Orphanet Report Series

Reports collection

2011 Activity Report

orphanet

2011 Activity Report

www.orpha.net

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

1. Overview

1.1. Objective

The general objective of Orphanet is to provide the community at large with a comprehensive set of information on rare diseases (RD) and orphan drugs (OD) in order to contribute to the improvement of the diagnosis, care and treatment of patients with rare diseases.

1.2. 2011 activities

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

The site gives access to:

- An inventory of diseases classified according to 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 comprehensive 'identity card').
- An encyclopaedia covering 3000 rare diseases, written by science writers and review by world-renowned experts. Abstracts are produced in English and are then translated into French, German, Italian, Portuguese and Spanish. For some selected diseases emergency guidelines are produced.
- An inventory of orphan drugs at all stages of development, from orphan designation to market authorisation.
- A directory of specialised services in the 36 partner countries, providing information on: specialised expert centres and centres of expertise, medical laboratories, research projects, clinical trials, registries, networks, technological platforms, patient organisations.
- A range of other services:
  - A support-to-diagnosis tool (search by signs and symptoms).
  - A newsletter in English covering both scientific and political news. This newsletter is also published in French and in Italian.
  - Thematic studies and reports on overarching subjects: "the Orphanet Report Series" published as PDF documents.

Currently Orphanet is the only project which establishes a link between the diseases, the textual information existing about them (including links to other informative websites) and the appropriate services for patients. Orphanet is thus the number one website dedicated to RD in terms of referenced documents.

1.3. 2011 main achievements

The main achievements of 2011 are:

- The launch of the Joint Action Orphanet Europe in April 2011.

Orphanet is now supported by the European Commission as a Joint Action between the European Member States.

- The governance of Orphanet has been further improved.

To ensure optimal governance of the Orphanet consortium and efficient management of the workflow, and also to reflect the new involvement of the health authorities of the Member States, the governance was reorganised in April 2011. Three boards (the Management Board, the Steering Committee and the International

Advisory Board) ensure the project's coherence, its evolution in relation to technological developments and to the needs of its end-users, as well as its sustainability.

- The translation of the international website and of the whole database content into Portuguese.

- The publication of front pages, specific to each country and in the national language(s).

The national pages offer new ways to access and present the information adapted to the needs of each Member State (MS) and of each type of stakeholder. They give access to the common core information but also to information specific to the country: news, events, RD policy and documents.

- The expansion of the network

Canada joined Orphanet in 2011 and negotiations have started with Argentina, Australia, Brazil, China and Japan.

- Free access to the Orphanet dataset on www.orphadata.org.

Because of the growing number of requests for data, to ensure dissemination of the Orphanet nomenclature of RD and to maximise the use of collected information on expert resources, orphadata.org was created. On this website, the whole Orphanet dataset is directly accessible in a reusable format since June 2011. The dataset is a partial extraction of the data stored in Orphanet and is updated monthly. It is freely accessible in six languages (English, French, German, Italian, Portuguese and Spanish). It is a great success as there are more than 1,000 downloads per month.

- The encyclopaedia of RD has been expanded and updated.

Since 2011, it is available in Portuguese and Polish in addition to English, French, German, Italian and Spanish. Requests for review articles in the OJRD have continued.

- The directory of expert centres, medical laboratories, clinical trials, research projects, networks, registries and patient organisations has been expanded and updated.

- The information provided on Orphan Drugs was published as an Orphanet Report Series for more effective communication.

2. Orphanet consortium evolution

2.1. Launch of the Orphanet Europe Joint Action

Orphanet is perceived at the international level as a valuable resource as it is the only source of validated data on rare diseases, and it is mentioned in the documents of the European Commission on Rare diseases ("Rare diseases: Europe's challenge" 11 November 2008 and "Recommendations of the Council on Rare Diseases" - 8 June 2009) as the source of current information on the situation of RD in the European Union and also as a strategic element of any national strategy on Rare Diseases that each MS is encouraged to develop by the end of 2013.
In 2011, an important step forward was taken with the launching of the Orphanet Europe Joint Action, an instrument that combines funding from the European Commission with each of the participating Member States (MS), as well as from Switzerland, a collaborating partner. The three-year €7.2 million Joint Action began on 1 April and the first Kick-Off Meeting, gathering all national country coordinators and MS health representatives to a one-and-a-half-day meeting in Paris took place on 7 and 8 June 2011. The overriding aim of the Joint Action is to improve and adapt the presence of Orphanet in each participating country. Objectives thus include improving the existing services and developing new tools and services (including building an Orphanet ontology and developing several new services). Priority initiatives that are part of the Joint Action include expanding the language availability of the Orphanet database and related documents.

To ensure optimal governance of the Joint Action and efficient management of the workflow, and also to reflect the new involvement of the health authorities of the Member States, Orphanet governance was reorganised in June 2011. In addition to the Management Board (composed of country coordinators), two new committees were created and nominated during the first annual meeting of the Joint Action:
The Steering Committee composed of representatives from the funding agencies/health authorities contributing to the funding of the core project (diseases database, encyclopaedia, database structure, infrastructures, and coordination of activities).
The International Advisory Board composed of international experts.
These boards ensure the project's coherence, its evolution in relation to technological developments and to the needs of its end-users, as well as its sustainability.
Within the framework of the Joint Action and the new governance, Orphanet seeks to become more cost-effective, more user-friendly and to achieve sustainability. Despite an immense number of complex challenges and opportunities that the Joint Action creates for the Orphanet country partners, the partners are moving forward, always keeping their eyes on the ultimate goal: improving conditions for rare disease patients and their families throughout Europe and beyond.

2.2. Expansion of the consortium

Since its creation, because of the quality of data and its reputation, Orphanet has grown as a European consortium, gradually widening to 35 neighbouring countries to the East and the South.
In 2011, Orphanet has gone further west to include Canada.
The number of countries wishing to join the consortium is growing every year as 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 2011, discussions took place with six new countries: Australia, Argentina, Brazil, China, Ireland and Japan.

2.3. List of partners and scope of their activity

2.3.1. Coordinating team

The coordination of the consortium is managed by the coordinating team, Orphanet France, located in the Service Commun 11 of INSERM (the French National Institute of Health and Medical Research). INSERM has been the coordinator of the Orphanet consortium since the beginning in 1997.
The coordinating team is responsible for coordination of the consortium activities, the hardware and software aspects of the project, the database of rare diseases and the production of the encyclopaedia, as well as quality control of the directory of resources in the participating countries.
The coordinating team is also in charge of updating the database regarding medicinal products in development, from the designation stage to the marketing authorisation and availability at country level.

2.3.2. Partners

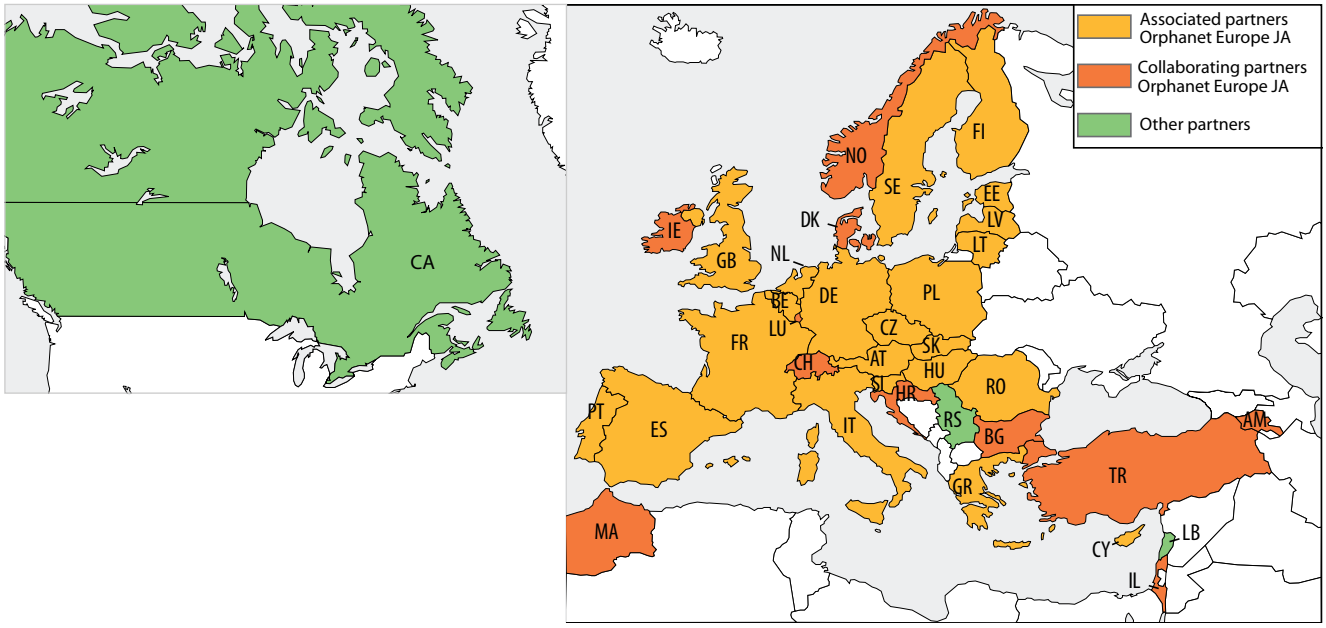
The establishment of a Directory of Services can only be achieved by consolidation of data collected at the MS level. The identification of expert resources requires a very good knowledge of the national research and health care institutions and pathways. All national coordinators are located in high-profile institutions which can provide a suitable environment for the information scientists to work, in terms of documentation, secretarial facilities and access to the network.

The partners are responsible for collecting, validating and submitting data on clinical trials, medical laboratories, expert centres, research projects, registries, and patient organistaions.

List of Orphanet partner institutions:

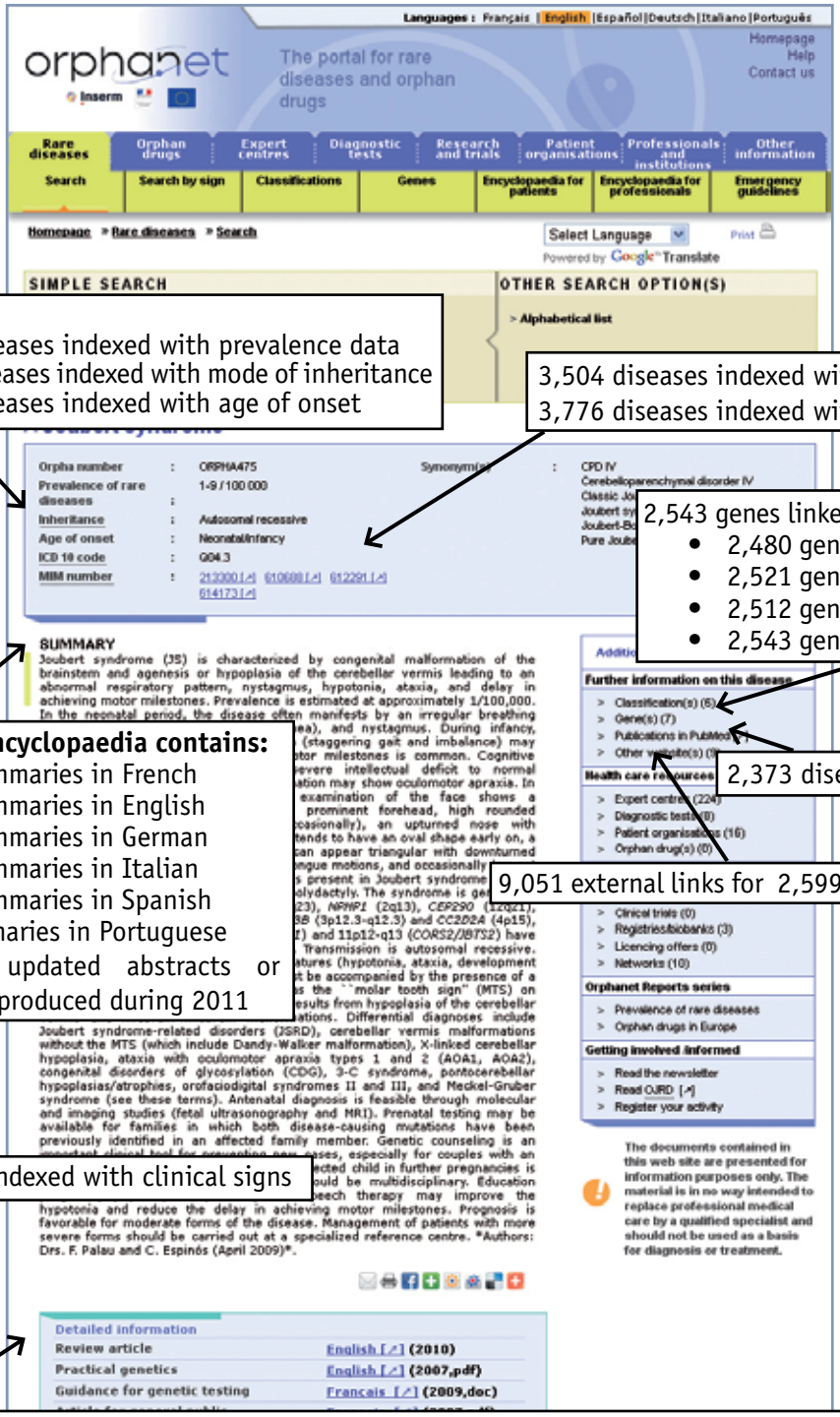
Armenia: Center of Medical genetics and Primary Health of Armenia
Austria: Gesundheit Osterreich GmbH
Medizinische Universitat Wien
Belgium: Federal Public Service health, Food Chain Safety and Environment
Wetenschappelijk Instituut Volksgezondheid, Institut de Santé Publique
Bulgaria: Bulgarian association for Promotion of Education and Science
Canada: Mc Gill University
Czech Republic: Univerzita Karlova v Praze, Charles University in Prague
Croatia: Zagreb University
Cyprus: Archibishop Makarios II Hospital
Denmark: University Hospital of Aarhus
Estonia: University of Tartu
France: Institut National de la Santé et de la Recherche Médicale (INSERM)
Germany: Medizinische Hochschule Hannover
Greece: Institute of Child Health Athens
Finland: The Family Federation of Finland - (Väestöliitto)
Hungary: National Center for Healthcare Audit and Improvement
Ireland: National Centre for Medical Genetics
Israel: Tel Aviv University
Italy: Hospital Bambino Gesù Roma
Lebanon: Université Saint Joseph Beyrouth
Lithuania: Vilnius University Hospital Santariskiu Klinikos Centre for Medical Genetics
Latvia: The National Health Service
Luxembourg: Ministère de la Santé du Luxembourg

Morocco: Department of Medical Genetics, Institut National d'Hygiène du Maroc
The Netherlands: Academisch ziekenhuis Leiden- Leids Universitair Medisch Centrum
Norway: Department for Rehabilitation and RD, Norwegian directorate of Health
Poland: Instytut Pomnik - Centrum Zdrowia Dziecka
Portugal: Instituto de Biologia Molecular e Celular
Romania: Universitatea de Medicina si Farmacie «Grigore. T. Popa»
Serbia: Institute of Molecular Genetics and genetic Engineering - Belgrade University
Sweden: Karolinska Institutet
Slovenia: Univerzitetni Klinicni Center Ljubljana, University Medical Centre Ljubljana
Slovakia: Children's University Hospital in Bratislava
Spain: Centro de investigation biomedica en Red de Enfermedades Raras
Ministry of Health and Social policy
Switzerland: Division of Genetic Medicine - University Hospitals Geneva
Turkey: Department of Human and Medical Genetics, University of Istanbul
United Kingdom: The University of Manchester



3. Evolution of database content

The disease and gene database contains **8461 diseases or groups of diseases** and their synonyms.



Epidemiology:
- 3,747 diseases indexed with prevalence data
- 3,800 diseases indexed with mode of inheritance
- 3,642 diseases indexed with age of onset

3,504 diseases indexed with ICD10
3,776 diseases indexed with OMIM

2,543 genes linked to 2,544 diseases, including:
- 2,480 genes interfaced with uniprot
- 2,521 genes interfaced with OMIM
- 2,512 genes interfaced with Genatlas
- 2,543 genes interfaced avec HGNC

The Orphanet encyclopaedia contains:
- 2,845 summaries in French
- 3,077 summaries in English
- 2,475 summaries in German
- 2,714 summaries in Italian
- 2,101 summaries in Spanish
- 268 summaries in Portuguese

1148 new or updated abstracts or definitions were produced during 2011

2,373 diseases indexed with Pubmed

9,051 external links for 2,599 diseases

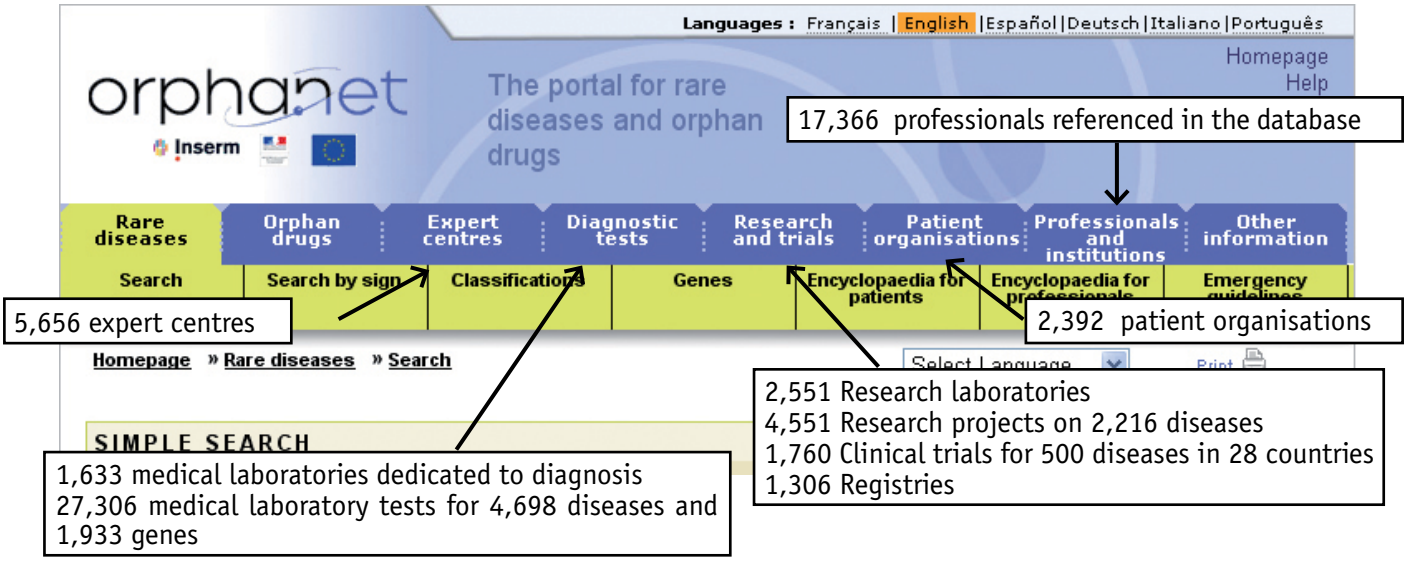
2,700 diseases indexed with clinical signs

573 articles in French of which 113 for the general public and 34 emergency guidelines
728 articles in English of which 9 emergency guidelines

The database of Orphan drugs and substances contains the following data:

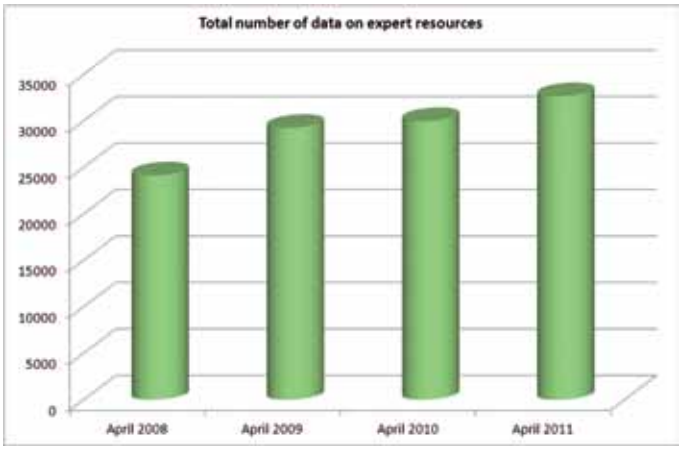
- 961 substances linked to more than 1137 orphan designations (EU and US)
- 133 European Marketing Authorisations (of which 62 after orphan designation and 71 with no previous orphan designation)
- 150 USA Marketing Authorisations
- These substances have a designation/indication for more than 550 rare diseases

The directory of services in 36 countries worldwide contains the following data:



The 36 countries in which Orphanet collects data are the following:
Armenia, Austria, Belgium, Bulgaria, Canada, 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/or annual updates are managed either by the teams at country level when they have sufficient funding for a dedicated professional, or by the coordinating team on behalf of the Orphanet national team. In 2011, countries managing both data collection and updates at country level are: Belgium, France, Germany, Italy, the Netherlands, Portugal, Spain, Switzerland and the United Kingdom (and Ireland).



Total amount of data on expert resources present in the Orphanet database per year.

Globally, the total amount of data on expert resources in the database increased in 2011 compared to the previous years. Moreover, the quality of the data present was also improved as, since April 2011, a Quality Manager is in charge of the global Quality assurance of the data and the data validation processes are agreed on with the national Health Authorities of the MS.

4. Orphanet products and services

4.1. New Functionalities of the Orphanet site in 2011

Implementation of 4 new functionalities of the Orphanet database in 2011 has improved the user friendliness and interactivity of the site and ensured a better dissemination of the data.

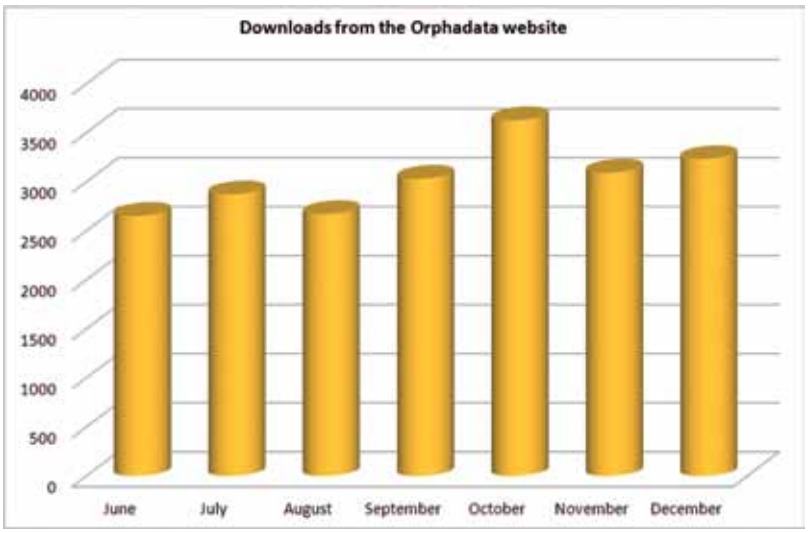
4.1.1. Orphadata

Since Orphanet is increasingly well-known as the reference source for documentation on rare diseases, a growing number of requests for data are received. To ensure dissemination of the Orphanet nomenclature of RD and to maximise the use of collected information on expert resources, orphadata.org was created. On this website, the whole Orphanet dataset has been directly accessible in a reusable format since June 2011.

Orphadata was developed within the context of the RD Portal and the Orphanet Europe Joint Action contracts funded by DG Sanco. Additional support is also provided by GSK. The dataset is a partial extraction of the data stored in Orphanet and is updated monthly.
Freely accessible in six languages (English, French, German, Italian, Portuguese and Spanish) the Orphadata dataset encompasses:

- An inventory of rare diseases, cross-referenced with OMIM, ICD-10 and with genes in HGNC, OMIM, UniProtKB and Genatlas
- A classification of rare diseases established by Orphanet, based on published expert classifications
- Epidemiology data related to rare diseases in Europe (class of prevalence, average age of onset, average age at death) extracted from the literature
- A list of signs and symptoms associated with each disease, with their frequency class within the disease
- It is also possible, on request, to access other types of Orphanet data, including:
- An inventory of Orphan Drugs at all stages of development, from EMA (European Medicines Agency) orphan designation to European marketing authorisation, cross-linked with diseases
- Summary information on each rare disease in six languages (English, French, German, Italian, Spanish, Portuguese)
- URLs of other websites providing information on specific rare diseases
- A directory of specialised services, providing information on expert centres, medical laboratories, research projects, clinical trials, patient registries, mutation registries, and patient organisations in the field of rare diseases, in each of the countries in the Orphanet consortium.

Orphadata provides a guide for users that defines and describes the elements of the dataset. Orphadata is intended to contribute to accelerating R&D and to facilitate global adoption of the Orphanet nomenclature.
Since June 2011, Orphanet products were downloaded more than 21,000 times, with an average of 3,000 times a month.



Total number of downloaded products from the Orphadata website

4.1.2. Translation of the Orphanet site into Portuguese

The international website and the database have been translated into Portuguese and since 23 February 2011, all the information is accessible to users from Portugal and Brazil in their own language.
The Portuguese version of the site has been heavily accessed since its launch.

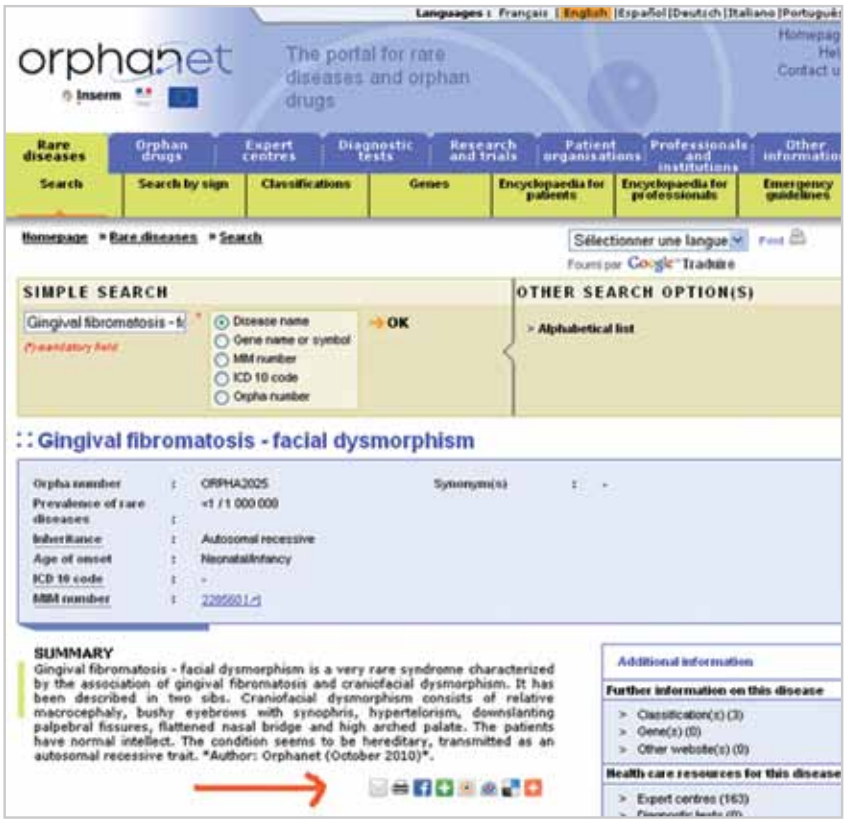
Overview of the 2011 views of the Orphanet website in Portuguese

Orphanet pages in Portuguese have been viewed 310,000 times since the launch in February 2011.

4.1.3. Interactivity

Interactivity for information sharing on Orphanet was implemented in 2011.
Share features are available on the disease pages of the international website in all languages. The react button has been implemented as a beta-test phase only on the French version of the website to evaluate the workflow

that it is going to generate before making it available for all the other languages. The comments generated using the react button are sent as a message to a coordinator and are not available on line.



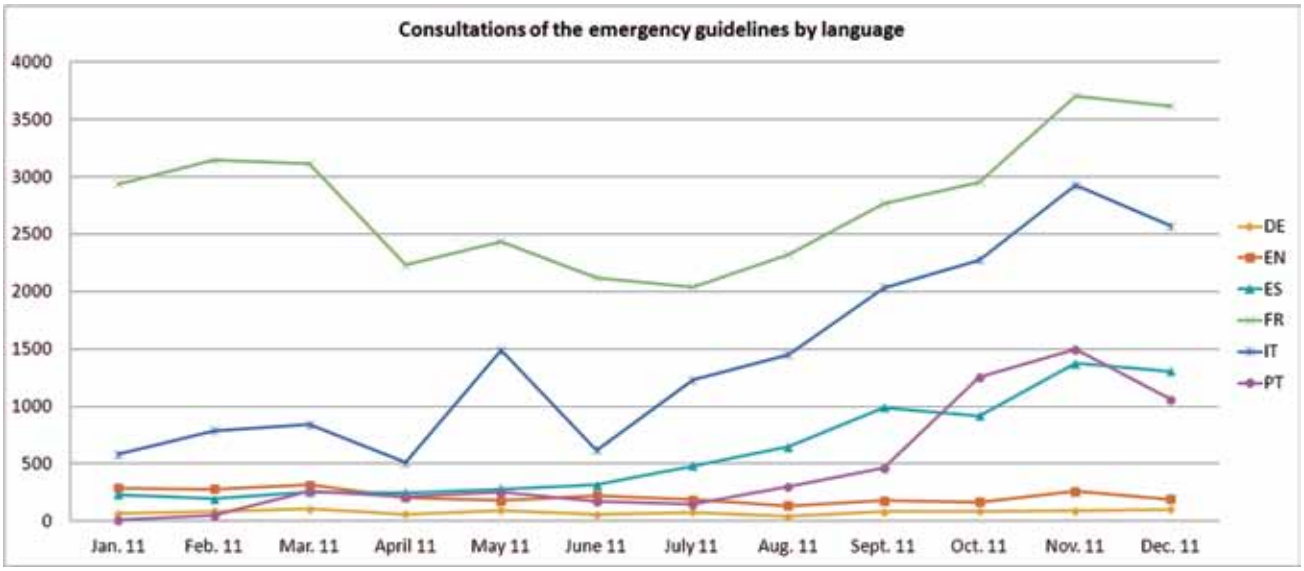
4.1.4. Therapy indications implementation

Therapy indications for the substances contained in Orphan drugs are available on-line in the detailed information section of the European Marketing Authorisation.

4.2. Evolution of previous products and services

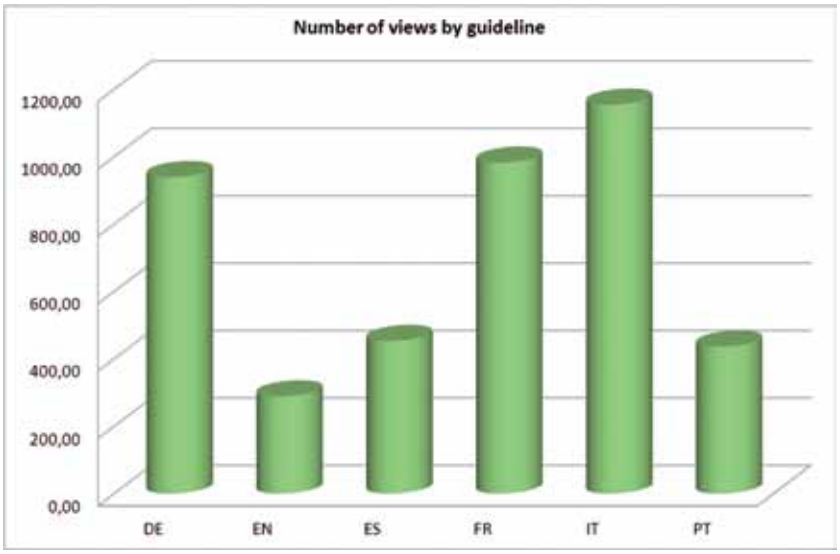
4.2.1. Extension of the Orphanet emergency guidelines

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 produced in collaboration with French reference centres and patient organisations, and are peer-reviewed by emergency health care physicians from learned societies: 34 emergency guidelines in French are now online. They are being translated into four languages (English, German, Italian and Spanish) since 2010 and Portuguese since 2011. Currently, 9 emergency guidelines are available in English, 16 in Spanish, 15 in Italian, 13 in Portuguese and one in German. There are 33 new guidelines compared to 2010.



Consultations of the emergency guidelines by language in 2011

Over 67,000 guidelines were viewed in 2011, versus less than 37,000 in 2010, representing an increase of 80% in a year. This global increase reflects the expansion of the collection in the different languages.



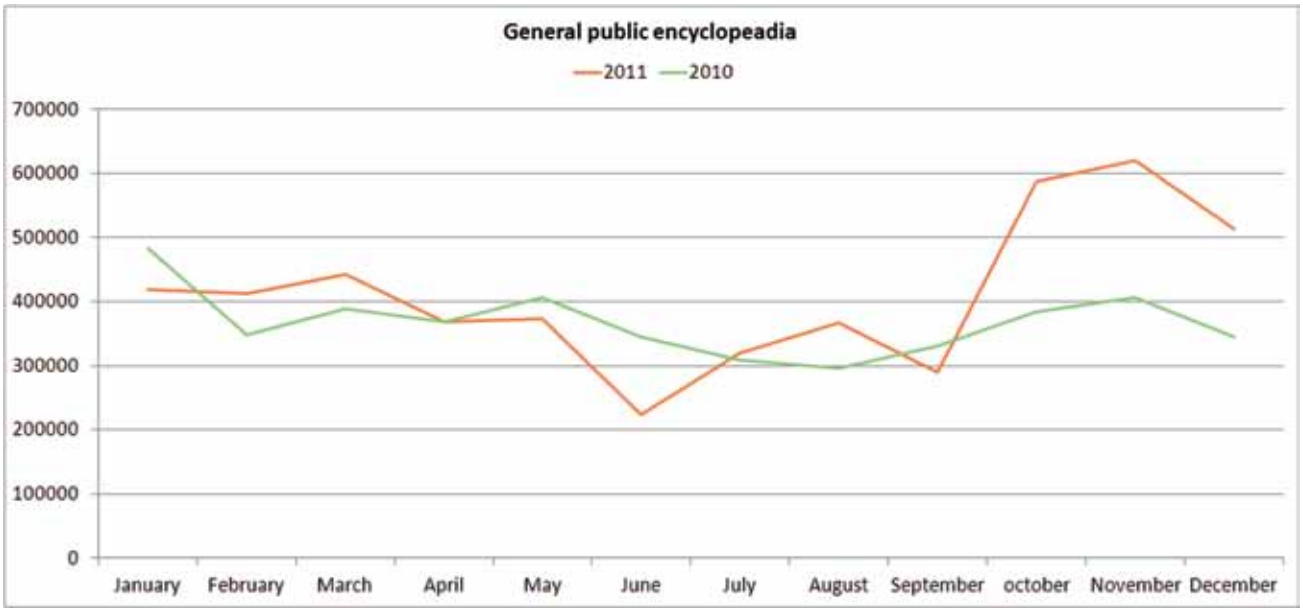
Language	Number of guidelines
German	1
English	9
Spanish	16
French	34
Italian	15
Portuguese	13

Number of views by language and by guideline

The number of consultations by guideline shows that this collection is a success in several languages such as French and Italian. Despite the fact that there is only one guideline in German, it is highly consulted. The collection of guidelines is still growing in Spanish and Portuguese and needs to be better identified by the rare disease community. On the other hand, the disappointing number of consultations in English can be explained by the existence of a larger number of equivalent documents in this language, which are produced by learned societies. To avoid dissipation of time and work, both precious for the rare disease community, Orphanet is currently establishing collaborations with these learned societies to provide links to these already existing and valuable resources.

4.2.2. Extension of general public encyclopaedia

The general public encyclopaedia was initially a French project intended to give complete, honest, up-to-date information to patients and their relatives on the diseases they are concerned by. Translation into other languages is planned for 2012 as well as the publication of general public-intended texts by expert centres or patient organisations (produced in compliance with a reliable methodology) in any other language.



Downloads from the general public encyclopaedia by month, comparison 2010-2011

As of 31 December 2011, 113 texts are online. Documents from this encyclopaedia have been downloaded more than 411,000 times per month, which corresponds to 4,950,000 downloads in 2011, representing a 12% increase compared to 2010.

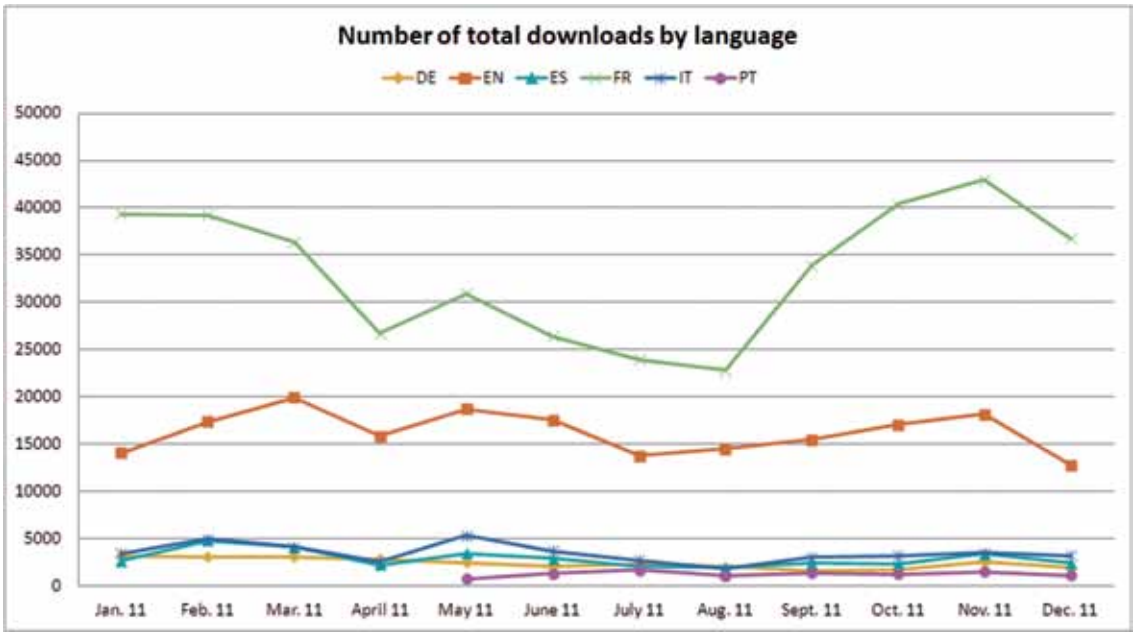
4.2.3. Orphanet Reports Series (ORS)

Orphanet reports are a series of texts providing aggregated data covering topics relevant to all rare diseases. New reports are regularly put online and are periodically updated. Acrobat Reader is required to read the texts as they are published as PDF documents accessible from the homepage page of the website. Here is the list of the ORS published in English:

- Prevalence or reported number of published cases listed in alphabetical order of disease (in six languages)
- Diseases listed by decreasing prevalence or number of published cases (in six languages)
- Patient Registries in Europe (in English)

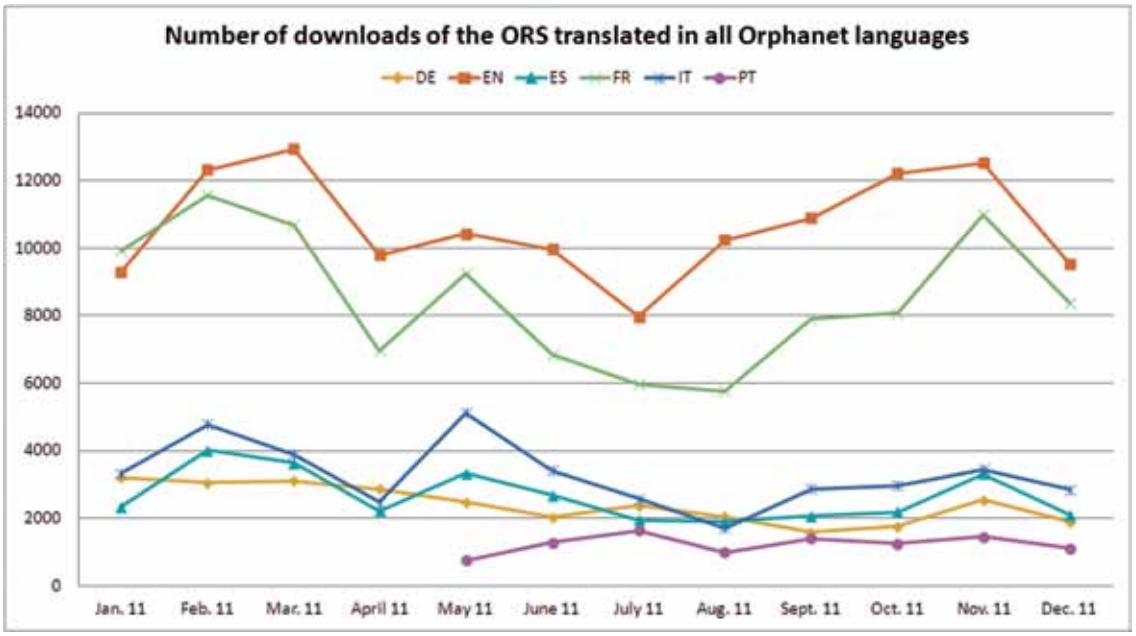
- European Research Projects and Clinical Networks (in English)
- Lists of orphan drugs in Europe (in six languages)
- Orphanet activity reports (in six languages)
- Orphanet Users Satisfaction Survey (in English)
- OrphaNews Europe Reader Satisfaction Survey (in English)

These publications are advertised in OrphaNews Europe, the newsletter of the EUCERD which has nearly 14,000 registered readers. All Orphanet Report Series are posted on the homepage of the website.



Number of total downloads by language

The Orphanet Report Series are heavily downloaded: in 2011, more than 710,000 Orphanet Report Series were consulted. This represents an increase of 25% compared to 2010 (around 570,000 downloads).



Number of downloads of the Orphanet Report Series translated in all Orphanet languages

Comparison of the number of downloads of the Orphanet Report Series translated in all 6 languages shows that this collection is heavily downloaded in French and English. The lower consultation rate in other languages could be explained by a lack of visibility of the ORS on the website. This issue should be resolved by the new Orphanet homepage that will be online in 2012.

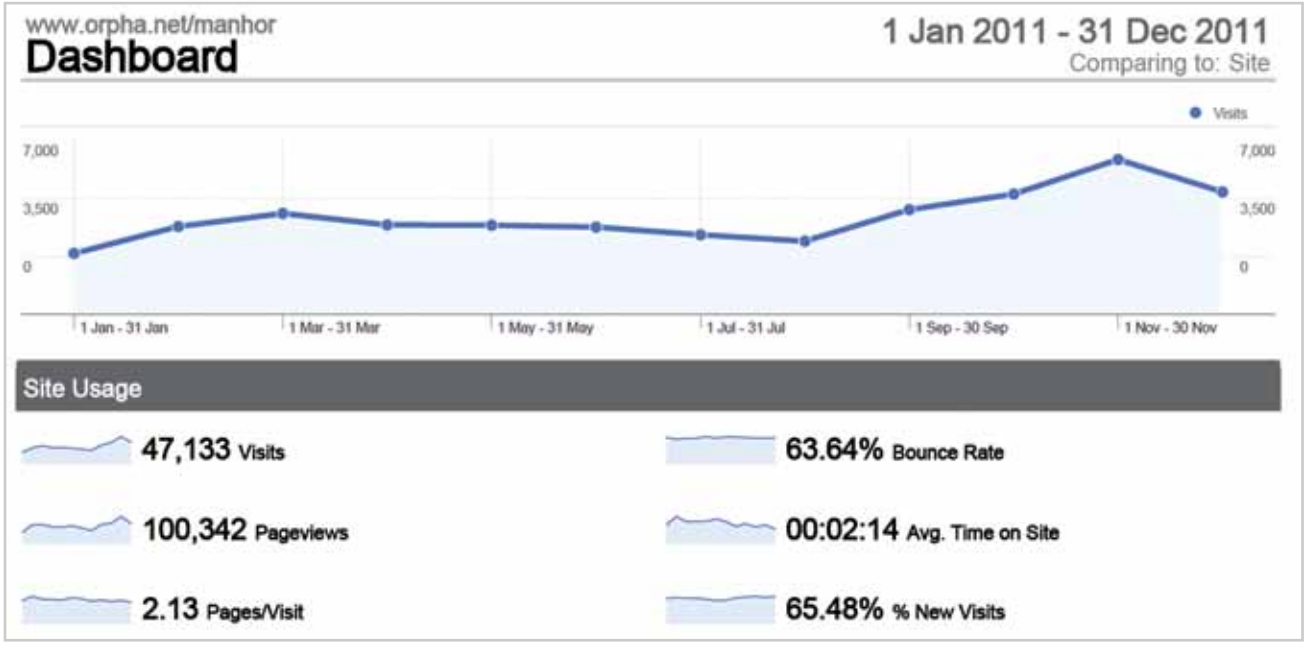
4.2.4. Success of the Orphanet Journal of Rare Diseases, an electronic, free-access journal on the Internet with BioMedCentral (www.ojrd.com)

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, 5.83 in June 2010 and 5.93 in 2011.
In 2011, 237 publications were submitted to the journal, double the number received in the previous year. Of these, 89 were accepted for publication.

4.2.5. Orphanet national websites

In order for Orphanet to become an instrument in national plans or strategies for rare diseases, the current international portal in five languages has evolved toward customised websites by country in national language(s).
National websites dedicated to each partner country enable them to have an entry point in their national language(s). The national pages include information on national events, news and will give access to national policy documents concerning RD and orphan drugs and to Orphanet Report Series in the 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 six languages. It is therefore a semi-solution pending the identification of resources for the true translation of the entire site.

As to 31 December 2011, 23 national websites in the national language are online: Austria, Bulgaria, Czech Republic, Estonia, Finland, France, Germany, Greece, Ireland, Israel, Italy, Lithuania, the Netherlands, Poland, Portugal, Serbia, Spain, Slovakia, Slovenia, Sweden, Switzerland and Turkey, and the United Kingdom.
There are 18 new sites compared to 2010.



Overview of visits to the 23 Orphanet national websites in 2011

The global increase of the number of visits, all websites together, reflects the growing number of published Orphanet national websites, and also the increased awareness of the existence of these national entry points by the users.

5. Users

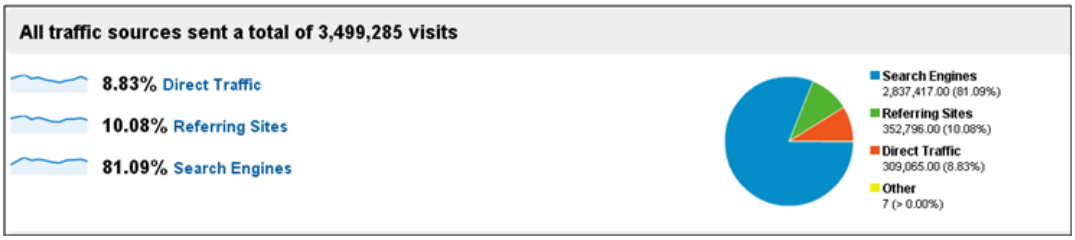
5.1. Indexation by search engines

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 2,060,000 responses. In comparison, NORD's site leads to 1,230,000 answers, GeneClinics 288,000 and INSERM 1,010,000.

The dissemination to the public is straightforward through the Orphanet website. In 2011, Google indexes 1,260,000 documents and pages originating from the web domain www.orpha.net, which is stable compared to one year ago for instance.

Users mainly access the Orphanet website through search engines (81.1% of visits according to Google Analytics) and Google alone accounts for 77.5% of queries. Other sites generating traffic to Orphanet represent 10.1% of visits. The remaining visits are made via direct access (bookmarks, 8.8%).

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 7.4% of visits. The indexation of our site is of the "long tail" type: more than 920,570 different keywords generate traffic to the site.



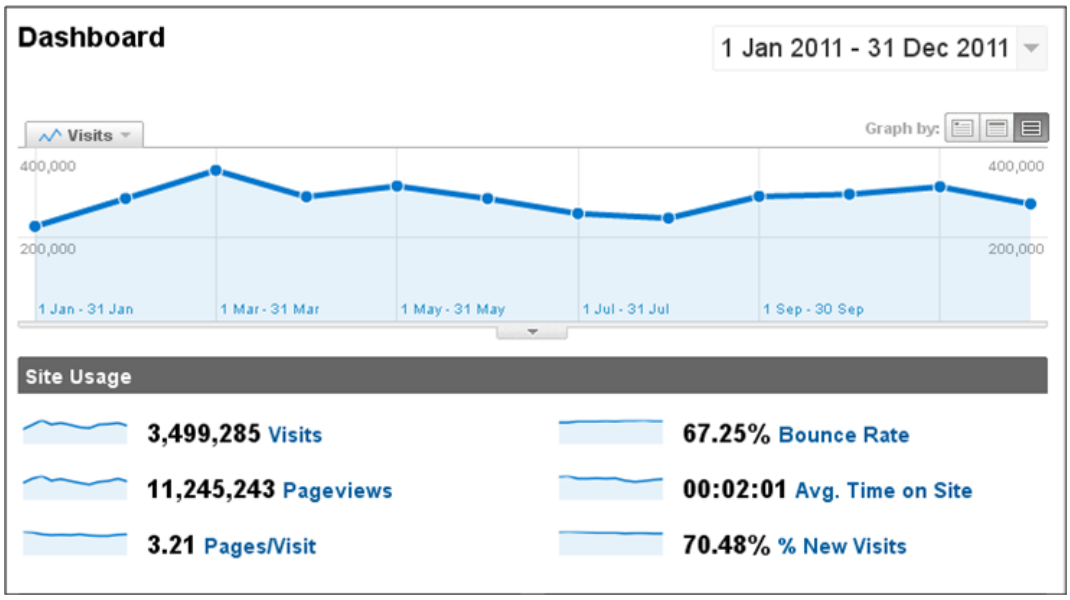
Distribution of the traffic sources
(Source: Google Analytics, 1st January 2011 to 31st December 2011)

Since November 2009, Google Analytics allows users to trace visits made from mobile devices (phones, iPad...). These visits represent 4.96% of all visits during 2011, i.e. 173,692 visits. In 2010, it only represented 1.56% (30,000 visits).

Consultations of the Orphanet website via a mobile device in 2011
(Source: Google Analytics, 1st January 2011 to 31st December 2011)

In conclusion, the indexation of Orphanet is satisfactory and the richness of our site explains its renown.

5.2. The website's audience



Orphanet website consultations in 2011
(Source: Google Analytics, 1st January 2011 to 31st December 2011)

In 2011, over 11,245,243 pages were viewed, thus on average around 30,800 pages viewed per day. This figure has increased from last year by 40% (8,000,000 page views in 2010).

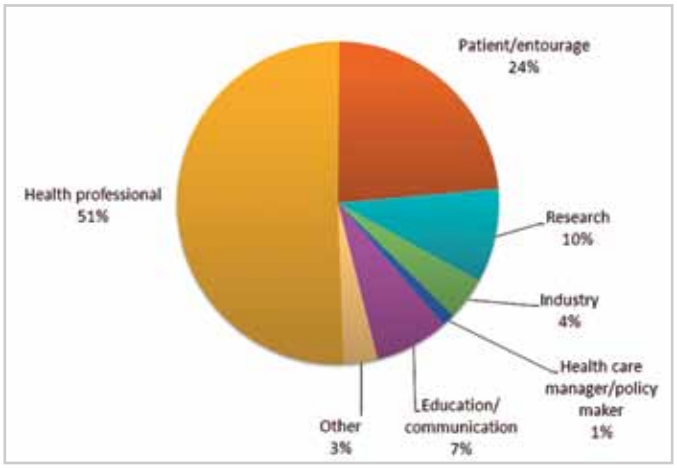
The Google Analytics tool does not include direct access to PDF documents. Yet this remains an entry point and generates a consistent volume of visits: each month, 875,000 PDF documents are consulted on the Orphanet website. This represents more than 10,500,000 downloads in 2011, which is around 20% more than in 2010 (870,000).
The geographical distribution of users still shows very varied sources (211 sources listed). The top ten countries are: France, Italy, Germany, Spain, United States, Brazil, Canada, Switzerland, Belgium, and Mexico.

5.3. Type of users and use

An online survey was carried out in December 2011. The satisfaction of the portal users was assessed by asking them to fill in a questionnaire until we obtain a thousand replies per language. This is the methodology which has been in place for the past 10 years at Orphanet. Interestingly, we had difficulty reaching the 1,000 answers to our questionnaire this year. We thus decided to stop the survey when 700 responses were collected.
The following results present the responses collected in English.

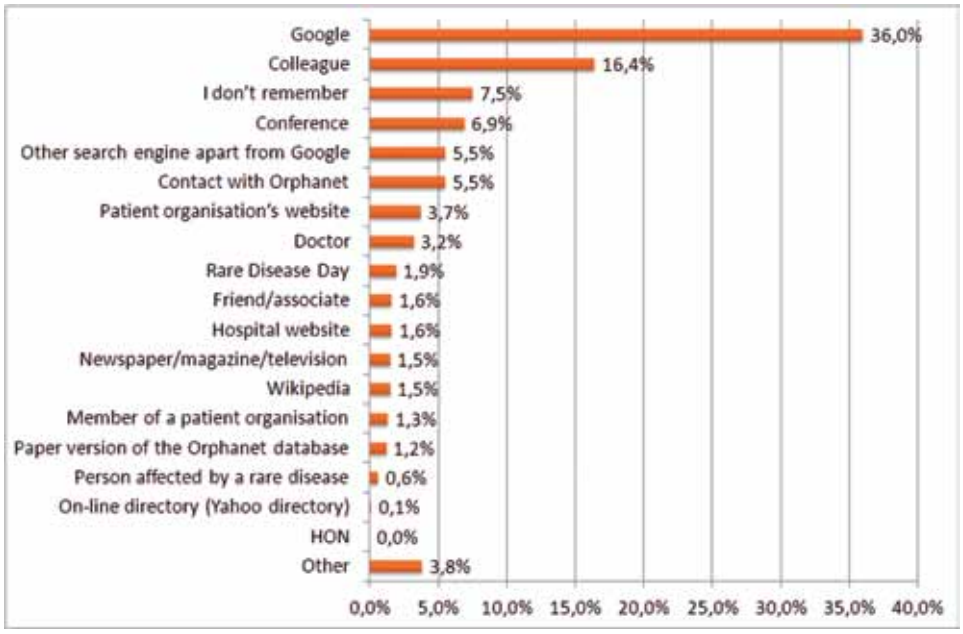
The site's users are:

Health professionals	50.7%
of which hospital specialists	29.3%
of which paramedics	7.8%
of which independent doctors	5.1%
of which biologists	2.8%
of which pharmacists	1.5%
Patient/relative or friend	23.4%
Researchers	9.4%
Industry	4.3%
Field of education/communication	7.4%
Health care managers/policy makers	1.3%
Other	3.4%



These results are consistent with 2010 figures.

Users discover the site in the following ways (one choice possible):



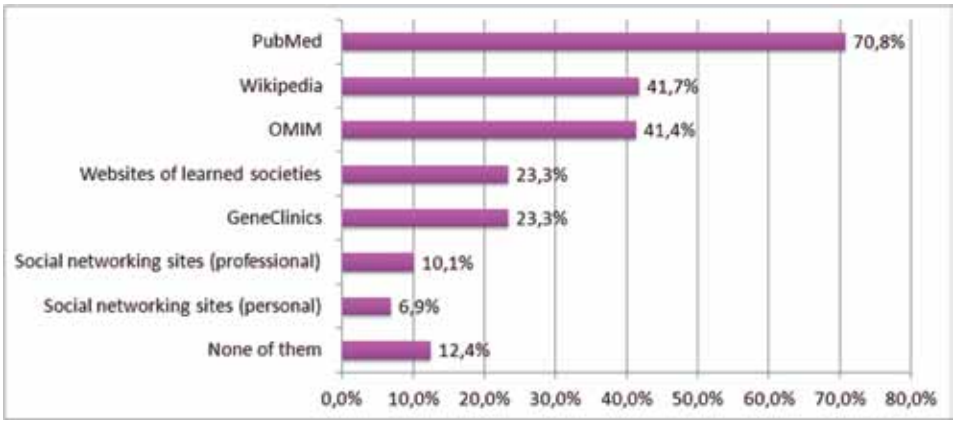
This question aimed to determine how respondents first learnt of Orphanet.
The results are consistent with 2010.

Frequency at which users visit the site:

- First visit 39.4%
- Over twice a year 19.2%
- Over twice a month 27.6%
- Over twice a week 13.7%

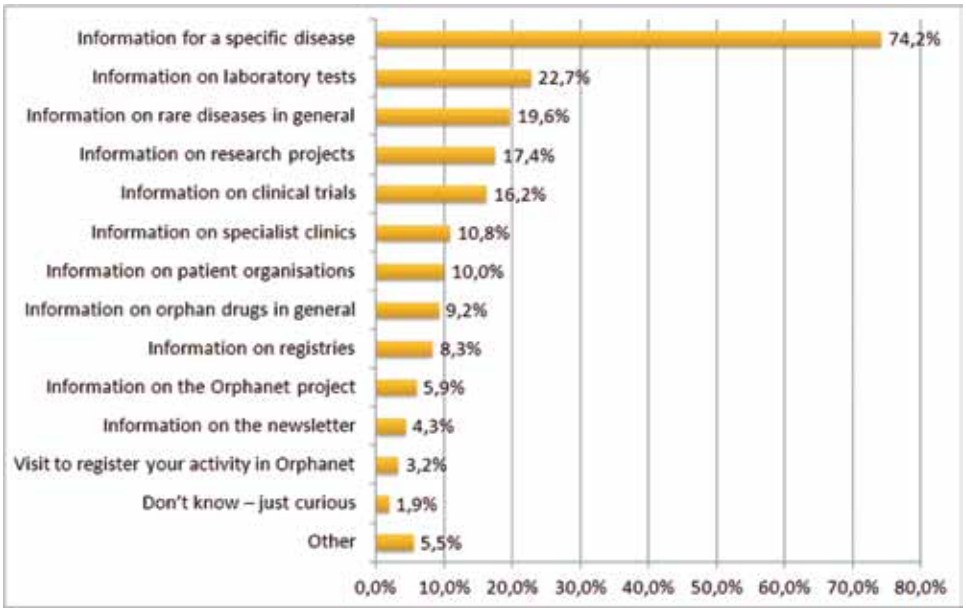
Our visitors seem to be more regular. Among the respondents, less than 40% were visiting the Orphanet website for the first time, which represents a 7% decrease compared to 2010. Those 7% were equally split between the 'Over twice a year' and 'Over twice a month' categories, that thus increased in 2011, while the number of our very frequent users remained the same (around 14%).

Other sites used to find information on rare diseases (more than one choice possible):



The respondent of this survey mainly use Pubmed as an additional source of information on rare diseases. Compared to 2010, the use of professional source of information like Pubmed or OMIM has increased by 7.5% and 6.5% respectively.

Information looked for on this particular visit (more than one choice possible):



The results show a clear trend: most of the users that answered our questionnaire were looking for information on a specific disease. Compared to 2010, our respondents seem to be looking more frequently for information on research projects and clinical trials.

Around 55 % of the users who answered the online questionnaire left comments, criticism and suggestions to improve the site, or words of thanks.

6. Network: the national and international collaborations of Orphanet

6.1. Collaboration with the European Commission

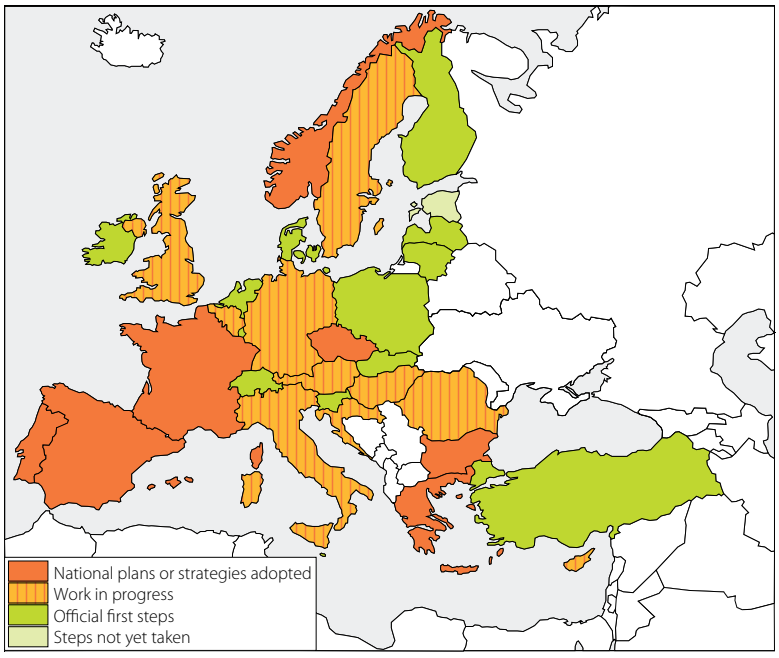
The Director of Orphanet chairs the European Union Committee of Experts on Rare Diseases (EUCERD, www.eucerd.eu) and provides the Scientific Secretariat.

6.2. 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, chaired by Ségolène Aymé, 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 ICD-11 related to rare diseases, which will be delivered in April 2012. The new version of the ICD should be adopted in 2014.

6.3. 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, Czech Republic, France, Greece, Norway, Portugal and Spain.

Work in progress: Austria, Belgium, Croatia, Cyprus, Germany, Hungary, Italy, Romania, Sweden and UK.

Official first steps: Denmark, Finland, Ireland, Latvia, Lithuania, Luxembourg, Malta, The Netherlands, Poland, Slovakia, Slovenia, Switzerland and Turkey.

Steps not yet taken: Estonia.

6.4. Scientific collaborations and partnerships with industry

Orphanet is also a partner of other European projects in the 7th Framework Programme (FP7): RDPlatform (end April 2011), CliniGene (end Dec 2011), Treat-NMD (end Dec 2011), ENCE (end April 2011) and EuroGentest 2 until 2013.

A major partnership was formed with GlaxoSmithKline (GSK) in 2010. This partnership was renewed in 2011. The company, which has recently opened a division dedicated to rare diseases, wishes to support the development of the disease database and of the Orphadata website, which are considered as strategic resources of interest to Industry.

A partnership was formed with the International Union of Basic and Clinical Pharmacology (IUPHAR) at the end of 2011 to cross-link Orphanet with the IUPHAR database of Receptors and associated Drugs.

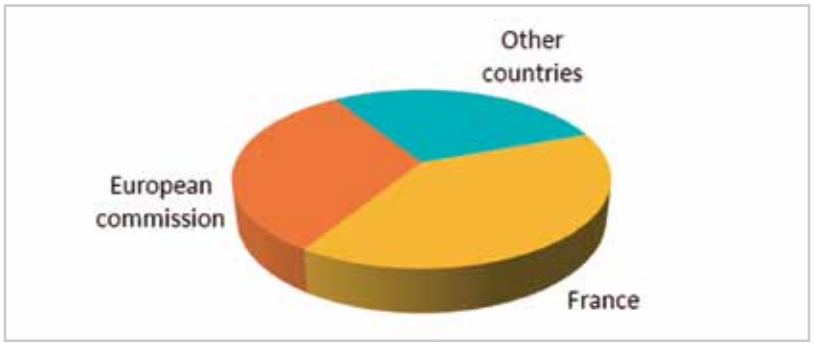
A partnership was formed with the European Bioinformatics Institute (EBI) at the end of 2011 to cross-link Orphanet's database with their genomic and their biological pathway data resources (Ensembl and Reactome).

A partnership was formed with the European Commission DG JRC Institute for Health and Consumer Protection, Molecular biology and genomics Unit in Ispra, Italy in 2011 to analyse the gene testing data collected by Orphanet (in collaboration with EuroGentest) to map the service offer in the MS and to analyse the test offer in comparison with the utility standards defined by the German Society of Human Genetics and the French Association of Molecular geneticists.

Partnerships were requested to use some Orphanet data through orphadata.org for research purposes, by the following organisations in 2011:
Profit: Bioxcel (DE), Booz Company GmbH (DE), CurrentMed (US), GSK (FR), Sanofi (FR)
Non profit: Division of Biomedical Informatics, Cincinnati Children's Hospital Medical Center Departments of Pediatrics and Biomedical Engineering (US), Centro de Investigacion Principe Felipe (ES), University of Copenhagen (DK), NIH Human Genome Research Institute (NHGRI) (US), Centro Superior de Investigación en Salud Pública (ES), University of Georgia (US), Ministry for Health- the Elderly and Community Care (MHEC) (MT), Nationales Aktionsbündnis für Menschen mit seltenen Erkrankungen National Action league for People with Rare Diseases – NAMSE(DE), Instituto de Biomedicina y Biotecnologia de Cantabria (IBBTEC) (ES), Joint Research Centre (JRC) (EC), Ciberer- Malaga University (ES).

7. Funding

Orphanet's budget was approximately €3M in 2011, originating from 13 different contracts in France and from various other contracts in some of the participating countries.

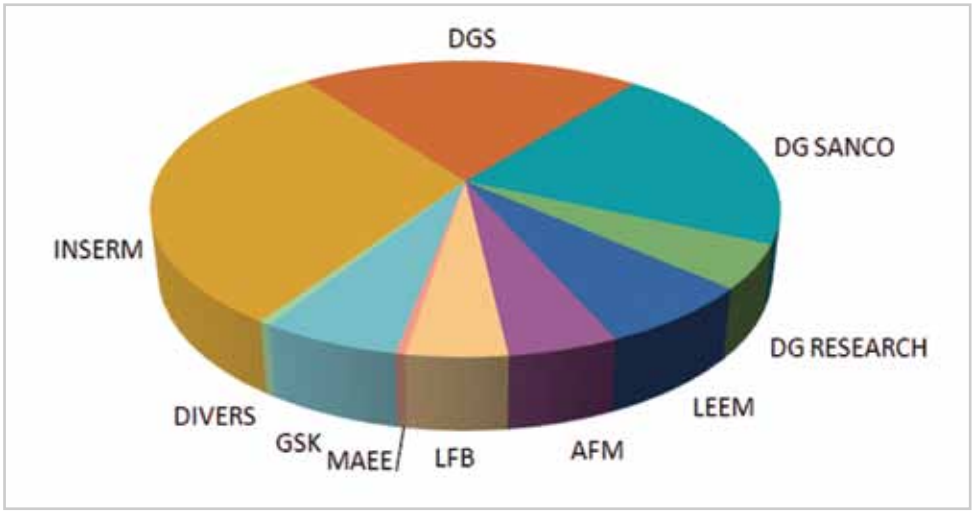


Orphanet global budget 2011

Globally, we can distinguish funding for the core activities and for national activities.

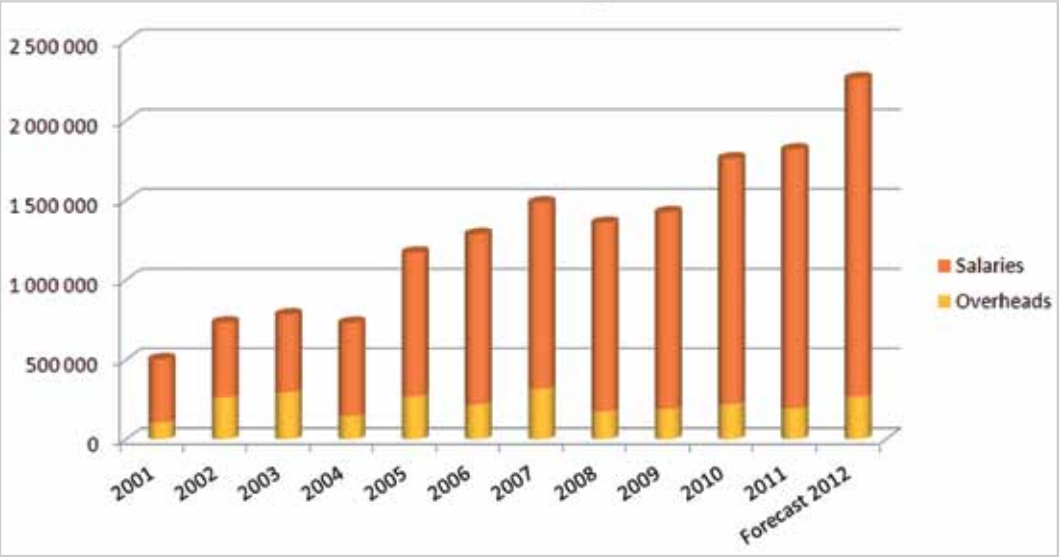
7.1. Orphanet's core activity funding

Orphanet's core activities represent the infrastructure, the coordination activities (management, management tools, quality control, rare disease inventory, classifications and production of the encyclopaedia) and communication.



Orphanet core activities funding 2011

In the last 10 years, the funding of Orphanet's core activities has quadrupled reflecting the growth of the project.



Core Activities funding evolution 2001-2012

Although there has been an extremely positive increase over the years, the current budget is still too limited in comparison to the needs of maintaining and updating a database of this size.

7.1.1. European funding

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

In 2011, Orphanet was funded by the following contracts:

DG Sanco
RDPortal2 - 20091215
Orphanet Europe (RDPortal3-JA) - 20112206
DG Research
RDPlatform - HEALTH-F2-2008-201230
Eurogentest2 - HEALTH-F4-2011-261469
Clinigene - LSHB-CT-2006-018933
ENCE - HEALTH-F2-2009-223355

7.1.2. Other current financial partnerships for core activity funding

Inserm	The "Institut National de la Santé et de la Recherche Médicale" 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 (DGS).
	The European Commission finances the database of diseases, the encyclopaedia in English, the coordination, the communication and IT of the project
CNSA	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	The "Association Française contre les Myopathies" finances OrphaNews France and OrphaNews Europe, as well as data collection on clinical trials.
leem Fondation	The "Fondation des Entreprises du Médicament" finances the collection of data on orphan drugs and clinical trials.
gsk GlaxoSmithKline	GlaxoSmithKline finances the extension of the database's disease annotation and free access to this data.
	The French "Ministère des affaires Etrangères" finances the cooperation with Canada

7.1.3. Current non financial partnerships for core activity funding

Non financial partners are those that provide services in kind and/or share their expertise for Orphanet core activities.

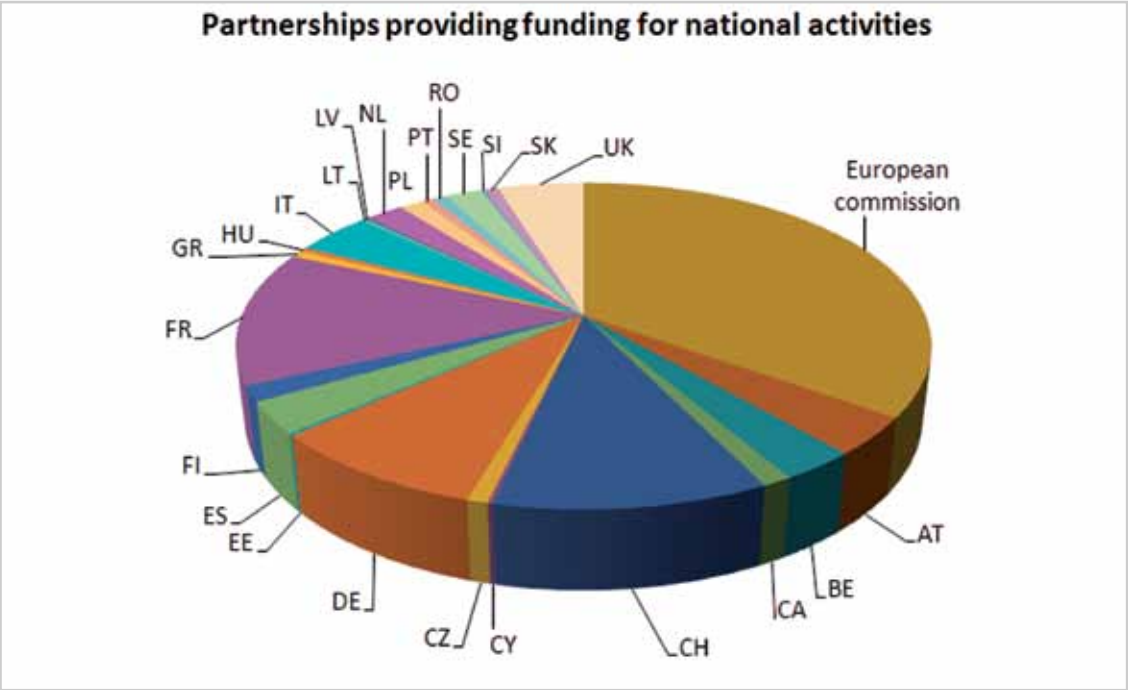
WHO	Orphanet collaborates with the World Health Organisation (WHO) in the process of revising the International Classification of Diseases.
GENATLAS	Genatlas collaborates with Orphanet in updating the data on genes involved in rare diseases.
UniProt	UniProt KB collaborates with Orphanet in updating the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

7.2. 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. In European countries, data collection at the national level is also supported by the European Commission.

7.2.1. Partnerships providing funding for national activities

Institutional partners host Orphanet national team activities and contribute to the project by allocating a budget and the time of some professionals. For European countries, this kind of partner is defined as an "Associated partner".



Partnerships providing funding for national activities in 2011

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.

CANADA	
CIHR IRSC	Canadian Institute of Health Research is the host institution of Orphanet Canada, finances a position for the project manager and provides some administrative support for the project.
RQMO	"Regroupement Québécois des Maladies Orphelines" finances a part time position for the deputy coordinator.
Centre universitaire de santé McGill McGill University Health Centre	Mc Gill University Health Centre finances a part time position for the deputy coordinator.
Relations internationales Québec	The Québec "Ministère des relations internatioanles" (ministry of international relations) finances data collection in Canada-Québec.
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.
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	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	
Bundesministerium für Gesundheit	The Federal Ministry of Health Germany provides funding to the Orphanet Europe Joint Action as of April 2011.
MHH Medizinische Hochschule Hannover	The Medical School of Hanover (MHH) supports data collection, and is an associated partner in 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	
	The Italian Health Ministry finances Orphanet-Italy activities through current research funding.
Bambino Gesù OSPEDALE PEDIATRICO	The Bambino Gesù Children's Hospital is an associated partner in the Orphanet Europe Joint Action as of April 2011.
FARMINDUSTRIA	Farmindustria finances Orphanet publications.
genzyme	Genzyme Italia finances OrphaNews Italia.
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. It hosts Orphanet Netherlands and co-funds the work of prof. dr. van Ommen.
CENTRE FOR Medical Systems Biology	The Centre for Medical Systems Biology is a joint activity of six institutions in the Netherlands, led by LUMC and including VUMC. The CMSB co-funds the rare disease work of the country coordinator, prof. dr. van Ommen, and, from April 2011 on, funds the work of the project manager.
VUmc	The VU Medical Centre funds the work of prof. dr. Cornel and, until the end of March 2011, co-hosted Orphanet Netherlands and funded part of the work of an information scientist.
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	
	The Spanish Ministry of Health, Social Services and Equality - Office for Health Planning and Quality is an associated partner in the Orphanet Europe Joint Action as of April 2011.
ciberer	The Centre for Biomedical Network Research on Rare Diseases (CIBERER) has been the partner for Orphanet in Spain since April 2010 and an associated partner in the Orphanet Europe Joint Action as of April 2011. CIBERER finances the main activities of the Spanish team.
SWEDEN	
Karolinska Institutet	The "Karolinska Institutet" is an associated partner in the Orphanet Europe Joint Action as of April 2011.
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	Since 2011, Orphanet Switzerland is funded by the Swiss Conference of the Cantonal Ministers of Public Health. The support finances the position for the coordinator (part-time), the positions for two information scientists (1 full-time from April 2011 and 1 part-time) and 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.
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.

7.2.2. Institutional partnerships providing services in kind for national activities

All the Institutions hosting the national Orphanet teams provide the office, all the necessary supplies to run the team activities, and allocate time of some of their professionals. For European countries, this kind of partner is defined as a "Collaborating partner".

ARMENIA	
	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.
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.
IRELAND	
	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.
LEBANON	
	The Saint Joseph University hosts Orphanet-Lebanon'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.
SERBIA	
	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.
TURKEY	
	The Istanbul University hosts Orphanet-Turkey's activities and contributes to the project by allocating the time of some professionals.

7.2.3. Non-financial partnerships in 2011

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.
RaDiOrg.be	A partnership was formed with RaDiOrg.be, which will coordinate the validation of the Belgian Patient Organisations for Rare Diseases.
IPG	The "Institut de Pathologie et de Génétique" provides the Belbis list which contains the most up to date information on genetic testing in Belgium and is used to verify the information in Orphanet.
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.
	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. The Czech National Plan is developed under the auspices of the Ministry of Health - Department of Medical Services.
MINISTRY OF HEALTH OF THE CZECH REPUBLIC	The Ministry of Health of the Czech Republic officially supports Orphanet.
ESTONIA	
sotsiaalministeerium	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	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 Ministry of Health officially supports Orphanet.
	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.
	The French Health Products Safety Agency (AFSSAPS) provides Orphanet with data on clinical trials in France.
	"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.
	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	
	The Federal Ministry of Health Germany officially supports Orphanet.
	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.
	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	
	The Ministry of Health of Hungary officially supports Orphanet.
ISRAEL	
	The Israeli Ministry of Health officially supports Orphanet.

ITALY	
	The “Istituto Superiore di Sanità“ officially supports Orphanet.
	Telethon collaborates with Orphanet in the collection of data concerning research projects.
	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 collaborates with Orphanet in the collection of data concerning clinical trials.
	Netgene collaborates with Orphanet in the diffusion of information on rare diseases.
	Mediart Promotion collaborates in the promotion of OrphaNews Italia.
	Italian Society for Paediatric Anaesthesia, Analgesia and Intensive Therapy (SIAATIP) collaborates in the revision of the Italian Emergency guidelines.
LATVIA	
	The Ministry of Health of the Republic of Latvia officially supports Orphanet.
	The Rare Diseases Society in Latvia which aims to promote equal rights and opportunities for patients with rare diseases.
	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	
	The Ministry of Health of the Republic of Lithuania officially supports Orphanet.
NETHERLANDS	
	The Ministry of Health, Welfare and Sport of the Netherlands officially supports Orphanet.
	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.
	Orphanet-Netherlands receives information from the “Vereniging Samenwerkende Ouder- en Patiëntenorganisaties“ (VSOP) on patient organisations dedicated to rare disease.
	Orphanet-Netherlands receives information from the Steering Committee on Orphan Drugs on patient organisations and orphan drugs dedicated to rare disease.
	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	The patient organisation, Ars Vivendi, provides patients and parents with information about Orphanet services and cooperates with Orphanet Poland.
PORTUGAL	
	The Portugese Ministry of Health officially supports Orphanet.
infarmed	INFARMED (National Authority of Medicines and Health Products, IP) collaborates in the update and validation of orphan drugs approved and available in Portugal.
Administração Central ACSS do Sistema de Saúde	ACSS (Central Administration of Health System) acknowledge that Orphanet-Portugal is the reference source of information on rare diseases and orphan drugs in Portugal.
ces	CES (Social Studies Center) of University of Coimbra collaborates in the update and validation of Portuguese Patient Associations.
N.E.D.R.	NEDR (Center for Study of Rare Diseases) of the Portuguese Society of Internal Medicine collaborates in the update and validation of activities about rare diseases ongoing in Portugal.
Aliança Portuguesa de Associações das Doenças Raras	The Portuguese Alliance of Rare Diseases Organizations collaborates in joint actions with Orphanet-Portugal and update and validation of Portuguese Patient Associations.
ROMANIA	
MINISTERUL SĂNĂTĂȚII	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.
ASOCIAȚIA 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 POLITICA 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	ProRaris, the Swiss Alliance of patients with rare diseases has established a close collaboration with Orphanet Switzerland 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.
IG Seltene Krankheiten CI Maladies rares CI Malattie rare	Orphanet Switzerland is member of the «Community of Interest for Rare Diseases» launched in August 2011. This community brings together all the relevant stakeholders in the field of rare diseases in Switzerland in order to develop, in collaboration with the Federal Office of Public Health, a national strategy for rare diseases.
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.
Ataxia UK	Ataxia UK and Orphanet cooperate in the exchange of information, in the validation and online publication of research projects regarding Ataxia and in the endorsement and boosting of Orphanet and Ataxia UK activities.
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.

8. Communication strategy

8.1. Communication documents

For the third 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 2011:

- 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 2011, we distributed A5-size flyers to present an overview of Orphanet and A5-size flyers presenting Orphadata were designed, printed and distributed.

The leaflets were distributed at approximately 20 events in 2011 for a total of 1,700 leaflets in French, and 34,000 leaflets in English.

8.2. Invitations to give lectures at conferences in 2011

Orphanet was invited to participate in more than 70 conferences, in Europe and abroad. 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 presenting of the Orphanet database (49), public health policies (29), classifications of diseases (8) or orphan drugs (2), medical and genetic approaches (45 presentations).

8.3. Booths at conferences in 2011

Orphanet booths were held in 6 different congresses in 2011 and 4 different events. Amongst these as in previous years, Orphanet had a booth at the annual meeting of the European Society of Human Genetics, which was held in Amsterdam (the Netherlands) from 28 to 31 May 2011 and for the first time at the International Congress of Human Genetics held in Montreal from 13 to 15 October 2011.

9. The Orphanet team as of December 2011

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