Orphanet: Advancing collaboration on Rare Diseases in Europe

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A unique initiative on Rare Diseases and Rare Cancers
Orphanet’s ID

- **Date of birth:** 1997
- **Nationality:** European
- **Main language:** English (and 9 other languages)
- **Founders:** INSERM and Ministry of Health
- **Size:** 41 countries
- **Domains:** Rare Diseases, Rare Cancers
- **Missions:**
  - Improve knowledge on RD
  - Provide info on RD
  - Ensure access to info on RD
  - Develop a database of RD
  - Develop a nomenclature for RD
Orphanet’s activities in a nutshell

- Coordination, consortium animation, Quality Assurance, IT Infrastructure & Development
- Encyclopedia of RD and RC
- Scientific activities: classification and codification of RD, epidemiological data, genotype, phenotypes, disabilities
- Inventory of Orphan Drugs
- Information on national centres of expertise, laboratories, patient associations, research & trials, registries, biobanks…
Orphanet in figures

- Annual budget 3M€
- 39 staff
- 86% of fixed-term contracts
- Mixed-funding: FR, EU, MSs

**Orphanet core activities funding**

- INSERM 45%
- DG SANTE 28%
- DGS 10%
- DG RESEARCH 2%
- AFM 8%
- CNSA 0%
- Valorisation 1%
- OJRD 3%

**Funding sources for national activities**

- EC 32%
- FR 16%
- AT BE BG CA CH HR 1%
- IT 4%
- UK 3%
- EE <1%
- ES 3%
- FI 1%
- HU 1%
- SI <1%
- SE 1%
- RO 2%
- PL NL NO LV LT <1%
- PT 1%
- RO 2%
- NL 1%
Orphanet is born to be European and International
20 years of fruitful European and international cooperation

- Massive expansion of the Consortium
  - 1999: Expansion of the Clinical Trials Network
  - 2003: Publication of Orphanet Europe
  - 2005: Orphanet gets its first makeover
  - 2008: Start of Orphanet Joint Action and creation of national entry points
  - 2011: A new home page and addition of Dutch
  - 2013: Start of RD-Action Joint Action on Rare Diseases
  - 2015: A new look for a new era

- Key dates:
  - 1997: Creation of Orphanet at INSERM with financing from the French Ministry of Health
  - 2000: Orphanet becomes the European and French action for orphans and becomes a leading player in the field
  - 2009: European Commission: Rare Diseases Joint Action
  - 2010: Orphanet joins the Horizon 2020 Work Program
  - 2014: Collaboration with the NBTH: Genes and Advances in Orphan Diseases
The European political context on RD

- Commission’s communication to European Parliament, Council, European Committee for regions and European Economic and Social Committee: Rare diseases, a common challenge for Europe (2008)
- Council Recommendation of 8 June 2009 on action for rare diseases
- Directive 2011/24/EU of 9 March 2011 on the application of patients’ rights in cross-border healthcare – art 13
- Commission Expert Group on RDs: Recommendation on ways to improve codification for RDs in health information systems (2014)
- 24 European Reference Networks

RARE DISEASES => STRONG ADDED VALUE FOR EUROPEAN COOPERATION (Research and healthcare)
Orphanet’s added value

- Unique nomenclature on Rare Diseases and Rare Cancers – International reference for interoperability between countries, and between health and research.

- Comprehensive information on provision of healthcare for RDs (incl. Centres of expertise, experts, laboratories, orphan drugs, etc)

- Robust, accessible and high quality information (reports, newsletters, info leaflets)

- Complemetarity with ERNs (knowledge generation, global visibility of ERNs activities…)

- Free of charge for users
Orphanet’s future is starting now
Main challenges

- Continue providing free access to high quality and reliable information on RDs to patients and citizens
- Develop a sustainable cooperation model between
  - France
  - EU Member States
  - EU institutions
  - International partners
- Define a status for Orphanet at the EU level
- Secure sustainable funding
Scenario-based future

Scenario 1
• France

Scenario 2
• #1
  • France
  • EU Member States
• # 2
  • France
  • EU Institutions

Scenario 3
• France
• EU Member States
• EU Institutions

Scenario 4
• France
• EU Member States
• EU Institutions
• International partners

Steering Group on Prevention and Promotion – Brussels – 11 April 2018
Possible cooperation model
# The scenario-based matrix

<table>
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<th>activities</th>
<th>France</th>
<th>EU MS</th>
<th>EU institutions</th>
<th>International Partners</th>
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<tr>
<td>Information on national provision and organization of care</td>
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Notes: TO BE DEFINED TOGETHER DEPENDING ON CHosen SCENARIO
Conclusion
Time for action

- France to propose a scenario-based concept paper
- EU MS to decide how and where to get involved
- EU to define/identify sustainable funding options

=> Shape the European agenda on Rare Diseases and Rare Cancers