Rare Disease Patient Solidarity

Presentation to the Rare Diseases Task Force

Rare Diseases Task Force
December 14th, 2006
WP 5
Eurordis survey on the provision of health care and social services (EurordisCare)

• WP5 leader: Rosa Sanchez de Vega
  - Federación Española de Enfermedades Raras

• Eurordis survey on patient's experience and expectations concerning access to health services in Europe - quantitative analysis of determinants

• 3rd EurordisCare survey

• Based on Eurordis Membership and networking

• WP working group
  - 8 members with
    - François Faurisson (Eurordis, Research Engineer)
    - Pierre Chauvin (Inserm U707 - Epidemiology)
    - Sarah Zohar (Inserm U717 - Biomathematics)

  - Co-sponsors (UGIM, Actelion, Sigma-Tau)
14 participating disease networks (proposed)

A wide diversity of determinism, genetic and multifactorial, age of onset, clinical manifestations, kind of handicap and prevalence:

- Marfan syndrome \((3 / 10,000)\)
- Fragile X syndrome \((1.4 / 10,000)\)
- Williams syndrome \((1.3 / 10,000)\)
- Ehlers-Danlos syndrome \((1.2 / 10,000)\)
- Cystic fibrosis \((1.2 / 10,000)\)
- Prader-Willi syndrome \((1.1 / 10,000)\)
- Epidermolysis bullosa (all) \((1 / 10,000)\)
- Tuberous sclerosis \((0.88 / 10,000)\)
- Myasthenia \((0.85 / 10,000)\)
- Osteogenesis imperfecta \((0.65 / 10,000)\)
- Huntington disease \((0.62 / 10,000)\)
- Friedreich ataxia \((0.25 / 10,000)\)
- Aniridia \((0.175 / 10,000)\)
- Pulmonary arterial hypertension \((0.150 / 10,000)\)

1-2 diseases with extremely low prevalence:

- 11q deletion syndrome \((150\ reported\ cases)\)
- Alternating hemiplegia \((200\ reported\ cases\ -\ about\ 30\ in\ France)\)
Commitment of patient organisations (POs)

- **Commitment of 78 PO to participate**
  - 48 PO involved in the questionnaire development.
  - Waiting for potential 30 other PO to respond

- **in 19 European Countries**
  - Austria, Belgium, Denmark, Finland, France, Germany, Greece, Italy, Ireland, Malta, Netherlands, Norway, Poland, Romania, Slovakia, Spain, Sweden, Switzerland, United Kingdom

- **Commitment of the European Chromosome 11q Network**
  - approximately 80 families with children with anomalies on the long arm of chromosome 11, 11q.
• questionnaire finalised
• Ongoing translation – 14 languages
  ▪ French, Italian, German, Finnish, English, Spanish, Danish, Greek, Norwegian, Dutch, Polish, Romanian, Slovak, Swedish
    ▪ Translation completed
      – French, English, Italian, Spanish
    ▪ Translation started
      – Danish, Dutch, Swedish, Finnish
    ▪ Translation planned
      – German, Norwegian, Polish, Romanian
  ▪ Volunteer translators needed for
    – Greek and Slovak
## Survey timelines

<table>
<thead>
<tr>
<th>Event</th>
<th>Date</th>
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<tbody>
<tr>
<td>Selection of diseases and medical services in survey</td>
<td>June 2006</td>
</tr>
<tr>
<td>Contact with POs, questionnaire development</td>
<td>June – Oct. 2006</td>
</tr>
<tr>
<td>Deadline for agreement of participation</td>
<td>30/11/2006</td>
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<tr>
<td>Questionnaire form available</td>
<td>Nov. 2006</td>
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<tr>
<td>Deadline for mailing of questionnaires to patients</td>
<td>31/01/2007</td>
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<tr>
<td>Deadline for responses</td>
<td>15/03/2007</td>
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<tr>
<td>Preliminary analysis</td>
<td>May - June 2007</td>
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</table>
WP8 – EU networks of centres of reference for rare diseases
WP8 – Centres of Reference

• Chaired by Birthe Holm, Rare Disorders Denmark (RDD)

• Agenda
  ▪ Preparation of national workshops on centres of reference: composition, questions to be addressed, materials to be distributed
  ▪ With the objective to report back to EU workshop in Prague, July 2007
WP 8 - Advisory committee members

- **Health policy makers**
  - Dr Alexandra Fourcade
    - France, Ministry of Health
    - EU High Level Group on Health Services and Medical Care
  - Dr Ségolène Aymé
    - France, Orphanet
    - EU Rare Diseases Task Force
  - Dr Edmund Jessop
    - UK, Department of Health, National Specialist Commissioning Advisory Group (NSCAG)
    - Working group on Centres of Reference of EU Rare Diseases Task Force
WP 8 - Advisory committee members

• Patient representatives
  ▪ Simona Bellagambi
    – Italy, Tuberous sclerosis Organisation and UNIAMO
  ▪ Christel Nourissier
    – France, Prader Willi France and Alliance Nationale Maladies Rares
  ▪ Rosa Sanchez de Vega
    – Spain, Aniridia and Federacion Española de Enfermedades Raras FEDER
• Health care professionals
  - Prof. Dian Donnai
    - UK, Medical Genetics,
      Clinical Head of Division of St Mary's Hospital
  - Prof Birgitta Strandvik
    - Sweden, Department of Paediatrics
      Institute of the Health of Women and Children
      Göteborg University
  - Prof Olaf Rieß
    - Germany, University of Tübingen / Department of Medical Genetics
Objectives of the national workshops:

- needs and expectations
- suggestions for the evaluation

Both for national centres of reference and European networks of centres of reference

in order to ultimately delineate recommendations for principles and criteria for the identification of such centres/networks.
National meetings: Preparatory materials

- Introduction letter, prior to the workshop:
  - EU process on the policy for centres of expertise (letter of the advisory committee, to be drafted by Ségolène Aymé, and endorsed by the committee)
- Synthesis of the document entitled “Report of the work of the High Level Group in 2006” and annexes (to be prepared by Martin Dorazil, EC)
- Synthesis of the Rare Diseases Task Force report on centres of reference (to be prepared by the RD Task Force)
The European political agenda and the state-of the-art

- **The agenda: political orientations of the High Level Group on Health Services and Medical Care**
  - A synthesis by the national representative of the High Level Group on Health Services and Medical Care (HLGHSMC) to present the proposed concepts of the European networks of centres of reference outlined in the 2005 report.
  - Options for a procedure for identification and development of European reference networks outlined in the 2006 report

- **State of the art of existing centres of reference or specialised care centres for rare diseases in Europe today**
  - Key finding of the DG SanCo Rare Disease Task Force report and rational for establishing EU collaboration, by the national representative of the task force or the Orphanet contact
  - Key figures and data, typology of specialised centres relevant to the national debate and comparison with other member states.
  - Recommendations of the Rare Disease Task Force
Agenda (2: debates)

1. **Question 1: Needs and expectations for rare diseases centres of reference**
   - What are the expected benefits for patients?
   - How the concept of centres of reference responds to these needs?
   - How the concept of centres of reference applies to the national health care system?
   - On which criteria to identify/designate them? How is it done already or how would you like to do it?
   - Which cooperation between centres of reference and specialised care centres? Are national networks the solution?
   - How to ensure long term sustainable funding?
   - What critical view do you have on the risks and benefits of the approach, expected advantages and potential risks?

2. **Question 2: Proposals for the evaluation of centres of reference in your country**
   - How would you measure if the centres of reference actually respond to your expectations?
   - How can we ensure that patients really benefit from them?

3. **Question 3: Cooperation with other countries**
   - How do you see centres cooperating at the EU level?
   - What role would play EU reference networks?
   - On which criteria to identify/designate them?
   - What would the expected specific benefits for patients be?
Composition of national workshops

• **Half patient representatives**
  - For National Alliances, to also invite groups that are not members of the national Rare Disease Alliance (for example if they participate to WP5 survey)

• **Half other:**
  - Health authorities (1-2 or more for MS with regionalised health care Σ)
  - Health care professionals
  - National member of the HLGHSMC (formal invitation through Martin Dorazil EC, secretariat of HLG?)

• **All together: 20 participants minimum (x 10 countries)**
  - 10 patient representatives (disease specific networks that participate to survey WP 5, but not exclusively)
  - 8 health care professionals
  - 2 health authorities or more, including member of HLGHSMC
Presentation of HLGHSNC report, by MS and local organiser

- **Belgium**
  - Pascal Meeus, MofH, for Flemish part
  - Belgische Organisatie voor Kinderen en Volwassenen met een Stofwisselingsziekte BOKS vzw
- **Czech Republic**
  - Milan Macek
  - Huntington organisation
- **Denmark**
  - Marianne Jespersen, Karen Brodum-Nielsen
  - Rare Disorders Denmark
- **France**
  - Alexandra Fourcade, Marc Brodin (pdt comité)
  - Alliance Maladies Rares
- **Germany**
  - Thomas Wagner
  - ACHSE
- **Italy**
  - Domenica Taruscio
  - UNIAMO
- **Luxembourg**
  - Yolande Wagener
  - ALAN
- **Spain**
  - Manuel Posada
  - FEDER
- **Sweden (date: March 16th, 2006)**
  - Anders Fasth
  - Riksförbundet Sällsynta diagnoser
- **The Netherlands**
  - Vereniging Samenwerkende Ouder en Patiëntenorganisaties VSOP
- **United Kingdom**
  - Edmund Jessop
  - Specialised Healthcare Alliance (tbc)
Timelines

• All national workshops: before end March 2007
• National reports sent: before May 1st 2007
• Meeting of Advisory Committee: June 11th 2007

(to prepare synthesis for European Workshop in Prague)
European Workshop, Prague 12-13 July 2007

- European Commission
  - DG SanCo
  - invitation to public health director Dr Andrzej Rys
  - DG Research
    - invitation to Head of Unit - Directorate for Health Research, Generic Activities and Infrastructures Manuel Hallen
    - and also Catherine Berens, Octavi Quintana Trias
  - DG Employment & Social Affairs, DG Enterprise Martin Terberger

- Czech Minister of Health
- Presidency of the HLGHSME
- Leader DG Sanco RDTF
- 6 networks selected Public Health Call for Proposals 2006
- COMP Chairperson Kerstin Westermark & EMEA representative
- ENVI committee at the EP (1 to 5)
- 30 representatives from national workshops
  - 10 reporters
  - 10 patient representatives
  - 10 co-opted
European Workshop, Prague 12-13 July 2007

- 14 representatives from rare diseases in WP 5 survey
- 25 MS representatives
  - In co-ordination with Martin Dorazil
  - For MS with no representatives at the HLG, ask Orphanet
- 15 representatives from MS with no national workshops
  - Health care professionals or patient representatives
- 9 members of the WP8 Advisory Committee
- 8 members of WP5 Advisory Committee

- Total: 125 /130
## EU workshop programme

<table>
<thead>
<tr>
<th>First day: starts at 11.00 am</th>
<th>Second day (ends 4.00 pm)</th>
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<tbody>
<tr>
<td><strong>Introduction</strong></td>
<td><strong>Synthesis of national workshops</strong></td>
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<tr>
<td>Workshop objectives</td>
<td>One speaker summarises responses to question 1</td>
</tr>
<tr>
<td>Presentation of the Task Force report, High Level Group</td>
<td>One speaker summarises responses to question 2</td>
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<tr>
<td><strong>Presentation of selected networks</strong></td>
<td>One speaker summarises responses to question 3</td>
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<tr>
<td>Dysmorphology</td>
<td>Discussion 30 min after each speaker</td>
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<tr>
<td>Cystic fibrosis</td>
<td></td>
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<tr>
<td>Commonalities and differences 6 selected projects</td>
<td></td>
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<tr>
<td><strong>WP5 survey results</strong></td>
<td><strong>Methodology to assess the outcomes of centres of expertise</strong></td>
</tr>
<tr>
<td>Reactions, debate with representatives of the selected diseases</td>
<td>Discussion based on task force meeting June 2007</td>
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</table>
November 27th-28th, 2007

- Part of the EU Presidency Programme
- Supported by Ministry of Health / Direcção- General de Saude
- Participation of Infarmed
- Meeting with Patient organisations: 20 October 2006 – Lisbon
  - Local organising committee
European context 2007

- 2nd Workshop on the Value of Innovation EPPOSI
  - 22-23 January 2007, Dublin
- CAPOIRA workshop (Gaining access to research resources)
  - 4-5 May 2007, Paris
- European workshop on centres of reference (part of Rapsody)
  - 12-13 July 2007, Prague
- DG Research workshop
  - September 2007, Brussels
- European Science Foundation seminar on rare diseases
- EPPOSI workshop on health technology assessment of OD
  - October 2007
- Health Technology Assessment Agencies workshop
Mains themes / sessions

• Opening Ceremony
  ▪ M. Markos Kyprianou, EC (tbc)
  ▪ M. Minister of health of Portugal (tbc)

• Member states policies/initiatives for RD

• Centres of reference and EU networks of centres of reference

• Framing healthcare pathways to patients’ needs
  ▪ inequalities, impact of migration, patient mobility…

• Assessing clinical utility to guarantee availability
  ▪ quality documents, centres of reference, genetic tests

• Genetic screening / testing

• Beyond medical care, addressing all patient needs
  ▪ paramedical & social care

• Research policies EU/National

• Drugs, safety, access (pre-authorisation, post-authorisation)
  Consolidating the environment / current framework
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This paper was produced for a meeting organized by Health & Consumer Protection DG and represents the views of its author on the subject. These views have not been adopted or in any way approved by the Commission and should not be relied upon as a statement of the Commission’s or Health & Consumer Protection DG’s views. The European Commission does not guarantee the accuracy of the data included in this paper, nor does it accept responsibility for any use made thereof.