



Orphanet: Advancing cooperation on Rare Diseases in Europe

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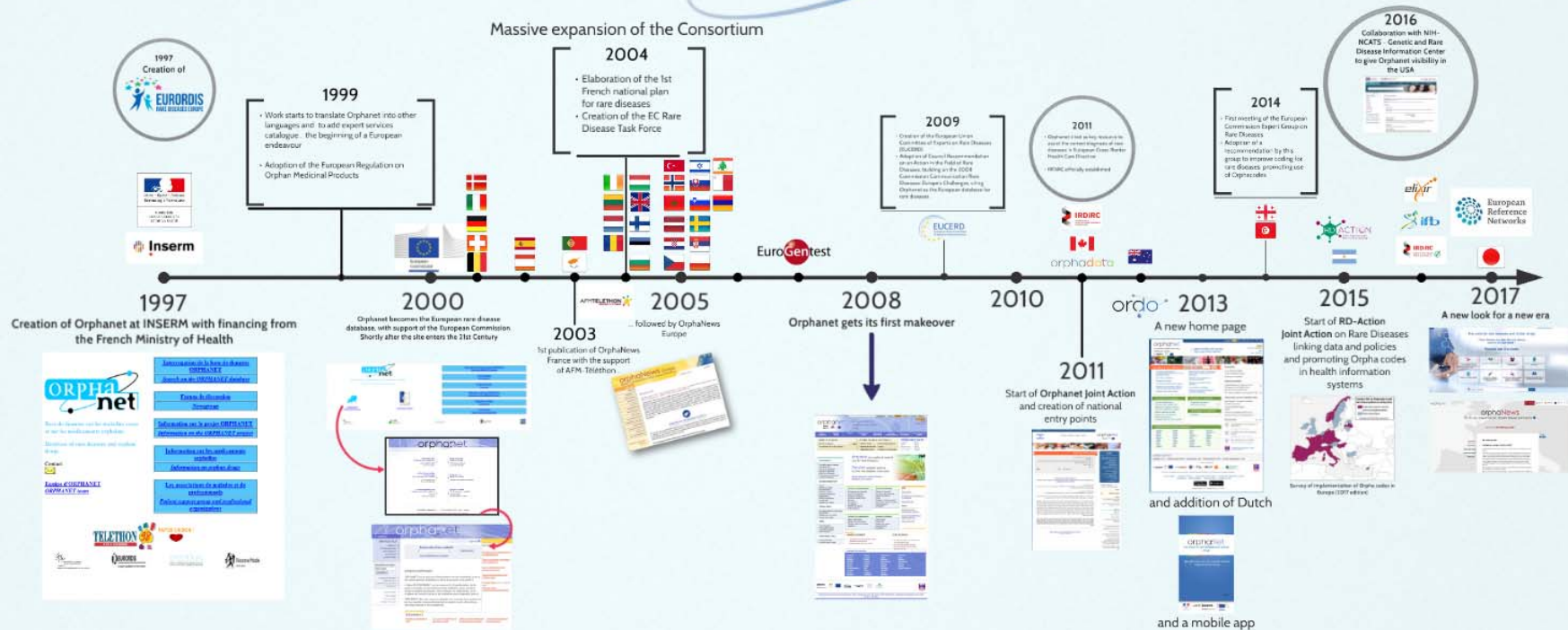
Rare diseases: a challenge for European countries

- ▶ RD patients and expertise are scattered but share the same problems: need for sharing solutions at the trans-national level
 - ▶ Commission Communication on Rare Diseases: Europe's Challenges (2008)
 - ▶ Council Recommendation in the field of rare diseases (2009)
 - ▶ Dedicated Commission Expert Group for RD : up to the end 2016
 - ▶ Commission Expert Group on Rare Diseases – Recommendation on Ways to Improve Codification of Rare Diseases (2014): **ORPHAcodes (Orphanet nomenclature)**
- ▶ **Orphanet as the reference knowledge base for data and information on RD**



20 years of fruitful European and international cooperation

20 years Orphanet



Orphanet missions

- ▶ Improve the **visibility** of rare diseases in the field of care and research through the development of the specific and comprehensive nomenclature on rare diseases (ORPHA codes),
- ▶ Identify and provide high quality information on RD accessible to anyone: ensuring **equal access to knowledge and expertise** for all the stakeholders, and orientating them in the mass of information available online
- ▶ Contribute to **improved knowledge on RD**: piecing together the parts of the puzzle to better understand RD from a medical, scientific and organisational point of view.



Orphanet's contribution to address EU and MS needs

- ▶ Improving the **visibility of rare diseases** in healthcare systems
 - ▶ Producing the Orphanet nomenclature (currently 9 languages)
 - ▶ Providing tools and support for the ORPHAcodes implementation
- ▶ Providing **high-quality information** on rare diseases and expertise and ensuring **equal access to knowledge** for all stakeholders orientating all of them in the field in the mass of information available online
 - ▶ Manually curated, expert reviewed encyclopaedia, including Emergency guidelines and Disability factsheets
 - ▶ Dissemination of quality assessed articles, including CPGs, at a single entry point
- ▶ Improving **access to the best standard of care**, cross-border healthcare and next-generation genetic testing for patients;
 - ▶ Directory of expert centres, diagnostic labs and tests, patient organisations in each Orphanet network country
 - ▶ Searchable by disease, genes, institutions
 - ▶ Cartographic representation of ERNs
- ▶ Improving universal access to appropriate **social services**;
 - ▶ Information on disabilities related to RD for a better evaluation of patients' needs
 - ▶ Access to the Social services cartographic representation (by EURORDIS)
- ▶ Promoting innovative strategies to ensure access to **rare diseases therapies**;
 - ▶ Data on the state of the art of research and development, including orphan designation and drugs
 - ▶ Collaboration with IRDiRC
- ▶ Allowing **access and data sharing** for databases, registries and projects on RD;
 - ▶ Database of research projects, clinical trials, registries, biobanks and infrastructures
 - ▶ Providing the interoperability backbone for registries, biobanks and databases in a computable way
- ▶ Providing useful **data to monitor Member States (MS) activity** in the field of RD.
 - ▶ Orphanet data are the core of the *State of the Art on RD activities in Europe since 2009*

Positive impact of Orphanet data and information

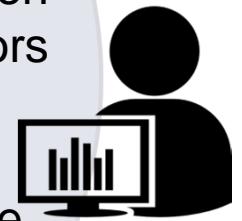


Visibility and recognition
Tackle isolation
Know better their disease
Identify experts and organisations
Identify research and treatments



Know better the disease
Access to best literature & guidelines
▶ Better care
Identify experts and labs
▶ Better referrals
Help patients

Quantifying burden
Measure indicators
Follow policies
State of art
Standards of care
Gap analysis

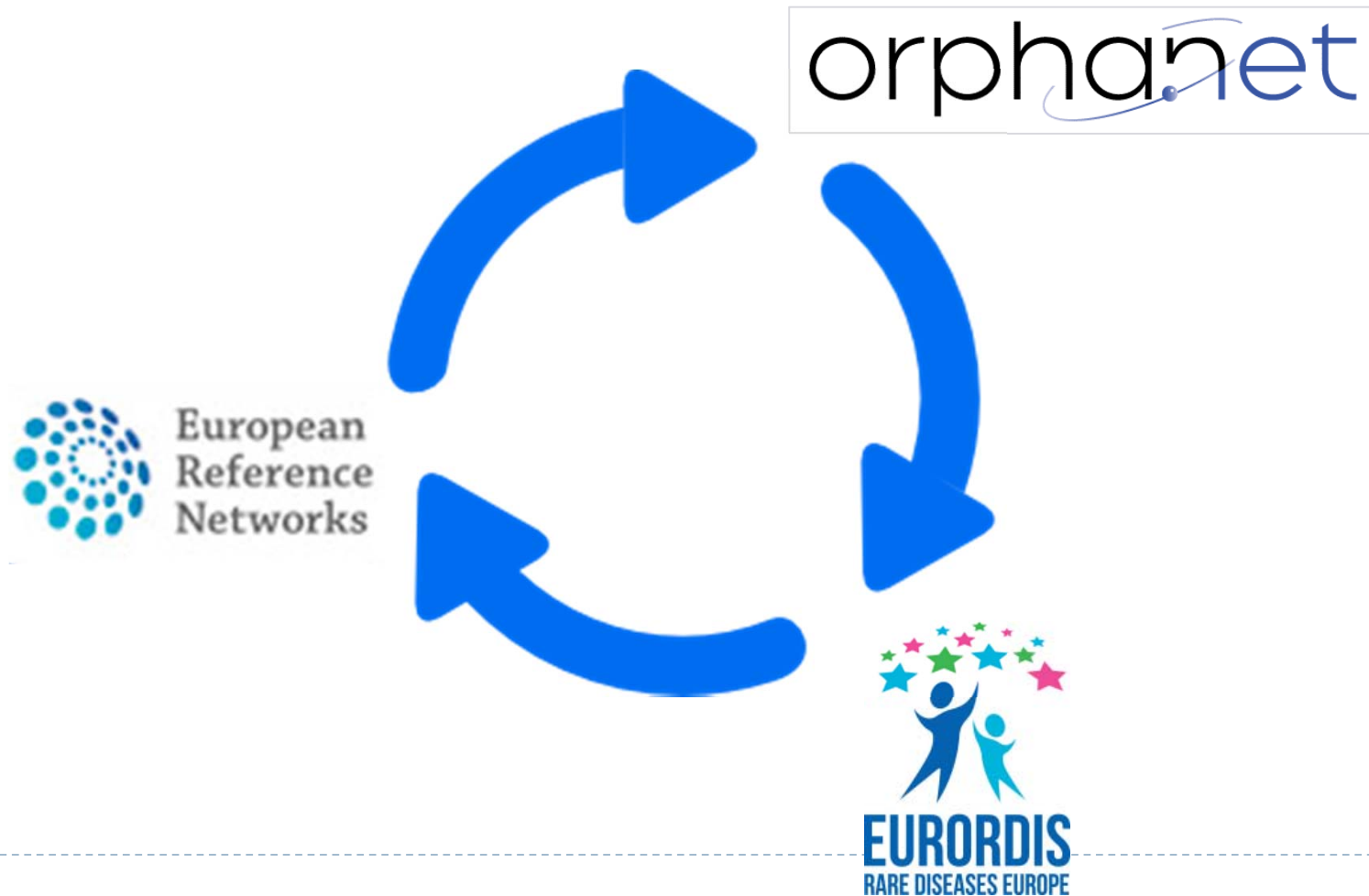


Data sharing and re-use
Patient recruitment
Networking

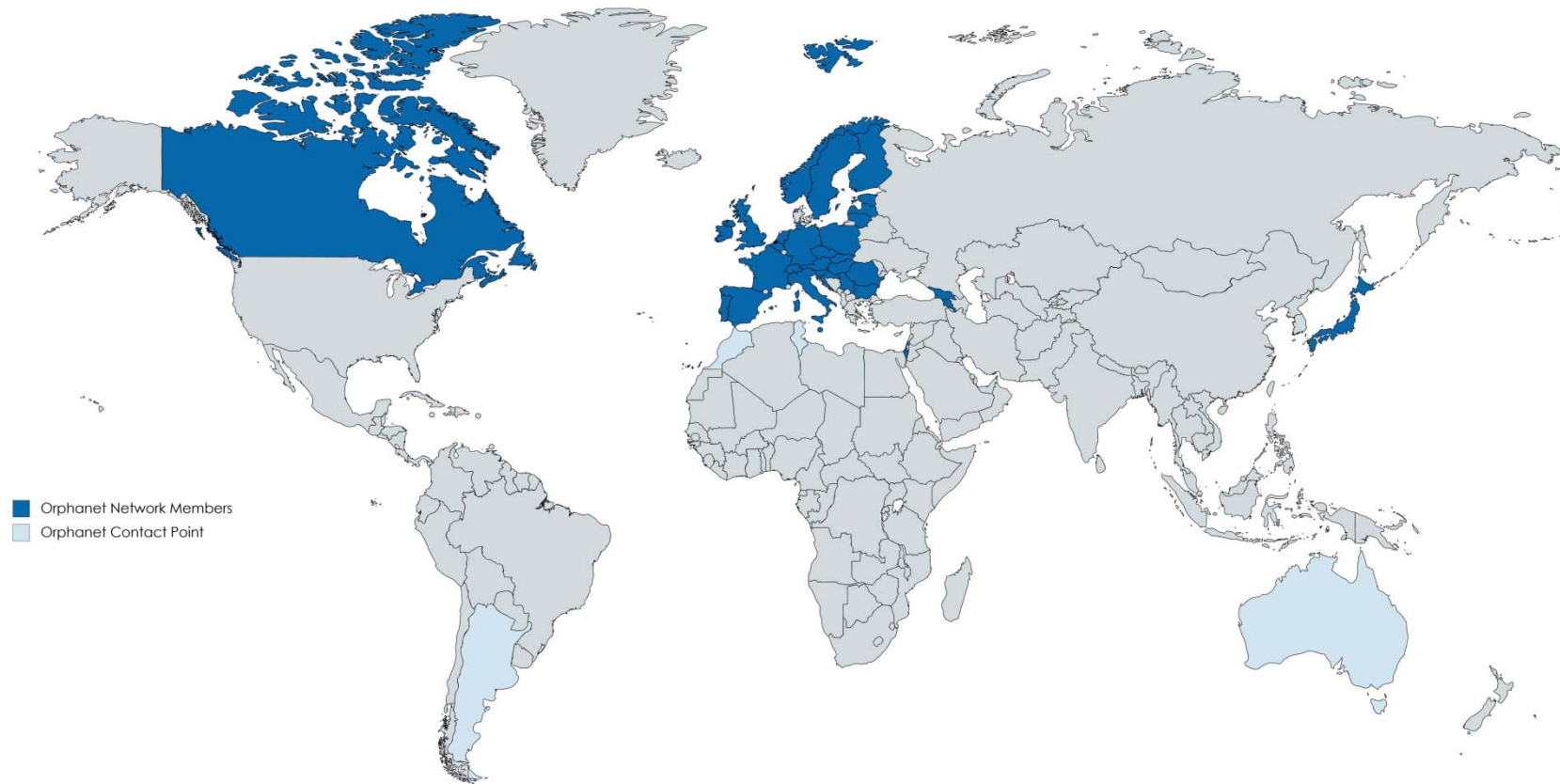
Patient recruitment
Market size
Re-use of data



A consistent European RD ecosystem



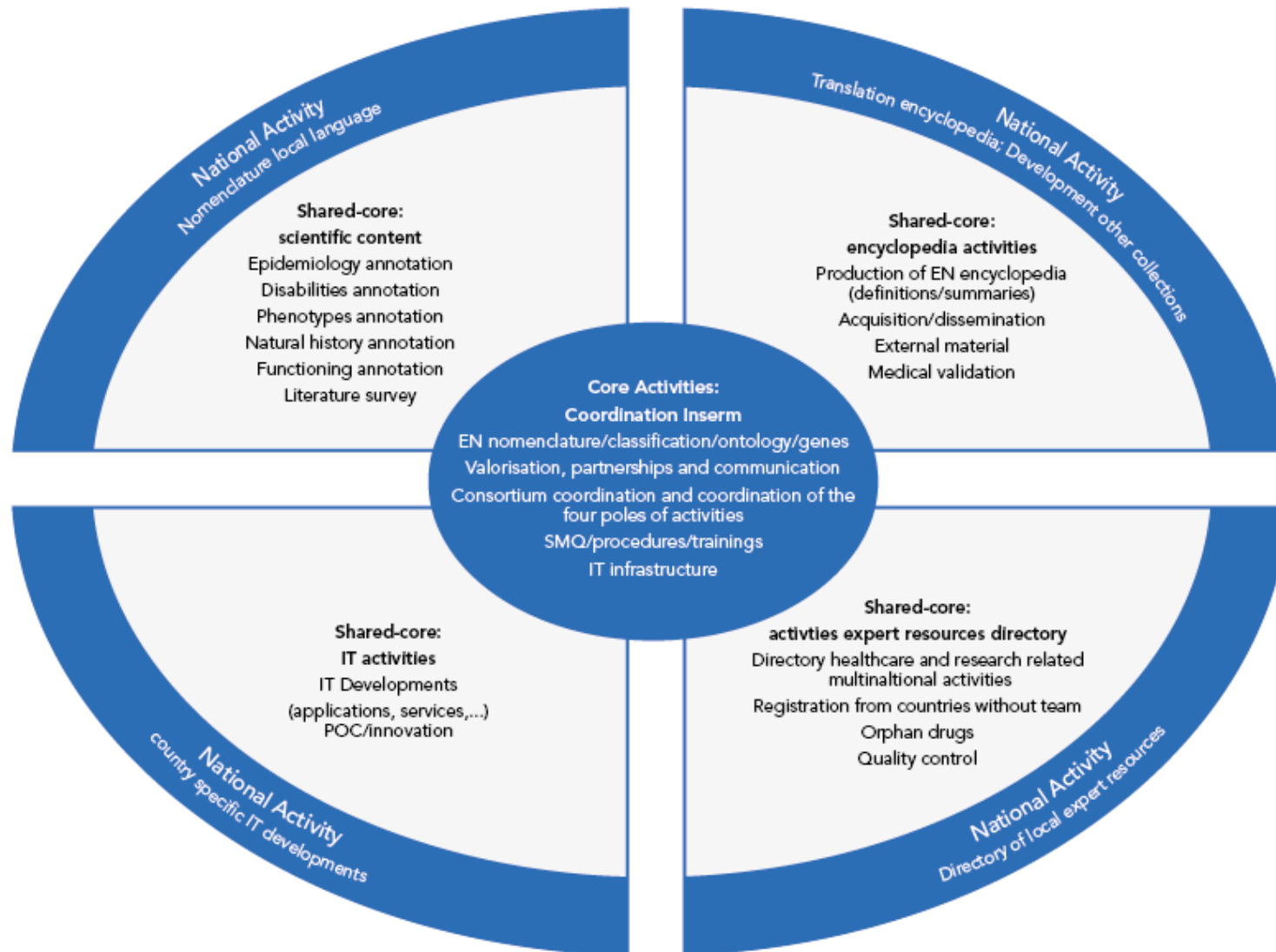
Orphanet Network



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Orphanet activities: a modular model that can be distributed



Orphanet current budget

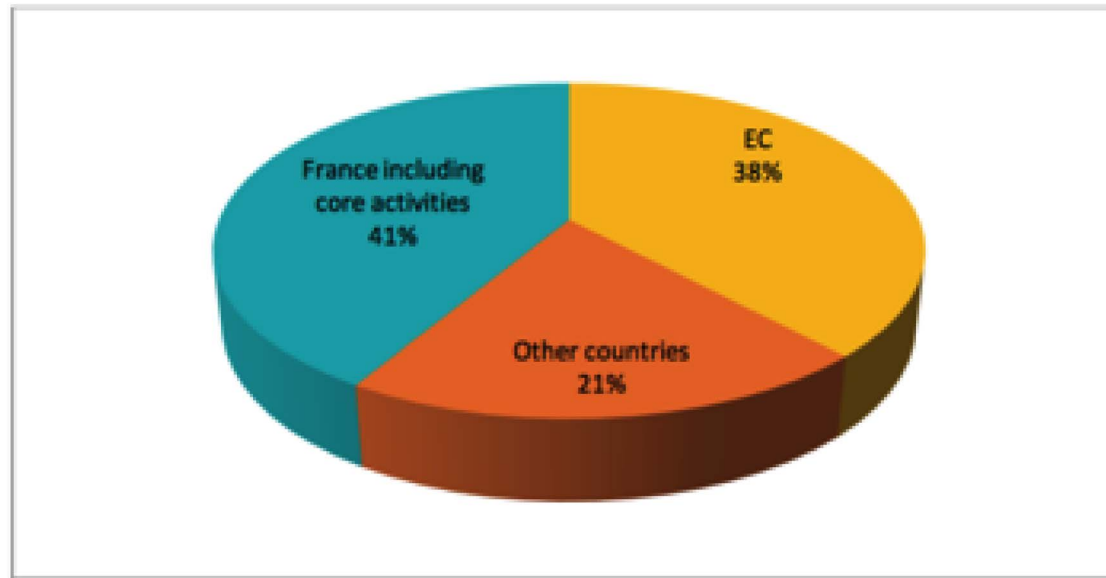


Figure 32 *Orphanet's global budget 2017*

- ▶ Currently short-medium term project-based funding
- ▶ No stable funding for core and national activities except for some coordination activities
- ▶ Valorisation of data to finance IT developments



Orphanet current costs

Activity	Annual Cost*	Impact
Coordination: coordination of the Orphanet consortium, definition of the common procedures, training for Orphanet National Teams, coordination of core and national activities, quality control, quality management system, communication, business development and partnerships strategies, IT infrastructure and database developments.	1,389,500 €	<i>For patients, doctors, researchers and industry:</i> Same level of quality whatever the country. <i>For policy-makers:</i> No-need to develop and maintain national databases: mutualisation of efforts, cost-saving.
Nomenclature and alignments Core nomenclature in English (coordination level activity) Alignments with medical terminologies	276,500€ 39,500 €	<i>For patients:</i> increased visibility/recognition in health pathways <i>For healthcare/social care professionals:</i> facilitated identification of RD patients <i>For policy-makers:</i> estimation of prevalence and burden of RD for policy planification <i>For researchers:</i> facilitated data sharing and patient recruitment <i>For industry:</i> facilitated estimation of the market size; facilitated recruitment
Nomenclature in local languages (per country)	39,500 €	
Scientific Annotations (epidemiology, phenotype, genotype, natural history, disabilities)	316,000 €	<i>For patients:</i> know the manifestations and disabilities of their disease <i>For professionals:</i> facilitated examination, diagnosis and evaluation of patients <i>For policy-makers:</i> having reliable figures on epidemiology for policy planning <i>For researchers:</i> re-use of combinations of data for hypothesis making <i>For industry:</i> facilitated knowledge of market size; re-use of combinations of data for hypothesis making in R&D (pre-competitive)

*Cost calculation: Adjusted average cost of the current team in terms of PMs (average salary considered 6,000 € monthly)+ plus HR costs (2,000€/person) and infrastructure costs (5,000€ costs).

Orphanet current costs

Encyclopedia in English	355,500 €	<i>For patients:</i> understanding of diseases; support for communication with professionals/institutions on their or their relative's disease
Encyclopaedia in local languages (per country)	79,000 € - 158,000 €	<i>For professionals:</i> increased knowledge, support for diagnosis and referral, access to best practice guidelines <i>For researchers:</i> rapid access to the literature and state of the art of knowledge <i>For policy-makers:</i> facilitated access to state of the art per disease and to guidelines (standards of care)
Orphan Drugs database	39,500 €	<i>For patients and professionals:</i> identification of possible treatments <i>For decision-makers:</i> facilitation of analysis, anticipation of costs and follow-up of the therapies to be provided; gap analysis <i>For researchers:</i> facilitated analysis and hypothesis-generation together with other elements of the knowledge base; gap analysis; <i>For industry:</i> facilitated market surveillance and analysis per disease/groups/domains
International/trans-national expert resources catalogue	152 500 €	<i>For patients:</i> identification of expertise including cross-borders; tackling isolation <i>For professionals:</i> facilitated referrals including cross-border <i>For decision-makers:</i> follow-up of the allocation of resources, patients' needs coverage and gap analysis on healthcare planification and research agenda
National expert resources catalogue (per country)	79,000€ - 158,000 €	<i>For researchers:</i> facilitated networking by identification of experts and groups <i>For industry:</i> facilitated identification of experts, researchers, patient organisations for collaboration and recruitment (i.e. for clinical research)
IT developments	204,000€	<i>All stakeholders:</i> availability of information in dedicated formats (query facilities; re-use formats)

▶ *Cost calculation: Adjusted average cost of the current team in terms of PMs (average salary considered 6,000 € monthly)+ plus HR costs (2,000€/person) and infrastructure costs (5,000€ costs).

Scenario	Brief description	PROs	CONs
France only (Inserm)	Inserm funds Orphanet alone (permanent positions and IT infrastructure)	Sustainable national model Likely English nomenclature will persist.	Risk of losing permanent staff and expertise Lack of resource for competing for further projects. Drastic reduction of activities with negative impact on service offered to European citizens. No supporting activities for ERNs (save on a service contract basis).
Consortium of member states/other countries	France + other MS + other extra-EU countries having agreed on cooperation and sustainable funding of mutual benefit.	Sustainable transnational core activities are defined, funded and are further developed according to participant countries plans.	Requires lengthy negotiation with the respective governments. Risk of decreasing support to ERNs. Lose of European Commission flagship status.
Member states and EC	MS and EC steadily co-fund Orphanet for mutual benefit and cross-borders actions.	Sustainable multiannual model Core activities persist and are further developed according to participant countries and EU plans. Full support provided to ERNs.	Requires lengthy negotiation with the respective governments and the European Commission. Orphanet new statute needs to be defined The European Commission's funding is project based and would not meet the needs of a sustainable funding solution.
EC only	Orphanet becomes a European structure, hosted and fully funded by the EC. No national funding provided.	Core activities are maintained when re-developed, according to resources.	Data and technology transfer agreement needed from Inserm to EC. Re-creation of a database, IT infrastructure and team needed, with risk of losing knowledge, know-how and interruption of service. A major implication for the JRC's resources and availability of staff for this kind of work.

Time for discussion ... and action

- ▶ **Question 1:** My country might envisage a sustained financial contribution to the Orphanet: *yes/no*. If the reply is yes, please provide an estimate of the financial contribution in monetary terms (euros).
- ▶ **Question 2:** My country may envisage a sustained non-financial contribution such as seconding an expert or providing in-kind contribution to the Orphanet: *yes/no*.
- ▶ If yes, please define the specific contribution.
- ▶ **Questions 3:** My country may envisage to support Orphanet in other way i.e. contribute to activities / core functions (*yes/no*).
- ▶ Please explain what specific form of support is considered.



Thank you for your attention

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