ICORD 2010

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Countries with Legislation

- 1983 USA
- 1993 Japan
- 1991 Singapore
- 1998 Australia
- 1999 European Union – 27 Member States:
  Austria, Belgium, Bulgaria, Cyprus, Czech Republic, Denmark, Estonia, Finland, France, Germany, Greece, Hungary, Ireland, Italy, Latvia, Lithuania, Luxemburg, Malta, Netherlands, Poland, Portugal, Romania, Slovakia, Slovenia, Spain, Sweden, United Kingdom;
  EEC countries: Iceland, Liechtenstein, Norway
- 2000 Taiwan

Identifying Needs of the Rare Diseases Community

- Patients, Their Families, and Advocacy Groups
- Research Investigators
- Health Care Providers
- Foundations
- National (Federal) Agencies
- Pharmaceutical Industry

Identifying Informational Needs of the Rare Diseases Community

- Information About Rare Diseases
- Approved and Investigational Treatments
- Location of Research Centers and Investigators
- Existing Patient Advocacy Groups
- Obtain Appropriate Diagnosis
- Payment or Reimbursement for Diagnosis, Prevention or Treatment
- Adequate and Stable Research Funding

Identifying Needs of Your Country

- **Each Country Identify Specific Needs**
- **Historical Survey Instruments Available**
- **Utilize Existing Resources from Numerous Sources**
- **Dedicated and Experienced Individuals and Organizations Are Available to Help**
- **Requests for Assistance**
- **Patients and Families are Recognized as Research Partners**
- **Centralized Coordinating Offices are Essential for Each Country**

Identifying Needs of Rare Diseases Community

- **Ready Access to Information Sources and Help Lines**
- **Patient Registries/Patient Recruitment**
- **Bio-Specimen Repositories**
- **Natural History Studies**
- **Personalized Medicine**
- **Prevalence of Rare Diseases**
- **Genetic Testing Sources and Testing Materials**
- **Whole Genome Sequencing**

Selected Programs Developed to Respond to Needs of Rare Diseases Community

- Improved Access to Clinical Trials
- Information about Rare Diseases
- Undiagnosed Diseases Programs
- WHO / ICD 11 Classification of Rare Diseases
- Expanded Role of PAG: Basic Research to Product – Grand Rx
- Re-Purposing of Approved and Investigational Drugs (TRND)
- Research Networks RDCRN/CTSA
- Course-Science of Small Clinical Trials – FDA / OOPD and NIH / ORDR

Selected Programs Developed to Respond to Needs of Rare Diseases Community

- Rare Disease Educational Module for Middle School Students
- Renewed Industry Interest in Rare Diseases and Orphan Products/ Niche Markets
- Increase in Virtual Drug Companies
- Emphasis on Tropical or Neglected Diseases in Developing Nations
- Future with Novel Delivery Systems, Nanomedicine, Stem Cells, Cell Therapy, Gene Therapy,

Patient and Public Driven Emphasis on Rare Diseases and Orphan Products

- "Public sentiment is everything. With public sentiment nothing can fail. Without it nothing can succeed…"
- Abraham Lincoln: August 21, 1858 - Lincoln-Douglas Debate at Ottawa, IL, USA


Finland
Sweden
Estonia
Latvia
Lithuania
Denmark
Ireland
United Kingdom
Netherlands
Belgium
Germany
Luxembourg
Poland
Czech Rep.
Slovakia
France
Austria
Slovenia
Hungary
Romania
Bulgaria
Italy
Spain
Portugal
Greece
Malta
Cyprus

Incentives for Research and Development "European Union Council Recommendation of June 8, 2009 on an action in the field of rare diseases" (2009/C 151/02)

- Around 5-8000 rare diseases in the EU – affecting 6-8% of the population at any time – corresponding to 29-36 million EU citizens

- "The specificities of rare diseases – a limited number of patients and a scarcity of relevant knowledge and expertise - single them out as a unique domain of very high added value of an action at (EU) Community level…"

EU Objectives to Respond to Needs of Rare Diseases Community

"Commission Communication …on Rare Diseases: Europe's challenges (11.11.2008)

- **Improving recognition and visibility of rare diseases** (coding and classification – ICD-11)
- **Supporting policies on rare diseases in the Member States** (National action plans, Fostering research , Ensuring access to high-quality healthcare – European Reference Networks; Gather national expertise – pool with European counterparts; Ensure empowerment and involvement of patients and patients' organizations; Ensuring appropriate provisions for sustainability over time
- **Developing European cooperation, coordination and regulation for rare diseases**

EU Operational Actions to develop European cooperation and improve access to high-quality healthcare for RDs

- European Reference Networks
- Access to specialised social services
- Access to orphan drugs
- Compassionate use programmes
- E-health
- Screening practices
- Quality management of diagnostic laboratories
- Registries and databases
- Research and Development - EU Framework Programmes

EU Operational Actions to improve recognition and visibility of rare diseases

- Definition of rare diseases prevalence - not more than 5/10.000 persons in the EU
- Classification and codification : WHO-ICD
- Dissemination of knowledge – e.g. Orphanet
- Disease information networks

Incentives for Research and Development "European Union Council recommendation of June 8 2009 on an action in the field of rare diseases" (2009/C 151/02)

- Plans and strategies in the field of rare diseases
- Adequate definition, codification and inventory of rare diseases
- Research on rare diseases
- Centres of expertise and European reference networks for rare diseases
- Gathering the expertise on rare diseases at European level
- Empowerment of patient organisations
- Sustainability

EU Operational Actions to develop European cooperation and improve access to high-quality healthcare for RDs

- International Cooperation

- Governance and Monitoring:

  EU Committee of Experts on Rare Diseases

  EUCERD, 2010

Incentives for Research and Development
The example of Orphan Drug Development

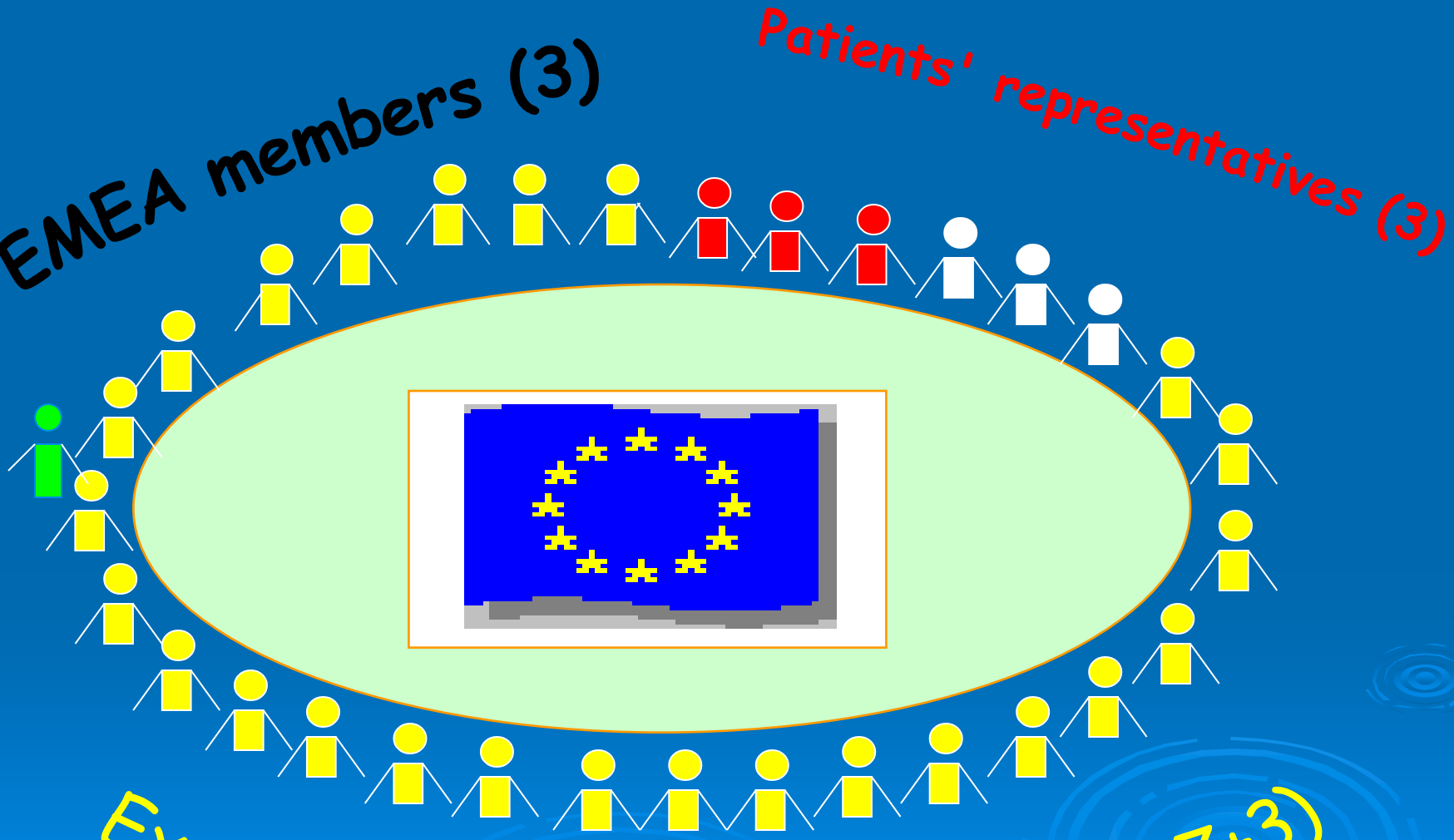
- 1983 the US Orphan Drug Act
- 1999 the EU Orphan Regulation

  Initiated by patient organisations –

  both in the US and the EU!
- Economic incentives, Research Grants, Regulatory incentives (by US/FDA and EU/EMA Medicines Agencies),
- Before – virtually no drugs for patients with rare diseases.
- After – Thousands orphan designated – Hundreds authorized
- Collaboration between the US and the EU – Harmonisation of requirements witihin respective regulatory frameworks

  → Augmentation of research and development!

Committee for Orphan Medicinal Products, COMP
European Medicines Agency, EMA



EU Incentives for Orphan designated Medicinal Products (OMP)

- **Fee reductions for OMP development**

  Application for OMP Designation: free of charge

  Protocol assistance (scientific advice for OMPS) from the EMA: free of charge

  Application for Marketing Authorisation: free of charge for Small and Medium-sized Enterprises (from 2009)

  + Extended incentives for SMEs in post authorisation

- **EU marketing authorisation** hrough unique centralised procedure
- **Market Exclusivity in the EU (all 27 Member States)**

  - 10 years for all orphan medicines (from marketing authorisation)

  + 2 years if Paediatrics Investigational Plan (PIP) results included in the MA and reflected in the SPC

- **Priority to EU Research - Framework programs** – clinical trial grants from 2009
- **Access to Member States Incentives** (EC inventory)

”Learning from the Rare”

“…Nature is nowhere accustomed more openly to display her secret mysteries than in cases where she shows traces of her workings apart from the beaten path; nor is there any better way to advance the proper practice of medicine than to give our minds to the discovery of the usual law of nature by the careful investigation of cases of rarer forms of disease”.

(Letter by William Harvey, year 1657)

ICORD
Buenos Aires 2010
marks the:

”Call for a GLOBAL approach based on special and combined efforts to prevent significant morbidity or avoid premature mortality and to improve the quality of life and socioeconomic potential of affected persons”