

orphanet

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



2017

ORDO patient associations
encyclopaedia clinical trials
classifications coding
laboratories
genes ontology
handicap
registries
data
Orphanet
database orphan drugs
mapping rare diseases
Orphacode biobanks
terminologies nomenclature
expert centres Orphadata
diagnostic tests epidemiology
guidelines research
registries experts

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

www.orpha.net



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the Health Programme
of the European Union



Inserm

La science pour la santé
From science to health



IRDiRC
RECOGNIZED
RESOURCES



**THE
HUMAN VARIOME
PROJECT**
RECOMMENDED SYSTEM



**Global Alliance
for Genomics & Health**
Collaborate. Innovate. Accelerate.

Orphanet in numbers

Database content and website

A freely accessible site available in **7 languages**

30 million pages viewed in 2017

7.8 million PDF documents downloaded in 2017

An **IRDiRC Recognized Resource** and **HVP Recommended System**



Diseases

6,151	rare diseases with unique identifiers : ORPHA numbers
3,898	genes for 3,739 rare diseases
2,963	diseases annotated with HPO terms
5,648	diseases annotated with prevalence/incidence data

Rare disease summaries in 12 languages

4,595	English
3,334	Italian
3,282	French
3,165	German
3,169	Spanish
1,180	Portuguese
662	Dutch
167	Finnish
831	Polish
423	Greek
103	Slovak
255	Russian

Directory of expert resources in 41 countries worldwide

22,184	professionals
7,400	expert centres
2,599	patient organisations
1,648	medical laboratories
44,129	diagnostic tests
1,745	research laboratories
1,997	research projects
1,792	clinical trials
727	patient registries
270	mutation databases
142	biobanks

Data from Orphanet 2017 Activity Report



Around **782'000 visitors per month** from **235 countries**

46 % health professionals

25 % patients, families and support groups

As well as **researchers, industry, policy makers, students**

Most appreciated products: **list of diseases, texts on diseases, epidemiological data** and **clinical guidelines** *

* Annual Orphanet Users' Survey January 2018

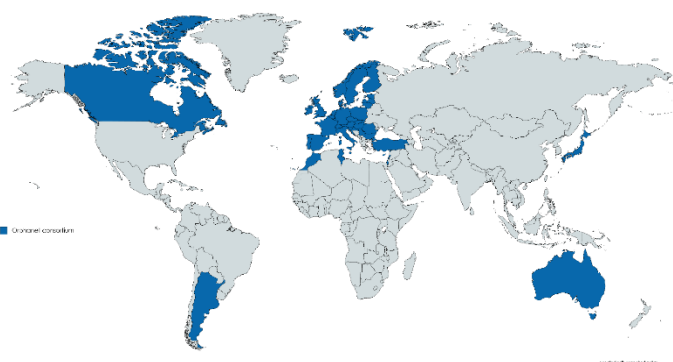
Users

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 (EC): Orphanet has gradually grown to a Consortium of 41 countries, within Europe and across the globe.

Orphanet continues to face 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 Consortium Members 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

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

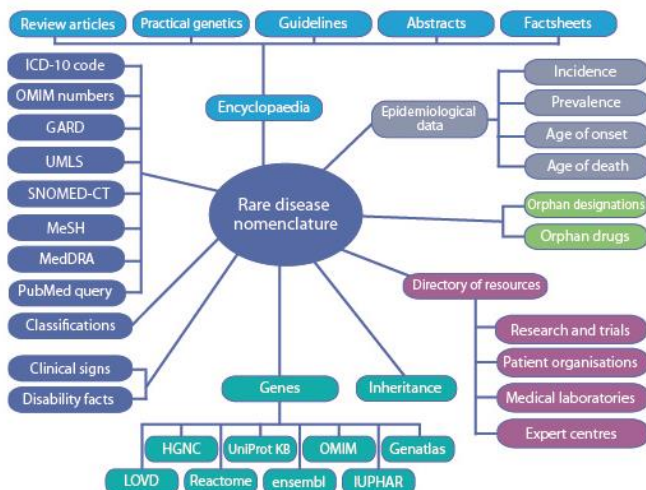
WORKING ACROSS BORDERS

Orphanet is a **multi-stakeholder, global consortium of 41 countries**, coordinated by the INSERM (French National Institute for Health and Medical Research) in Paris.

The INSERM **coordinating team maintains the database** as well as core resources such 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 and HVP Recommended System)**. Countries in the consortium contribute to the database of **expert resources** in each country and to the **encyclopaedia**.

Orphanet's core activity is funded by the INSERM, the French Ministry of Health, and the EC. Orphanet's national activities are financed by the EC, 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.



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

2017 Highlights

ORPHANET CELEBRATES ITS 20TH ANNIVERSARY

2017 marked the **20th anniversary of the creation of Orphanet**.

To celebrate the occasion, Orphanet teams present and past, as well as institutional representatives

and Orphanet's strategic partners were invited to a dinner in Paris. The representatives of the European Commission and INSERM gave opening speeches, reaffirming their support of this essential resource for the rare disease community. Ségolène Aymé, the evening's guest of honour, was presented by Ana Rath, on behalf of Orphanet teams both past and present, a symbol of recognition and gratitude for her action and involvement as founder of Orphanet.



A NEW LOOK & FEEL FOR THE ORPHANET WEBSITE

The 20th anniversary of Orphanet was also the perfect occasion to give the Orphanet website a complete makeover.

The new look and feel of the Orphanet website was launched in March 2017. The new site aims to **improve the navigation**. The design is **responsive**, making it easier to consult on a range of different devices, and the lay out makes data easier to read.

Video tutorials were made available to ease the use of the new site.



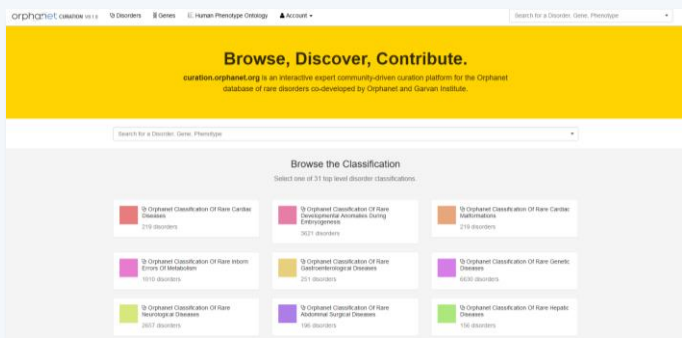
At the start of 2017, the **OrphaNews newsletter also experienced a complete makeover**. A new front and back office have improved the look and feel of the newsletter and also allow new functionalities.

Navigation has been improved through the construction of distinct sections, articles can be shared more easily, and the design is responsive on a range of different mobile devices. Users can now search the archives using an in-house thesaurus of terms, making it easier to find information concerning a certain subject or disease.

2017 Highlights

LAUNCH OF THE ORPHANET KNOWLEDGE MANAGEMENT PLATFORM

The **Orphanet Knowledge Management Platform** <https://curation.orphanet.org/> was launched in early 2017. Developed 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. A RD-Action workshop was held in April 2017 to present the platform to European Reference Networks, who will be key end-users of this tool.



The curation platform provides a different visualisation of the data in the Orphanet database, with icons next to editable data to **ease the sourcing of expert input**. Curators at Orphanet are alerted of the suggestion and a discussion between experts can be initiated using the platform. For the moment, experts have the possibility of registering via the platform, stating the diseases for which they have expertise, and providing suggestions to **curate the nomenclature, definitions and abstracts in the Orphanet database**. Other types of scientific data, such as genes, phenotypes and epidemiological data will be open for suggestions in a second step.

ERNs have been formally invited to curate data concerning their field of expertise using the platform as part of collaborative activities with Orphanet in 2017. The main fields of collaboration initiated include the Orphanet nomenclature and classifications, as well as the update of the expert services directory.

ORPHANET BECOMES A PARTNER IN SOLVE-RD: HORIZON 2020 PROJECT TO SOLVE UNSOLVED RARE DISEASES

Led by the University of Tübingen, the Solve-RD consortium www.solve-rd.eu, allocated €15 million by the European Commission Horizon 2020 programme, will work from 2018 to **improve the diagnosis of rare diseases**, hand in hand with the new European Reference Networks (ERN) on rare diseases.

The Orphanet coordinating team at INSERM is leading the work package in charge of **collecting standardised genetic and phenotypic information** concerning unsolved rare diseases cases from ERN cohorts and the European Genome Archive, and **developing an ontology of unsolved cases** that will work alongside the Orphanet Rare Disease Ontology (ORDO) and the HPO-ORDO Ontological Module (HOOM) in order to make **new diagnostic hypotheses**.



ORPHANET YOUTUBE TUTORIAL CHANNEL

In 2017 Orphanet launched a YouTube channel to host video tutorials aimed at **helping users understand the Orphanet nomenclature of rare diseases, and search for a disease/gene** on the website. Videos are available in English and French, with other languages (subtitles) added progressively.



JAPAN JOINS THE ORPHANET CONSORTIUM

The Orphanet Consortium officially extended its reach to Asia and welcomed its 41st member country, **Japan**. The Agency for Medical Research and Development (AMED) through the Translational Research Informatics Center (TRI) has been designated to fulfil this role, and is in the process of building the Orphanet Japan team.

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 **35 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 **8 languages** of the database (English, French, Spanish, Italian, German, Dutch, Portuguese, Polish) with texts also freely available in Greek, Slovak, Finnish and Russian 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)**. In the context of the HIPBI-RD (www.hipbi-rd.net) E-Rare3 ERA-NET project aimed at the harmonisation of phenomics information for a better interoperability in the RD field, the **HPO-ORDO Ontological Module (HOOM)** provides a bridge between the Human Phenotype Ontology and 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) www.rd-action.eu, 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.
- In 2017, Orphanet was recognised as having a **de facto monopoly** in its field in the 2018 Work Plan of the European Union's Third Health Programme (2014-2020), notably for its unique nomenclature of 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.

Orphanet will assume this role in the upcoming **European Joint Programme Co-Fund on Rare Diseases**, due to start in 2019, coordinated by INSERM.

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



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 can contribute and give their feedback on our data at <http://curation.orpha.net>
- **Sustain:** sponsor one of Orphanet's activities
- **Give your feedback:** participate in our annual users' survey

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