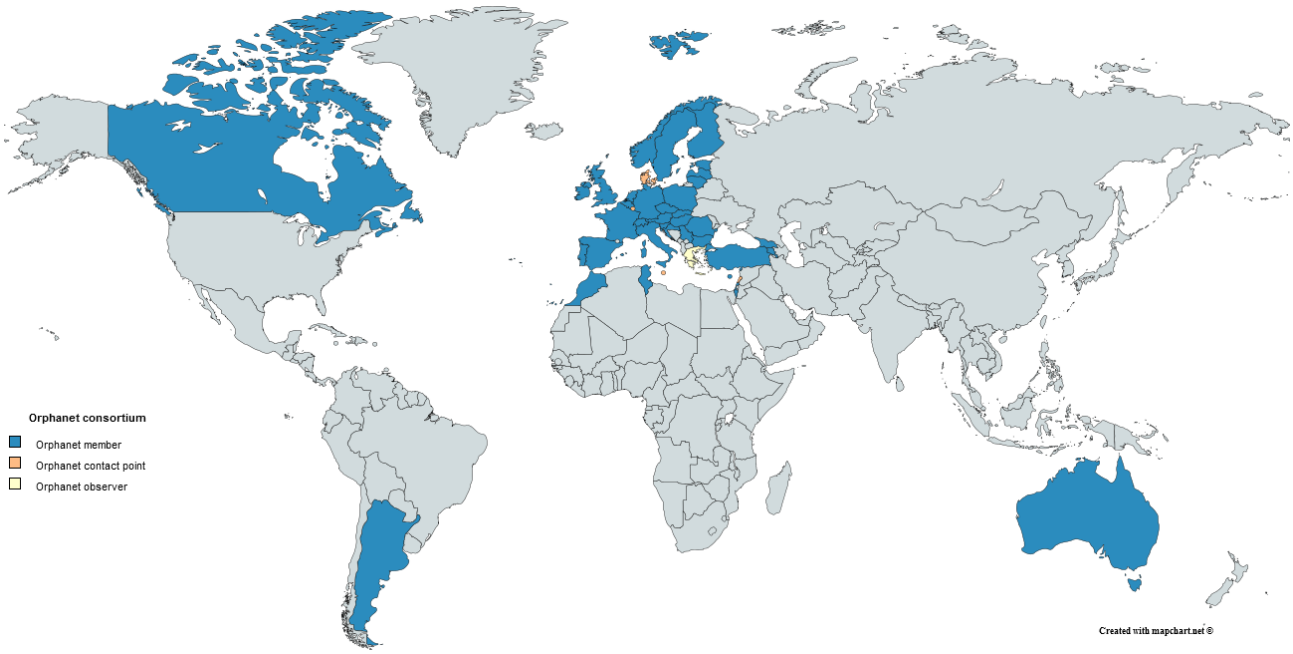
http://www.orpha.net/consor/cgi-bin/Education_AboutOrphanet.php?lng=EN

Orphanet: The portal for rare diseases and orphan drugs

About Orphanet

- Our missions
- Our services
- The Orphanet Consortium
- Our governance
- Our funders
- Our quality commitment



Our missions

Orphanet was established in France in 1997 at the advent of the internet in order to gather scarce knowledge on rare diseases so as to improve the diagnosis, care and treatment of patients with rare diseases. This initiative became a European endeavour from 2000, supported by grants from the European Commission: Orphanet has gradually grown to a Consortium of 40 countries, within Europe and across the globe.

Over the past 20 years, Orphanet has become the reference source of information on rare diseases. As such, Orphanet is committed to meeting new challenges arise from a rapidly evolving political, scientific, and informatics landscape. In particular, it is crucial to help all

audiences access quality information amongst the plethora of information available online, to provide the means to identify rare disease patients and to contribute to generating knowledge by producing massive, computable, re-usable scientific data.

Orphanet works towards meeting three main goals:

- **Improve the visibility of rare diseases in the fields of healthcare and research by maintaining the Orphanet rare disease nomenclature (ORPHAnumbers): providing a common language to understand each other across the rare disease field.**
  In a global community, we need to understand each other, although we may not speak the same language. A stable nomenclature, cross-referenced with other international terminologies is therefore essential. In order to improve the visibility of rare diseases in information systems, Orphanet has developed, and maintains, a unique, multi-lingual nomenclature of rare diseases, around which the rest of our relational database is structured. Each disease is assigned a unique ORPHAnumber: integrating this nomenclature in health and research information systems is essential in ensuring that rare diseases are visible. This nomenclature is aligned with other terminologies: OMIM, ICD, SNOMED-CT, MedDRA, UMLS, MeSH, GARD. This cross-referencing is a key step towards the interoperability of databases.

- **Provide high-quality information on rare diseases and expertise, ensuring equal access to knowledge for all stakeholders: orientating users and actors in the field in the mass of information online.**
  Rare diseases patients are scattered across the globe, as are rare disease experts. Orphanet provides visibility to experts and for patients by providing access to a directory of expert services in 40 countries by disease, such as centres of expertise, laboratories and diagnostic tests, patient organisations, research projects and clinical trials. This data promotes networking, tackles isolation and helps foster appropriate referrals. Orphanet draws on the expertise of professionals from across the world to provide scientific data on rare diseases (gene-disease relationship, epidemiology, phenotypic features, functional consequences of the disease, etc.). In addition, Orphanet produces an encyclopaedia of rare diseases, progressively translated into the 7 languages of the database (English, French, Spanish, Italian, German, Dutch, Portuguese) with texts also currently available in Polish, Greek, Slovak, Finnish and Russian, freely available online. Orphanet integrates and provides access to quality information produced around the world, such as clinical practice guidelines and information geared to the general public.

- **Contribute to generating knowledge on rare diseases: piecing together the parts of the puzzle to better understand rare diseases.**
  To develop and curate the scientific data in the Orphanet database, Orphanet works with experts from around the globe, from health care professionals and researchers, to patient representatives and professionals from the medical -social sector. The wealth of data in Orphanet and the way this data is structured allows additional knowledge to be generated, helping

to piece together data that at times can resemble pieces of an irresolvable puzzle. Integration of this data adds value and renders it interpretable. Orphanet provides standards for rare disease identification, notably via the Orphanet nomenclature, an essential key for interoperability. Orphanet provides integrated, re-usable data essential for research on the www.orphadata.org platform and as a structured vocabulary for rare diseases, the Orphanet Ontology of Rare Diseases (ORDO). These resources contribute to improving the interoperability of data on rare diseases across the globe and across the fields of health care and research. They are being integrated in several bioinformatics projects and infrastructures around the world in order to improve diagnosis and treatment. Orphanet is committed to networking with partners across the globe order to help piece together the parts of this puzzle.

The integral role played by Orphanet in the research and care spheres has led to its recognition as an IRDiRC Recognised Resource, and integration in the French node of ELIXIR, a European Research Infrastructure Consortium uniting Europe's leading life science organisations. Orphanet and the ORPHA nomenclature are also cited as key resources in every European legislative text on rare diseases and as key measures in many national plans/strategies for rare diseases.

Our services

Orphanet offers a range of freely accessible services:

- An inventory of rare diseases mapped with resources as OMIM, ICD10, MeSH, MedDRA, GARD and UMLS and a classification of diseases elaborated using existing published expert classifications. Diseases are also annotated with phenotypic features and frequency using HPO.
- An encyclopaedia of rare diseases in English, progressively translated into the other languages of the website.
- An inventory of orphan drugs at all stages of development.
- A directory of expert resources, providing information on expert clinics, medical laboratories, ongoing research projects, clinical trials, registries, networks, technological platforms and patient organisations, in the field of rare diseases, in each of the countries in Orphanet's consortium.
- An assistance-to-diagnosis tool allowing users to search by signs and symptoms.
- An encyclopaedia of recommendations and guidelines for emergency medical care and anaesthesia.
- A fortnightly newsletter, OrphaNews, which gives an overview of scientific and political current affairs in the field of rare diseases and orphan drugs, in English, French and Italian.
- A collection of thematic reports, the Orphanet Reports Series, focusing on overarching themes, directly downloadable from the website.
- A platform, Orphadata, providing high-quality datasets related to rare diseases and Orphan Drugs, in a reusable and computable format.

- The Orphanet Rare Disease Ontology (ORDO), a structured vocabulary for rare diseases derived from the Orphanet database, capturing relationships between diseases, genes and other relevant features. ORDO provides integrated, re-usable data for computational analysis.

Orphanet and ORDO are IRDiRC Recognized Resources.

The Orphanet Consortium

Orphanet is a multi-stakeholder, global consortium of 40 countries, coordinated by the core resource team at the French National Institute of Health and Medical Research (INSERM) in Paris.

List of Consortium Members

Orphanet national team

National teams are responsible for the collection of information on expert centres, medical laboratories, ongoing research and patient organisations in their country and translation in their language if funding available. All Orphanet teams work according to the Orphanet Standard Operating Procedures.

Each national team maintains a national entry point to Orphanet, providing latest news and updates concerning national activities, in the national language of the country concerned.

Orphanet national websites

Orphanet coordinating team

The coordinating team (Inserm US14) is responsible for the coordination of consortium activities, the hardware and software aspects of the project, the database of rare diseases and the production of the encyclopaedia, as well as the training of all members of the consortium and the quality control of the directory of resources in the participating countries.

The coordinating team is also in charge of updating the database in regards to medicinal products in development, from their designation stage to their marketing authorisation.

Our governance

International Level

- The Management Board, composed of country coordinators, is in charge of identifying funding opportunities, guiding the project to provide an optimum service for the end-users, and considering the inclusion of new teams as well as ensuring the continuity of the project. In the framework of the RD-Action Joint Action 2015-2018 most of the Orphanet activities are co- funded by the EC. As a result, the Orphanet Management board also refers to the General

Assembly of the RD-Action project (please refer to www.rd-action.eu for additional information).

External boards

- The International Advisory Board, composed of international experts , is in charge of advising the Management Board regarding the overall strategy of the project.
  Rules of procedures available here.
- The Genetic Advisory Board, is in charge of advising Orphanet on topics related to the gene database and the database of genetic tests and laboratories.
  Rules of procedures available here.

These boards discuss the evolution of the project in scope and depth; ensuring its coherence and its evolution, in relation to technological developments and to the needs of its end-users, as well as its sustainability.

National level

- The National Advisory Board is composed by members nominated by the appropriate legitimate institutions which are defined at country level. The board members contribute with their expertise to Orphanet at country level.

Our funders

The infrastructure and coordination activities are funded jointly by Inserm (the French National Institute of Health and Medical Research), the French Directorate General for Health, and the European Commission through RD-Action (677024), a Joint Action of the 3rd Health Programme of the European Union. Certain services are specifically funded by other partners. Orphanet's national activities are financed by national institutions and/or specific contracts.

Read more

Our quality commitment

Our quality control program

Data in Orphanet are manually curated and expert validated, following formalised procedures.

Orphanet is committed to providing quality data to its users, and as such a pre and/or post release quality control is performed at Orphanet in order to insure the quality of all the data registered. This quality control is performed according to a predetermined program, updated every year.

Our quality management system

In order to improve our operations and allow our users to assess the way in which we work, Orphanet is working towards ISO 9001 certification for its activities. In this frame work, Orphanet will publish procedures explaining the way in which we work, our workflow and exclusion/inclusion criteria for each type of data.

List of published procedures

- Quality charters
- About orphan drugs
- About Orphanet
- About rare diseases
- Orphanet Reports series/Procedures
- Mobile application
- Orphanet Tutorials

- Homepage
- Other information
- About Orphanet

- About Orphanet
- Orphanet funding
- Orphanet national websites