Orphanet is the reference portal for information and documentation on rare diseases (RD) and orphan drugs (OD). Orphanet provides freely available, user-friendly web-based access to medically validated, comprehensive information, thus improving knowledge and contributing to the accurate diagnosis and appropriate care and treatment of patients with RD. Orphanet was originally established in 1997 by the French Ministry of Health and the French National Institute of Health and Medical Research (Institut National de la Santé et de la Recherche Médicale, INSERM) as a pilot project. Today, it is a broad partnership of public and private institutions[1] including short-term funding from the European Commission, currently via the Orphanet Europe Joint Action 2011-2014 (20102206).

Orphanet is run by a consortium of European partners. The coordination team, located at the same Rare Disease Platform as EURORDIS in Paris, is in charge of the infrastructure and produces content for the free access portal including:

- Inventory of over 6,000 RD cross-referenced with other nomenclature systems
- Online encyclopedia for over 3,000 diseases
- Hierarchical disease classification system (according to literature or established in-house)
- Unique and stable Orpha number, to code each disease in the database
- Search facility by clinical signs and symptoms
- Inventory of drugs for RD
- Emergency and clinical guidelines for RD.

Currently available in six languages (English, French, German, Italian, Portuguese and Spanish), the portal will continually be translated into additional languages including Dutch, Finnish, Polish and Russian in the near future.

Each of the national Orphanet teams collects data in their country and contributes to the directory of expert services including:

- Expert clinics
- Medical laboratories
- Patient organisations
- Research projects
- Clinical trials
- Registries and biobanks
- Research and reference networks
- Experts
- Specific national websites

Since 2011, Orphanet has become global, with the arrival of Canada and Australia. Negotiations are underway with Argentina, Brazil, Chile, China, Japan and Russia. Currently, nearly 40 countries take part in the consortium.

WHY IS THIS TOOL REQUIRED?

As stated in the European Commission's Communication, "Rare Diseases: Europe's Challenges"[2], one key element for improving diagnosis and care in the field of RD is to provide and disseminate accurate information on RD and OD in a format adapted to the needs of professionals and patients. The Commission ensures that this information will continue to be available at the European level, in particular by building on the Orphanet database.

The Commission's Council Recommendation[3] on an action in the field of RD following this communication recommends that Member States (MS):

- Ensure that RD are adequately coded and traceable in all health information systems, encouraging recognition of the disease in the national healthcare and reimbursement systems based on the International Classification of Diseases (ICD) while respecting national procedures.
- Contribute actively to the development of the EU inventory of RD based on the Orphanet network and other existing networks

1 - Orphanet is financially supported by : French Ministry of Health (Direction Générale de la Santé), INSERM (Institut National de la Santé et de la Recherche Médicale), European Commission (DG Public Health and Consumers Protection and DG Research), French Muscular Dystrophy Association (Association Française contre les Myopathies - AFM), the Fundation of the French pharmaceutical companies association (Les Entreprises du Médicament; LEEM), the Caisse Nationale des Solidarités pour l'Autonomie (CNSA), and GSK.
2 - COM (2008) 679 Communication from the Commission to the European Parliament, the Council the Economic and Social Committee and the Committee of the Regions on Rare Diseases: Europe's challenges.
3 - Council Recommendation of 8 June 2009 on an action in the field of rare diseases.

The European Project for Rare Diseases National Plans Development (EUROPLAN) recommendations[4] further underscore the importance of promoting and contributing to a common EU inventory of RD like Orphanet in the context of national plans for RD. It is additionally recommended that "the cross-referencing of RD is carried out across the different classification systems in use in each country, ensuring coordination and coherence with European initiatives, such as reference to the Orpha-code[5]". EURORDIS supports these recommendations.

Why is Orphanet important to rare disease patients?

For RD in particular, validated information and expertise do not always exist or information is often scattered. Orphanet was established to improve knowledge on RD with the ultimate aim of improving diagnosis, management and treatment of patients with RD.

Centralisation of RDs and OD information and resources via the Orphanet portal brings added-value to the collection of data by:

- Establishing a critical mass of financial support and expertise to maintain the documentation of RD and guarantee accessible information on expert resources in participating countries
- Allowing each Member State and its citizens to benefit from the core Orphanet infrastructure and an opportunity to access a national portal at a marginal cost
- Improving the coverage and quality of information on RDs
- Improving information, identification and knowledge on RDs necessary to improve diagnosis and care of patients
- Providing evidence for priority setting in research and health care services at the European and national levels
- Strengthening cooperation between MS
- Contributing to a common approach towards prevention, diagnosis and care of patients with RDs through the sharing of expertise
- Improving the traceability of RDs in health information systems and the recognition of each RDs in national health and reimbursement systems by revising coding and classification. Orphanet has, therefore, developed its own coding (Orpha code), composed of a unique and stable Orpha number for each rare disease. Since December 2012, the Orpha code system has been implemented in the French hospital system database. In addition Orphanet was commissioned by the World Health Organisation[6] to provide data for the revision of the next ICD.

Lack of knowledge and expertise remains an obstacle for RD patients to receive timely diagnosis, appropriate follow-up care and access to social assistance. In reality, no general practitioner is expected to, or capable of, identifying all 5,000- 8,000 RD. In support, Orphanet serves as the only centralised European reference to help improve diagnosis and follow-up care of RD patients.

The role of patient organisations in delivering and processing information on RD resources alongside health professionals is of the utmost importance. Patient groups play a pivotal role as legitimate and trusted sources of information of patient-generated knowledge and rely strongly on the existence of and access to information from such a reliable and validated source such as Orphanet. Amongst Orphanet website users[7], over two-thirds are professionals and nearly one-third patients and representatives. Users most frequently seek information on a specific RD, on RD in general or information on RD testing laboratories and most report the information "useful" to "very useful".

How to support Orphanet

- Continued and sustainable European Commission support for the core infrastructure of Orphanet remains imperative. Additional partnerships with relevant databases to improve the coverage and quality of data should also be encouraged and supported.
- Commitment of national teams, not only fully responsible for collecting and validating information on expert services, but also for dissemination of information on national policies and initiatives has been formalised through the Orphanet Europe Joint Action and should be sustained.
- Support of national governments, other local public (and private) partnerships, and national strategies to support contribution to an EU inventory of RD such as Orphanet, will contribute to an adequate definition, recognition, and classification of RD.

National and local patient groups are particularly well-placed to identify local experts and resources contributing to the centralised RD database and to advocate for the support of such activities in the context of a national plan on RD.

Related Issues

- Orphanet is not an institution; it is a dynamic, academic network currently largely supported by short-term grants and partnerships.
- Lack of long-term EU funding affects the sustainability of Orphanet staff and work.
- Existing funding is not enough to sufficiently update data. With its ever-evolving content, it is challenging to ensure the most up-to-date information.
- Maintaining security of data is a challenging responsibility.
- Translation of the Orphanet content is especially important for patients, as well as professionals.
- Quality control is important and is improving with increasing partnerships that allow cross referencing.

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4 - Recommendations for the development of strategic plan for rare diseases including methodological guidance. 8 July, 2010.
5 - Each rare disease identified in the Orphanet database is given an Orpha-code corresponding to the 10th international classification of diseases established by the World Health Organization (WHO) where available.
6 - Revision of the International Classification of Diseases. http://www.who.int/classifications/icd/ICDRevision/en/index.html
7 - « Orphanet - 2012 Activity Report », Orphanet Report Series, Reports collection, February 2013. http://www.orpha.net/orphacom/cahiers/docs/GB/ActivityReport2012.pdf

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