Adamski, Jakub
Aymé, Ségolène
Devillé, Walter
Donadieu, Jean
Fregonese, Laura
Jansen, Herwig
Jessop, Edmund
Landais, Paul
Lavery, Christine
Lehenanff, Guillaume
Macek, Milan
Mehta, Anil
Miteva, Tsonka
Oestli, Elin
Palau, Francesc
Parker, Samantha
Posada, Manuel
Rodwell, Charlotte
Sandor, Janos
Schuster, Ralph
Stokke, Bodil
Stuhrmann, Manfred
Tchernia, Gil
Van Weely, Sonja
Vittozzi, Luciano
Voigtländer, Till

Observers:

Ashton, Charlotte
Guesdon, Benjamin
Jovanovic, Mariana
Taylor, Louise