NORDIC CONFERENCE
ON RARE DISEASES
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RARE DISEASES IN EUROPE

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30 years back in Europe…

• A winding road to diagnosis
• No best practice of communicating a diagnosis
• Rarity of the disease made it difficult to find a specialist
• No internet, little information available
• Few research projects on rare diseases
• Parents of patients with rare diseases were left with lots of uncertainty and no network
• Major inequalities amongst European countries
“Out of the darkness, into the light”

- Creation of patients’ organisations by parents of patients driven by frustration, determination to act and hope to change the predicted outcome.
  - Find existing patient groups
  - Meet other parents of patients
  - Share experiences
  - Raise awareness
  - Connect with medical doctors, researchers
  - Advocate for proper organisation of care and support services
  - Outline the need for good data collection and registries
  - Genetic counselling, hospital services
  - Raise funds for research, development of treatments
  - Participate in healthcare policy making
1980-2000: First National initiatives in Europe

- **Nordic collaboration on Rare Diseases 1984-1989**
- **Norway:** RD initiatives from the late 1970’s. Priority area from 1990 to 1993. Individual plans for support; Designation of national centers
- **Sweden:** 1988: Socialstyrelsen – large scale support for development projects. 1989 Information and Competence centre, Ågrenska
- **France:** 1997: Launch of EURORDIS. Creation of OrphaNet
- **Spain:** 1999: Sistema de Información sobre Infermedadas Raras
- **Italy:** 2001: rare diseases become a health care priority in the 3 years. National health plan: national network for prevention, surveillance, diagnosis and treatment: over 250 regional Centres designated & National and regional registries
- **Netherlands:** 2001: Dutch Steering Committee on orphan drugs, national web-based registry
Gamechangers

The World Wide Web:
A revolution for the dissemination of information, expertise, e-health, strengthening communication among patient groups and other stakeholders

The advance in genetic sciences & biotech:
New diagnostic tools, new insights into diseases, hope for development of therapies

Cultural changes:
Acknowledgement of patients rights, the equal opportunities principles and the role of patient groups in cocreation of welfare and innovation (empowerment)

A broader vision of Europe:
From the open market to a social Europe based on the added value of collaboration for citizens
Orphan drugs: Initiatives outside Europe

• **1983:** The Orphan Drug Act (ODA) passed in the USA

  upon proposal by the National Organization for Rare Disorders (NORD)

  ⇒ The impulse for introducing laws to promote drug development for rare diseases in other countries:

• **1991:** SINGAPORE

• **1993:** JAPAN

• **1997:** AUSTRALIA
1999: The European Regulation for Orphan Drugs: a corner stone

- 16 December 1999: Adoption of the European Regulation of the European Parliament and of the Council on orphan medicinal products – strongly advocated by patients through EURORDIS and other stakeholders

  ➢ One of the main incentives: market exclusivity for 10 years after Marketing Authorization

  ➢ 2000: Creation of the COMP - Committee for Orphan Medicinal Products at the European Medicines Agency (EMA):
    3 patients’ representatives for the first time

  ➢ Defining the field of action & providing a regulatory environment for fostering research & development

  ➢ Development of biotech SMEs in the field of ODs
2000-2010: Emergence of a European patients’ movement for advocating rare disease policies

Shaping rare diseases policies at local, regional, national, EU and international levels

Important role in
• promoting rare diseases as a public health priority
• advocating for the EU Regulation on Orphan Drugs
• contributing to the adoption of other regulations, directives, strategies and services for improving the life of patients

- EURORDIS: the Voice of patients at the EU level
  By 2012: Over 500 member organisations
- National Alliances of RD Patients Organisations – active at Nat level - 28 National Alliances today
- European Federations for specific rare diseases – 33 in 2012
- Many patient organisations, an active support for specific groups
Raising awareness

Dedicated people and pioneers can move mountains!

• Awareness raising of the public:

• **The International Rare Disease Day** – 29th February
  Initiated by EURORDIS: www.rarediseaseday.org

• **RARE CONNECT**: Online Rare Disease Patient Communities
  Partnership NORD – EURORDIS
  connect with each other, offer support, share experiences,
  sometimes even contribute to deeper insight into the natural
  history of RD’s
Empowerment of Patients’ Advocates


• Organisation of capacity building workshops – e.g. EURORDIS Summer School for Patients’ Advocates in clinical trials and drugs development


• Active participation of Patients’ Advocates in scientific committees of EMA, experts committees of EC and national competent authorities
2000-2010: European Commission’s initiatives

Support to fund projects for rare diseases research, healthcare policies through:

Community Action Programmes

Research Framework Programmes
Eurordis led surveys: “The Voice of 12,000 Patients”

- EurordisCare 2:
  - Access to diagnosis
  - 2003-2006
  - 5980 patients and families
    - 8 rare diseases, 70 patient groups
    - 16 countries, 12 languages

- EurordisCare 3:
  - Access to medical and social services
  - 2006-2008
  - 5995 patients and families
  - 16 rare diseases, 130 patient groups
  - 22 countries, 15 languages
2000 - 2010: Consolidating a broader EU regulatory framework for RD therapies

• EU Regulation on Paediatric Use Medicines
  ▪ Adopted in 2006, implemented since 2008
  ▪ Creation of the PDCO - Paediatric Committee - at the European Medicines Agency (including 3 patients’ representatives + alternates)

• EU Regulation on Advanced Therapies
  ▪ Adopted in 2007, implemented since 2009
  ▪ Creation of the CAT - Committee for Advanced Therapies at the European Medicines Agency (including 2 patients’ representatives + alternates)

• EU Directive on Cross-Border Healthcare in 2011
  ▪ Adopted in March 2011
  ▪ Deadline for transposition in the Member States: 25 October 2013
2010-2020: Rare disease policy framework shaping European actions for the next decade

• Communication from the European Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions:
  « Rare Diseases, Europe’s challenge »
  ▪ Adopted on November 11th, 2008

• Council Recommendation on an action in the field of Rare Diseases
  ▪ Adopted on June 9th, 2009
  ▪ Launch of the process for the development & adoption of National Plan on Rare Diseases in all 27 EU Member States by the End of 2013
  → Creation of the EU Committee of Experts on Rare Diseases (EUCERD)
EUCERD - European Committee of Experts on Rare Diseases

- EUCERD is an advisory committee on rare diseases of the European Commission. All stakeholders in the field of Rare Diseases participate.

- Objectives: Implement the Commission Communication on Rare diseases and Council Recommendations.

- Representation: 27 EU MS + candidate countries, EEA countries, Switzerland, EC, EMA COMP, ECDC, industry, academia, 8 patients’ representatives.

All 8 patients’ representatives are members of EURORDIS.

They cover main rare disease patient support groups and different European regions. They coordinate advocacy work through regular internal meetings.
EUCERD STATE OF THE ART ON RARE DISEASES

I. Overview of Rare Disease Activities in Europe and Key Developments in 2010 - July 2011

II. European Commission and Other European Activities – July 2011

III. Activities in EU Member States and Other European Countries – July 2011

Updated on yearly basis

www.eucerd.eu
This Joint Action will address the following priority areas:

a. Enhancing visibility and recognition of RD;

b. Contributing to the development and dissemination of knowledge on RD, from specialised research, through to the support of the healthcare professionals and the empowerment of patients;

c. Contributing to improvements in access to quality services and care, from diagnosis, through to care and social support and innovative therapies.

8 Work Packages including:

WP on support of RD plans/strategies at MS level – organisation of 20 National Conferences in Europe

WP on specialised services and integration of RD into social policies and services
2014-2020: European Commission’s programmes

- **2014-2020: “Health For Growth” - Health Community Action Programme**
  - Budget: 446 million euros
  - 4 main objectives:
    - Developing innovative and sustainable health systems;
    - Increasing access to better and safer healthcare for citizens;
    - Promoting health and preventing disease; and
    - Protecting citizens from cross-border health threats.
  - The Programme is expected to be adopted by the Health Council at the earliest in Autumn 2012.

  - Budget: 80 billion euros
2010-2020: A strategic approach to promote Rare Disease National Policies

- Plans or strategies are to be established and implemented “preferably by the end of 2013”

- The health authorities of the 27 EU member states (MS) signed the Council Recommendation, stating their willingness to fulfil this deadline.

- National strategies or plans will feed the European dynamics across member-states and future European policies
6 main priorities for National strategies or plans

• Adequate definition, inventory and codification of rare diseases

• Research

• Centres of Expertise and EU Reference Networks

• Gather expertise at EU level

• Empowerment of patient organisations

• Comprehensive approach and sustainability
2010-2020: The International Rare Diseases Research Consortium (IRDiRC)

An initiative for global cooperation on RD research

• Fostering transatlantic cooperation on rare disease research ⇒ utmost importance for the patients

• IRDiRC teams up researchers, funders and other stakeholders to deliver by the year 2020:
  • 200 new therapies for rare diseases
  • diagnostic tools for most rare diseases

European RD Patients’ representatives via EURORDIS will participate to the Executive Committee and the Scientific Committee on Therapies (including pre-clinical and clinical development)
Empowering Patient Advocates and Researchers 2010-2020: Research, a joint collaborative efforts

Translating research into therapies and care

EURORDIS’ patient advocates priorities

• Allocate more funds to basic, translational and clinical research
• Develop public – private partnerships
• Further develop disease registries and harmonise data collection
• Further develop biobanks networks and links with registries
• Reinforce multidisciplinary networks of experts relying on Centres of Expertise
• Further develop training on RDs for Researchers
• Explore broad strategy trials
• Develop research in social sciences for RDs
Main challenges from the patients’ perspective

- Decentralised health care systems
- Insufficient resources in all countries
- Need for continued supportive policies at EU level
- Further development of cross national collaboration
- Sustainability
- Measuring the real impact on patients’ quality of life and health
Main challenges from the patients’ perspective

1. Will we be able to ensure improved access to care and introduction of state of the art treatment and new therapies?

2. Will we be capable of further development and renewal of information and support systems?

3. Will involvement and empowerment of patient groups remain a key element in innovation?

4. Will networks of reference centres result in generally approved guidelines for comprehensive care?

5. Will we adapt to a prospectively changing agenda such as an ageing population of RD-patients with complex problems?
The need for a combined and global effort

- Rare diseases will continue to be a serious challenge to the health and welfare of EU citizens for decades to come: Continuity is a precondition for success.

- No Member State will be able to manage this challenge alone.

- By their complex nature Rare Diseases will be a set off for increased collaboration and innovation both in science and technology as in provision of services.

- Rare Diseases will be a test case for a modern approach to disease management leading to comprehensive, intelligent and empowering solutions.

- In their own interest Member States must pool resources and support collaboration. EU must secure continuity.

- Patient organisations will work actively to support creation of synergies, promote idea generation and maximise available resources and assess outcome!
What lies around the corner...

- Innovative solutions, new breakthroughs in diagnostics and therapies
- Intensified exchange in networks of centres of reference and more cross border collaboration
- A sharp increase in global cooperation
- A smarter use of internet based information systems
- Increased involvement of patient networks in mutual support, patient education, data collection and in gaining depth understanding of issues and needs

But also:

- A stronger focus on outcome measurements and health technology assessment (HTA)
- New ethical questions and dilemmas
The strong need for sustainability

5 or 10 year strategies and action plans will only partially address the needs of citizens affected by rare diseases

• Necessity to build sustainability into National Plans and strategies: Centres of Expertise, proper funding of clinical pathways, information and support services and clinical research infrastructures:

• Support co-operation between Member States (ex: E Rare, Rarelink..) and active participation in Reference Networks

• Necessity to build sustainability at European level: information networks, patient associations networks, EU Reference Networks of specialised centres, databases and biobanks, other basic and clinical research infrastructures, continue to build best practice in comprehensive care

[Website: www.eurordis.org]
Thank you!

www.eurordis.org