Orphanet Report Series

Reports collection

2012 Activity Report

orphanet

2012 Activity Report

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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 and orphan drugs in order to contribute to the improvement of the diagnosis, care and treatment of patients with rare diseases.

1.2. 2012 activities

Orphanet is a 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, OMIM, MeSH, SNOMED CT, UMLS, MedRA and its 'identity card' includes the relevant prevalence class, age of onset class, mode of inheritance and associated genes. At the moment, some 'identity cards' have yet to be completed.

- An encyclopaedia covering more than 4000 rare diseases, written by science writers and reviewed by world-renowned experts. Abstracts are produced in English and are then translated into French, German, Italian, Portuguese, Spanish, Polish and Finnish. For some selected diseases, emergency guidelines and articles for general public are produced and translated.

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

- A directory of expert resources in the 37 partner countries, providing information on: specialised expert centres and centres of expertise, medical laboratories, research projects, clinical trials, registries, networks, technological platforms and 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.
- Quality articles published by other journals or learned societies. More than 830 articles have been published with the authors' and editors' permission, comprising national and international clinical guidelines produced by learned societies that are not always published in peer-reviewed journals but available as reports.

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 rare diseases in terms of referenced documents.

1.3. Main achievements of 2012

- The new Orphanet homepage was launched in the summer of 2012. This new version is intended to meet the needs of site users who requested a less cluttered, more user-friendly homepage.

- All the countries that constitute the Orphanet consortium have a national website that is either published entirely in their national language or with a layout published in English and the mandatory texts in the national language. This represents 14 new national websites in 2012.

- The consortium has expanded toward Australasia: Western Australia joined Orphanet in 2012. Negotiations were initiated with Argentina, Brazil, China, Chile, Japan and Russia.

- The Orpha number has been implemented in the French information systems for all hospitalised patients so as to better identify patients in the healthcare system and to improve knowledge of their healthcare pathways, as of 1 December 2012.

- The servers hosting Orphanet have moved from Paris to the largest civil data centre in France which is based in Montpellier, the CINES, to improve the architecture of the servers and upgrade security.

- A "suggest an update" button is available at the bottom of disease pages to allow experts and/or users to suggest modifications and comments to disease information available online.

- Diseases are now cross-referenced with UMLS, MeSH, MedDRA and SNOMED CT.

- Genes are now cross-referenced with Ensembl, Reactome and IUPHAR.

- The Orphanet ontology of rare diseases is now available on BioPortal.

- The encyclopaedia of rare diseases has been expanded and updated. As of 31 December 2012, some abstracts are available in Finnish and Polish in addition to English, French, German, Italian, Spanish, and Portuguese.

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

- A survey of the centres of expertise listed on Orphanet has been organised to check whether they match the quality criteria defined by the European Union Committee of Experts on Rare Diseases (EUCERD).

- The list of rare diseases (in English and French) has been published as an Orphanet Report Series for more effective communication.

- Most of the Orphanet Report Series have been updated (List of rare diseases, Prevalence of Rare Diseases, Lists of Orphan Drugs, Orphanet Activity Reports, and Satisfaction Surveys).

- The Orphanet Activity report 2011 has been translated into French, Italian, Spanish and Polish.

2. Orphanet consortium

2.1. The Orphanet Europe Joint Action

Orphanet 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 rare diseases in the European Union and also as a strategic element of any national plan/strategy on rare diseases that each Member States is encouraged to develop by the end of 2013. In 2011, the Orphanet Europe Joint Action was launched, an instrument that combines funding from the European Commission with each of the participating Member States, as well as from Switzerland, a collaborating partner. The three-year Joint Action began on 1 April 2011. The overriding aim of the Joint Action is to improve and adapt the presence of Orphanet in each participating country.

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 is organised in three different boards:

- The **Management Board** composed of country coordinators;
- 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.

On 23 May 2012, the Joint Action Orphanet Annual Partners' Meeting was held in Brussels. Future funding instruments for Orphanet were discussed with the European committee representatives. Several options were analysed but the subject remains pending at the European level.

2.2. Expansion of the consortium

Since its creation, the quality of the data has built its reputation and Orphanet has grown as a European consortium, gradually expanding into 35 neighbouring countries to the East and the South. In 2011, Orphanet has gone further West to include Canada. In 2012, the consortium expanded towards Australasia with the joining of Western Australia.

The number of countries wishing to join the consortium is growing every year. Joining Orphanet rather than creating a system *de novo* allows new members to reap the benefits from the investment already made in the infrastructure. During 2012, discussions took place with six new countries: Argentina, Brazil, China, Chile, Japan and Russia.
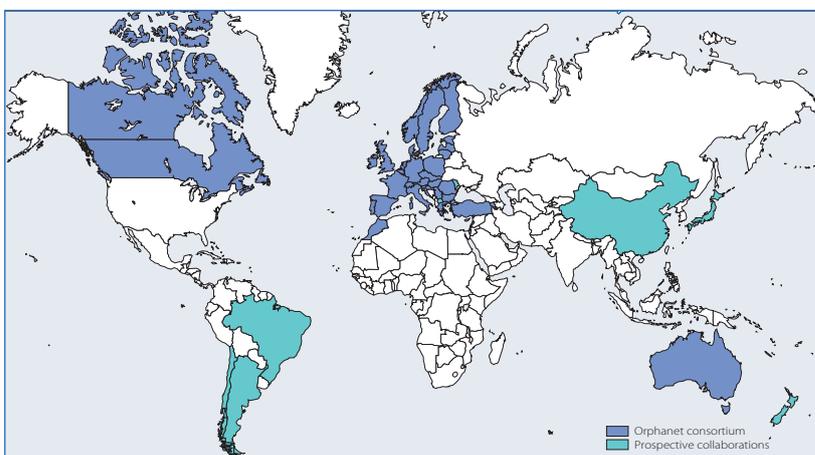
2.3. List of partners and scope of their activity

2.3.1.COORDINATING TEAM

The coordination of the consortium is managed by the coordinating team, Orphanet France, located in the Unit of Service 14 of INSERM (the French National Institute of Health and Medical Research). INSERM has been the coordinator of the Orphanet consortium since 2001.
The coordinating team is responsible for the coordination of consortium activities, the hardware and software aspects of the project, the database of rare diseases and the production of the encyclopaedia, as well as 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 in each country.

2.3.2.PARTNERS

The establishment of a Directory of Services can only be achieved by consolidation of data collected at the country level. The identification of expert resources requires a very good knowledge of the national research and health care institutions and their organisation. 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 organisations.
Translations of the Orphanet content in the national language are also managed by the national teams when they have a sufficient budget. Currently Belgium, Germany, Italy, Spain and Portugal are undertaking the translation of the entire website's content into their national language, while the Polish and Finnish teams are translating the encyclopaedia.

Management of the national website/entrypoint to the Orphanet portal is also carried out by every national team in their national language.

2.3.3.LIST OF ORPHANET PARTNER INSTITUTIONS

Armenia:	Center of Medical genetics and Primary Health of Armenia
Austria:	Gesundheit Österreich GmbH / Austrian Health Institute
	Medical University of Vienna
Australia	Office of Population Health Genomics Departement of Health, Governement of Western Australia
Belgium:	Federal Public Service Health, Food Chain Safety and Environment
	Institute of Public Health, WIV-ISP
Bulgaria:	Bulgarian Association for Promotion of Education and Science
Canada:	CIHR Institute of Genetics
Czech Republic:	2nd Faculty of Medicine, Charles University in Prague
Croatia:	Zagreb University
Cyprus:	Archibishop Makarios II Hospital
Denmark:	University Hospital of Aarhus
Estonia:	University of Tartu
France:	National Institute of Health and Medical Research
Germany:	Hannover Medical School
Greece:	Institute of Child Health Athens
Finland:	The Family Federation of Finland (Väestöliitto)

Hungary: National Institute for Health Development
Ireland: National Centre for Medical Genetics
Israel: Schneider Children's Memorial Medical Hospital
Italy: Bambino Gesù Hospital, Rome
Lebanon: Saint Joseph Beirut University
Lithuania: Vilnius University Hospital, Centre for Medical Genetics
Latvia: Centre for Disease Prevention and Control
Luxembourg: Ministry of Health of Luxembourg
Morocco: Department of Medical Genetics, National Institute of Hygiene in Morocco
The Netherlands: University Hospital Leiden, Leiden University Medical Center
Norway: Department for Rehabilitation and RD, Norwegian Directorate of Health
Poland: The Children's Memorial Health Institute
Portugal: Institute of Molecular and Cell Biology
Romania: Medical University of Pharmacy « Gregory T. Popa »
Serbia: Institute of Molecular Genetics and Genetic Engineering-Belgrade University
Sweden: Karolinska Institute
Slovenia: University Medical Centre Ljubljana
Slovakia: Children's University Hospital in Bratislava
Spain: Centre for Biomedical Network Research on Rare Diseases
Ministry of Health and Social Policy
Switzerland: CMU, Institute of Medical Genetics
Turkey: Department of Human and Medical Genetics, University of Istanbul
United Kingdom: The University of Manchester
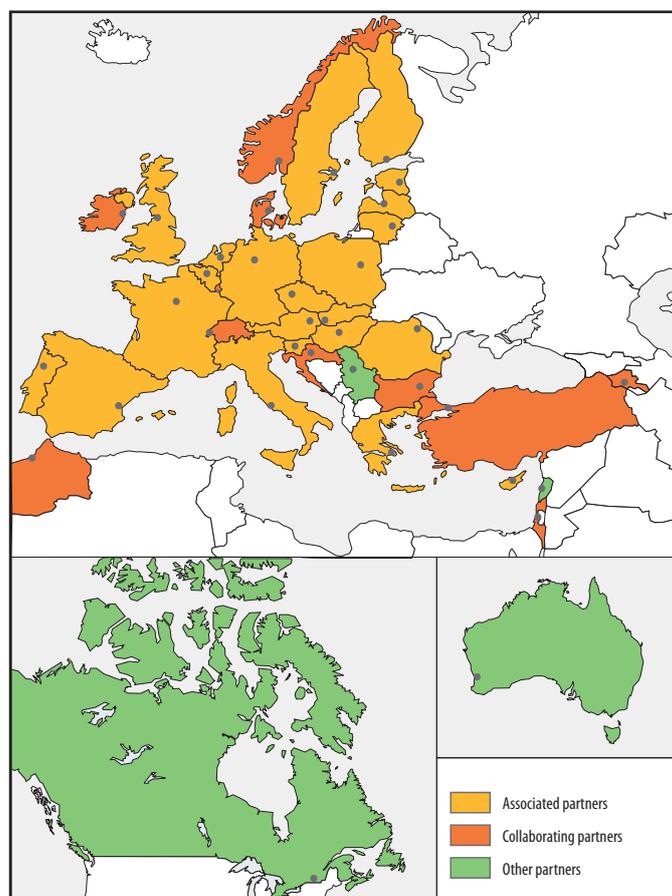
The Orphanet consortium partners

3. Orphanet products and services

Entries in the Orphanet diseases database correspond to rare diseases, rare forms of common diseases, or, in some particular cases, non-rare diseases considered to be orphan because their diagnosis or management remains particularly difficult or they are very often requested of Orphanet.

The updating of the database is based on a literature watch of international journals that helps to identify new syndromes, genes or treatments, update classifications of diseases, and is the basis for the production of various texts (encyclopaedia, recommendations ...). All texts are produced in collaboration with internationally recognised experts, learned societies and patient organisations.

All the teams that make up the Orphanet consortium are responsible for the collection, validation and submission of data on clinical trials, diagnostic laboratories, expert centres, ongoing research projects, registries and national patient organisations.

To publish data which are relevant and accurate (complete, valid, consistent with other data from the database), a validation and quality control is performed by the coordinating team, and regular updates are performed with other country teams via an intranet.

3.1. The Orphanet website

The Orphanet website provides a user-friendly homepage with ergonomics that have been designed to provide easier access to the numerous services offered by Orphanet and to enhance usability, with specific emphasis on improving accessibility for visually impaired users. Indeed, the typeface is magnified and information is organised in easy-to-spot blocks that allow users to more readily navigate the site. The disease search function is in the center of the homepage, while the tabs for Orphanet's other principal resources are organised into a table of contents. The popular Orphanet Report Series, which address relevant rare disease and orphan drug topics, are now highlighted in a specific area. Finally, OrphaNews, the official newsletter of the European Union Committee of Experts on Rare diseases (EUCERD), is easily identified in the top right.

To help users navigate the website, a list of our principal services is proposed on the "Help" page. Services are categorised to accommodate different user profiles.

3.1.1. ORPHANET NATIONAL WEBSITES

In order for Orphanet to become an instrument in national plans or strategies for rare diseases, the international portal in six languages has evolved towards customised websites by each country in their 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 access to policy documents concerning rare diseases and orphan drugs. Beyond the scope of national information, these pages provide access to the international database in six languages.

As of 31 December 2012, 37 national websites are online. These include 14 new sites compared to 2011: Armenia, Australia, Belgium, Canada, Croatia, Cyprus, Denmark, Hungary, Latvia, Lebanon, Luxembourg, Morocco, Norway and Romania. Some of these national websites are published completely in their national language while for other countries, the layout of the national webpage is in English and the mandatory texts (General Information) are in their respective national language.
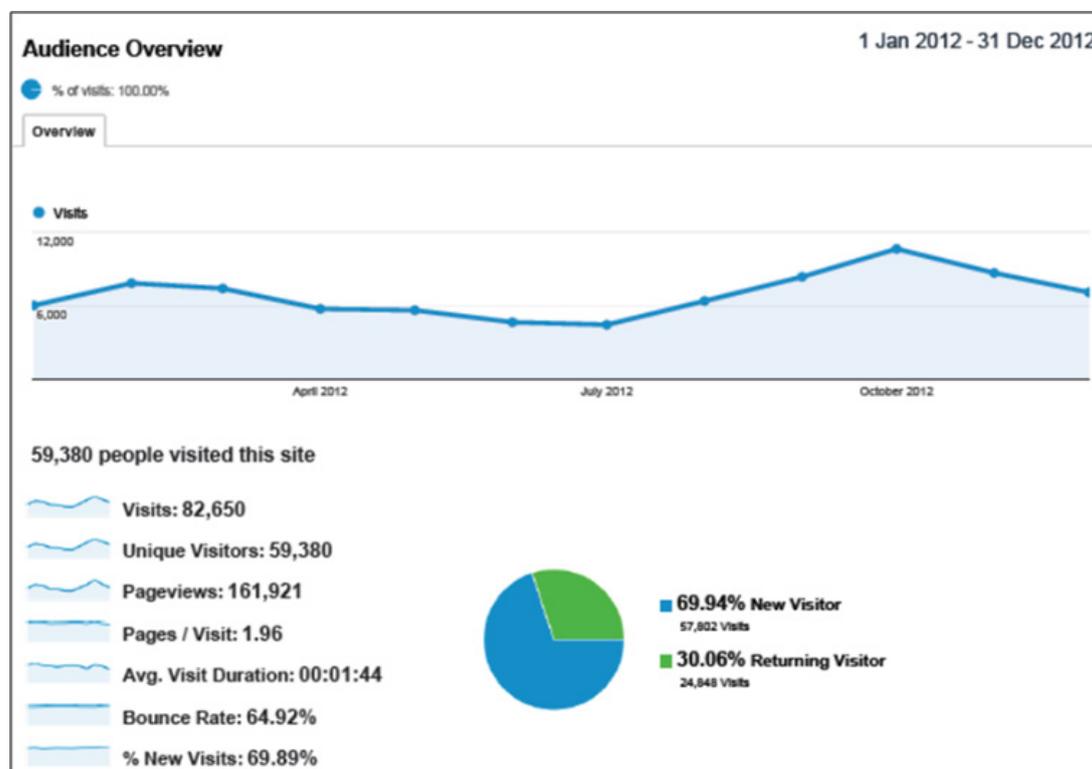
Overview of visits to the 37 Orphanet national websites in 2012

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.

3.1.2. THE ORPHANET SERVERS

From 2007 to 2012, the servers needed to run all the sites and services that Orphanet offers were hosted by the DSI (Department of Information Systems) of INSERM. Growing demand and an increased workload (more than one million pages per month are viewed on the site www.orpha.net), along with the need to develop new services in an appropriate technical architecture, led to a complete reorganisation that required the purchase of software licences and new, more powerful servers.

We decided to increase our capacity by installing new machines and to host the production servers at the largest civil data centre in France, the CINES (Centre Informatique National de l'Enseignement Supérieur). To ensure structural security we opted for a low-cost solution by housing development servers at INSERM structures that are geographically close to CINES and which are linked by a fiber optic connection. This allowed us to have an excellent connectivity between production servers, development servers and backup environments.

This organisation now has many production environments: backoffice, pre-production, preservation and development environments. This enabled us to improve our Activity Recovery Plan (PRA) of the Orphanet website.

Migration operations took place between March 2012 and September 2012. The migration of services happened without any interruption of access to the Orphanet website, with less than a week of inconvenience for all

other services. We have also secured the access to back-office tools used by the coordinating team in France and internationally. To do this we set up VPN (Virtual Private Network) servers and deployed VPN clients across the teams in the 37 countries.
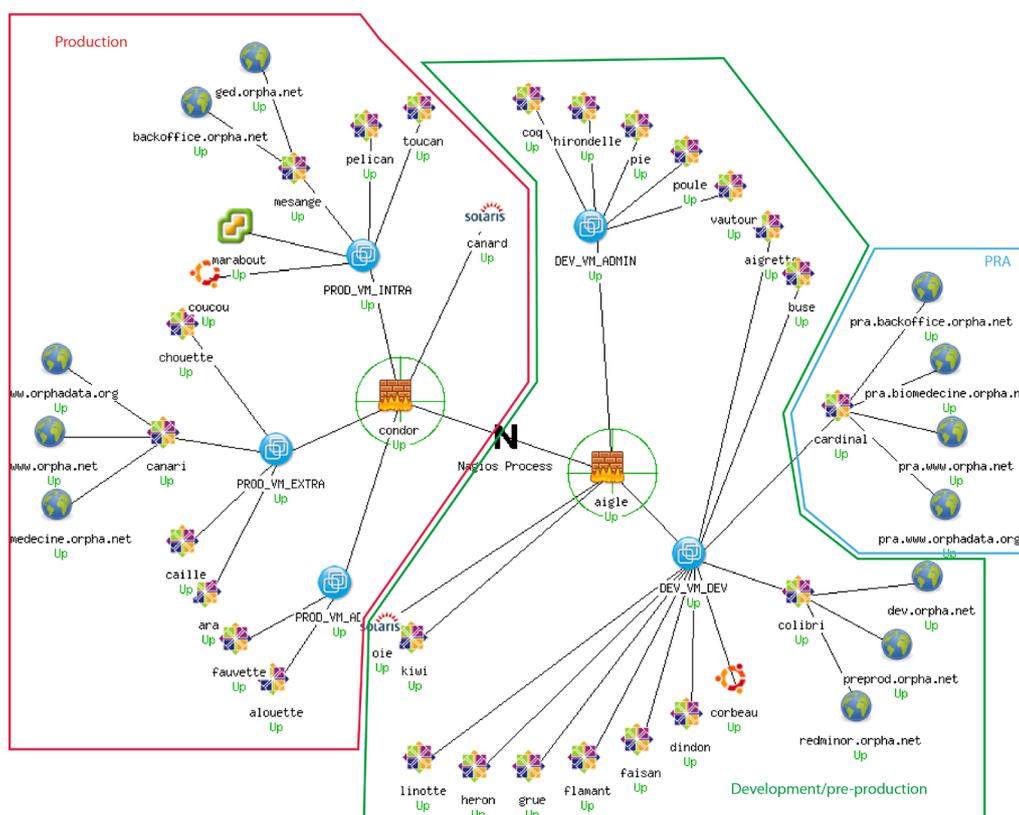
The Orphanet IT architecture

3.2. Orphanet inventory of rare diseases

Orphanet provides a comprehensive inventory of rare diseases which is updated on a monthly basis according to new publications.

The inventory of rare diseases is classified according to a polyhierarchic classification system of rare diseases. This system consists of classifications organised according to the medical and/or surgical speciality that manages specific aspects of each rare disease within a healthcare system. When relevant, a distinction is made between childhood and adult disorders. The diseases have been classified within each speciality according to clinical criteria or etiological criteria when diagnostically or therapeutically relevant. The Orphanet classification system employs two non-exclusive sources: documented sources or expert advice. An evolutionary process that is updated regularly and can be viewed on the website and extracted on Orphadata in XML format (www. orphadata.org/cgi-bin/inc/product3.inc.php).

As of 2012, the ontology of rare diseases (OntoOrpha) can also be viewed from the BioPortal website (http://purl.bioontology.org/ontology/OntoOrpha).

3.2.1. INDEXATION OF RARE DISEASES

Rare diseases are indexed with ICD-10 codes.
Indexation by clinical signs uses an in-house thesaurus of phenotype terms and is carried out with the aim of feeding the Orphanet assistance-to-diagnosis tool.

Diseases are also linked to one or more OMIM (Online Mendelian Inheritance in Man) numbers and to the list of indexed publications in scientific journals via a specific PubMed query, resulting from mapping Orphanet terms with MeSH descriptors.

Disease 'identity cards' have been improved with additional cross referencing and epidemiological data. The information regarding the ongoing mapping between the Orpha nomenclature and Unified Medical Language System (UMLS), MeSH, MedDRA and SNOMED CT is now available online and downloadable at Orphadata (http://www.orphadata.org/cgi-bin/inc/product1.inc.php).

3.2.2. Additional functionalities in 2012

To allow the experts and/or the users to suggest modifications and comments to the disease information available online, a "suggest an update" button is now available at the bottom of the disease pages.
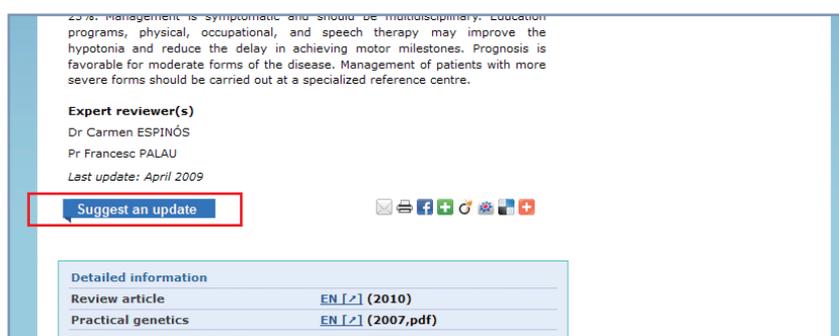
3.3. Orphanet inventory of genes

Genes involved in rare diseases (pathogenic and susceptibility genes) are entered in the database. Genes are associated with one or more diseases, to one or more genetic tests, mutation registries and/or research projects in the database. The registered data include: the main name and symbol of the gene (from HGNC), its synonyms, and its HGNC, UniProtKB, Genatlas and OMIM references (in order to cross-reference these websites). Moreover, genes are now cross-referenced with Ensembl (an EBI database that maintains automatic annotation on selected eukaryotic genomes), Reactome (an EBI open-source, open access, manually curated and peer-reviewed pathway database) and IUPHAR (The International Union of Basic and Clinical Pharmacology).

3.3.1. Additional functionalities in 2012

The relationship between a gene and a disease is now qualified according to the role the gene plays in the pathogenesis of a disease. Genes are annotated as causative, modifiers (both from germline or somatic mutations), major susceptibility factors or playing a role in the phenotype (for chromosomal anomalies). These annotations represent a unique, added-value service for researchers.

The disease database contains 8,945 diseases or groups of diseases and their synonyms.

Epidemiology:
- 4,251 diseases indexed with prevalence data
- 4,177 diseases indexed with mode of inheritance
- 4,161 diseases indexed with age of onset

5,749 diseases indexed with ICD10
4,006 diseases indexed with OMIM
2,200 diseases indexed with UMLS
961 diseases indexed with MedRA
1,689 diseases indexed with SNOMED CT
1,759 diseases indexed with MeSH

2,876 genes linked to 2,904 diseases, including:
- 2,798 genes interfaced with UniProt KB
- 2,841 genes interfaced with OMIM
- 2,827 genes interfaced with Genatlas
- 2,876 genes interfaced with HGNC

2,300 diseases indexed with Pubmed

4,969 external links for 2,910 diseases

The Orphanet encyclopaedia contains:
- 3,557 summaries in English
- 2,950 summaries in French
- 2,785 summaries in German
- 2,950 summaries in Italian
- 2,642 summaries in Spanish
- 480 summaries in Portuguese

2,705 diseases indexed with clinical signs

386 articles in French of which 123 for the general public and 45 emergency guidelines
1,213 articles in English of which 13 emergency guidelines.

3.4. Orphanet encyclopaedia

Two distinct encyclopaedia are provided on the Orphanet website: one for health professionals and one for the general public.

3.4.1. HEALTH PROFESSIONALS ENCYCLOPAEDIA

- **Summary information**

The Orphanet abstracts are unique, originally written in English by a member of the Paris-based editorial team and reviewed by an invited expert. Additional information on the diseases (i.e. annotations on epidemiological data, clinical signs, functional consequences of the disease, genes, etc.) is requested from the expert if not already documented in the database, or validation of the existing data is requested. As of December 2012, abstracts are structured in sections according to the Definition of the disease – Epidemiology – Clinical description – Etiology – Diagnostic methods – Differential diagnosis – Antenatal diagnosis (if relevant) – Genetic counselling (if relevant) – Management and treatment – Prognosis.
They are systematically translated into the five other languages (French, Italian, Spanish, German and Portuguese). In addition, as of 31 December 2012, 100 abstracts each are available in Finnish and Polish.

- **Practical genetics articles**

These articles are co-produced by Orphanet and the *European Journal of Human Genetics* (EJHG), the official journal of the European Society of Human Genetics. Freely-accessible articles are published in the EJHG (Nature Publishing Group) and accessible from Orphanet.

- **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: 45 emergency guidelines in French are now online. They are being translated into five languages (English, German, Italian, Portuguese and Spanish). Currently, 13 emergency guidelines are available in English, 23 in Spanish, 19 in Italian, 18 in Portuguese and one in German. There are 31 new guidelines since 2011.
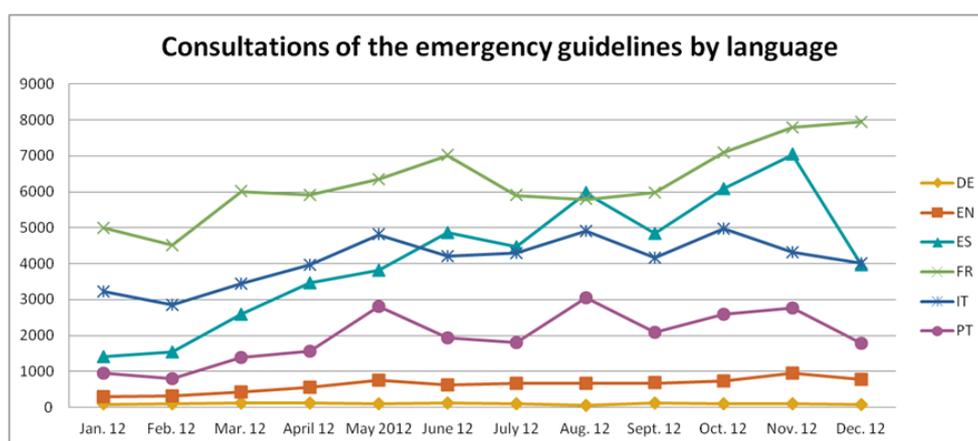
Consultations of the emergency guidelines by language in 2012

Over 200,000 guidelines were viewed in 2012, versus 67,000 in 2011, representing an increase of 200% in one year. This global increase reflects the expansion of the collection into the different languages.
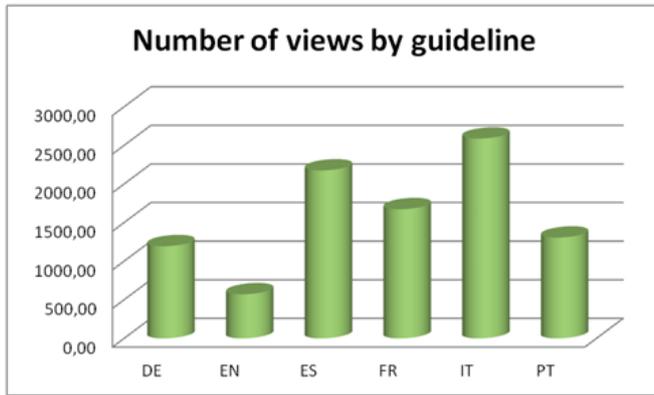
Language	Number of guidelines
German	1
English	13
Spanish	23
French	45
Italian	19
Portuguese	18

Number of views by language and by guideline

The ratio of the number of consultations for each language to the number of guidelines shows that this collection is a success in several languages such as French, Italian, Spanish and Portuguese. Despite the fact that there is only one guideline in German, it is highly consulted. On the other hand, the disappointing number of consultations in English can be explained by the existence of a larger number of equivalent documents elsewhere 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.

3.4.2. LINKS TO EXTERNAL RESOURCES

With the purpose of expanding the number of review articles available online and to disseminate articles matching Orphanet quality criteria, the editorial team is also in charge of identifying articles suitable for publication on the website produced by other journals or learned societies. Authorisations from the copyright holder are requested so as to give access to the full text.

We can distinguish three distinct external resources accessible from the Orphanet website:

- **Review articles**

Review articles of good quality published in peer-reviewed journals can be linked to a disease and published on the Orphanet website with the permission of the copyright holder. As of 31 december 2012, 5 review articles (excluding those published in the Orphanet Journal of Rare Diseases) were available on the website.

- **Clinical Genetics Review**

These are peer-reviewed disease descriptions focused on genetic aspects with an implication in the diagnosis, management, and genetic counseling of patients and families with specific inherited conditions.
These review articles can be linked to a disease and displayed on the Orphanet website with the permission of their copyright owners. As of 31 December 2012, the clinical genetic review collection comprises 547 articles from GeneReviews.

- **Best practice guidelines**

These guidelines are recommendations for the management of patients, issued by official organisations. There are two kinds of best practice guidelines: anaesthesia guidelines and clinical practice guidelines. They are both produced by learned societies and published either in scientific journals or in learned societies or health agencies websites. A methodology of assessment has been developed to review the guidelines and link only the most accurate ones. These guidelines can be linked to a disease and displayed on the Orphanet website with the permission of their copyright owners. As of 31 december 2012, 112 best practice guidelines were available on the website.

- **Guidance for genetic testing**

This collection comprises summary recommendations intended to desseminate best practice in genetics testing. They include Gene Cards (published in the European Journal of Human Genetics) and charts from the ANPGM (Association nationale des praticiens en génétique médicale). As of 31 December 2012, 138 recommendations were available on the website.

3.4.3. 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 that concern them. In 2012 it has begun to be translated into Spanish.

General public-intended texts externally produced by expert centres or patient organisations (produced in compliance with a reliable methodology) in any other language are now selected and, if permission is granted, the texts are loaded on the Orphanet website.

As to 31 December 2012, 123 texts in French are online. Documents from this encyclopaedia have been downloaded more than 382,000 times per month, which corresponds to more than 4 million downloads in 2012.

3.5. Orphanet directory of expert resources

Orphanet provides a directory of:

- Centres of expertise
- Medical laboratories
- Patient organisations
- Clinical trials

- Patient registries
- Mutation registries
- Biobanks
- Ongoing research projects

Data collected from non-official sources passes through a pre-release validation that is defined by each country, to ensure data relevance for the rare disease community.

For official sources, no pre-release validation is required.

The aim of this multi-step process is to generate high-quality and accurate data: complete, valid, consistent, uniform with other data of the database and unique. New procedures have been introduced to allow a follow-up of the validation process. At the country level, there is a validation prior to publication of data following rules established at the national level with health authorities. A second round of validation is performed centrally on a monthly basis against the criteria of relevance to the project, coherence with data from other countries and proper indexing with disease classification systems. And a third round of quality control is carried out on online published data following a process defined at national level.

Once the information meets Orphanet quality standards, it is published online. Minor changes are possible on a continuous basis.

Regularly, published data is verified and updated (at least once a year).

The 37 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, the United Kingdom and Western Australia.

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 2012, countries managing both data collection and updates at country level are: Austria, Belgium, Canada-Quebec, Czech Republic, Finland, France, Germany, Ireland, Italy, the Netherlands, Portugal, Spain, Switzerland and the United Kingdom.

The directory of expert resources in 37 countries worldwide contains the following data:

17,728 professionals referenced in the database

6,078 expert centres

2,467 patient organisations

2,704 Research laboratories
4,861 Research projects on 1,712 diseases
2,611 Clinical trials for 618 diseases in 29 countries
   634 Patient registries
   616 Mutation registries
   112 Biobanks

1,645 Medical laboratories dedicated to diagnosis
32,344 Medical laboratory tests for 3,205 diseases and 2,196 genes

3.5.1. ADDITIONAL FUNCTIONALITIES IN 2012

To sort activities in the international Orphanet webpage, a new tool, "sort by specificity" (framed in red in the screen shot below), was developed that sorts expert resources (expert centres, research and trials, registries and patient organisations) by specificity and displays the activities specifically linked to the disease (framed in blue in the screen shot below) and shows separately the activities which are linked to upper and lower levels of classification (framed in green in the screen shot below). This is the default presentation of all the activities except for diagnostic tests which are sorted by quality management. The former geographical sorting is also available as an optional research facility.

3.5.2.DATA QUALITY VALIDATION OF MEDICAL LABORATORIES

Medical laboratories listed in Orphanet are those offering tests for the diagnosis of a rare disease or a group of rare diseases, and those performing genetic testing whatever the prevalence of the disease. These medical laboratories must be accredited and this involves a procedure by which an authoritative body gives formal recognition that a body or person is competent to carry out a specific task (ISO 9000, 2000 Quality management systems – fundamentals and vocabulary). Information on accreditation of medical laboratories is reviewed by Eurogentest and experts in the field.

Moreover, medical laboratories can undergo an external quality assessment (EQA) whereby a set of reagents and techniques are assessed by an external source and the results of the testing laboratory are compared with those of an approved reference laboratory (WHO). It allows a laboratory to compare its performance for an individual test or technique against that of other laboratories.

Information on EQA participation is provided annually by CF Network, CEQA and EMQN with the consent of the concerned laboratories. For other EQA providers, information is validated, as of December 2012, by Orphanet upon reception of an EQA participation certificate; otherwise the information provided on the Orphanet website is deemed 'not validated' and associated with a [!] sign.

3.6. Orphanet directory of Orphan Drugs

The list of Orphan Drugs includes all those substances that have been granted an orphan designation for a disease considered as rare in Europe, whether they were further developed to become drugs with marketing authorisation (MA) or not. The Orphanet database also includes drugs without an orphan designation as long as they have been granted a marketing authorisation issued by the European Medicines Agency (EMA - centralised procedure) with a specific indication for a rare disease. Some drugs (substance and/or trade name) are included in the database because they are tested in a clinical trial performed on a rare disease, but they do not have a regulatory status.

Drugs with a regulatory status in Europe are collected from reports issued by the two Committees of the EMA: the COMP (Committee for Orphan Medicinal Products) and the CHMP (Committee for Medicinal Products for Human use).

Orphan drugs are published on the Orphanet website, under the Orphan drugs tab and data is also released within an Orphanet Report Series that is issued every three months.

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

- 1,090 substances linked to more than 1,280 orphan designations (EU and US)
- 144 European marketing authorisations (of which 68 after orphan designation and 76 with no previous orphan designation)
- More than 150 USA marketing authorisations
- These substances have a designation/indication for more than 550 rare diseases

3.7. 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 meet the needs of massive data extraction, orphadata.org was created (www.orphadata.org). Orphadata is intended to contribute to accelerating R&D and to facilitate global adoption of the Orphanet nomenclature.

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 Rare Diseases Portal project funded by DG Research and Innovation (RTD) and the Orphanet Europe Joint Action contract funded by DG Sanco. 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:

Products freely accessible on Orphadata:
1. Diseases and cross-referencing
2. Epidemiological data
3. Orphanet classifications
4. Diseases with their clinical signs
5. Thesaurus of clinical signs
6. Diseases with their associated genes

Other products such as textual information, patient organisations, expert clinics, clinical laboratories, orphan drugs and research activities are available either freely (Academia) or for a fee (Industry) after signature of a Material Transfer Agreement (MTA).

Orphadata provides a guide for users that defines and describes the elements of the dataset.

Since January 2012, Orphadata products were downloaded more than 57,000 times, with an average of 4,800 times a month.
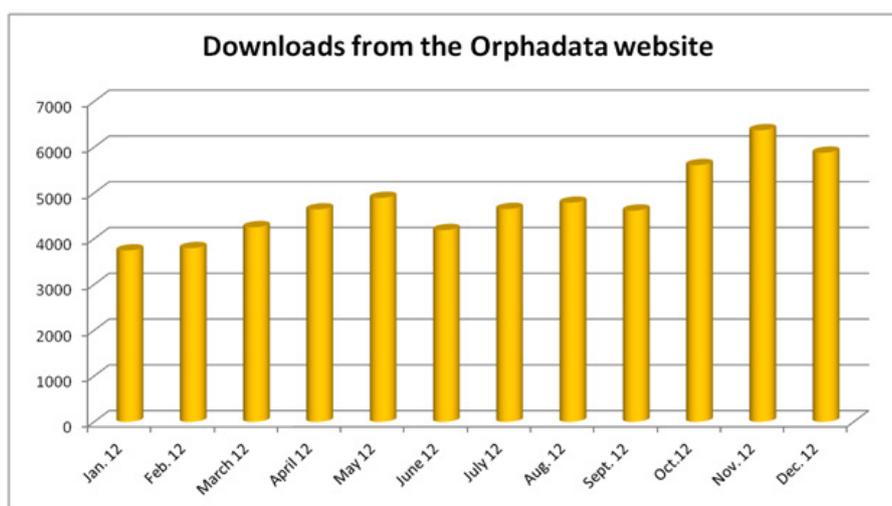
Total number of downloaded products from the Orphadata website

3.8. Orphanet Report 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. These texts are published as PDF documents accessible from the homepage page and from every page of the website.

The ORS available in 2012 were:
- List of rare diseases (in English and French)
- Prevalence or reported number of published cases listed in alphabetical order of disease (in the six languages of the website: English, French, German, Italian, Portuguese and Spanish)
- Diseases listed by decreasing prevalence or number of published cases (in the six languages)
- Patient Registries in Europe (in English)
- European Research Projects and Clinical Networks (in English)
- Lists of orphan drugs in Europe (in the six languages)
- Orphanet activity reports (in English, French, Italian, Spanish and Polish)
- Orphanet Users Satisfaction Survey (in English)
- OrphaNews Europe Reader Satisfaction Survey (in English)

New versions of these publications are advertised in OrphaNews Europe.
The Orphanet Report Series are heavily downloaded: in 2012, more than 810,000 Orphanet Report Series were consulted. This represents an increase of 15% compared to 2011 (approximately 710,000 downloads).
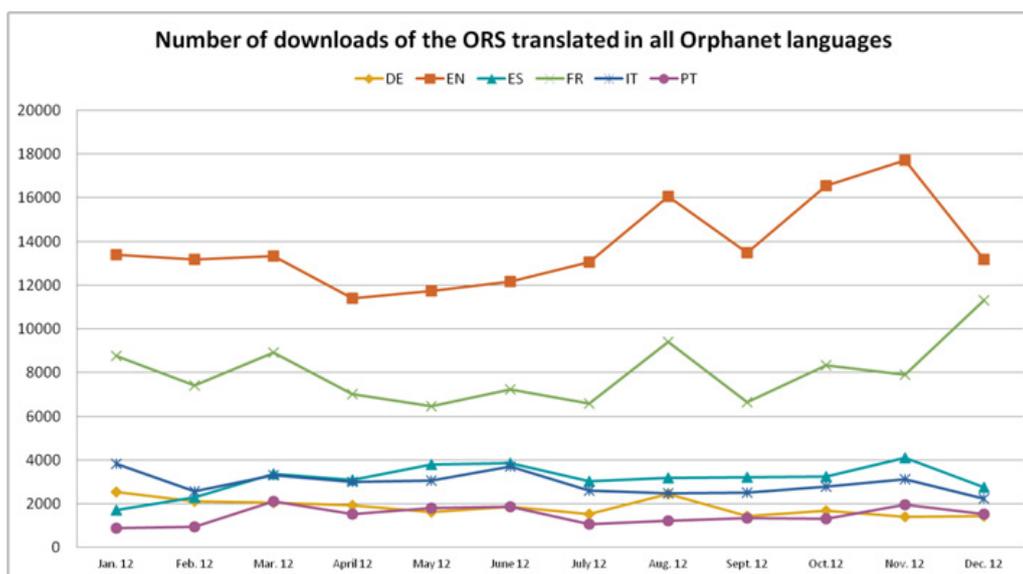
Number of downloads of the Orphanet Report Series translated in all Orphanet languages

Comparison of the number of downloads of the Orphanet Report Series translated in all 6 languages shows that this collection is most heavily downloaded in English and French.

3.9. The Orphanet Newsletter

The updating of the database is based on a literature review and continuous innovations are published in a bi-monthly electronic newsletter, OrphaNews to which the subscription is free. OrphaNews presents an overview of scientific and political news about rare diseases and orphan drugs as well as the progress of French and European work. It is the communication tool between Orphanet and the wider scientific community.

OrphaNews Europe is the fortnightly newsletter in English and counts 14,500 subscribers. It is the official newsletter of the European Union Committee of Experts on Rare Diseases (EUCERD). In addition, Orphanews is also available in French (OrphaNews France, 8,500 subscribers) and as of December 2011 in Italian (OrphaNews Italia, 2,500 subscribers).

3.9.1. ADDITIONAL FUNCTIONALITIES OF THE NEWSLETTERS IN 2012

To resolve the issue of information dispersion and to address specific needs of the different stakeholders, the Google Web Search API is now available on the OrphaNews Europe website (framed in red in the screenshot below). This application is a simple and dynamic search box that displays search results of the OrphaNews web pages and allows the simple retrieval of articles from the newsletter archives.

3.10.   Orphanet Journal of Rare Diseases (OJRD)

Orphanet Journal of Rare Diseases is an open access, online journal that encompasses all aspects of rare diseases and orphan drugs. The journal publishes high quality reviews on specific rare diseases. In addition, the journal may consider articles on clinical trial outcome reports, either positive or negative, and articles on public health issues in the field of rare diseases and orphan drugs. 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. Its current impact factor is 5.07. In 2012, 285 publications were submitted to the journal. Of these, 103 were accepted for publication.

4. Users

4.1. 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 1,230,000 responses.

Users mainly access the Orphanet website through search engines (83.6% of visits according to Google Analytics) and Google alone accounts for 95.1% of queries. Other sites generating traffic to Orphanet represent 8.6% of visits. The remaining visits are made via direct access (bookmarks, 7.8%).

Distribution of the traffic sources
(Source: Google Analytics, 1st January 2012 to 31st December 2012)

The wealth of information available on our site attracts a substantial number of visits due to a sizable corpus of keywords (rather than just certain predominant keywords). The keyword primarily used to access our site is simply "Orphanet" which represents 5.1% of visits. The indexation of our site is of the "long tail" type: more than 958,500 different keywords generate traffic to the site.

Since November 2009, Google Analytics allows users to trace visits made from mobile devices (smartphones, tablets). These visits represented 12% of all visits during 2012, i.e. 525,769 visits. In 2011, it represented only 4.96% (173,692 visits).

4.2. The website's audience

In 2012, over 12.2 million pages were viewed, thus on average around 33,350 pages views per day. This figure has increased by 8% in comparison to 2011 (11,245,243 page views in 2011).

The Google Analytics tool does not include direct access to PDF documents. However, PDF documents are an important entry point and generate a consistent volume of visits: each month, 850,000 PDF documents are consulted on the Orphanet website. This represents more than 10,000,000 downloads in 2012, which is approximately the same amount as in 2011 (10,500,000).

The geographical distribution of users continues to show very varied sources (211 sources listed). The top ten countries are: France, Italy, Germany, Spain, United States, Brazil, Canada, Switzerland, Belgium and Mexico.
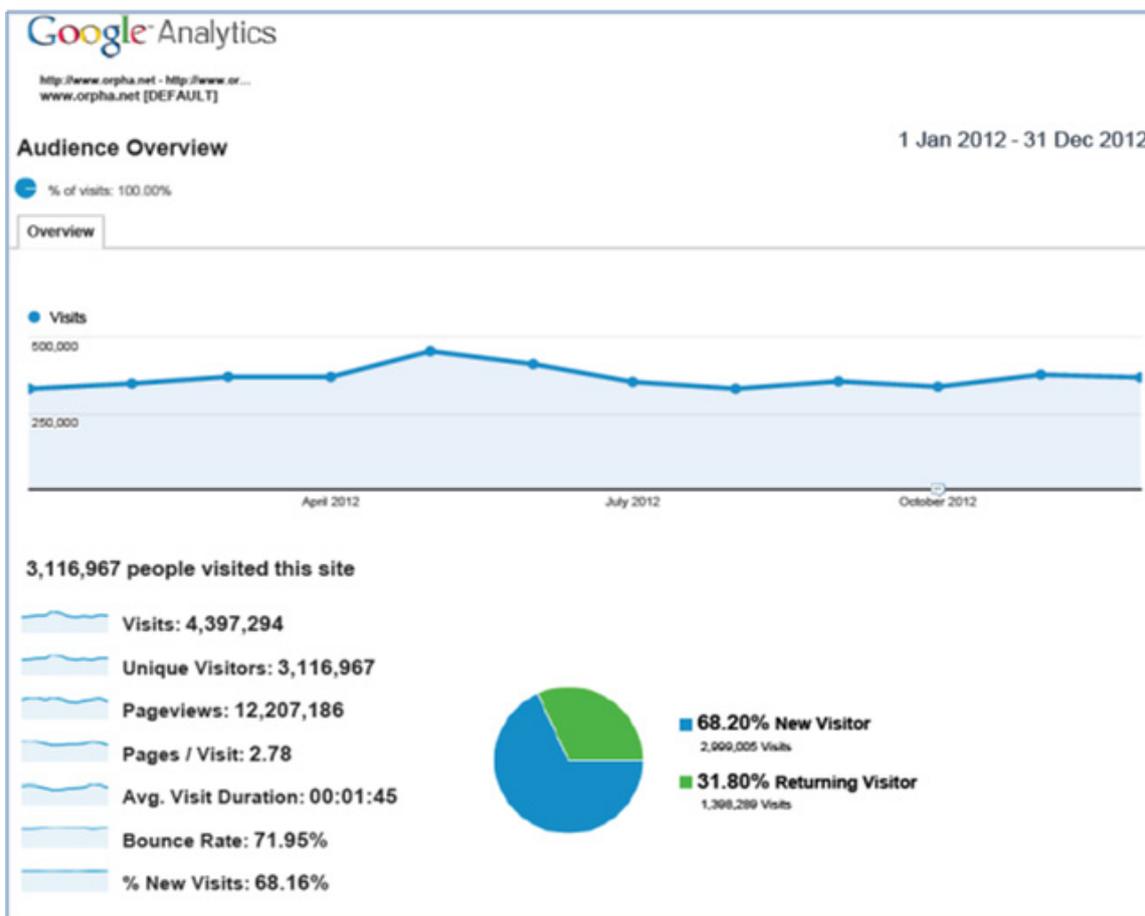
Orphanet website consultations in 2012
(Source: Google Analytics, 1ˢᵗ January 2012 to 31ˢᵗ December 2012)

4.3. Type of users and use

2012 Orphanet English users satisfaction survey

An online survey was carried out in December 2012. The satisfaction of the portal users was assessed by asking users to respond to a questionnaire until we obtained one thousand replies per language. This is the methodology which has been in place for the past 10 years at Orphanet.

The following results present the responses collected in English.

QUESTION 1

BY WHAT CAPACITY ARE YOU CONSULTING THE ORPHANET WEBSITE TODAY? PLEASE SELECT A CATEGORY.

This question aimed to determine the profile of Orphanet's users.

Seven categories were proposed (i.e. health professional, patients/entourage, researcher, industrial, health care manager/policy maker and, for the first time, students), and a free text field was included for other types of users to enter their profession. Only one response was possible.

The table below shows the distribution of respondents amongst these categories:

Answer Options	Response Percent	Response Count
Health professional	40,7%	407
Patient/entourage	24,5%	245
Research	12,9%	129
Industry	5,7%	57
Health care manager/policy maker	1,3%	13
Education/communication	3,6%	36
Student	5,6%	56
Other	5,7%	57
Answered Question		1000
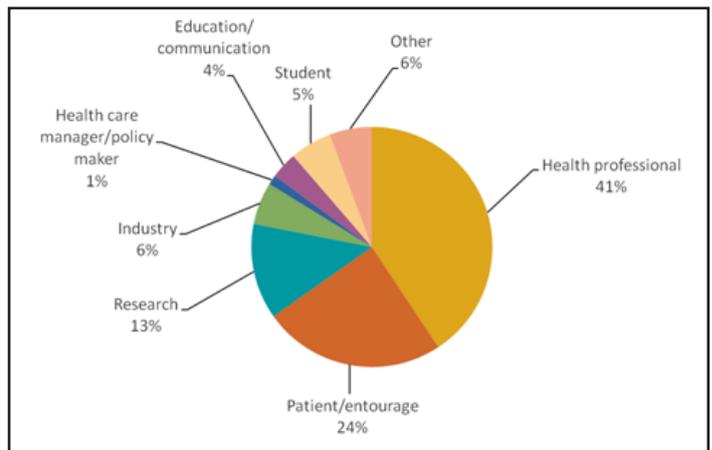
The largest category of respondents is the health professionals category (41%). The second largest category of respondents is patients and their entourage (including patient organisations, alliances and support groups) with 24% of responses. These results are consistent with 2011 figures for the Patient category, but the health professionals category has decreased by 10 points, mostlikely due to the creation of the Student category.

Amongst the "Other" types of users in the table above, were several health professionals, attorneys, medical translators and clinical coding technicians, but the large majority (28) were patients or patients' relatives.

Then, for each category, respondents were asked to choose the sub-category that would best describe them.

Health professionals (n=399):
Hospital specialists represent by far the main category of respondents (45%). All together, 57% are specialists. Healthcare professional (nurses and others) stand for more than 11% of the health professionals, genetic counsellors for 7.5%, general practitioners for 6.5%.
In the "Other" category, a third of the respondents are clinical or medical geneticists.
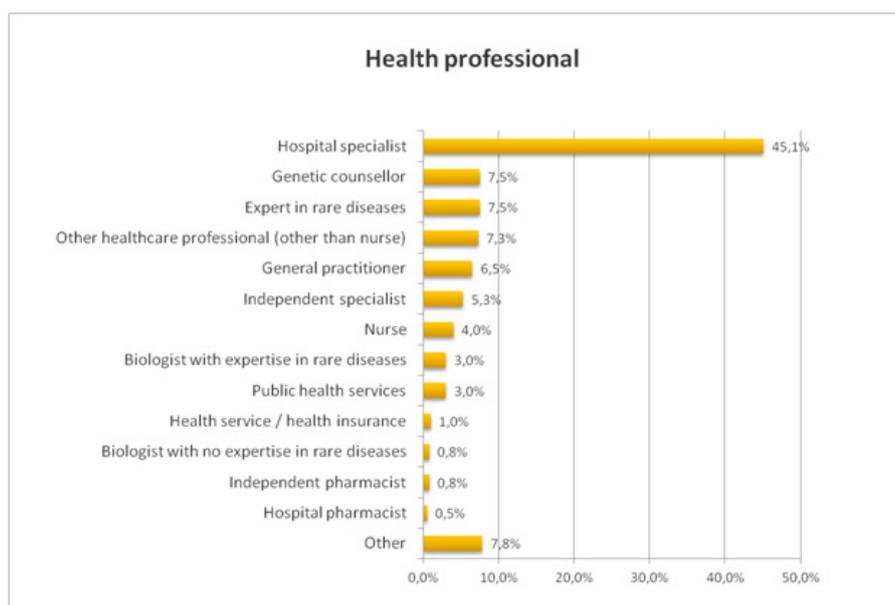
Patient/entourage (n= 243):
Most of the people who selected this category are patients (52%). More than 38% are family members of a patient with rare diseases.

Patient/entourage

|	
Patient	52,7%
Mother/father/child of a patient	28,8%
Other family member	9,5%
Patient organisation administration	3,3%
Friend of a patient	2,9%
Member of a patient organisation	0,0%
Other	2,9%
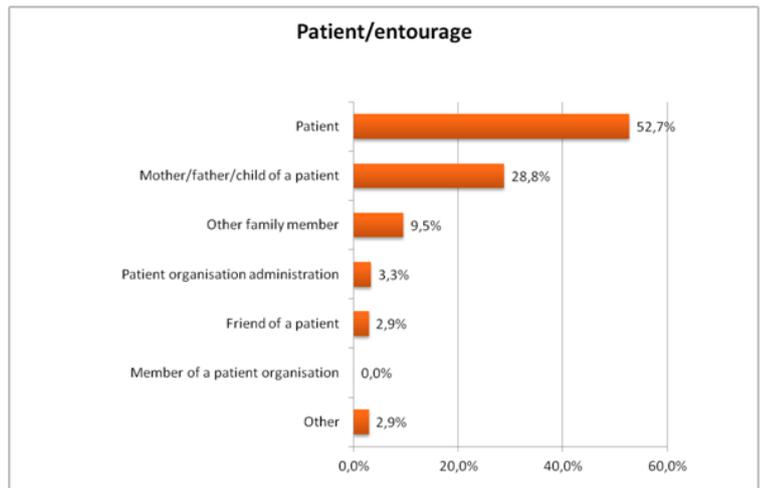
Research (n= 122):
Academic researchers represent 48% of respondents of the research category, and are equally divided between basic and clinical research. The newly created 'Social sciences' category gathers 10% of researchers, underlining the emergence of this domain in the rare diseases field.

Research

Academic/basic researcher	23,8%
Academic/clinical researcher	23,8%
Industry researcher	13,1%
Social sciences	9,8%
Bioinformatician	6,6%
Health economist	3,3%
Other	19,7%
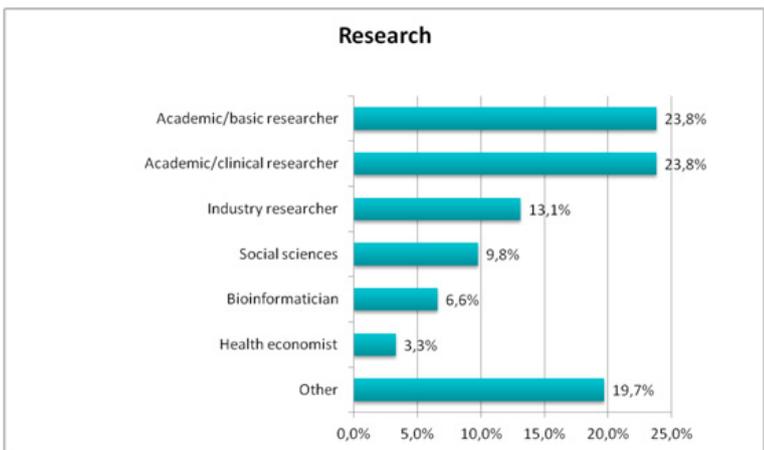
Industry (n=55):
More than 70% of respondents in this category work in biotechnology or pharmaceutical industry. Others are mainly consultants for industry (25.5%).

Industry

Biotechnology and pharmaceutical	70,9%
Consultant for Industry	25,5%
Investor / business developer	1,8%
Private health insurance	1,8%
Other	0,0%
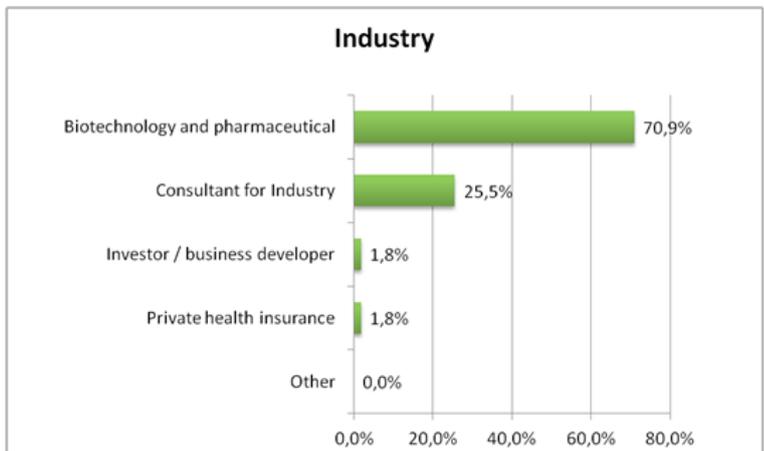
Health care manager/policy maker (n=13):
In this category, 8 individuals work for governmental administration (61%) and 2 work for a hospital administration (15%).

Health care manager/policy maker

Governmental administration	61,5%
Hospital administration	15,4%
European administration	0,0%
Other	23,1%
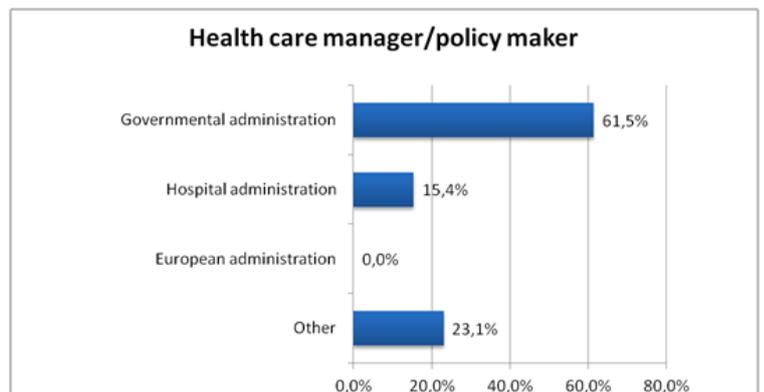
Education/communication (n=33):
Instructors represent 27% of this category and journalists 15%. In the Other category, there were several students, teachers and medical writers.

Education/communication

|	
Teacher (higher education)	15,2%
Journalist	15,2%
Teacher (primary/secondary education)	12,1%
Librarian	6,1%
Webmaster	0,0%
Other	51,5%
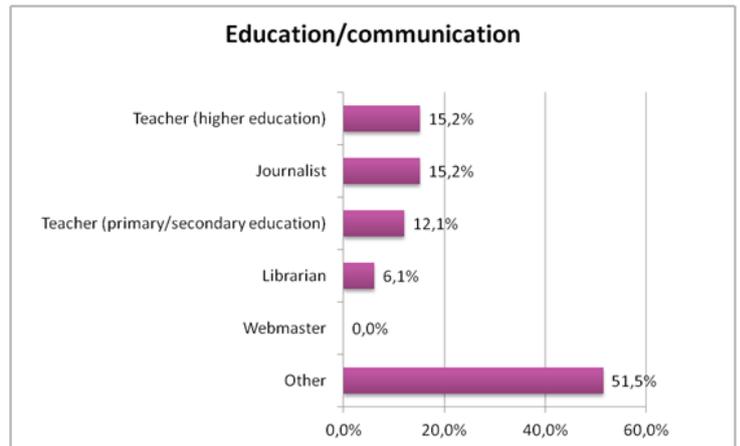
Students (n=55):
Medical students represent 62%. Other students are mainly biology students, but also biotechnology, bioinformatics, pharmacy or nursing students.

Students

Other than medical student	38%
Medical student	61,8%
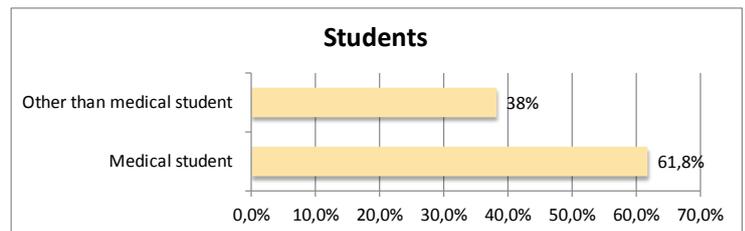
Question 2

How often do you visit Orphanet?

Around 38% of those who answered this survey are regular users whereas 47% were visiting Orphanet for the first time.

How often do you visit Orphanet?

- Over twice a week 15%
- First visit 47%
- Over twice a month 23%
- Over twice a year 15%
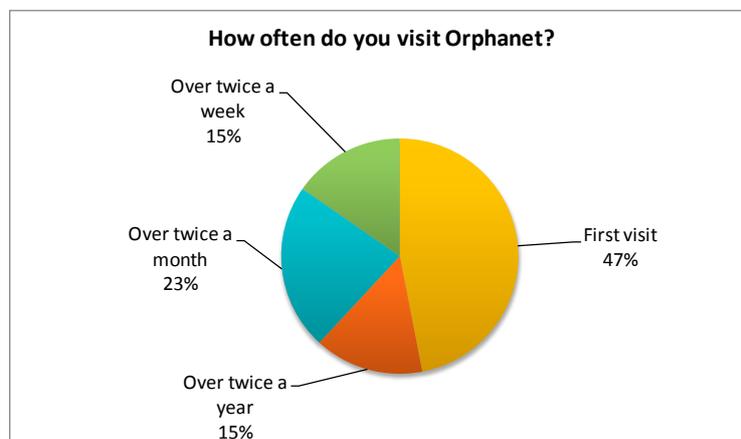
Health professionals are more than 58% to visit the Orphanet website more than twice a month, industrials are 49%, whereas patients are less than 11%.

Question 3

What sort of information are you looking for during THIS CONNECTION to Orphanet?

This question aims to determine which kind of information visitors sought on Orphanet. More than one choice was possible.

What sort of information are you looking for during this connection to Orphanet?

|	
Information for a specific disease	78,3%
Information on rare diseases in general	24,9%
Information on laboratory tests	18,8%
Information on research projects	14,8%
Information on clinical trials	14,6%
Information on patient organisations	12,5%
Information on specialist clinics	12,1%
Information on orphan drugs in general	10,8%
Information on registries	7,1%
Information on the Orphanet project	5,1%
Information on the newsletter	3,2%
Don't know – just curious	2,6%
Visit to register your activity in Orphanet	1,8%
Other	4,7%

The results show a clear trend: most of the respondents were looking for information for a specific disease (78%). Our visitors also look for information on rare diseases in general (25%), on laboratory tests (19%), on research projects (15%) or on clinical trials (15%).

32% of health professionals are looking for information on a laboratory test; 40% of students are looking for information on rare diseases in general. As expected, those in industry are looking for information on orphan drugs (33%) and research data (research projects, clinical trials, registries), and also on patient organisations. Compared to 2011, our respondents seem to be looking more frequently for information on rare diseases in general.

QUESTION 4

DO YOU REGULARLY USE THE FOLLOWING SITES WHEN DEALING WITH RARE DISEASES?

This question aimed to determine which other websites are visited by people looking for information on rare diseases. More than one choice was possible.

Do you regularly use the following sites when dealing with rare diseases?

PubMed	74,0%
Wikipedia	50,3%
OMIM	41,2%
Websites of patient organisations or foundations	41,1%
Websites of learned societies	27,0%
GeneClinics	24,0%
Social networking sites (personal)	11,2%
Social networking sites (professional)	10,1%
None of them	8,7%
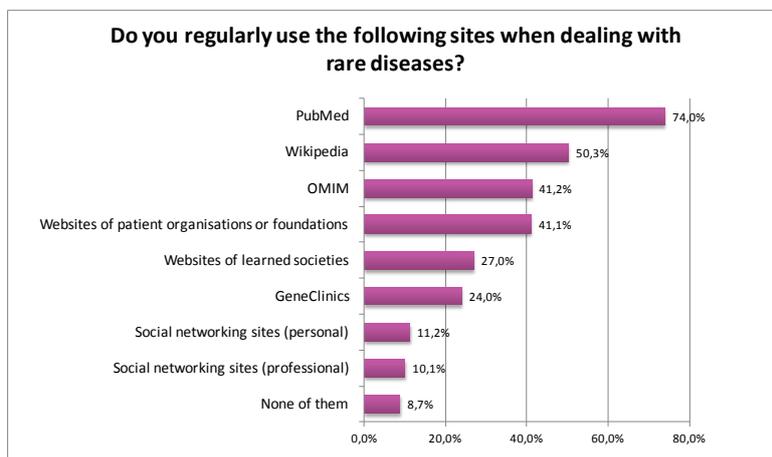
To obtain information on rare diseases, Pubmed is used by 74% of those who answered this question; 50% of them find information on Wikipedia; the OMIM website reaches the third position with 41%, as well as websites of patient organisations or foundations. Social networks represent less than 21% of the total.

Pubmed is massively consulted by healthcare professionals (88%), researchers (73%) and industry (85%) but patients also use this website as a source of information on rare diseases (46%). OMIM is mainly used by healthcare professionals (64%), researchers (39%) and not by patients (12%) or industrials (15%). Websites of patient organisations or foundations are more often consulted by industry (60%) or patients (54%).

This analysis also underlines that Wikipedia remains a main source of information for all categories of respondents.

QUESTION 5

HOW USEFUL WOULD YOU RANK THE FOLLOWING ORPHANET SERVICES FOR **YOUR OWN** USE?

The usefulness of Orphanet products was evaluated through this question. Only one response was possible for the 810 respondents. The results show that Orphanet products are highly appreciated but not sufficiently well known:

Answer Options	Very useful	Useful	Fairly useful	Not useful	No opinion	I didn't know Orphanet offered this service
List of diseases and classifications	**370**	210	53	8	67	102
Texts on diseases	**324**	259	75	5	59	88
Emergency guidelines	126	169	95	33	171	**216**
Search by sign facility	111	184	86	37	174	**218**
Directory of clinics	116	179	105	47	178	**185**
Directory of medical laboratories	139	175	89	44	178	**185**
Directory of patient organisations	144	**205**	119	30	139	173
Directory of research projects	134	**212**	104	31	146	183
Directory of clinical trials	151	**193**	95	38	144	189
Directory of registries	114	181	106	35	177	**197**
Directory of orphan drugs	149	**182**	112	30	161	176
Orphanet Report Series on epidemiology of Rare Diseases	**192**	191	84	17	142	184
Orphanet Report Series on Orphan Drugs	132	171	92	26	181	**208**
OrphaNews newsletter	102	164	84	29	190	**241**
Orphanet national websites	139	163	87	33	177	**211**
Orphadata (downloadable Orphanet datasets)	118	138	68	25	179	**282**

The following graph presents the usefulness of Orphanet products ('very useful' and 'useful' answers). The 'no opinion' and 'I didn't know Orphanet offered this service' answers were subtracted from the results to more faithfully represent the utility of the products, according those aware of these Orphanet services.

How useful would you rank the following Orphanet services for your own use?

Service	Percentage
List of diseases and classifications	90,5%
Texts on diseases	87,9%
Orphanet Report Series on epidemiology of Rare Diseases	79,1%
Orphadata (downloadable Orphanet datasets)	73,4%
Directory of clinical trials	72,1%
Orphanet Report Series on Orphan Drugs	72,0%
Directory of research projects	71,9%
Orphanet national websites	71,6%
Search by sign facility	70,6%
Directory of medical laboratories	70,2%
OrphaNews newsletter	70,2%
Directory of patient organisations	70,1%
Directory of orphan drugs	70,0%
Emergency guidelines	69,7%
Directory of registries	67,7%
Directory of clinics	66,0%

The key Orphanet service remains the encyclopaedia: the texts on diseases (88%) and the list of diseases and the classifications (91%). The Orphanet Report Series collection is also highly appreciated.

An analysis of the newly introduced category 'I didn't know Orphanet offered this service' is presented in the following graph, highlighting the fact that our users are not sufficiently informed about our range of products.

Orphanet services usefulness: I didn't know Orphanet offered this service

Service	Percentage
Orphadata (downloadable Orphanet datasets)	34,8%
OrphaNews newsletter	29,8%
Search by sign facility	26,9%
Emergency guidelines	26,7%
Orphanet national websites	26,0%
Orphanet Report Series on Orphan Drugs	25,7%
Directory of registries	24,3%
Directory of clinical trials	23,3%
Directory of medical laboratories	22,8%
Directory of clinics	22,8%
Orphanet Report Series on epidemiology of Rare Diseases	22,7%
Directory of research projects	22,6%
Directory of orphan drugs	21,7%
Directory of patient organisations	21,4%
List of diseases and classifications	12,6%
Texts on diseases	10,9%

Orphadata, the website that allows researchers to download Orphanet datasets, is well appreciated but also not very well known by our respondents. This is understandable as this service is recent (2011), research oriented and therefore not useful for the vast majority of users.

Patients and their entourage is the category that seems to know the least about Orphanet services: often more than 40% declare that they did not know about a service provided by Orphanet.

In particular, nearly a third of the respondents did not know about the OrphaNews newsletter even though health professionals, researchers and industrials seem to be more often aware of the existence of this product compared to patients.

The search by sign facility suffers from a lack of exposure in our user community, as well as our collection of emergency guidelines (which is better known in the health professionals category than in all others). These products are mainly dedicated to clinicians, who are more aware of these products than the average.

5. Network: the national and international collaborations of Orphanet

5.1. Collaboration with the WHO

The World Health Organisation (WHO) and Orphanet collaborate on the revision of the International Classification of Diseases (ICD-11).
Orphanet has been entrusted as the operating institution to prepare the proposal for ICD-11 related to rare diseases. The proposal is now accessible online for review by the community (http://apps.who.int/classifications/icd11/browse/f/en). All the experts' opinions collected for this revision process were implemented in the Orphanet classification.

5.2. Collaboration with Health Authorities

5.2.1. NATIONAL PLANS

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

5.2.2. ADOPTION OF THE ORPHANET NOMENCLATURE IN HEALTH INFORMATION SYSTEMS

To improve the traceability of rare diseases in health information systems and to increase the recognition of each rare disease in national health and reimbursement systems, Orphanet has developed its own coding (Orpha code), composed of a unique and stable Orpha number for each rare disease. Since December 2012, it has been implemented in the French hospital system database where the ORPHA number is used to code all hospitalised patients with rare diseases. The goal is to better identify patients in the healthcare system so as to improve knowledge of their healthcare pathways. Germany and Latvia are also elaborating their national plan and foresee a similar initiative.

5.3. Scientific collaborations and partnerships

Orphanet is a partner of EuroGentest 2 until 2013. A workshop was held 27 and 28 September 2012 so as to assess the standards to be used in order to make rare diseases visible in health information systems and to propose standards to the clinical research community for the description of phenotypes. It was agreed that given the multitude of needs and applications in the field of rare diseases, one terminology for all applications is unrealistic. The participants recommended to consider the Orphanet and OMIM codes as the standards of the rare disease community but to continue cross-referencing with ICD and SNOMED CT so as to ease the interoperability between databases. The expert group proposed to set up an International Consortium of Phenotype Terminologies (ICPT).

The partnership formed with GlaxoSmithKline (GSK) was renewed in 2012. The company, which has 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 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) and is now effective.

A partnership was formed with IUPHAR at the end of 2011 to cross-link Orphanet with the IUPHAR database and since 30 May 2012 the crosslink is effective.

The Orphanet team is a partner of an FP7 research support action entitled "Support IRDIRC".
The International Rare Diseases Research Consortium (IRDiRC) was launched in April 2011 to foster international collaboration in rare diseases research. IRDiRC affiliates researchers and organisations investing in rare disease research in order to achieve two main objectives, namely to deliver 200 new therapies for rare diseases and the means to diagnose most rare diseases by the year 2020. Orphanet will host data on research projects funded by IRDiRC members which are research funding agencies expanding data coverage to new countries such as the United States.

6. Funding

Orphanet's budget was approximately 3M Euros in 2012, originating from 9 different contracts for the core activity funding and from various other contracts in some of the participating countries.
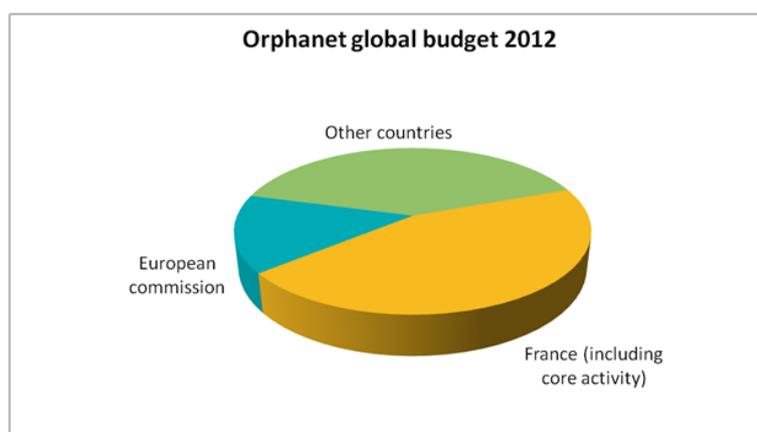
Orphanet global budget 2012

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

6.1. 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. It excludes the collection of data on expert services in the participating countries.
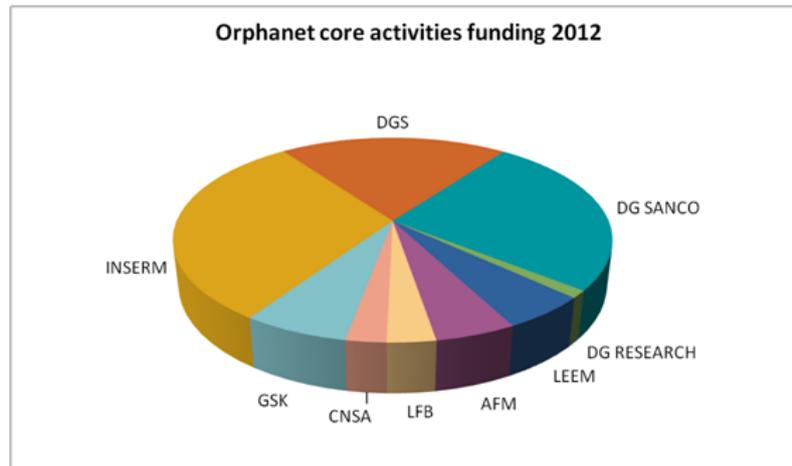
Orphanet core activities funding 2012

This budget excludes the costs of infrastructure (office space) which are essentially supported by INSERM.

In the last 10 years, the funding of Orphanet's core activities has quadrupled reflecting the growth of the project.
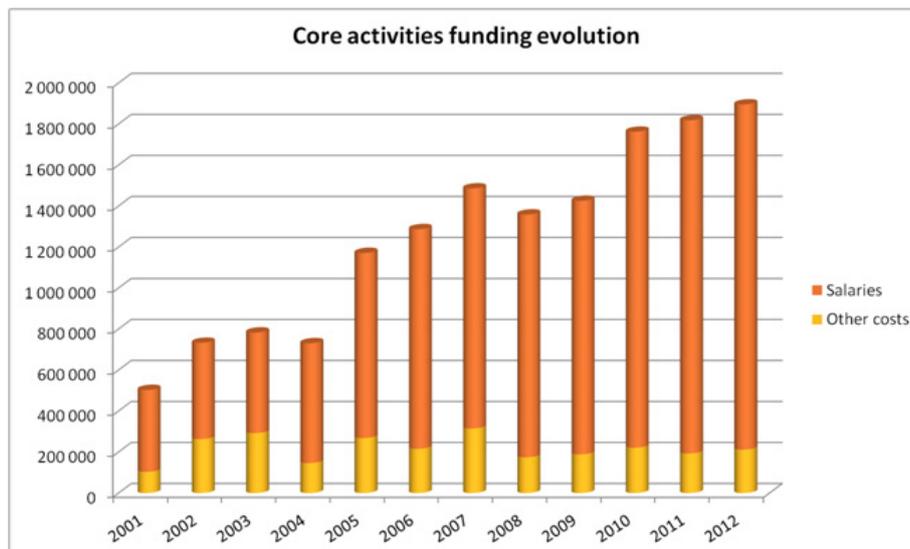
Core Activities funding evolution 2001-2012

Although there has been an extremely positive increase over the years, the current budget remains limited in comparison to the needs of maintaining and updating a database of this size. What is clear from the figure above is that apart the salaries, the budget required for running the activities has dramatically decreased in proportion when compared to the needs and to previous years.

6.1.1. EUROPEAN FUNDING

The European Commission funds the inventory of rare diseases, the encyclopaedia and the collection of data on expert services 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, HEALTH-F2-2009-223355).

In 2012, Orphanet was funded by the DG Sanco grant 20102206 (Orphanet Europe Joint Action) and the DG Research grant HEALTH-F4-2010-261469 (Eurogentest2).

6.1.2. OTHER CURRENT FINANCIAL PARTNERSHIPS FOR CORE ACTIVITY FUNDING

Instituts thématiques **Inserm** — Institut national de la santé et de la recherche médicale	The "Institut National de la Santé et de la Recherche Médicale" finances Orphanet's core activities. Inserm Transfert is in charge of ensuring licencing benefits and intellectual property transfer concerning Orphanet data.
RÉPUBLIQUE FRANÇAISE — MINISTÈRE DES AFFAIRES SOCIALES ET DE LA SANTÉ	The French Directorate General for Health finances Orphanet's core activities (DGS).
European Union flag	The European Commission finances the database of diseases, the encyclopaedia in English, coordination, communication and IT of the project.
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 Fondation — Sous l'égide de la Fondation de France	The "Fondation des Entreprises du Médicament" finances the collection of data on orphan drugs and clinical trials.
gsk GlaxoSmithKline	Glaxo Smith Klein finances the extension of the database's disease annotation and free access to this data.
RÉPUBLIQUE FRANÇAISE — MINISTÈRE DES AFFAIRES ETRANGÈRES ET EUROPÉENNES	The French "Ministère des affaires Etrangères" finances the cooperation with Canada.
EuroGentest	EuroGentest financed the creation of a thesaurus of clinical signs to harmonise international phenotype nomenclatures.

6.1.3. 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 logo)	Collaboration with the World Health Organisation (WHO) in the process of revising the International Classification of Diseases.
GENATLAS	Cross-referencing with Genatlas which collaborates in updating the data on genes involved in rare diseases.
UniProt	Cross-referencing with UniProt KB which collaborates in updating the data on genes linked to proteins involved in rare diseases.
HGNC (HUGO Gene Nomenclature Committee)	Cross-referencing with HGNC which collaborates in updating the data on genes involved in rare diseases.
OMIM® Online Mendelian Inheritance in Man® An Online Catalog of Human Genes and Genetic Disorders	Cross-referencing with OMIM (The Online Mendelian Inheritance in Man), and OMIM has added Orphanet to the list of external links provided on its website.
REACTOME A CURATED PATHWAY DATABASE	Cross-referencing with Reactome.
Browsing Genes & Genomes with e!Ensembl	Cross-referencing with Ensembl.
IUPHAR International union of basic and clinical pharmacology	Cross-referencing with The International Union of Basic and Clinical Pharmacology DataBase (IUPHAR-DB).
LOVD Leiden Open Variation Database	The LOVD (Leiden Open Variation Database) platform has been updated with links to Orphanet's gene pages.
EuroGentest	EuroGenetest collaborates with Orphanet in the field of quality management of medical laboratories.

6.2. 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.

6.2.1. 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".
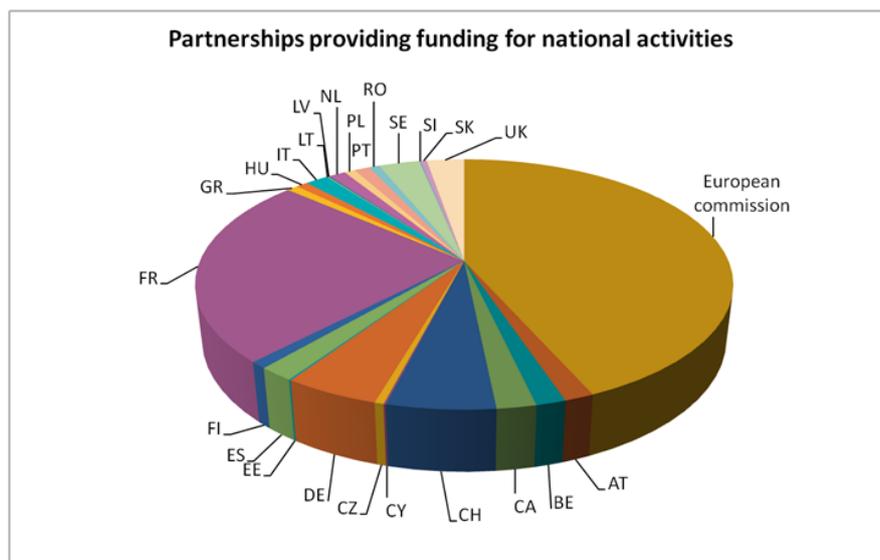
Partnerships providing funding for national activities in 2012

AUSTRIA	
Gesundheit Österreich GmbH	The "Gesundheit Österreich GmbH" (GÖG) is an associated partner in the Orphanet Europe Joint Action as of April 2011.
MEDIZINISCHE UNIVERSITÄT WIEN	The Medical University of Vienna is an associated partner in the Orphanet Europe Joint Action as of April 2011 and hosts Orphanet Austria since 2005. It further provides part-time funding (in kind) for the work of the country coordinator.
BUNDESMINISTERIUM FÜR GESUNDHEIT	The Austrian Ministry of Health provides funding to 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 additional administrative support for the project.
COOPÉRATION FRANCE-QUÉBEC	La "Commission permanente de coopération franco-québécoise" finances the teams' missions between France and Québec .
Santé et Services sociaux Québec	The Québec "Ministère de la Santé et des Services sociaux" finances a project manager position in Quebec and some administrative support.
Centre universitaire de santé McGill McGill University Health Centre	The Department of Medical Genetics of the McGill University Health Centre is the host institution of Orphanet-Quebec and provides the medical coordinator.
RQMO	Le "Regroupement québécois des maladies orphelines" provides the project coordinator and administrative support.

CYPRUS	
	The Department of Medical and Public Health Services is an associated partner in the Orphanet Europe Joint Action as of April 2011.
CZECH REPUBLIC	
	The Charles University Prague - 2nd School of Medicine is an associated partner in the Orphanet Europe Joint Action as of April 2011.
	Czech Association of rare diseases finances the activity of the Czech team since April 2012.
ESTONIA	
	The University of Tartu is an associated partner in the Orphanet Europe Joint Action as of April 2011.
FINLAND	
	The Family Federation of Finland ("Väestöliitto ry") is an associated partner in the Orphanet Europe Joint Action as of April 2011.
FRANCE	
	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.
	The "LFB Biomédicaments" helps finance the development and update of emergency guidelines and the French encyclopaedia for the general public.
	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.
	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.
	The "Institut National du Cancer" (INCa) supports the development of the Orphanet encyclopaedia on rare cancers.
GERMANY	
	The Federal Ministry of Health Germany provides funding to the Orphanet Europe Joint Action as of April 2011.
	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") was an associated partner in the Orphanet Europe Joint Action from April 2011 until August 2012.
	The National Institute for Health Development ("*Országos Egészségfejlesztési Intézet*") is an associated partner in the Orphanet Europe Joint Action as of August 2012.
ITALY	
	The Italian Health Ministry finances Orphanet-Italy activities through current research funding.
	The Bambino Gesù Children's Hospital is an associated partner in the Orphanet Europe Joint Action as of April 2011.
	Genzyme Italia finances OrphaNews Italia.
LATVIA	
	"Centre for Disease Prevention and Control of Latvia" (Slim bu profilakses un kontroles centrs) is an associated partner in the Orphanet Europe Joint Action as of April 2012.
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.
NETHERLANDS	
	The "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.
	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.
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 - Institute for Molecular and Cell Biology is the host institution of Orphanet-Portugal since 2009 and is a partner in the Orphanet Europe Joint Action as of April 2011; it partly funds the national activities, namely the full salary of the project coordinator.
	ICBAS - Instituto de Ciências Biomédicas Abel Salazar, the Biomedical Sciences Institute at the University of Porto, is an Orphanet partner since 2009; it is responsible for the full salary of the national coordinator, a full-time Professor at that institution.
	DGS - The Directorate-General of Health, at the Portuguese Ministry of Health, officially supports Orphanet-Portugal, and provided a specific grant for 2012, including part of the salary of the information scientist, as well as divulgation materials and activities, among others.

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.
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
The "Karolinska Institutet" is an associated partner in the Orphanet Europe Joint Action as of April 2011.
Karolinska University Hospital of Stockholm supports the Orphanet Sweden activities.

SWITZERLAND
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.
Since 2011, Orphanet Switzerland is funded by the Swiss Conference of the Cantonal Ministers of Public Health. The support finances the position of the coordinator (part-time), 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
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 supported 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
The University of Manchester is an associated partner in the Orphanet Europe Joint Action as of April 2011.
Nowgen in Manchester hosts Orphanet-UK's activities and contributes to the project by allocating the time of some professionals.

6.2.2. INSTITUTIONAL PARTNERSHIPS PROVIDING SERVICES IN KIND FOR NATIONAL ACTIVITIES

All the Institutions hosting the national Orphanet teams provide their office space, all the necessary supplies to run the team activities and allocate time for 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.
AUSTRALIA	
	The Office of Population Health Genomics, Department of Health, Western Australia hosts Orphanet-Australia'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.
	Nowgen in Manchester, UK hosts Orphanet-Ireland's activities and contributes to the project by allocating the time of some professionals.
ISRAEL	
	The Schneider 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-Morocco'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.

6.2.3. NON-FINANCIAL PARTNERSHIPS IN 2012

AUSTRIA	
	The Federal Ministry of Health of Austria officially supports Orphanet.
BELGIUM	
	The Federal Public Service Health, Food Chain Safety and Environment of Belgium officially supports Orphanet.
	A partnership exists with RaDiOrg.be, a EURORDIS member, which continued to play a role in the validation of data on Belgian patient organisations 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 on the Orphanet database.
CZECH REPUBLIC	
	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.
	The Ministry of Health of the Czech Republic officially supports Orphanet.
ESTONIA	
	The Ministry of Social Affairs of Estonia officially supports Orphanet.

FINLAND	
	The Ministry of Social Affairs and Health of Finland officially supports Orphanet.
	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 for the online publication of National Protocols for Diagnosis and Care (NHDP) produced by the HAS.
	The "Agence nationale de sécurité du médicament et des produits de santé" (ANSM) 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 State Secretary of Health within the Ministry of Human Resources 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 for 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 for the collection of data concerning clinical trials.
	Netgene collaborates with Orphanet for the diffusion of information on rare diseases.
	Mediart Promotion collaborates for the promotion of OrphaNews Italia.
	Farmindustria promotes Orphanet publications.
	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 aims to promote equal rights and opportunities for patients with rare diseases.
	Palidzesim.lv is a non-governmental organisation in Latvia which financially supports children and families to confirm a diagnosis of a rare disease 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.
	The VSOP (Vereniging Samenwerkende Ouder- en Patiëntenorganisaties) provides information regarding patient organisations dedicated to rare disease and participates in the validation of information on Dutch expert centres.
	The Dutch Federation of University Medical Centres and Orphanet NL collaborate in establishing a comprehensive list of Dutch expert centres.

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 rare disease with all stakeholders and improving access to orphan drugs.
Polskie Stowarzyszenie Pomocy Chorym na Fenyloketonurię ARS VIVENDI PKU	The patient organisation, Ars Vivendi, provides patients and parents with information about Orphanet services and cooperates with Orphanet Poland.
PORTUGAL	
infarmed Instituto Nacional da Farmácia e do Medicamento	INFARMED - the National Authority for Medicines and Health Products, collaborates with a regularly updated list of orphan drugs approved and available in Portugal, as well as its quantities used.
Administração Central ACSS do Sistema de Saúde	ACSS - the Central Administration of Health System acknowledges that Orphanet-Portugal is the reference source of information on rare diseases and orphan drugs in Portugal.
ces	CES - the Social Studies Center, at the School of Economics at the University of Coimbra, has been collaborating in the update and validation of the list of bona fide Patient Associations in the country.
N.E.D.R.	NEDR - the Nucleus 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	Aliança - the Portuguese Alliance of Rare Diseases Organizations has been collaborating in several joint actions with Orphanet-PT, including the update and validation of Patient Associations and the joint organization of the Rare Diseases Day each year.
FCT Fundação para a Ciência e a Tecnologia MINISTÉRIO DA CIÊNCIA, TECNOLOGIA E ENSINO SUPERIOR	FCT - The "Fundação para a Ciência e a Tecnologia" collaborates by updating information on research projects and clinical trials taking place in the field of rare diseases and/or orphan drugs ongoing in Portugal. The Orpha number is requested in every project in the life sciences dealing with rare diseases.
SPGH Sociedade Portuguesa de Genética Humana	SPGH - The Portuguese Society of Human Genetics collaborates by updating information on professionals, genetic counselling clinics, medical labs and diagnostic tests available in the country.
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 Centres of Medical Genetics and to promote collaboration with associations of people with genetic/malformative diseases.
ASOCIATIA 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	
	The Ministry of Health of the Slovak Republic officially supports Orphanet.
SLOVENIA	
	The Ministry of Health of Slovenia officially supports Orphanet.
	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	
	The Ministry of Health and Social Affairs of Spain officially supports Orphanet.
SWEDEN	
	The Ministry of Health and Social Affairs of Sweden officially supports Orphanet.
SWITZERLAND	
	The Health On the Net Foundation supports 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, the Swiss Alliance of patients with rare diseases has established a close collaboration with Orphanet Switzerland in order to identify relevant information services for patients and professionnals and in the organisation and promotion of events dedicated to rare diseases, in order to increase public awareness on this particular issue.
	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	
	The Department of Health officially supports Orphanet.
	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 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.
	Orphanet collaborates with Rare Disease UK in the sharing of data and expertise, in the endorsement and promotion of Orphanet and Rare Disease UK activities, and in the development of the UK Strategy for Rare Diseases.

7. Communication

7.1. Communication documents

For the fourth year since the creation of Orphanet, 4-page leaflets in A4 format, in four colours, each aimed at a different target audience, were distributed in 2012 at congresses:
- 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 2012, A5-size flyers to present an overview of Orphanet in 3 languages (English, French and German) and A5-size flyers presenting Orphadata were designed, printed and distributed.

7.2. Invitations to give lectures at conferences in 2012

Orphanet was invited to participate in more than 120 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 (80), public health policies (19), classification of diseases (6) or orphan drugs (3), medical and genetic approaches (16 presentations).

7.3. Booths at conferences in 2012

Orphanet booths were held in 15 different congresses in 2012 (see below). 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 Nuremberg (Germany) from 23 to 26 June 2012.
List of congresses where Orphanet booths were held:
- ProRaris – International Rare Disease Day, Lausanne, Switzerland, 25 February 2012
- RaDiOrg Information booth, Belgian University Hospitals, Belgium, 27 February – 2 March 2012
- RE(ACT) Conference – International Congress On Research of Rare an Orphan Diseases, Basel, Switzerland, 29 February 2012
- 3rd Rare disease symposium, Hannover, Germany, 29 February 2012
- 1st meeting of Orphanet-Portugal about Rare Diseases and Orphan Drugs and Regulation of Genetic Testing. Porto, Portugal, 20 April 2012.
- 6th European Conference on Rare Diseases, Brussels, Belgium, 23-25 May 2012
- Congress of the French Societies of Paediatry and Surgical Paediatry, Bordeaux, France, 6-9 June 2012
- European Society of Human Genetics meeting, Nuremberg, Germany, 23-26 June 2012
- Biospain 2012 – 6th International Meeting on Biotechnology, Bilbao, Spain, 19-21 September 2012
- National conference of medical genetics, Iasi, Romania, 4-6 October 2012
- Canadian College of Medical Geneticists Annual Scientific Meeting, Saskatoon, Canada, 18-20 October 2012
- Meeting of the Sickle Cell Disease Association of Canada, Montréal, Québec, Canada, 2 November 2012
- The American Society of Human Genetics (ASHG) congress, San Francisco, USA, 6-10 November 2012
- Orphanet promotional stand at the Research and Innovation Conference, Manchester, UK, 3 December 2012
- International Migrants Day, Hannover, Germany, 18 December 2012

8. The Orphanet team as of December 2012

COORDINATING TEAM
INSERM US14 - FRANCE

Coordinator of the Orphanet Joint Action
Ségolène Aymé

Administration, finances and communication

Financial officer
Christiane Lajeune

Assistant
Mariane Bellanger

Communications officer
Céline Angin

Chief Technical Officer
Marc Hanauer

Technical project manager
Céline Rousselot

Developers
Vincent Benoit
Laurent Corbin
Alexis Ulrich

Information systems architecture / networks / DBMS
Bruno Urbero
Sylvain Gérard

Client project manager
Valérie Lanneau

Bioinformatician
Laetitia Chanas

Ontologies
Xavier Aimé

Webmaster
Julie Christ

Chief Medical Officer
Ana Rath

Coding and classifications
Bertrand Bellet
Myriam de Chalendar
Janine-Sophie Giraudet-Le Quintrec
Catherine Gonthier
Natacha Marpillat
Annie Olry

Pharmaceutical affairs and expert resources
Virginie Hivert
└ Claude Barrère
└ Bénédicte Belloir
└ Natalia Ginaldi
└ Paul Mourlhou

Quality assurance
Charlotte Gueydan

Encyclopaedia
Clara Jaramillo
└ Natasha Barr
└ Stephanie Bee
└ Raphaël Demonchy
└ Sonja Janmaat
└ Christopher Jarvis
└ Sonia Pavan
└ Virginie Tsilibaris

Literature surveillance
Catherine Pouzat

International coordination
Sylvie Maiella
Véronique Newton

ARMENIA
Center of Medical Genetics and Primary Health Care

Coordinator
Kristine Hovhannesyan

AUSTRALIA
Office Population Health genomics

Coordinator
Hugh Dawkins

Project manager
Caron Molster

AUSTRIA
Medizinische Universität Wien & Gesundheit Österreich GmbH

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Project manager
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Sandro Coelzer
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BELGIUM
Scientific Institute Public Health & Federal Public Service Health

Project manager
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Bulgarian Association for Promotion of Education and Science

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Prof. Rumen Stefanov

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Radoslav Stilianov
Georgi Iskrov

CANADA
Mc Gill University

Coordinator
Micheil Innes (Can)
Serge Melançon (Qc)

Project manager
Gail Ouelette (Qc)

Information scientists
Mayss Naccache (Can)
Nancy Anoja (Qc)

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CZECH REPUBLIC
Charles University Prague - 2nd School of Medicine

Coordinator
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Information scientist
Marek Turnovec

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University Hospital of Aarhus

Coordinator
John Rosendahl Oestergaard

ESTONIA
Tartu University

Coordinator
Prof. Vallo Tillmann

Project manager
Rita Teek

Information scientist
Sille Vahtra

FINLAND
Väestöliitto ry - The Family Federation of Finland

Coordinator
Dr. Riitta Salonen

Information scientist
Leena Toivanen

FRANCE
INSERM US14

Coordinator
Prof. Odile Kremp

Information scientist
Myriam De Chalendar

French encyclopaedia
Arielle Lemasne
└ Marie Daniel
└ Florence Mousson

Science writer (newsletter)
Marie-Paule Hamon

Translator
Marie Charter

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Coordinator
András Becskeházi

Website administrator
Geza Ivanovits

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Coordinator
Prof. Andrew Green

Project manager
Idoia Gomez-Paramio (Manchester, UK)

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Schneider Children's Medical Center

Coordinator
Dr. Lina Basel

ITALY
Bambino Gesù Children's Hospital

Coordinator
Prof. Bruno Dallapiccola

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Coordinator
Jana Lepiksone

Project manager
Iveta Ciruke

Information scientist
Santa Rozite-Pildava

LEBANON
Université Saint Joseph

Coordinator
Prof. André Mégarbané

LITHUANIA
Vilnius University Hospital

Coordinator
Prof. Vaidutis Kucinskas

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