Rare Diseases Task Force

Established in January 2004
Secretariat funded since June 05

www.rdtf.org

Chair: Ségolène Aymé (Paris)
Deputy chair: Helen Dolk (Belfast)
RDTF was established in Jan 04...

- To advise and assist the EC Public Health Directorate in promoting the optimal prevention, diagnosis and treatment of RD in Europe

- To provide a forum for discussion and exchange of views and experience on all issues related to rare diseases

- In recognition of the unique added value gained for RD through European Co-ordination
RDTF objectives

- To wide access to high quality information
- To assist in the diffusion of good and best practice
- To promote the exchange of ideas and information regarding quality of life issues and patients preferences and choices
- To promote the availability of high quality epidemiological data
- To promote the development of a classification and a coding system to supplement the ICD
- To promote effective surveillance, early warning…
- To promote the creation of reference centres
- To facilitate the consideration of different models of cross-border health care
RDTF duration and composition

- Established for the duration of the Public Health programme 2003-2008

- Composed of:
  - Current and past leaders of projects funded by DG SanCo
  - Representative of member states nominated by their member state
  - Representatives of relevant International organisations
  - Additional members nominated on an Ad-Hoc basis

- Total number:
  - Cannot exceed 35 persons of which 20 have their expenses covered by EC services
RDTF Secretariat

- Established through an EC contract in June 05 for 3 years
- Based in Paris, at Orphanet office

- Specific objectives
  - Coordination of TF meetings
  - Preparation of background documents
  - Overall coordination of related projects under the Public Health programme
  - Identification of morbidity and mortality indicators taking into account of the European Community health indicators project
  - Assisting the Commission in setting priorities in the field of RD
  - Contributing to the dissemination of information to all stakeholders in order to encourage continuity of work and transnational cooperation
RDTF Secretariat achievement

- Publication of an electronic newsletter and of a website

- Establishment of three working groups:
  - WG Health Indicators
    - Workshop on January 06
  - WG Standards of Care
    - Workshops on June 05, September 05 and September 06
    - Reports of Centres of Reference in the EU
  - WG Coding and Classification
    - Workshop on 11 October 06
Activity Report

Communication
Rare Diseases Task Force

About the Rare Diseases Task Force

Rare diseases are life-threatening or chronic, debilitating diseases which are of such low prevalence that specific combined efforts are needed to address them. A guide, low prevalence is taken as prevalence of less than 1 per 10,000 in the Community.

The Rare Diseases Task Force (RDTF) was set up in January 2004 by the European Commission's Public Health Directorate. Its aims are:

- To advise and assist the European Commission's Public Health Directorate in promoting the optimal prevention, diagnosis and treatment of rare diseases in Europe, in recognition of the unique added value to be gained from rare diseases through European co-ordination;
- To provide a Forum for discussions and exchange of views and experience on all issues related to rare diseases;
- To provide information and guidance to rare disease patients and their representatives.

The Task Force is led by Dr. Giuseppe Pene, a medical geneticist and Director of the Orphanet database of rare diseases. The Deputy Leader is Professor Helen Dolk, director of the concerted programme on congenital disorders.

It currently has 16 members, comprising current and former project leaders of European research projects related to rare diseases, member state experts and representatives from relevant international organisations.

Read the mandate of the Rare Diseases Task Force

Log In for Task Force Members

Login
Password
To Subscribe...
Electronic newsletter

- OrphaNews Europe
- Opt-in service: registration on www.rdtf.org
- 6,700 registered readers from 28 countries
- Start: June 05  16 issues so far
- High satisfaction of the readership
  – On-line survey in May 06
OrphaNews Europe Editorial board

- Editor-in-chief: Ségolène Aymé
  Editor: Kathy Beuzard-Edwards
- Editorial Board:
  - Ségolène Aymé, Aurélie Bedin, Catherine Berens, Helen Dolk, Anders Fasth, Edmund Jessop, Jordi Llinares-Garcia, Jérôme Parisse-Brassens, Helmut Popper, Elena Prats, Antoni Montserrat, Janos Sandor, Arrigo Schieppati, Rumen Stefanov, Domenica Taruscio, Joan Luis Vives Corrons
OrphaNews Europe topics of interest

- European News
  - European Policy
  - from EC funded projects
- News from Member States initiatives
- Scientific news:
  - new RD
  - New genes
  - Diagnostic and therapeutic break through
- Announcements
  - Major publications / reports
  - Events/ congresses/workshops…
  - Call for collaboration / enrollment
  - Job opportunities
Invited lectures at meetings
Second semester 2006

- Second Balkan conference on rare diseases, Plovdiv, 7-9 September
- Italian Conference on rare diseases, Rome, 18-19 September
- European meeting on childhood disability, Barcelona, 18 October
- ICORD meeting, Madrid, 25 October
- EPPOSI meeting, Madrid 26-27 October
Activity report

Working Groups
The rare diseases task force was requested to provide an overview to the High Level Group for Health Services and Medical Care
« Centre of reference » is a denomination used only in a few MS

<table>
<thead>
<tr>
<th>Centres of reference for rare diseases</th>
<th>Centres of Reference not specifically for rare diseases</th>
</tr>
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<tbody>
<tr>
<td>1990: Sweden</td>
<td>Belgium,</td>
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<tr>
<td>2001: Denmark,</td>
<td>Czech Republic,</td>
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<td>2002: Italy</td>
<td>Finland,</td>
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<tr>
<td>2004: France</td>
<td>Greece,</td>
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<tr>
<td>?2007: Bulgaria, Spain</td>
<td>Ireland,</td>
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<td></td>
<td>United Kingdom</td>
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« Centres of Reference » are established by reputation in all MS

Countries with identified unofficial « centres of reference » established by reputation

Austria, Cyprus, Estonia, Germany, Hungary, Lithuania, The Netherlands, Poland, Portugal, Romania, Serbia, Switzerland, Turkey
The definition of what is a CR differs from one country to another...

- Definition of rare disease differs
- Size of the population to serve differs
- Definition of the task differs
- Number of diseases to be covered differs
- Process to identify CR differs
- Financial support differs
- Purpose for establishing CR differs
- All expert centres are listed in Orphanet
Recommendations from the RDTF expert group
Centres of reference for RD seem to be needed…. 

- Patients are rare but experts are rare as well…. 
  - Need to identify them
- Expertise may be found only at International level 
  - Impossible for most countries to offer appropriate services to all patients 
- Clinical research is badly needed 
  - Requires cohorts large enough 
  - Systematic collection of data 
  - Production of clinical recommendations 
- Optimisation of OD prescription is desirable
But patients should not have to travel....

- **Financial constraints**
  - Cost of travel + accommodation

- **Communication constraints**
  - 20 languages in Europe + cultural differences

- **Logistical constraints**
  - Expert departments have a limited number of beds and of out patient clinics
Centres of reference are needed...

- Centres of reference are necessary for 2 main reasons:
  
  Reason 1: As a label of « quality » for the patients and the care providers. Quality has to be defined according to the missions of the centre. Rating approach.
  
  Reason 2: As a resource centre where to concentrate man power and funding to speed up research at lowest possible cost: cost/efficiency.
Have a common label

- Criteria to be fulfilled by CR
  - Appropriate capacity to diagnose and manage
  - Attractivity
  - Capacity to provide expert advice
  - Demonstration of multidisciplinary approach
  - High level of expertise + research capacity
  - Close links with other expert groups + support groups
Reference networks are already in place...

- Funded at EU level or national level (Germany, Spain, France)
- Real added value as
  - Diversity of expertise is shared
  - Rational provision of highly specialised services
  - Share case management systems
  - Common repository of cases
  - Paediatrics/ adult centres: continuity of care
Recommendations to Member States

To explore all possible forms of cooperation between MS in the field of health services and medical care for patients with RD

For MS having official CR:
- To agree on the definition / mission / evaluation
- To share experience and outcome measures

For MS not having official CR:
- Establish CR or find other appropriate way to meet the needs of patients (by contracting with foreign CR)
- Develop electronic communication with « CR »
Recommendations to Member States (2)

Contribute to the identification of expert centres and support them as much as possible

Organise health care pathways for patients
  - through the establishment of cooperation with all necessary expert centres from the country
  - and from abroad when necessary

To recognise and fund the activity of expert opinion
Recommendations to European Commission

♦ To fund reference networks of centres of expertise for rare diseases

♦ To open its call for proposals to the definition of a methodology to assess the benefit from such networks from the perspective of the different stakeholders

♦ To encourage the development of electronic services in the field of RD
Conclusion

- A step forward for improving the delivery of appropriate care
- Will contribute to concentrating expertise and resources
- A model for other sectors in medicine (severe chronic diseases)
- An experience which needs to be assessed
To follow up on this issue...

- The rare diseases task force website
  www.rdtf.org
- The newsletter of the task force
  OrphaNews Europe
- Participate in the working group of the RDTF
Working Group

Coding and Classification
WG Coding and Classification

- Issues to be tackled
  - State of art of existing coding systems regarding rare diseases: ICD, Snomed, MeSH, MedDRA
  - Plans for contributing to improve these systems
  - Establishment of a database of expert classifications of rare diseases
Establishment of the working group
  – Members of the RDTF who volunteered
  – Experts identified by RDTF members
    • Experts of RD directly involved in the classification effort
    • Experts of coding in the field of genetic diseases
    • Experts of coding for Death certificates
WG Coding and Classification

- Meeting of the working group 11 Oct 06
- Agenda:
  - Presentation of RD coding initiatives
    - NHS project « Connecting for health »
    - Dutch Central information system for hereditary diseases and synonyms
    - Padua registry of patients with RD
    - Coding for mortality data at DIMDI in Germany
    - Orphanet contribution to coding and classification
  - WHO view on the revision of ICD 10
  - Points for action
WHO plans towards ICD-11

Key drivers
- Respond to consumer needs
- Keep-up with scientific knowledge
- Compatibility with previous systems
- Better implementation within health information systems

Revision process
- Drafting: definition, diagnosis and indexing
- Overseeing the total ICD: structured comments + field testing
- Final draft and WHA approval
WG Coding and Classification

- WHO knowledge portal for ICD revision
- Evidence based systematic reviews
  - Clinical used
  - Epidemiology, demography and public health
- Knowledge sharing
  - Structures discussion forums
  - Field trials: global practice networks to test the proposed codes, definitions…
  - Classifications on-line journal
WG Coding and Classification

- WHO ICD revision: involving RDTF
- Involvement strategy
  - International: multiple countries + multiple languages + multiple health care systems
  - Multiple parties and professionals: different health care professionals and sectors + consumers
- Procedures: wikipedia tool
  - ICD-10+ application: scientific group review
  - ICD-11 draft
WHO ICD revision

- RD working Group to be established with active input providers NOW
- April 07: Official launch of the Revision
  - Meeting of leaders of WG + media event
  - Tokyo
  - 2012 Final version for public viewing
  - 2015 WHA approval
Contribution of Orphanet to classification
Contribution of Orphanet to Coding and Classification

- **Orphanet mission**
  - Establish a list of rare diseases and provide up-to-date information about them, including a directory of services in Europe

- **Orphanet achievements**
  - Database of 4,300 rare phenotypes
  - Total of 7,200 with included diseases
  - Encyclopaedia: 1,713 short articles and 420 Review articles (peer-reviewed)
Contribute to defining diseases

- Exemple: Ehlers-Danlos disease
  - Dominant form: type 1, 2, 3, 4, 7, 8
  - X-linked form: type 5
  - Recessive form: type 6 and 7

? 1 disease or 8 diseases?

- A disease is a phenotype which is recognisable clinically and which has a unique clinical course and a unique management approach
Classification of chanelopathies

  - Neurological inherited / autoimmunes /

- **Classification 2**: Nature. 2006 Mar 23;440(7083):440-7. From molecule to malady. Ashcroft FM.
  - Pore-loop / Non-pore-loop channelopathies / cys-loop receptors
Indexation of RD: an on-going process…

- **ICD 10**
  - 324 diseases have a specific code
  - 1,586 have a generic code

- **MeSH**
  - MeSH terms attributed to 1,149 diseases

- **PubMed automatic search facility**
  - Available so far for 1,407 diseases
2007 Plans

Working Groups
Working Group on indicators

- Meeting of WG in Paris on 30 Jan 06
  - Identification of areas for action:
    - Priorisation of RD for surveillance
    - Feasibility of using death certificates for RD
    - List of macro-indicators
    - Mapping of existing sources of epidemiological data

- 07 meeting:
  - date and agenda to be set up
  - Proposal:
    - Follow-up on going projects
    - Preparation of a report to be discussed during the Workshop
Working Group on Centres of Reference

- Publication of the 2006 Report
  - Dissemination through the Newsletter
  - Through the High Level Group on Health Services and Medical Care members

- Preparation of a new report
  - on the methods to assess the added-value of CR
  - and of the added-value of Reference networks
  - Publication: November 07
Working Group on coding and classification

- Identify issues to be tackled
  - State of art of existing coding systems regarding rare diseases: ICD, Snomed, MeSH, MedDRA
  - Plans for contributing to improve these systems
  - Establishment of a database of expert classifications of rare diseases
  - Contribute to the Phenome project

- Establish a working group
- Liaise with WHO
- Organise a workshop
  - Paris, 11 October 06
Initiatives at the secretariat level

- Establishment of an updated version of the Inventory of national incentives
  – Annual report
- Addition of a permanent section in the newsletter about genetic testing
  – In collaboration with EuroGenTest (NoE)
- Preparation of the RDTF work programme 2008-2010