orphanet

The international rare disease and orphan drug database bridging healthcare & research

A global consortium improving visibility, awareness, information and knowledge in the field of rare diseases

www.orpha.net

IRDiRC RECOGNIZED RESOURCES

Inserm

Co-funded by the Health Programme of the European Union

RD ACTION

Orphanet in numbers

Database content and website

A freely accessible site available in **7 languages**
47 million pages viewed in 2016
12 million PDF documents downloaded in 2016
An **IRDiRC Recognized Resource**

Diseases

6,084	rare diseases with unique identifiers : ORPHA numbers
3,715	genes for 3,566 rare diseases
2,630	diseases indexed with HPO terms
5,299	diseases annotated with prevalence/incidence data

Rare disease summaries in 11 languages

4,089	English
3,314	Italian
3,299	French
3,159	German
3,074	Spanish
1,182	Portuguese
663	Dutch
167	Finnish
647	Polish
424	Greek
103	Slovak

Directory of expert resources in 40 countries worldwide

21,791	professionals
7,230	expert centres
2,537	patient organisations
1,676	medical laboratories
42,982	diagnostic tests
1,856	research laboratories
2,475	research projects
2,455	clinical trials
744	patient registries
621	mutation databases
142	biobanks

Data from Orphanet 2016 Activity Report

Users

Around **1,100,000 visitors** per month from **232 countries**

48% health professionals
22% patients, families and support groups
As well as **researchers, industry, policy makers, students**
Most appreciated products: **texts on diseases, list of diseases, epidemiological data** and **clinical guidelines** *

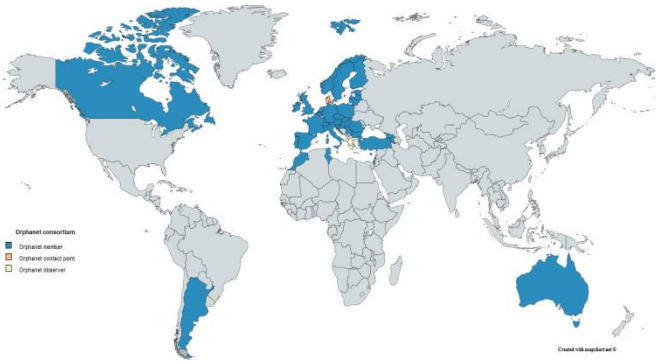
* Annual Orphanet Users' Survey 2017

Our mission statement

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.

2017 marks Orphanet's 20th anniversary: with age comes new challenges resulting from a rapidly evolving political, scientific, and informatics landscape. It is now 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.

* A rare disease in the Orphanet database is one with a prevalence of not more than 1 per 2000 inhabitants in Europe, as defined in Regulation (EC) N°141/2000 of the European Parliament and of the Council of 16 December 1999 on orphan medicinal products



Orphanet members and contact points around the globe

Composition of Orphanet Consortium Members

50%	University hospital/hospital
14%	Ministry of Health
14%	Research centre
12%	Public health institute
7%:	Foundation/association other than patient organisation
3%:	Patient organisations

WORKING ACROSS BORDERS

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

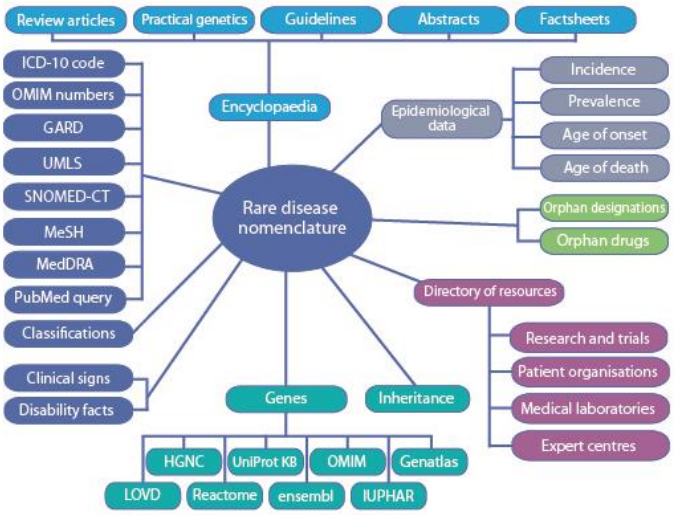
The INSERM **core resources team maintains the database** as well as the Orphanet rare disease **nomenclature** (each with a unique ORPHA number), **classifications**, **encyclopaedia**, and the **Orphanet Ontology of Rare Diseases (ORDO, an IRDiRC Recognized Resource)**. Countries in the consortium contribute to the database of **expert resources** in each country and to the **encyclopaedia**.

Orphanet's core resources are funded by the INSERM, the French Ministry of Health, and the European Commission. Orphanet's national activities are financed by national institutions and/or specific contracts.

Orphanet is governed by a Management Board of Consortium members, overseen by a multi-stakeholder International Scientific Advisory Board.

OUR COMMITMENT TO THE RARE DISEASE COMMUNITY

- Contribute to improving knowledge on rare diseases
- Provide high-quality information on rare diseases, and ensure equal access to knowledge for all stakeholders
- Maintain the Orphanet rare disease nomenclature (ORPHA numbers), essential in improving the visibility of rare diseases in health and research information systems



A relational database centred around a unique, clinical rare disease nomenclature, cross-referenced with other resources

2016 Highlights

PARTNERSHIP WITH NIH-NCATS GENETIC AND RARE DISEASE INFORMATION CENTER

A partnership between Orphanet and the Genetic and Rare Disease Information Center (GARD), hosted by the National Institutes of Health – National Center for Advancing Translation Sciences (NIH-NCATS) was established in 2016.

GARD Genetic and Rare Diseases Information Center

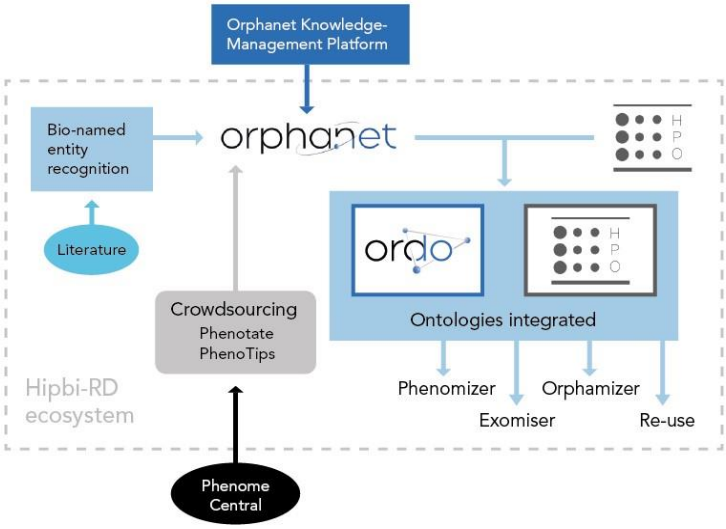
The aim of this partnership is to **mutualise efforts so as to provide the audiences of both sites with the most complete and up-to-date information on rare diseases**.

In a first step, the **Orphanet and GARD nomenclatures are being aligned**, so as to allow cross-referencing between the two resources. In a second step, **summary texts from Orphanet (along with a link to the relevant Orphanet disease page and the Orphanet logo) will be included in GARD for the diseases for which GARD does not have a text**. This cross-Atlantic partnership will improve the visibility of Orphanet in the United States and is just one of the ongoing efforts to integrate the Orphanet nomenclature, in particular, into the different rare disease-related resources maintained by the NIH (e.g. UMLS, ClinVar, MedGen, GTR).

HARMONISING PHENOMICS INFORMATION: COORDINATION OF E-RARE3 PROJECT HIPBI-RD

From January 2016, Orphanet coordinates the three-year ERare3 ERANET project HIPBI-RD - **Harmonising phenomics information for a better interoperability in the RD field**: www.hipbi-rd.net

Harmonisation of phenomics information including disorders and phenotype traits that are stored in different supports (patient records, databases, registries) in a non-standardised way, is a cornerstone for the production of sound data necessary to foster research.



It is aimed to **provide the community with an integrated, RD-specific informatics ecosystem that will harmonise the way phenomics information is stored in databases** and in patient files worldwide, and thereby contribute to interoperability. This ecosystem will consist of a suite of tools and ontologies, optimised to work together, and available to clinicians and scientists through commonly used software repositories.

Additionally, the ecosystem will improve and streamline the interpretation of variants identified through exome and full genome sequencing by harmonising the way phenotypic information is collected.

2016 Highlights

ORPHANET AT THE LAUNCH OF THE UN NGO COMMITTEE FOR RARE DISEASES

In November 2016, the **NGO Committee for Rare Diseases** was launched at the United Nations in New York under the patronage of the Conference of NGOs in Consultative Relationship with the United Nations (CoNGO).

Orphanet was presented as a global resource for rare diseases at this meeting at which the Orphanet Consortium affirmed their support of the goals of the Committee.

Ana Rath, on behalf of the Orphanet Consortium signed the **Founding Act on "Rare Diseases and the UN Sustainable Development Goals".** Orphanet will soon formally join the committee.

NGO COMMITTEE FOR RARE DISEASES

LAUNCH OF THE EUROPEAN JOINT ACTION ON RARE CANCERS

Orphanet is now member of the **European Joint Action on Rare Cancers** (www.jointactionrarecancers.eu) that aims to integrate and maximise efforts of the European Union (EU) Commission, EU Member States and all stakeholders to advance quality of care and research on rare cancers.

Orphanet is involved in a number of the work packages, in particular on issues such as the collection of **epidemiological data of rare cancers, information on expert centres for rare diseases, clinical practice guidelines, and nomenclature and classifications.**

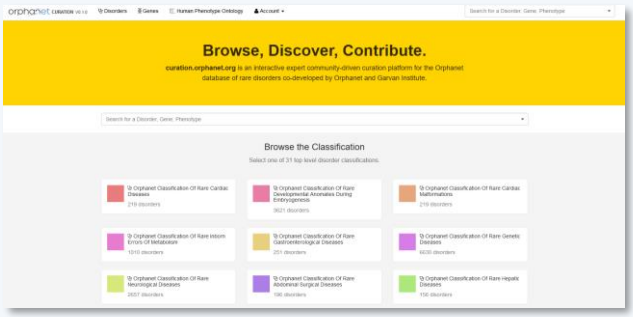
JARC

Cross-talk with the European Joint Action on Rare Diseases, RD-Action, coordinated by Orphanet, is also assured.

ORPHANET KNOWLEDGE MANAGEMENT PLATFORM

The development of the **Orphanet Knowledge Management Platform** (https://curation.orphanet.org/) was carried out in 2016 by partners of the RD-Action consortium in Australia at the Garvan Institute.

This platform allows experts in the field of rare diseases to **help curate the scientific data** in the Orphanet database in a traceable and transparent manner.



NEW REPRESENTATION OF DIAGNOSTIC TESTS

A new representation of diagnostic tests went live on the Orphanet website 2016, expanding this dataset to **give more information to users** and **allowing them to filter results** by **speciality/objective, technique, purpose, quality management data** (accreditation and participation in EQA schemes), lab and test quality (accredited/participating in EQA), as well as the country in which the laboratory is located.

TRANSITION TO A DISTRIBUTED MODEL OF PRODUCTION

From 2016, the transition to a more distributed model for the production of the encyclopaedia was launched, with **members of the Orphanet Slovakia and Ireland teams assuming responsibility for part of the production of texts in English.** Other countries will join this effort.

Our key objectives

IMPROVING VISIBILITY

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 **ORPHA number** (or ORPHA code): integrating these codes in health and research information systems is essential in ensuring rare diseases are visible and that different systems can work together. 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.**

Rare diseases patients are scattered across the globe, as are rare disease experts. Orphanet **provides visibility to experts and 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.**

PROVIDING INFORMATION

To **serve the needs of a global audience**, Orphanet draws on the **expertise of professionals from across the world**, 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 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 tailored to the general public.

GENERATING KNOWLEDGE

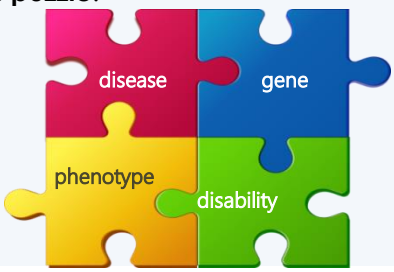
To **develop and curate the scientific data** in the Orphanet database, Orphanet works with **experts from around the globe**, from health care professionals and researchers, to patient representatives and professionals from the medical - social sector.

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 can sometimes seem like pieces of an irresolvable puzzle. **Integration of 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 key 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**.



A key policy resource

ORPHANET DATA INFORMING RARE DISEASE POLICY – ORPHANET DATA RESPONDING TO POLICY NEEDS

Orphanet coordinates the on-going European Joint Action on Rare Diseases (RD-Action), and contributes to the work of the IRDiRC Scientific Secretariat: **Orphanet and its data is key to informing healthcare and research policy.** The presence of Orphanet in our partner countries has also proved to be an important catalyst in the improving the awareness of rare diseases, in particular as regards the national political agenda.

A **virtuous, symbiotic cycle has evolved, connecting Orphanet's data and rare diseases policy**:

- Orphanet **informs policy** by providing aggregated data (Orphanet Report Series), and data for annual **reports on the State of the Art of rare disease policies in Europe**, and the **State of Play of rare disease research.** Orphanet data has proved indispensable in **implementing policy measures**, such as the official designation of centres of expertise for rare diseases at national level by identifying where experts and expertise is located in a country.
- Orphanet's data can **respond to policy needs**: for example, data on laboratories carrying out diagnostic tests for rare diseases in Orphanet has evolved in order to facilitate cross-border testing.
- In 2009, the Council of the European Union recommended that all EU Member States develop **national plans or strategies** to structure their response to the challenges presented by rare diseases. National contribution to the Orphanet network was recommended, and as a result many **national plans or strategies developed to date mention Orphanet and Orpha codification as key measures.**
- Orphanet is also cited in the **EU Cross-Border Healthcare Directive (2011)** as a key resource to assist *« health professionals […] in correct diagnosis of rare diseases».*
- **Orphanet's activities and resources are also supported by a number of key policy recommendations** issued by expert committees at the European Commission, on topics such as codification of rare diseases, centres of expertise, European Reference Networks and cross-border genetic testing.

www.rd-action.eu

RD ACTION
DATA AND POLICIES FOR RARE DISEASES

AT THE CROSSROADS OF THE RARE DISEASES WORLD: ORPHANET AS AN INTEGRATED PLATFORM

Orphanet is at the crossroads of multiple spheres implicated in issues arising from the rare disease challenge. Its role as an integrated, global platform is to **build the bridges necessary to help advance policy, healthcare and research by delivering global information, data and knowledge.** As such, Orphanet's networking capacity is key in **bringing together different countries and different languages**, as well as the **different types of expertise** emanating from different domains (healthcare, research, genetics, medical – social sector, patient organisations).

The scientific and informatics revolution underway offers **possibilities to further integrate data in Orphanet to facilitate the connection between the worlds of healthcare and research**. This same revolution also offers opportunities for Orphanet to become truly global and better respond to needs of users' across the world and across stakeholder groups.

Our commitment to the global rare disease community

Rare diseases know no borders, and the challenges faced by people living with rare diseases share commonalities across many different diseases and regions of the world. No nation, no continent can tackle the challenges around the diagnosis, treatment and care of people living with rare diseases alone.

This challenge is a global one, and all stakeholders must cooperate at a global level in order to address the challenge and provide answers. The synergies resulting from cooperation across all stakeholder groups and across all continents are essential in this effort.

Global data is essential for global policies. Orphanet will continue to fulfil its missions at the service of the global community by:

- **Ensuring equality of access to high quality information, essential in the empowerment of people living with rare diseases, and in the generation and improvement of scientific knowledge on rare diseases**
- **Promoting a common language and the means to integrate data on rare diseases in order to tackle the fragmentation of research into rare diseases**
- **Providing data essential for rare disease policy development, and remaining attentive to the needs of stakeholders in terms of rare disease data**

"The Orphanet Consortium supports the work of the NGO Committee for Rare Diseases, and will, to the best of its ability, support the endeavour to improve the recognition and inclusion of rare diseases into the UN Sustainable Development Goals, and to strengthen the voice of research and medical institutions in all subsequent UN policy discussions around rare diseases."

Orphanet Management Board, 4 November 2016

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For more information about our objectives, achievements and partnerships, consult the annual Orphanet Activity Report at www.orpha.net

Get involved!

There are different ways you can help Orphanet improve its services for the rare disease community:

- **Contribute: make your activities related to RD visible and register via Orphanet's homepage**
- **Curate: experts will soon be able to contribute and give their feedback on our data at https://curation.orpha.net**
- **Sustain: sponsor one of Orphanet's activities**
- **Give your feedback : participate in our annual users' survey**

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This Leaflet is part of joint action '677024 / RD-ACTION' which has received funding from the European Union's Health Programme (2014-2020).The content of leaflet represents the views of the author only and is his/her sole responsibility; it can not be considered to reflect the views of the European Commission and/or the Consumers, Health, Agriculture and Food Executive Agency or any other body of the European Union. The European Commission and the Agency do not accept any responsibility for use that may be made of the information it contains.
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