orphanet

2010 Activity Report

Table of contents

For any questions or comments, please contact us: *contact.orphanet@inserm.fr*

Overview of activities in 2010

Orphanet has become the reference portal for information on rare diseases and orphan drugs.

The site gives access to:

- An inventory of diseases including 5,832 diseases and classifications of these diseases developed using existing published expert classifications. Each disease is indexed with ICD10 and OMIM, and its 'identity card' includes the relevant prevalence class, age of onset class, mode of inheritance and associated genes. (At the moment, not every disease has a completed 'identity card').

- An encyclopaedia covering 2,878 rare diseases, written by world-renowned experts and peer-reviewed. Systematically produced in both English and French, this encyclopaedia is partly translated into German, Italian and Spanish.

- An inventory of orphan drugs at all stages of development, from EMA orphan designation to European market authorisation.

- A directory of specialised services in the 36 European partner countries, providing information on:
    - Specialised clinics and centres of expertise
    - Medical laboratories
    - Ongoing research projects
    - Clinical trials
    - Registries
    - Networks
    - Technological platforms
    - Patient organisations
    - Orphan drugs

- A range of other services for specific stakeholders:
    - For health care professionals: an assistance-to-diagnosis tool (search by signs and symptoms)
    - For professionals in the field of emergency health care: an encyclopaedia of emergency guidelines
    - For researchers and the pharmaceutical industry: availability of data from the database for research purpose
    - For all: a newsletters with both scientific and political content
    - For all: regularly published thematic studies and reports on overarching subjects, downloadable from the site: "the Orphanet Report Series"

1. Orphanet's audience

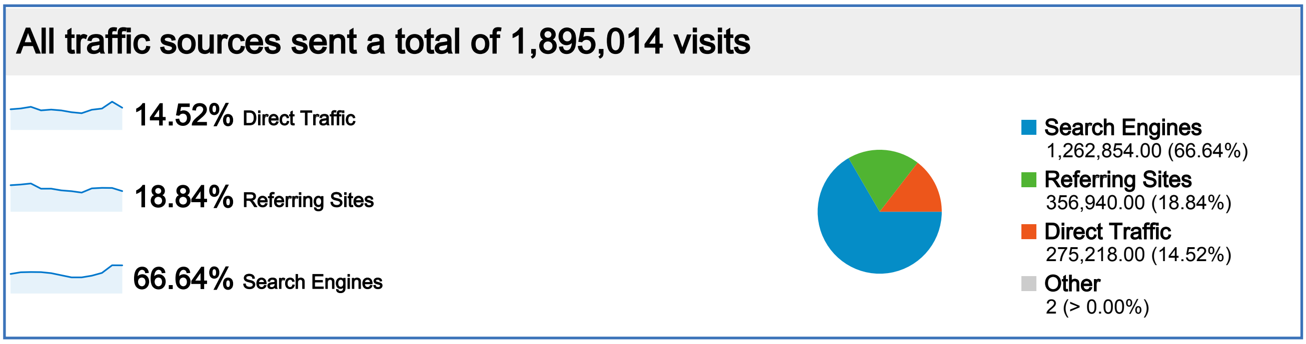
1.1. INDEXATION BY SEARCH ENGINES

According to Google, the prominence of the www.orpha.net site can be assessed by the number of results obtained by using the site name as a query, which is 457,000 responses. In comparison, NORD's site leads to 368,000 answers, GeneClinics 19,800 and INSERM 2,160,000.

Users mainly access the Orphanet website through search engines (66.6% of visits according to Google Analytics) and Google alone accounts for 62.2% of queries. Other sites generating traffic to Orphanet represent 18.8% of visits. The remaining visits are made via direct access (bookmarks, 14.5%).

The richness of our site means that a substantial quantity of visits is brought in through a sizable corpus of keywords (rather than just certain predominant keywords). The keyword primarily used to access our site is simply «Orphanet» which represents 13.7 % of visits. The indexation of our site is of the "long tail" type: more than 400,000 different keywords generate traffic to the site.

Distribution of the traffic sources



All traffic sources sent a total of 1,895,014 visits

14.52% Direct Traffic

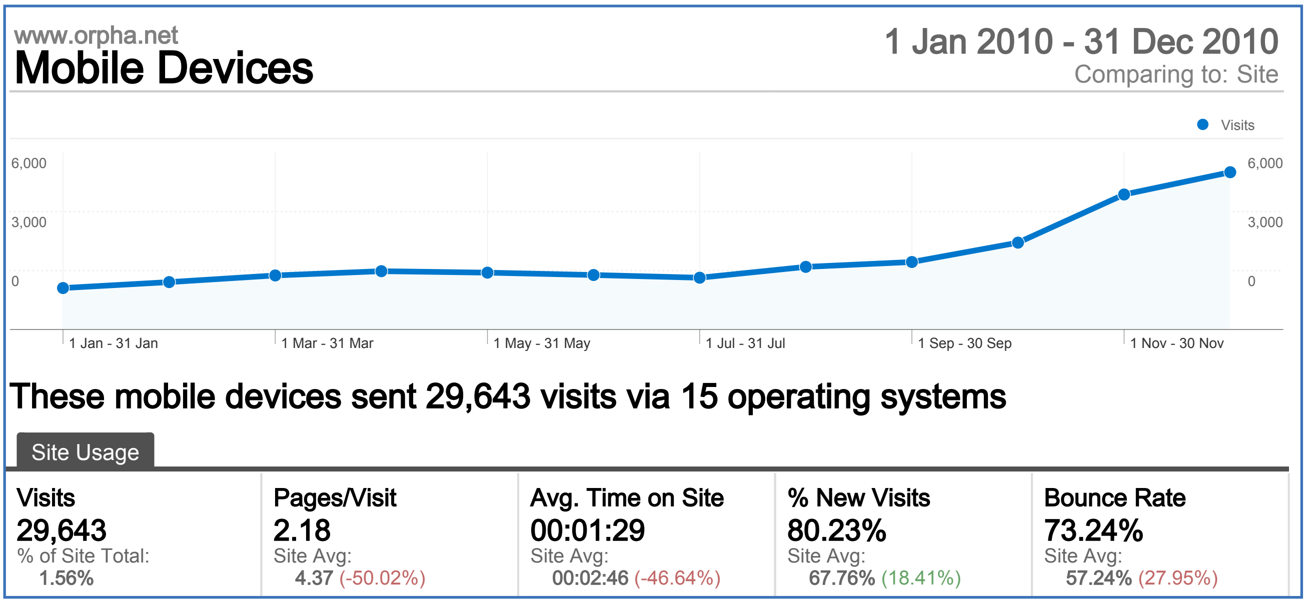
18.84% Referring Sites

66.64% Search Engines

🟦 Search Engines
1,262,854.00 (66.64%)
🟩 Referring Sites
356,940.00 (18.84%)
🟥 Direct Traffic
275,218.00 (14.52%)
⬜ Other
2 (> 0.00%)

(Source: Google Analytics, 1 January 2010 to 31 December 2010)

Since November 2009, Google Analytics allows users to trace visits made from mobile devices (phones, iPad...). These visits represent 1.56% of all visits during 2010, i.e. nearly 30,000 visits.



www.orpha.net
Mobile Devices

1 Jan 2010 - 31 Dec 2010
Comparing to: Site

These mobile devices sent 29,643 visits via 15 operating systems

Site Usage

Visits	Pages/Visit	Avg. Time on Site	% New Visits	Bounce Rate
29,643 % of Site Total: 1.56%	2.18 Site Avg: 4.37 (-50.02%)	00:01:29 Site Avg: 00:02:46 (-46.64%)	80.23% Site Avg: 67.76% (18.41%)	73.24% Site Avg: 57.24% (27.95%)

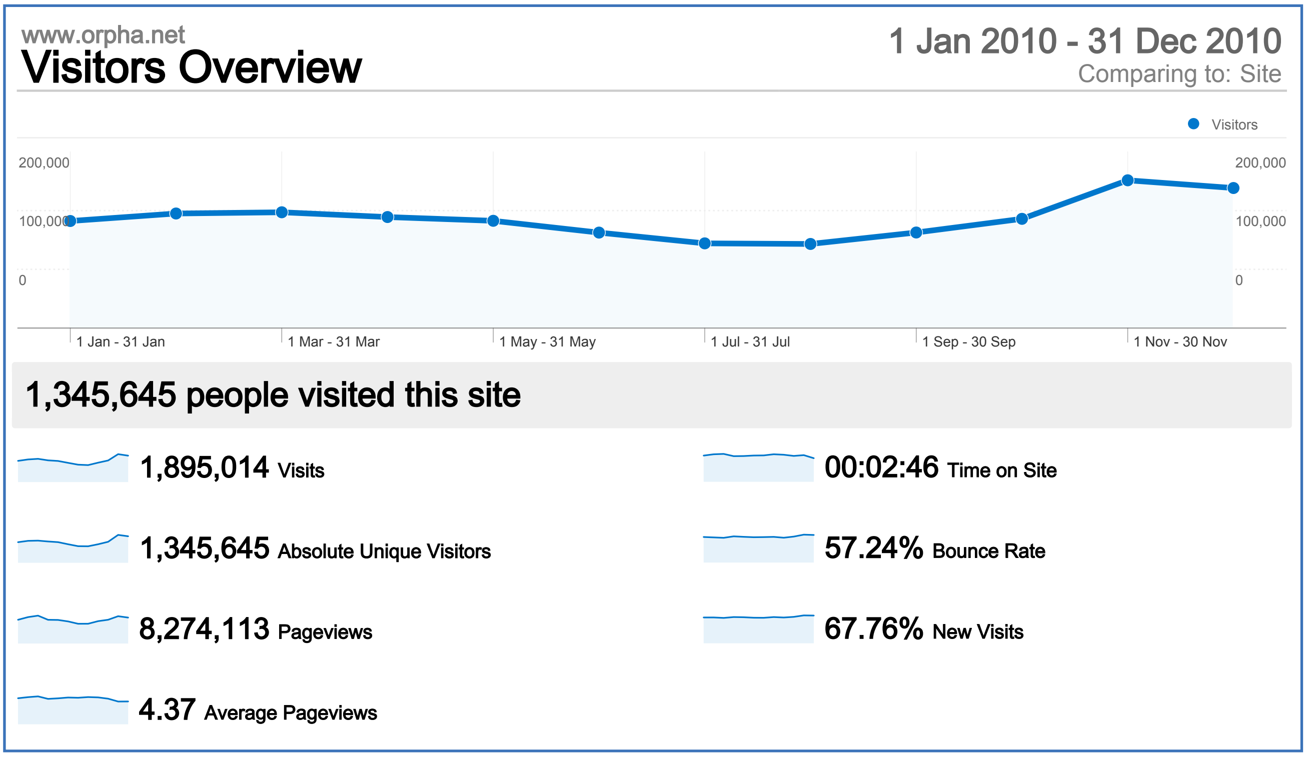
(Source: Google Analytics, 1 January 2010 to 31 December 2010)

In conclusion, the indexation of Orphanet is satisfactory and the richness of our site explains its renown. However, we could optimise the indexation of Orphanet if there were a dedicated member of the team (traffic manager) for this task.

1.2. THE WEBSITE'S AUDIENCE

Over the period 1 January to 1 December 2010, over 8,000,000 pages were viewed, thus on average around 22,000 pages viewed per day. The rate of pages viewed per visit has increased by 10 %. Similarly, the average time spent per visit increased by 17 % and the bounce rate (immediate exit from a user after viewing a single page) has decreased by 5 %.

The Google Analytics tool does not include direct access to PDF documents. Yet this remains an entry point and generates a consistent volume of visits: each month, 650,000 PDF documents are consulted on the Orphanet website.


www.orpha.net
Visitors Overview
1 Jan 2010 - 31 Dec 2010
Comparing to: Site

1,345,645 people visited this site

1,895,014 Visits
1,345,645 Absolute Unique Visitors
8,274,113 Pageviews
4.37 Average Pageviews
00:02:46 Time on Site
57.24% Bounce Rate
67.76% New Visits

(Source: Google Analytics, 1 January 2010 to 31 December 2010)

The geographical distribution of users still shows very varied sources (207 sources listed).

1.3. TYPE OF USERS AND USE

An online satisfaction survey of 1,000 users of the site in English was carried out in September 2009. The results show that Orphanet is used by all types of audience, and about half of these visitors are regular, satisfied users. Two thirds are professionals.

A new online satisfaction survey was launched in November 2010. An Orphanet Report Series document will be published to present the full results.

1.4. The audience of OrphaNews Europe, EUCERD’s newsletter

OrphaNews Europe is the electronic newsletter of the European Union Committee of Experts on Rare Diseases (formerly the European Commission’s Rare Diseases Task Force), which is published on-line, and sent to over 12,000 subscribed readers, twice a month. OrphaNews Europe was launched on 15 June 2005 and over 80 issues of the newsletter have since been published.

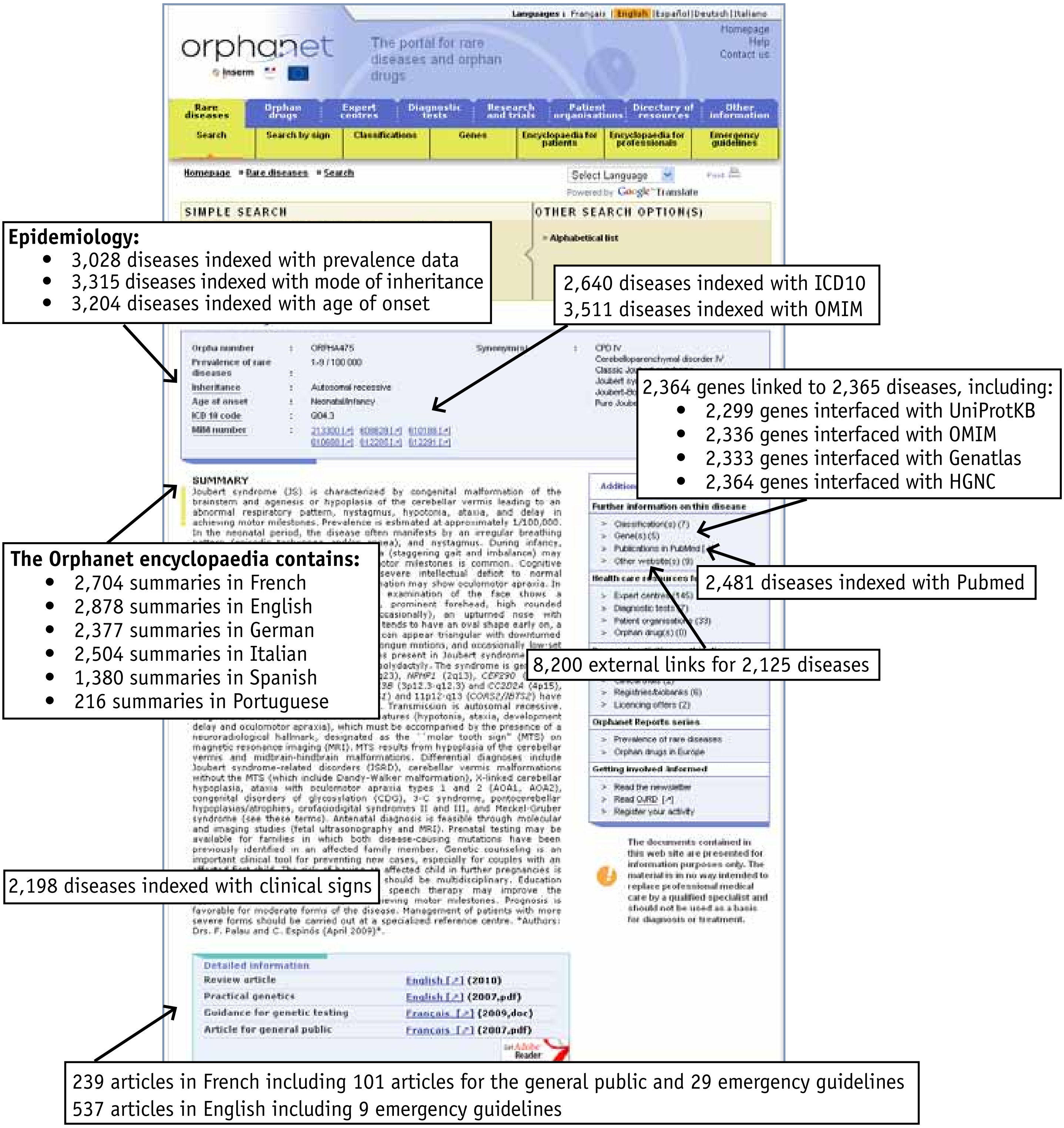
An online survey was launched at the end of May 2010. The overwhelming majority of readers who responded to it reported being either “satisfied” or “very satisfied” with the contents and quality of the newsletter.

OrphaNews Europe readers represent a wide variety of professions and interests – including scientists and researchers, members of the biopharmaceutical industry, government representatives, patients, families, and patient organisation members. Medical professionals accounted for the majority of readers who replied – almost 50%. The survey also revealed that amongst the sections most frequently read are the new syndromes, new genes, orphan drug news, and political and national/international news sections. The calendar of upcoming events was also cited as being regularly consulted, along with new publications.

For further details, please consult the OrphaNews Europe Reader Satisfaction Survey.

2. The evolution of database content

The disease and gene database contains 7,786 diseases or groups of diseases and their synonyms.



Epidemiology:
- 3,028 diseases indexed with prevalence data
- 3,315 diseases indexed with mode of inheritance
- 3,204 diseases indexed with age of onset

2,640 diseases indexed with ICD10
3,511 diseases indexed with OMIM

2,364 genes linked to 2,365 diseases, including:
- 2,299 genes interfaced with UniProtKB
- 2,336 genes interfaced with OMIM
- 2,333 genes interfaced with Genatlas
- 2,364 genes interfaced with HGNC

The Orphanet encyclopaedia contains:
- 2,704 summaries in French
- 2,878 summaries in English
- 2,377 summaries in German
- 2,504 summaries in Italian
- 1,380 summaries in Spanish
- 216 summaries in Portuguese

2,481 diseases indexed with Pubmed

8,200 external links for 2,125 diseases

2,198 diseases indexed with clinical signs

239 articles in French including 101 articles for the general public and 29 emergency guidelines
537 articles in English including 9 emergency guidelines

Orpha number	:	ORPHA475	
Prevalence of rare diseases	:	1-9 / 100 000	
Inheritance	:	Autosomal recessive	
Age of onset	:	Neonatal/infancy	
ICD 10 code	:	G04.3	
MIM number	:	213300 [↗] 608091 [↗] 610188 [↗] 610688 [↗] 611560 [↗] 612291 [↗]	

Synonym(s) : CPD IV
Cerebelloparenchymal disorder IV
Classic Jo…
Joubert sy…
Joubert-Bo…
Pure Joube…

SUMMARY

Joubert syndrome (JS) is characterized by congenital malformation of the brainstem and agenesis or hypoplasia of the cerebellar vermis leading to an abnormal respiratory pattern, nystagmus, hypotonia, ataxia, and delay in achieving motor milestones. Prevalence is estimated at approximately 1/100,000. In the neonatal period, the disease often manifests by an irregular breathing …

… delay and oculomotor apraxia), which must be accompanied by the presence of a neuroradiological hallmark, designated as the "molar tooth sign" (MTS) on magnetic resonance imaging (MRI). MTS results from hypoplasia of the cerebellar vermis and midbrain-hindbrain malformations. Differential diagnoses include Joubert syndrome-related disorders (JSRD), cerebellar vermis malformations without the MTS (which include Dandy-Walker malformation), X-linked cerebellar hypoplasia, ataxia with oculomotor apraxia types 1 and 2 (AOA1, AOA2), congenital disorders of glycosylation (CDG), 3-C syndrome, pontocerebellar hypoplasias/atrophies, orofaciodigital syndromes II and III, and Meckel-Gruber syndrome (see these terms). Antenatal diagnosis is feasible through molecular and imaging studies (fetal ultrasonography and MRI). Prenatal testing may be available for families in which both disease-causing mutations have been previously identified in an affected family member. Genetic counseling is an important clinical tool for preventing new cases, especially for couples with an …

… favorable for moderate forms of the disease. Management of patients with more severe forms should be carried out at a specialized reference centre. *Authors: Drs. F. Palau and C. Espinós (April 2009)*.

Further information on this disease
- Classification(s) (7)
- Gene(s) (5)
- Publications in PubMed
- Other website(s) (9)

Health care resources
- Expert centres (145)
- Diagnostic tests (7)
- Patient organisations (33)
- Orphan drug(s) (0)
- Registries/biobanks (6)
- Licencing offers (2)

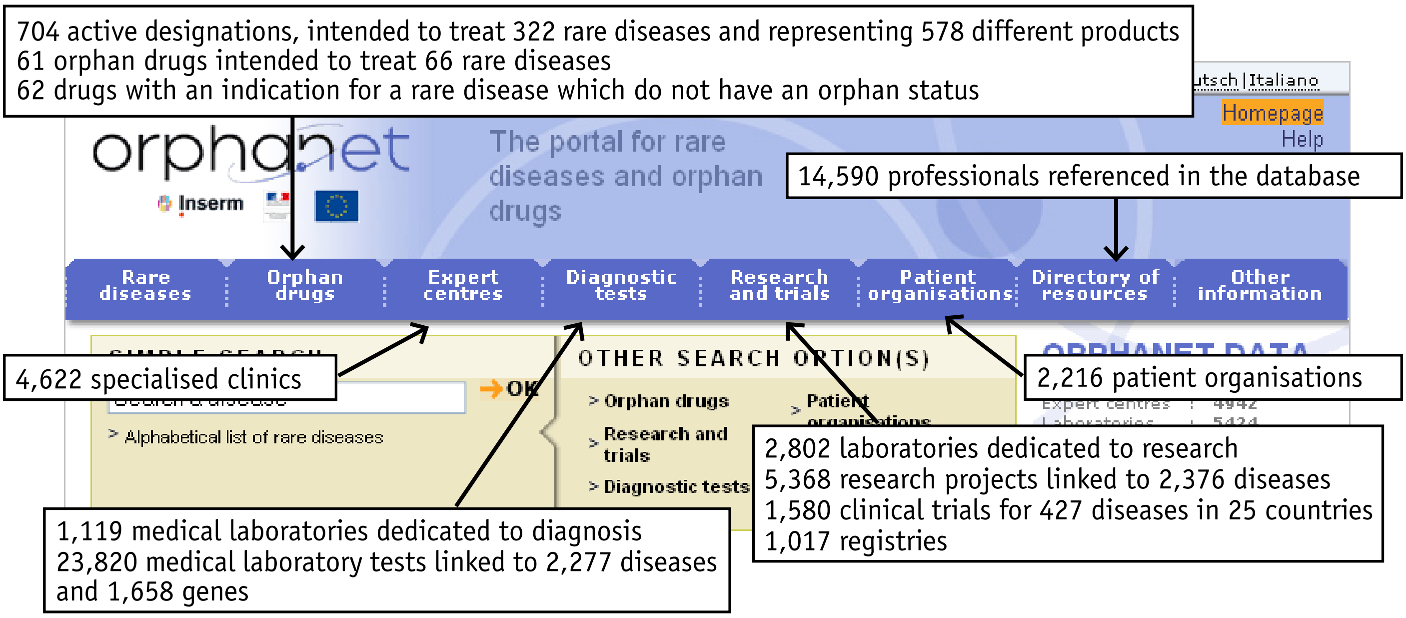
Orphanet Reports series
- Prevalence of rare diseases
- Orphan drugs in Europe

Getting involved informed
- Read the newsletter
- Read OJRD [↗]
- Register your activity

The documents contained in this web site are presented for information purposes only. The material is in no way intended to replace professional medical care by a qualified specialist and should not be used as a basis for diagnosis or treatment.

Detailed information	
Review article	English [↗] (2010)
Practical genetics	English [↗] (2007,pdf)
Guidance for genetic testing	Français [↗] (2009,doc)
Article for general public	Français [↗] (2007,pdf)

The directory of services in 36 European and surrounding countries contains the following data:



704 active designations, intended to treat 322 rare diseases and representing 578 different products
61 orphan drugs intended to treat 66 rare diseases
62 drugs with an indication for a rare disease which do not have an orphan status

14,590 professionals referenced in the database

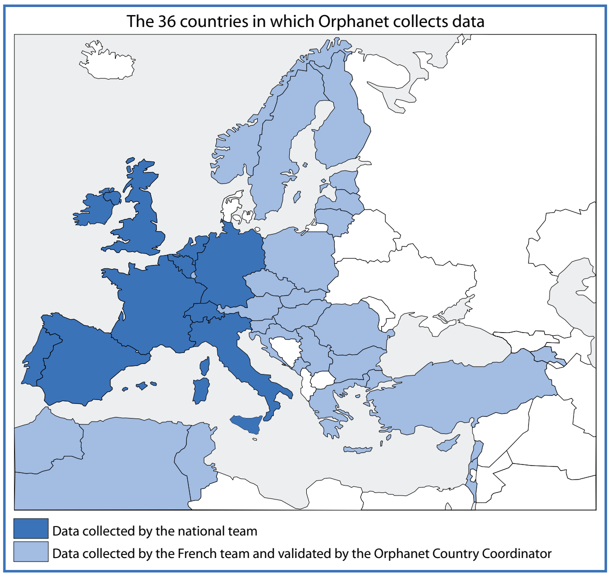
4,622 specialised clinics

2,216 patient organisations

2,802 laboratories dedicated to research
5,368 research projects linked to 2,376 diseases
1,580 clinical trials for 427 diseases in 25 countries
1,017 registries

1,119 medical laboratories dedicated to diagnosis
23,820 medical laboratory tests linked to 2,277 diseases and 1,658 genes

The 36 countries in which Orphanet collects data are the following:


The 36 countries in which Orphanet collects data

Data collected by the national team

Data collected by the French team and validated by the Orphanet Country Coordinator

Algeria, Armenia, Austria, Belgium, Bulgaria, Croatia, Cyprus, Czech Republic, Estonia, Finland, France, Germany, Greece, Hungary, Ireland, Israel, Italy, Latvia, Lebanon, Lithuania, Luxembourg, Morocco, the Netherlands, Norway, Poland, Portugal, Romania, Serbia, Slovakia, Slovenia, Spain, Sweden, Switzerland, Tunisia, Turkey and the United Kingdom.

Data collection and annual updates are managed by the teams at country level which have sufficient funding for a dedicated professional. This is the case for the following countries: Belgium, France, Germany, Italy, the Netherlands, Portugal, Spain, Switzerland and the United Kingdom (and Ireland).

For all other countries, data is collected by France and submitted to the Orphanet Coordinator in each country for validation. The countries in this category are: Algeria, Armenia, Austria, Bulgaria, Croatia, Cyprus, Czech Republic, Estonia, Finland, Greece, Hungary, Israel, Latvia, Lebanon, Lithuania, Luxembourg, Morocco, Norway, Poland, Romania, Serbia, Slovakia, Slovenia, Sweden, Tunisia and Turkey.

Since its creation, Orphanet has grown as a European network, gradually widening to neighbouring countries to the East and the South. But borders have no meaning for Orphanet's users. It was inevitable that other regions of the world would want, at one time or another, to join the network in order to translate the site into their language and publish information on the expert resources of their country. The advantage of joining Orphanet rather than creating a new system 'de novo', is to benefit from the investment already made in the infrastructure.

During 2010, discussions have therefore taken place with five new countries to join the network: Russia, Canada, Australia, Argentina and Japan, which should be finalised in 2011.

3. Evolution of products and services

3.1. FUNCTIONALITIES OF THE ORPHANET WEBSITE DEVELOPED IN 2010

3.1.1. Orphanet pages now translatable online through the Google translation tool.

The Orphanet website is now equipped with this new functionality. This is a translation tool, developed by Google that allows the user to translate any page on the website into many languages.

Following a search in the Orphanet database, any result page can be translated with this tool. The search itself will nevertheless take place in one of the "official" languages of Orphanet.

Although the machine-generated translation is not perfect, it should at least allow the "allophone" user to access information provided by Orphanet.

3.1.2. Implementation of autocomplete

A system called "auto-complete" has been implemented. It is designed to facilitate searches by providing appropriate terms, as they are entered into the database. The outline is simple: the system will suggest terms present in the database that may correspond to the letters / words typed, and displays them in alphabetical order. Once a choice has been made in the dropdown menu, the search engine does its job and proposes a list of answers. No more problems with syntax or spelling!

3.2. OPENING OF COUNTRY WEBSITES

Access to Orphanet is now available through websites dedicated to each partner country in order to enable them to have an entry point in their national language(s). We have developed a tool for creating and updating specific pages for each country, in any language and any alphabet. Beyond the scope of national information, these pages provide access to the international database in five languages. It is therefore a semi-solution pending the identification of resources for the true translation of the entire site.

The following websites were available online at the end of 2010:
Czech Republic: http://www.orphanet.cz;
France: http://www.orphanet-france.fr;
Germany: http://www.orpha.net/national/DE-DE;
Israel: http://www.orpha.net/national/IL-HE;
Spain: http://www.orphanet-espana.es;
Sweden: http://www.orphanet.se;

3.3. CONTINUATION OF THE ORPHANET EMERGENCY GUIDELINES

These guidelines are intended for pre-hospital emergency health care professionals (a dedicated section is included for their use) and for hospital emergency departments. These practical guidelines are devised in collaboration with reference centres and patient organisations, and are peer-reviewed by emergency health care doctors from learned societies (SFMU, the French SAMU, SFP and SNFMI). 28 emergency guidelines in French are now online. They are being translated into four languages (English, German, Italian and Spanish). Currently, 9 emergency guidelines are available in English, 8 in Spanish, 9 in Italian and one in German.

3.4. SUCCESS OF THE ORPHANET JOURNAL OF RARE DISEASES, AN ELECTRONIC, FREE-ACCESS JOURNAL ON THE INTERNET WITH BIOMEDCENTRAL (www.ojrd.com)

OJRD was indexed in Medline at the end of its first year of existence (2006) and was selected by Thompson Scientific after only two years in publication, which led to the OJRD receiving an impact factor of 1.3 in June 2008, 3.14 in June 2009 and 5.83 in June 2010.
181 publications were submitted to the journal in 2010, double the number received in the previous year. Of these, 40 were accepted for publication.

4. Communication

4.1. COMMUNICATION DOCUMENTS

For the second year since the creation of Orphanet, four types of 4-page leaflets in A4 format, in four colours, each aimed at a different target audience, were distributed in 2010:

- A leaflet for all audiences on Orphanet's role as an information portal
- A leaflet for medical biologists on Orphanet as the source of information on biological tests for the diagnosis of rare diseases
- A leaflet for researchers and the pharmaceutical industry on all the services offered by Orphanet to support R&D in the field of rare diseases
- A leaflet for the information systems community on Orphanet as a documentation tool.

Each leaflet was produced in 5 languages (French, English, German, Spanish and Italian). In addition, an A5 format leaflet was produced in Swedish.

In 2010, we designed, printed and distributed A5-size flyers to present an overview of Orphanet. These flyers are available in French and English.

The leaflets are freely available in the offices of the "Plateforme Maladies Rares" (Rare Disease Platform) located at the Broussais Hospital in Paris. The leaflets were distributed at 18 events in 2010 for a total of 1,700 leaflets in French, and 34,000 leaflets in English.

4.2. Invitations to give lectures at conferences in 2010

Orphanet participated in 43 conferences, of which 17 were in France and 25 abroad (Germany, Argentina, Austria, Belgium, Germany, Ireland, Italy, Luxembourg, Morocco, Netherlands, Poland, Qatar, Romania, Russia, Spain, Sweden, Switzerland, United Kingdom). These presentations were mostly given at scientific conferences, where Orphanet played the role of specialist in the field of rare diseases. These lectures focused on medical and genetic approaches (13 presentations), public health policies (11), presenting of the Orphanet tool (5), classifications of diseases (3), or orphan drugs (3).
In addition to this, 3 teaching courses were run by Orphanet in 2010.

4.3. Booths at conferences in 2010

As in previous years, Orphanet had a booth at the annual meeting of the European Society of Human Genetics, which was held in Gothenburg (Sweden) from 12 to 15 June 2010.

5. The national and international collaborations of Orphanet

5.1. Collaboration with the European Commission

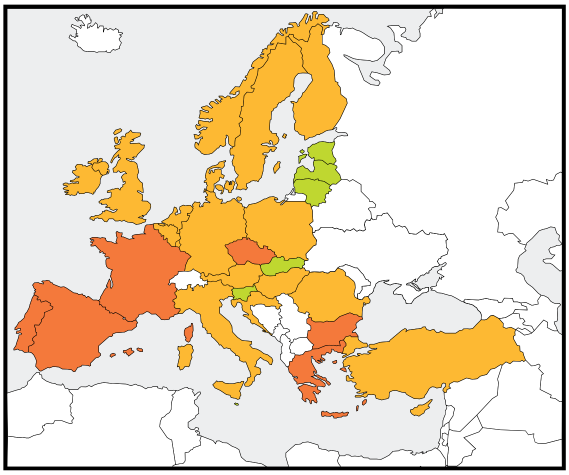
The Director of Orphanet leads the European Union Committee of Experts on Rare Diseases (EUCERD) and provides the Scientific Secretariat.

5.2. Collaboration with the WHO

The World Health Organisation (WHO) and Orphanet collaborate on the revision of the International Classification of Diseases (ICD-11).
The World Health Organisation is currently revising the International Classification of Diseases (ICD-10). This system " is intended to categorise diseases, health related problems and external causes of disease and injury in order to compile useful health information in terms of death, disease and injury (mortality and morbidity)".
However, currently, only 240 rare diseases are included in ICD-10 with their own code.
A group of experts on rare diseases, of which Ségolène Aymé is the President, was established by WHO in April 2007 to discuss the status of these diseases in the classification system. Orphanet has been entrusted with collecting the information necessary in order to establish the alpha draft of the ICD-11 which will be delivered in April 2011 as planned. The new version of the ICD should be adopted in 2014.

5.3. Collaboration with Health Authorities

Orphanet teams actively participate in the preparation of National Plans on Rare Diseases as they are recognised as experts at the national level.



🟧 **National plans or strategies on rare diseases adopted:** Bulgaria, Czeck Republic, France, Greece, Portugal, Spain.

🟨 **Work in progress:** Austria, Belgium, Croatia, Cyprus, Denmark, Finland, Germany, Hungary, Ireland, Italy, Luxembourg, Norway, Poland, Romania, Sweden, The Netherlands, Turkey, UK.

🟩 **Work to be done:** Estonia, Latvia, Lithuania, Malta, Slovakia, Slovenia.

5.4. Scientific collaborations and partnerships with Industry

Orphanet is also a partner of other European projects in the 7th Framework Programme (FP7): CliniGene, Treat-NMD and ENCE.

A major partnership was formed with GlaxoSmithKline (GSK). The company, which has recently opened a division dedicated to rare diseases, wishes to support the development of the diseases database which is considered as a strategic resource of interest to Industry.

Due to the fact that Orphanet is increasingly well-known as the reference source for documentation on rare diseases, we receive more and more requests to access our data from researchers, institutions, Industry and private companies (consultants for the pharmaceutical industry and software companies specialised in hospital management software, medical clinics, laboratories…)

The provision of data to these parties is free for public institutions and available for a fee for Industry and private companies. All situations require a Data Transfer Agreement to be signed.

Partnerships were requested by the following organisations in 2010:

Public/Non-profit private: APREHP Association for research in pediatric hepatology (FR), ARS Haute Normandie (FR), ARS Languedoc-Roussillon (FR), Center for Rare and Neglected diseases; University of Notre Dame (USA), Cincinnati Children's Hospital Medical Center (US), Genethon (FR), University Hospital of Udine (IT), Imperial College London (UK), NIH/NLM/LHC (US), Office of Rare Diseases Research (ORDR), National Institutes of Health (US), OMIM (US), Padua University (IT), Norwegian Directorate of Health (NO), GentoPhen Network; University of Nijmegen (NL), Cytogenetic department, Necker Hospital (FR), Steve Waugh Foundation Australia (AU), University Hospital Leuven (BE), University of Copenhagen (DK), University of Minnesota (USA), University of Ulm (DE), WGM (Dutch Steering Committee on Orphan Drugs) (NL).

Private for profit: Alexion Pharmaceuticals, Inc. (US), AVI BioPharma (US), GlaxoSmithKline (UK), MEDIWARE (FR), Pfizer (US), SimulConsult (US).

Funding

1. Orphanet's core activity funding

1.1. European funding

The European Commission funds the encyclopaedia and the collection of data in European countries (since 2000 DG Public Health and Consumers Protection grants N°s S12.305098; S12.324970; SPC.2002269-2003220, 2006119, 20091215 and since 2004 DG Research grants N°s LSSM-CT-2004-503246; LSHB-CT-2004-512148; LSHB-CT-2006-018933; Health-F2-2008-201230).

In 2010, Orphanet was funded by the following European contracts:

DG Sanco
RDPortal - 2006119
RDPortal2 - 20091215
DG Research
RDPlatform - HEALTH-F2-2008-201230
Eurogentest - LSHB-CT-2004-512148
Clinigene - LSHB-CT-2006-018933
ENCE - HEALTH-F2-2009-223355

In 2011, Orphanet will be funded by the following contracts:

DG Sanco
RDPortal2 - 20091215
Orphanet Europe (RDPortal3-JA) - 20102206
Eurowabb - 20101205
DG Research
RDPlatform - HEALTH-F2-2008-201230
Eurogentest2 - HEALTH-F4-2010-261469
Clinigene - LSHB-CT-2006-018933
ENCE - HEALTH-F2-2009-223355

1.2. OTHER CURRENT FINANCIAL PARTNERSHIPS FOR CORE ACTIVITY FUNDING

Instituts thématiques Inserm Institut national de la santé et de la recherche médicale	Inserm finances Orphanet's core activities. The Inserm Department of Information Services (DSI) hosts Orphanet's servers. Inserm Transfert is in charge of ensuring beneficial licencing and intellectual property transfer concerning Orphanet data
	The French Directorate General for Health finances Orphanet's core activities.
	The European Commission finances the encyclopaedia and data collection in the European countries
CNSA Caisse nationale de solidarité pour l'autonomie	The Caisse nationale de solidarité pour l'autonomie supports the indexing of rare diseases with the International Classification of Functioning, Disability and Health (ICF).
AFM Association Française contre les Myopathies	The Association Française contre les Myopathies finances OrphaNews France and OrphaNews Europe, as well as data collection on clinical trials.
LEEM les entreprises du médicament	Les Entreprises du Médicament (LEEM) finances the collection of data on orphan drugs and clinical trials.
gsk GlaxoSmithKline	GSK finances the extension of the database's disease annotation and free access to this data.

1.3. CURRENT NON FINANCIAL PARTNERSHIPS FOR CORE ACTIVITY FUNDING

WHO	Orphanet collaborates with the WHO (World Health Organisation) in the process of revising the International Classification of Diseases.
GENATLAS	Genatlas collaborates with Orphanet in updating the data on genes involved in rare diseases.
UniProt	UniProt KB collaborates with Orphanet in updating the data on genes linked to proteins involved in rare diseases.
EuroGentest	EuroGentest validates the information collected by Orphanet in the field of quality management of medical laboratories.

2. Financial and non-financial partnerships for national activities

Orphanet's national activities are also supported by national institutions, specific contracts and/or contributions in kind.

2.1. INSTITUTIONAL PARTNERSHIPS PROVIDING FUNDING FOR NATIONAL ACTIVITIES

FRANCE	
Fondation Groupama pour la santé Vaincre les maladies rares	The Fondation Groupama pour la santé finances the support service provided to patient organisations in France for the creation and development of their own website.
LFB L'ENGAGEMENT ETHIQUE	The LFB Biomédicaments helps finance the development and update of emergency guidelines and the French encyclopaedia for the general public.
agence de la biomédecine	The Agence de la biomédecine finances the monitoring of the list of laboratories, the creation of tools for collecting, managing and monitoring annual activity, as well as funding the compilation of data collected in France.
CNSA Caisse nationale de solidarité pour l'autonomie	The Caisse nationale de solidarité pour l'autonomie supports the development of the French encyclopaedia for the general public with information on the functional consequences of rare diseases, as well as the production of fact sheets on rare disabilities not necessarily related to rare diseases.
INSTITUT NATIONAL DU CANCER	The Institut National du Cancer (INCa) supports the development of the Orphanet encyclopaedia on rare cancers.
GERMANY	
MHH Medizinische Hochschule Hannover	The MHH, Medical School of Hanover supports data collection, and is a partner of RDplatform.
ITALY	
Ministero della salute	The Italian Health Ministry finances Orphanet-Italy activities through current research funding.
FARMINDUSTRIA	Farmindustria finances Orphanet publications.
SPAIN	
er ciberer GOBIERNO DE ESPAÑA MINISTERIO DE CIENCIA E INNOVACIÓN	The Centre for Biomedical Network Research on Rare Diseases (CIBERER) has been the partner for Orphanet in Spain since April 2010. CIBERER finances the main activities of the Spanish team.
SWITZERLAND	
HUG Hôpitaux Universitaires de Genève	University Hospitals of Geneva is the host institution of Orphanet Switzerland and finances a part-time position for the coordinator and provides some administrative support for the project.
GDK CDS CDS	Based on the recommendation of the Swiss Conference of the Cantonal Ministers of Public Health, the 26 cantons contribute to the funding of Orphanet Switzerland. The support finances a part-time position for the coordinator and a part-time position of an information scientist, and of a webmaster from the Health On the Net Foundation (HON).
TURKEY	
AiFD	The Association of Research-Based Pharmaceutical Companies gives non-restricted support for the Turkish translation of the Orphanet webpage and the document including over 10.000 rare genetic diseases together with their detailed description. They support the creation of the Orphanet-Turkey website and help Orphanet Turkey to prepare and print leaflets representing Orphanet, Orphanet-Turkey and their activities for health care professionals and the general public.

2.2. INSTITUTIONAL PARTNERSHIPS PROVIDING SERVICES IN KIND FOR NATIONAL ACTIVITIES

ARMENIA	
Center of Medical Genetics and Primary Health Care	The Center of Medical Genetics and Primary Health Care hosts Orphanet-Armenia's activities and contributes to the project by allocating the time of some professionals.
AUSTRIA	
MEDIZINISCHE UNIVERSITÄT WIEN	The Medical University Vienna hosts Orphanet-Austria's activities and contributes to the project by allocating the time of some professionals.
BELGIUM	
KATHOLIEKE UNIVERSITEIT LEUVEN	The University of Leuven hosts Orphanet-Belgium's activities and contributes to the project by allocating the time of some professionals.
BULGARIA	
	The Bulgarian Association for Promotion of Education and Science (BAPES), hosts Orphanet-Bulgaria's activities and contributes to the project by allocating the time of some professionals.
CROATIA	
	The Zagreb University hosts Orphanet-Croatia's activities and contributes to the project by allocating the time of some professionals.
CYPRUS	
	The Archbishop Makarios III Hospital hosts the Orphanet-Cyprus team activities and contributes to the project by allocating the time of some professionals.
CZECH REPUBLIC	
	The Charles University Prague - 2nd School of Medicine (UCPRA-2SM) and UH Motol hosts Orphanet-Czech Republic's activities and contributes to the project by allocating the time of some professionals.
ESTONIA	
Estonian Biocentre	The Estonian Biocentre hosts Orphanet-Estonia's activities and contributes to the project by allocating the time of some professionals.
FINLAND	
Väestöliitto	The Family Federation of Finland (VAESTOLIITTO) hosts Orphanet-Finland's activities and contributes to the project by allocating the time of some professionals.
GREECE	
	The Institute of Child Health (ICH) hosts Orphanet-Greece's activities and contributes to the project by allocating the time of some professionals.
HUNGARY	
	The National Centre for Healthcare Audit and Inspection hosts Orphanet-Hungary's activities and contributes to the project by allocating the time of some professionals.
IRELAND	
Our Lady's Children's Hospital, Crumlin	The Our Lady's Children's Hospital, Crumlin hosts Orphanet-Ireland's activities and contributes to the project by allocating the time of some professionals.

ISRAEL	
	The Schnedier Children's Medical Center of Israel hosts Orphanet-Israel's activities and contributes to the project by allocating the time of some professionals.
ITALY	
	The Casa Sollievo della Sofferenza (IRCCS-CSS) hosts Orphanet-Italy's activities and contributes to the project by allocating the time of some professionals.
LATVIA	
	The Children's University Hospital hosts Orphanet-Latvia's activities and contributes to the project by allocating the time of some professionals.
LEBANON	
	The Saint Joseph University hosts Orphanet-Lebanon's activities and contributes to the project by allocating the time of some professionals.
LITHUANIA	
	The Vilnius University hosts Orphanet-Lithuania's activities and contributes to the project by allocating the time of some professionals.
LUXEMBOURG	
	The Ministry of Health of Luxembourg hosts Orphanet-Luxembourg's activities and contributes to the project by allocating the time of some professionals.
MOROCCO	
	The National Institute of Hygiene hosts Orphanet-Marocco's activities and contributes to the project by allocating the time of some professionals.
NORWAY	
	The Norwegian Directorate of Health hosts Orphanet-Norway's activities and contributes to the project by allocating the time of some professionals.
NETHERLANDS	
VUmc	The VU University Medical Centre hosts the Orphanet-Netherlands team activities and contributes to the project by allocating the time of some professionals.
CENTRE FOR Medical Systems Biology	The Centre for Medical Systems Biology hosts the Orphanet-Netherlands team activities and contributes to the project by allocating the time of some professionals.
LUMC Leiden University Medical Center	The Leiden University Medical Centre hosts the Orphanet-Netherlands team activities and contributes to the project by allocating the time of some professionals.
POLAND	
	The Children's Memorial Health Institute (IP-CZD) hosts Orphanet-Poland's activities and contributes to the project by allocating the time of some professionals.
PORTUGAL	
IBMC	The Instituto de Biologia Molecular e Celular, University of Porto (IBMC) hosts Orphanet-Portugal's activities and contributes to the project by allocating the time of some professionals.

ROMANIA	
	The University of Medicine and Pharmacy «Gr.T.Popa» (UMF-Iasi) hosts Orphanet-Romania's activities and contributes to the project by allocating the time of some professionals.
SERBIA	
IMGGE	The Institute of Molecular Genetics and Genetic Engineering hosts Orphanet-Serbia's activities and contributes to the project by allocating the time of some professionals.
SLOVAKIA	
	The University Children's Hospital, Comenius University Medical School in Bratislava hosts Orphanet-Slovakia's activities and contributes to the project by allocating the time of some professionals.
SLOVENIA	
	The University Medical Centre Ljubljana hosts Orphanet-Slovenia's activities and contributes to the project by allocating the time of some professionals.
SPAIN	
Vall d'Hebron Institut de Recerca	The Vall d'Hebron Research Institute was the partner of Orphanet until April 2010. Since then, the VHIR hosts Orphanet-Spain's activities and contributes to the project by allocating the time of some professionals.
IBV CSIC	The Institute of Biomedicine of Valencia (IBV) managed the contract of one member of the team (RD Platform) until May 2010. Since then the IBV hosts Orphanet-Spain's activities and contributes to the project by allocating the time of some professionals.
SWEDEN	
Karolinska Institutet	The Karolinska Institutet (KI) hosts Orphanet-Sweden's activities and contributes to the project by allocating the time of some professionals.
TUNISIA	
	The Charles Nicolle Hospital hosts Orphanet-Tunisia's activities and contributes to the project by allocating the time of some professionals.
TURKEY	
	The Istanbul University hosts Orphanet-Turkey's activities and contributes to the project by allocating the time of some professionals.
UNITED KINGDOM	
MANCHESTER 1824 The University of Manchester	The University of Manchester (UNIMAN) hosts Orphanet-UK's activities and contributes to the project by allocating the time of some professionals.

2.3. NON-FINANCIAL PARTNERSHIPS IN 2010

AUSTRIA	
BUNDESMINISTERIUM FÜR GESUNDHEIT	The Federal Ministry of Health of Austria officially supports Orphanet.
BELGIUM	
federal public service HEALTH, FOOD CHAIN SAFETY AND ENVIRONMENT	The Federal Public Service Health, Food Chain Safety and Environment of Belgium officially supports Orphanet.
TREFPUNT zelfhulp	Trefpunt Zelfhulp provided information regarding patient associations for rare diseases.
UZ LEUVEN	The Laboratory of Molecular Diagnostics provided a national consensus list of molecular tests for rare diseases (available at the Belgian genetic centres).
BULGARIA	
	The Association of Medical Students in Plovdiv has been actively promoting Orphanet use in its community. Together, BAPES and ASM-Plovdiv have organised a series of workshops, dedicated to Orphanet.
БЪЛГАРИЯ	The Bulgarian National Alliance of People with Rare Diseases has partnered with BAPES in order to promote Orphanet among rare diseases patients in Bulgaria, as well as to list the Bulgarian patient associations in the Orphanet database.
CYPRUS	
REPUBLIC OF CYPRUS	The Department of Medical and Public Health Services of Cyprus officially supports Orphanet.
CZECH REPUBLIC	
SPOLEČNOST LÉKAŘSKÉ GENETIKY ČLS JEP	The Czech Medical Genetics society helps Orphanet CZ in the collection of information on DNA diagnostic laboratories in the country, information on rare diseases clinics - dysmorphology, genetic counselling, and information on patient support groups. They have a joint partnership for the development of the Czech National Plan for rare diseases following the Czech National Strategy from 2009.
MINISTRY OF HEALTH OF THE CZECH REPUBLIC	The Ministry of Health of the Czech Republic officially supports Orphanet.
ESTONIA	
sotsiaal ministeerium	The Ministry of Social Affairs of Estonia officially supports Orphanet.
FINLAND	
MINISTRY OF SOCIAL AFFAIRS AND HEALTH	The Ministry of Social Affairs and Health of Finland officially supports Orphanet.

Terveys portti DUODECIM Medical Publications Ltd	Terveysportti (www.terveysportti.fi) is a web service for medical professionals published by Duodecim Medical Publications Ltd, which is owned by The Finnish Medical Society Duodecim. Orphanet was included in Terveysportti's searches concerning the 300 "most common rare diseases". As a result, Orphanet will have a higher profile among the Finnish health care professionals.
FRANCE	
	The French Directorate General for Health officially supports Orphanet.
HAS HAUTE AUTORITÉ DE SANTÉ	The French High Authority for Health (HAS) and Orphanet cooperate in the online publication of National Protocols for Diagnosis and Care (NHDP) produced by the HAS.
afssaps	The French Health Products Safety Agency (AFSSAPS) provides Orphanet with data on clinical trials in France.
AIRFRANCE	Air France provides patients and professionals with a quota of airline tickets for patients to travel to medical experts or experts to patients with rare diseases. Orphanet provides expertise on the merits of applications.
Maladies Rares Info Services 0 810 63 19 20 N° Azur, prix appel local	Orphanet has delegated to Maladies Rares Info Services, the French helpline for information on rare diseases - 0810 69 19 20, the role of replying to unsolicited electronic messages received by Orphanet.
GERMANY	
Bundesministerium für Gesundheit	The Federal Ministry of Health Germany officially supports Orphanet.
achse	The Allianz Chronischer Seltener Erkrankungen e.V. (ACHSE) works together with Orphanet Germany on informational services for patients.
	The Kindernetzwerk e.V. - für Kinder, Jugendliche und (junge) Erwachsene mit chronischen Krankheiten und Behinderungen provides data on associations in Germany.
gfh	The Deutsche Gesellschaft für Humangenetik e.V. supports Orphanet by supplying the German team with addresses and information on laboratories and diagnostics.
GREECE	
	The Ministry of Health and Social Solidarity of the Hellenic Republic officially supports Orphanet.
HUNGARY	
MINISTRY OF HEALTH	The Ministry of Health of Hungary officially supports Orphanet.

ISRAEL	
משרד הבריאות MINISTRY OF HEALTH	The Israeli Ministry of Health officially supports Orphanet.
ITALY	
ISTITUTO SUPERIORE DI SANITÀ	The Istituto Superiore di Sanità officially supports Orphanet.
TeleThon	Telethon collaborates with Orphanet in the collection of data concerning research projects.
UNIAMO FEDERAZIONE ITALIANA MALATTIE RARE ONLUS	Uniamo, the Italian Federation of support groups on rare diseases, collaborates with Orphanet in the organization and promotion of events dedicated to rare diseases, in order to increase public awareness on this particular issue.
AIFA	AIFA collaborates with Orphanet in the collection of data concerning clinical trials.
netgene	Netgene collaborates with Orphanet in the diffusion of information on rare diseases.
LATVIA	
	The Ministry of Health of the Republic of Latvia officially supports Orphanet.
CALADRIUS	The Rare Diseases Society in Latvia which aims to promote equal rights and opportunities for patients with rare diseases.
PALĪDZĒSIM.LV	Non-governmental organisation in Latvia which financially supports children and families to confirm a diagnosis of rare diseases by sending patients or medical samples abroad.
LITHUANIA	
LIETUVOS RESPUBLIKOS SVEIKATOS APSAUGOS MINISTERIJA	The Ministry of Health of the Republic of Lithuania officially supports Orphanet.
NETHERLANDS	
Ministry of Health, Welfare and Sport	The Ministry of Health, Welfare and Sport of the Netherlands officially supports Orphanet.
erfocentrum	The Erfocentrum provides information to the general public on genetic, mainly rare, disorders. Collaboration has been established to increase the number of Dutch texts available on the Orphanet.
VSOP	Orphanet-Netherlands receives information from the Vereniging Samenwerkende Ouder- en Patiëntenorganisaties (VSOP) on patient organisations dedicated to rare disease.
wgm	Orphanet-Netherlands receives information from the Steering Committee on Orphan Drugs on patient organisations and orphan drugs dedicated to rare disease.
NFU NEDERLANDSE FEDERATIE VAN UNIVERSITAIR MEDISCHE CENTRA	The Netherlands Federation of University Medical Centres has begun an inventory of expertise on rare diseases. The discussion on the definition of “centres of expertise” has been initiated.

POLAND	
Ministerstwo Zdrowia	The Polish Ministry of Health officially supports Orphanet.
	The CMHI supports Orphanet Poland in all activities inside and outside the institution; e.g. organising conferences for professionals, parents and media, discussions on RD with all stakeholders and improving access to orphan drugs.
ARS VIVENDI PKU	The patient organisation, Ars Vivendi, provides patients and parents with information about Orphanet services and cooperates with Orphanet Poland.
PORTUGAL	
Direcção-Geral da Saúde www.dgs.pt Ministério da Saúde	The Portugese Ministry of Health officially supports Orphanet.
ROMANIA	
MINISTERUL SĂNĂTĂŢII	The Ministry of Health collaborates with Orphanet Romania in updating data on the Romanian medical system. It officially supports Orphanet.
CMR	Orphanet Romania collaborates with the Romanian Medical Association in updating data on health professionals.
SRGM Societatea Romana de Genetica Medicala	Orphanet Romania collaborates with the Romanian Society of Medical Genetics to set up programs for the development of a national network of diagnosis, investigation and prevention in Centers of Medical Genetics and to promote collaboration with associations of people with genetic/malformative diseases.
ASOCIAŢIA PRADER WILLI din ROMÂNIA	Orphanet Romania collaborates with Romanian Prader Willi Association in order to bring together the efforts of patients, specialists and families to ensure a better life for all people with genetic diseases.
SLOVAKIA	
Ministerstvo zdravotníctva SR	The Ministry of Health of the Slovak Republic officially supports Orphanet.
SLOVENIA	
REPUBLIC OF SLOVENIA MINISTRY OF HEALTH	The Ministry of Health of Slovenia officially supports Orphanet.
IGRE INŠTITUT ZA GENOMSKE RAZISKAVE IN EDUKACIJO	Orphanet Slovenia collaborates with the Institute of Genomic Research and Education IGRE with the aim of disseminating information about the Orphanet project and web services on the national level.
SPAIN	
MINISTERIO DE SANIDAD Y POLÍTICA SOCIAL	The Ministry of Health and Social Affairs of Spain officially supports Orphanet.
SWEDEN	
REGERINGSKANSLIET	The Ministry of Health and Social Affairs of Sweden officially supports Orphanet.

SWITZERLAND	
HON Health On the Net Foundation Non Governmental Organization www.healthonnet.org	The Health On the Net Foundation provides the technical aspect of the project by developing online forms to collect data. In addition, it hosts the website www.orphanet.ch and helps to update the homepage.
ProRaris	Orphanet Switzerland was instrumental in the creation, in June 2010, of ProRaris, the Swiss alliance of rare diseases. A close collaboration has been established in order to identify relevant informational services for patients and professionnals and in the organization and promotion of events dedicated to rare diseases, in order to increase public awareness on this particular issue.
TURKEY	
	The Turkish Ministry of Health officially supports Orphanet. It collaborates with Orphanet Turkey for data collection and the dissemination of Orphanet in Turkey.
UNITED KINGDOM	
DH Department of Health	The Department of Health officially supports Orphanet.
dyscerne	Dyscerne and Orphanet cooperate in endorsing and boosting Dyscerne and Orphanet activities, raising standards in the diagnosis and management of rare dysmorphic conditions, improving dissemination of information on these conditions, developing and sharing information and educational tools for healthcare professionals.
RARE DISEASE UK	Orphanet collaborates with Rare Disease UK in the sharing of data and expertise, in the endorsement and boosting of Orphanet and Rare Disease UK activities and in the development of the UK Strategy for Rare Diseases.

3. Funding outlook for national activities

3.1. NEW INSTITUTIONAL PARTNERSHIPS PROVIDING FUNDING FOR NATIONAL ACTIVITIES

AUSTRIA	
Gesundheit Österreich GmbH	The Gesundheit Österreich GmbH (GÖG) is an associated partner in the Orphanet Europe Joint Action as of April 2011.
	The Institute of Neurology, Medical University of Vienna, is an associated partner in the Orphanet Europe Joint Action as of April 2011.
BELGIUM	
federal public service HEALTH, FOOD CHAIN SAFETY AND ENVIRONMENT	The Federal Public Service Health, Food Chain Safety and Environment is an associated partner in the Orphanet Europe Joint Action as of April 2011.
isp wiv	The Wetenschappelijk Instituut Volksgezondheid – Institut Scientifique de Santé Publique is an associated partner in the Orphanet Europe Joint Action as of April 2011.

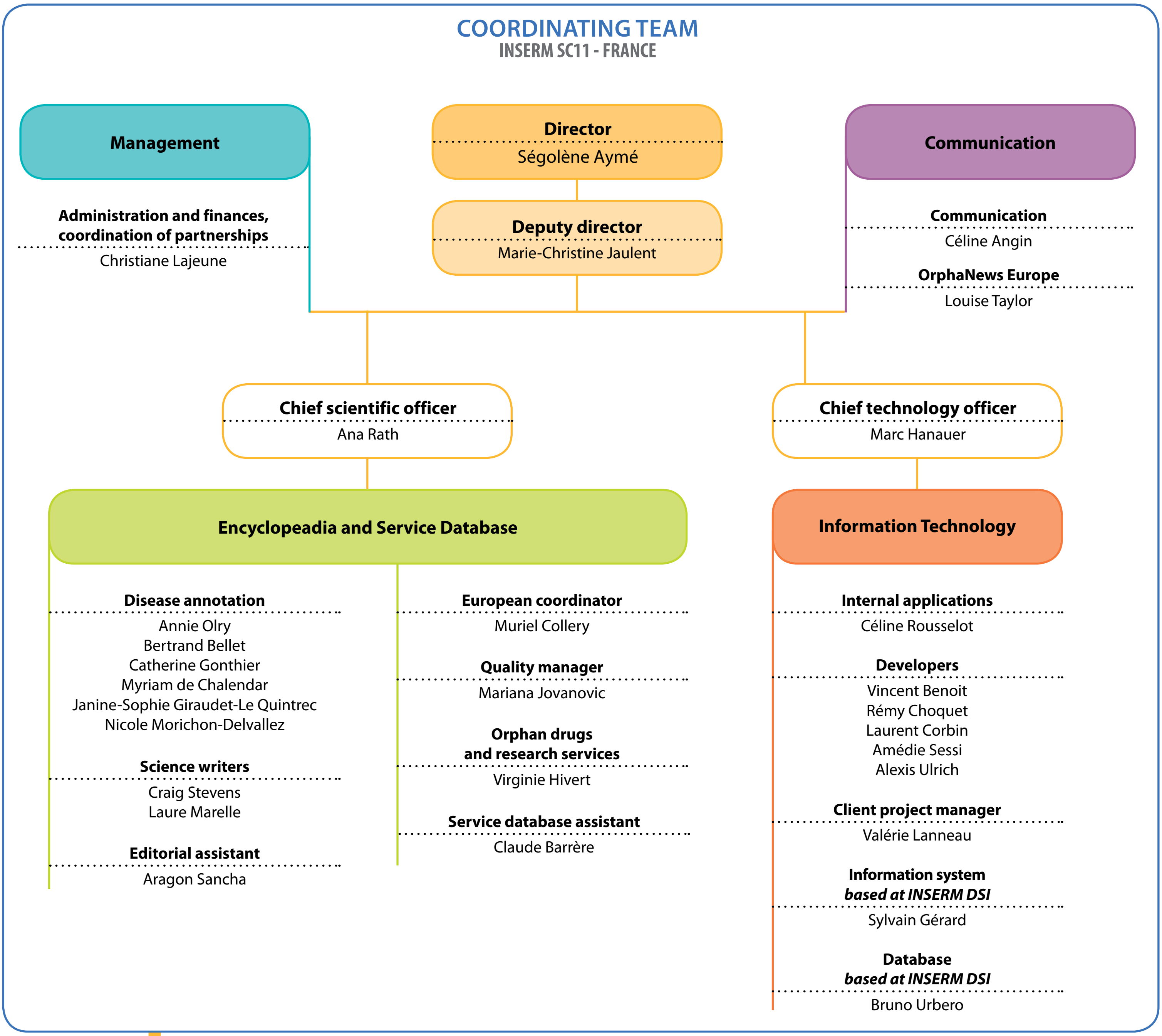
CYPRUS	
REPUBLIC OF CYPRUS	The Department of Medical and Public Health Services is an associated partner in the Orphanet Europe Joint Action as of April 2011.
CZECK REPUBLIC	
	The Charles University Prague - 2nd School of Medicine is an associated partner in the Orphanet Europe Joint Action as of April 2011.
ESTONIA	
	The University of Tartu is an associated partner in the Orphanet Europe Joint Action as of April 2011.
FINLAND	
Väestöliitto	The Family Federation of Finland (Väestöliitto ry) is an associated partner in the Orphanet Europe Joint Action as of April 2011.
GERMANY	
Bundesministerium für Gesundheit	The Federal Ministry of Health Germany provides funding to the Orphanet Europe Joint Action as of April 2011.
GREECE	
	The Institute of Child Health, Athens is an associated partner in the Orphanet Europe Joint Action as of April 2011.
HUNGARY	
	The National Centre for Healthcare Audit and Inspection (Orszagos Szakfelugyeleti Modszertani Kozpont) is an associated partner in the Orphanet Europe Joint Action as of April 2011.
ITALY	
Bambino Gesù OSPEDALE PEDIATRICO	The Bambino Gesù Children's Hospital is an associated partner in the Orphanet Europe Joint Action as of April 2011.
LATVIA	
	The Centre of Health Economics (Veselibas ekonomikas centrs) is an associated partner in the Orphanet Europe Joint Action as of April 2011.
LITHUANIA	
	The Vilnius University Hospital Santariškių Klinikos Centre for Medical Genetics is an associated partner in the Orphanet Europe Joint Action as of April 2011.
NETHERLAND	
LUMC Leiden University Medical Center	The Academisch ziekenhuis Leiden - Leids Universitair Medisch Centrum is an associated partner in the Orphanet Europe Joint Action as of April 2011.

POLAND	
	The Instytut Pomnik Centrum Zdrowia Dziecka (Children's Memorial Health Institute) is an associated partner in the Orphanet Europe Joint Action as of April 2011.
PORTUGAL	
IBMC	The Instituto de Biologia Molecular e Celular is an associated partner in the Orphanet Europe Joint Action as of April 2011.
ROMANIA	
	The Universitatea de Medicina si Farmacie "Gr.T.Popa" Iasi is an associated partner in the Orphanet Europe Joint Action as of April 2011.
SLOVAKIA	
	The Children's University Hospital in Bratislava is an associated partner in the Orphanet Europe Joint Action as of April 2011.
SLOVENIA	
	The University Medical Centre Ljubljana is an associated partner in the Orphanet Europe Joint Action as of April 2011.
SPAIN	
MINISTERIO DE SANIDAD Y POLÍTICA SOCIAL	The Spanish Ministry of Health and Social Policy - Office for Health Planning and Quality is an associated partner in the Orphanet Europe Joint Action as of April 2011.
SWEDEN	
Karolinska Institutet	The Karolinska Institutet is an associated partner in the Orphanet Europe Joint Action as of April 2011.
UNITED KINGDOM	
MANCHESTER 1824 The University of Manchester	The University of Manchester is an associated partner in the Orphanet Europe Joint Action as of April 2011.

3.2. New Institutional partnerships providing services in kind for national activities

DENMARK	
Århus Universitetshospital SKEJBY SYGEHUS	The University Hospital of Aarhus is a collaborating partner in the Orphanet Europe Joint Action as of April 2011.
MALTA	
	The University of Malta is a collaborating partner in the Orphanet Europe Joint Action as of April 2011.

The Orphanet team - April 2011



COORDINATING TEAM

INSERM SC11 - FRANCE

Director
Ségolène Aymé

Deputy director
Marie-Christine Jaulent

Management

Administration and finances, coordination of partnerships
Christiane Lajeune

Communication

Communication
Céline Angin

OrphaNews Europe
Louise Taylor

Chief scientific officer
Ana Rath

Chief technology officer
Marc Hanauer

Encyclopeadia and Service Database

Disease annotation
Annie Olry
Bertrand Bellet
Catherine Gonthier
Myriam de Chalendar
Janine-Sophie Giraudet-Le Quintrec
Nicole Morichon-Delvallez

Science writers
Craig Stevens
Laure Marelle

Editorial assistant
Aragon Sancha

European coordinator
Muriel Collery

Quality manager
Mariana Jovanovic

Orphan drugs and research services
Virginie Hivert

Service database assistant
Claude Barrère

Information Technology

Internal applications
Céline Rousselot

Developers
Vincent Benoit
Rémy Choquet
Laurent Corbin
Amédie Sessi
Alexis Ulrich

Client project manager
Valérie Lanneau

Information system *based at INSERM DSI*
Sylvain Gérard

Database *based at INSERM DSI*
Bruno Urbero

ARMENIA

Center of Medical Genetics and Primary Health Care

Coordinator
Prof. Tamara F. Sarkisian

AUSTRIA

Medizinische Universität Wien

Coordinator
Dr. Till Voigtländer

Information scientist
Christine Schragel
Ursula Unterberger

BELGIUM

Scientific Institute Public Health & Federal Public Service Health

Coordinator
Herwig Jansen

Information scientist
To be identified

BULGARIA

Bulgarian Association for Promotion of Education and Science

Coordinator
Prof. Rumen Stefanov

Information scientist
Ralitza Jordanova
Lilia Popova

CROATIA

Zagreb University

Coordinator
Prof. Ana Stavljenic-Rukavina

CYPRUS

Archbishop Makarios III Hospital

Coordinator
Dr. Violetta Anastasiadou

Information scientist
Turem Delikurt

CZECH REPUBLIC

Charles University Prague - 2nd School of Medicine and UH Motol

Coordinator
Prof. Milan Macek

Information scientist
Marek Turnovec

DENMARK
University Hospital of Aarhus

Coordinator
John Rosendahl Oestergaard

ESTONIA
Tartu University

Coordinator
Dr. Vallo Tillmann

FINLAND
Väestöliitto ry - The Family Federation of Finland

Coordinator
Dr. Riitta Salonen

Information scientist
Leena Toivanen

FRANCE
INSERM SC11

Coordinator
Dr. Segolène Aymé

Assistant
Ana Rait

Information scientist
Bénédicte Belloir

Science writers
Raphaël Demonchy
Florence Mousson
Catherine Pouzat
Virginie Tsilibaris

GERMANY
Medizinische Hochschule Hannover

Coordinator
Prof. Manfred Stuhrmann-Spangenberg

Information scientists
Kathrin Rommel
Elisabeth Nyoungui

GREECE
Institute of Child Health

Coordinator
Prof. Michael B. Petersen

Information scientists
Sofia Douzgou
Haris Kokotas

HUNGARY
National Centre for Healthcare Audit and Inspection

Coordinator
Janos Sandor

Information scientist
Peter Bencsik

IRELAND
Our Lady's Children's Hospital, Crumlin

Coordinator
Prof. Andrew Green

Project manager
Idoia Gomez-Paramio (Manchester, UK)

ISRAEL
Schnedier Children's Medical Center

Coordinator
Dr. Lina Basel

ITALY
IRCSS Bambino Gesù Paediatric Hospital

Coordinator
Prof. Bruno Dallapiccola

Project manager
Rita Mingarelli

Information scientists
Elena Cocchiara
Martina Di Giacinto
Sonia Festa
Tiziana Lauretti

LATVIA
Centre of Health Economics

Coordinator
Jana Lepiksone

Information scientist
Santa Rozite

LEBANON
Université Saint Joseph

Coordinator
Prof. André Mégarbané

LITHUANIA
Vilnius University

Coordinator
Prof. Vaidutis Kucinskas

Information scientist
Birute Tumiene

LUXEMBOURG
Ministère de la Santé du Luxembourg

Coordinator
Dr. Yolande Wagener

MALTA
University of Malta

Coordinator
Prof. Alfred Cuschieri

MOROCCO
Institut National d'Hygiène

Coordinator
Prof. Abdelaziz Sefiani

Information scientist
Imane Cherkaoui-Jaouad

NETHERLANDS
Leiden University Medical Center

Coordinator
Prof. Gert-Jan van Ommen
Prof. Martina Cornel (main advisor)

Information scientists
Judith Carlier - de Leeuw van Weenen
Petra van Overveld

NORWAY
Norweigan Directorate of Health

Coordinator
Stein Are Aksnes

POLAND
Children's Memorial Health Institute

Coordinator
Krystyna Chrzanowska
Malgorzata Krajewska-Walasek

Information scientist
Aleksandra Jezela-Stanek

PORTUGAL
Instituto de Biologia Molecular e Celular, University of Porto

Coordinator
Prof. Jorge Sequeiros

Project manager
Jorge Pinto Basto

Information scientist
Sandra Peixoto

ROMANIA
Universitatea de Medicina si Farmacie «Gr.T.Popa» Iasi

Coordinator
Ass. Prof. Cristina Rusu

Information scientists
Elena Braha
Monica Panzariu
Cristina Gavrilovici

SERBIA
Institute of Molecular Genetics and Genetic Engineering

Coordinator
Dr. Dragica Radojkovic

Information scientist
Maja Stojiljković

SLOVAKIA
University Children's Hospital, Comenius University Medical School

Coordinator
Prof. László Kovács

Information scientist
Dr. Anna Hlavatá

SLOVENIA
University Medical Centre Ljubljana

Coordinator
Prof. Borut Peterlin

Information scientist
Luca Lovrecic
Gorazd Rudolf

SPAIN
CIBERER

Coordinator
Dr. Francesc Palau

Project manager
Dr. Virginia Corrochano

Information scientists
Martin Arles Soler
María Elena Mateo

SWEDEN
Karolinska Institutet

Coordinator
Dr. Désirée Gavhed

Information scientist
Annika Larsson

SWITZERLAND
Service of Genetic Medicine - Geneva University Hospitals

Coordinator
Dr. Loredana D'Amato Sizonenko

Information scientist
Béatrice Geissbuhler

TURKEY
University of Istanbul

Coordinator
Prof. Ugur Özbek
Meral Ozguk

Information scientists
Fatmahan Atalar
Tufan Acuner

Assistant Scientist
Ibrahim Kisakesen
Ozge Ozgen

UNITED KINGDOM
University of Manchester

Coordinator
Prof. Dian Donnai

Project manager
Idoia Gomez-Paramio

For any questions or comments, please contact us: *contact.orphanet@inserm.fr*

Editor-in-chief: Ségolène Aymé ● Visual design : Céline Angin

The correct form when quoting this document is :
« Orphanet - 2010 Activity Report », Orphanet Report Series, *Reports collection*, March 2011,
http://www.orpha.net/orphacom/cahiers/docs/GB/ActivityReport2010.pdf