Orphanet Report Series

Reports collection

2009 Activity Report

orphanet

2009 Activity Report

Table of contents

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

Overview of activities in 2009

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

The site gives access to:

- An inventory of diseases including 5,857 diseases and a classification of these diseases elaborated using existing published expert classifications. Each disease is indexed with ICD10 and OMIM and the relevant prevalence class, age of onset class, mode of inheritance and associated genes.
- An encyclopaedia covering 2,687 rare diseases, written by world-renowned experts and peer-reviewed. Systematically produced in both English and French, this encyclopaedia is partly translated into German, Italian and Spanish.
- An inventory of orphan drugs at all stages of development, from EMA orphan designation to European market authorisation.
- A directory of specialised services in the 37 European countries, providing information on:
  - Specialised clinics and centres of expertise
  - Medical laboratories
  - Ongoing research projects
  - Clinical trials
  - Registries
  - Networks
  - Technological platforms
  - Patient organisations
  - Orphan drugs
- A range of other services for specific stakeholders:
  - For health care professionals: an assistance-to-diagnosis tool (search by sign and symptoms)
  - For professionals in the field of emergency health care: an encyclopaedia of emergency guidelines
  - For researchers and the Industry: a service to help build partnerships between researchers and the pharmaceutical industry through information on licence offers
  - For all: two newsletters with both scientific and political content, one covering European news, and the other French news
  - For all: regularly published thematic studies and reports on overarching subjects, downloadable from the site: “the Orphanet Report Series”
  - For the general public: instructive on-line learning modules covering overarching topics concerning rare diseases (OrphaSchool).

1. Orphanet's audience

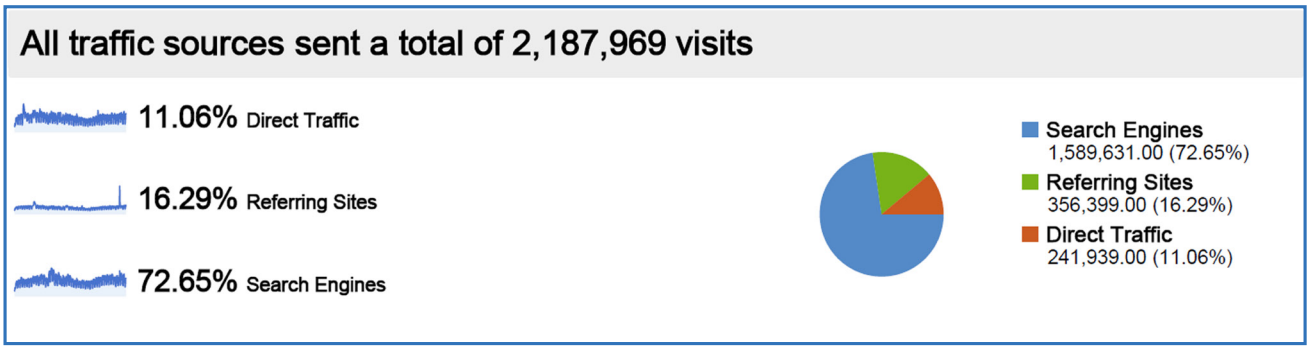
Indexation by search engines

Today (December 2009) Google indexes 880,000 documents and pages originating from the web domain www.orpha.net, thus 4 times more documents and pages than in June 2008 which gives an indicator of the depth and richness of the site. By way of comparison, the website of NORD (the website of reference for American patient organisations) has 5,460 documents referenced and the site GeneClinics, run by the National Library of Medicine, has 12,700.

The prominence of the site www.orpha.net can be evaluated by considering the number of results obtained when using the name of the site in a Google search, which amounts to 515,000 results. By way of comparison, NORD's website gives 62,400 results, GeneClinics' website gives 17,100 and INSERM's website gives 3,170,000. The Orphanet website is predominantly accessed via search engines (72.6% of visits, according to Google Analytics) and via Google alone in 66.8% cases. Other sites represent 16.3% of traffic directed towards Orphanet, the remaining visits to Orphanet are direct (i.e. bookmarks, 11.1%).

The richness of the site means that a substantial quantity of visits are brought in through a sizeable corpus of key words (rather than just certain predominant key words). The key word which is primarily used for accessing our site is simply "Orphanet", which represents 11.4% of visits. The indexation of our site is of the type "long tail": more than 287,000 different key words generate traffic to the site.

Distribution of the sources of traffic

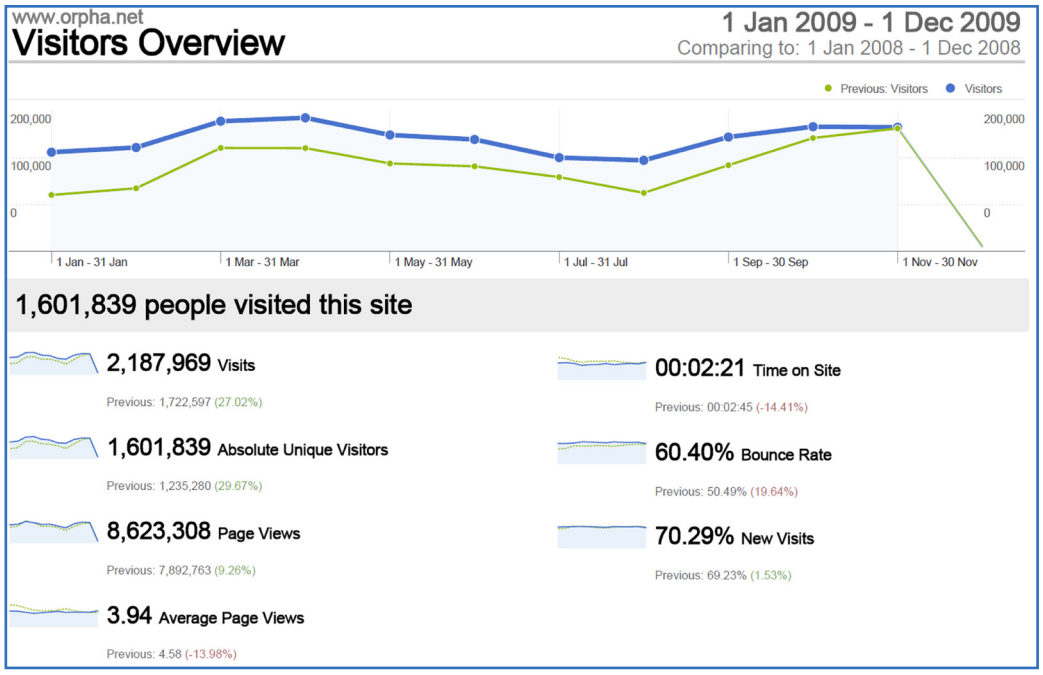


(Source: Google Analytics, 1 January to 1 December 2009)

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

The website's audience

Over the period 1 January to 1 December 2009, more than 8.6 million pages were viewed, thus on average around 24,000 pages were viewed per day. This figure has increased from last year (+9.26%), highlighting the increasing renown of the website and reinforcing Orphanet's indexed position as a referent.



(Source: Google Analytics, 1 January to 1 December 2008, compared with 2009)

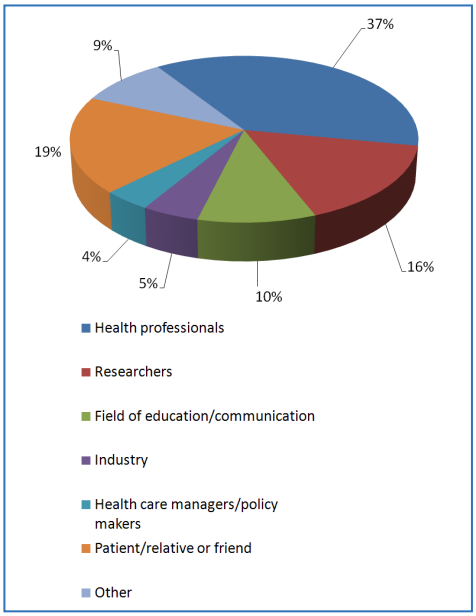
Type of users and use

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

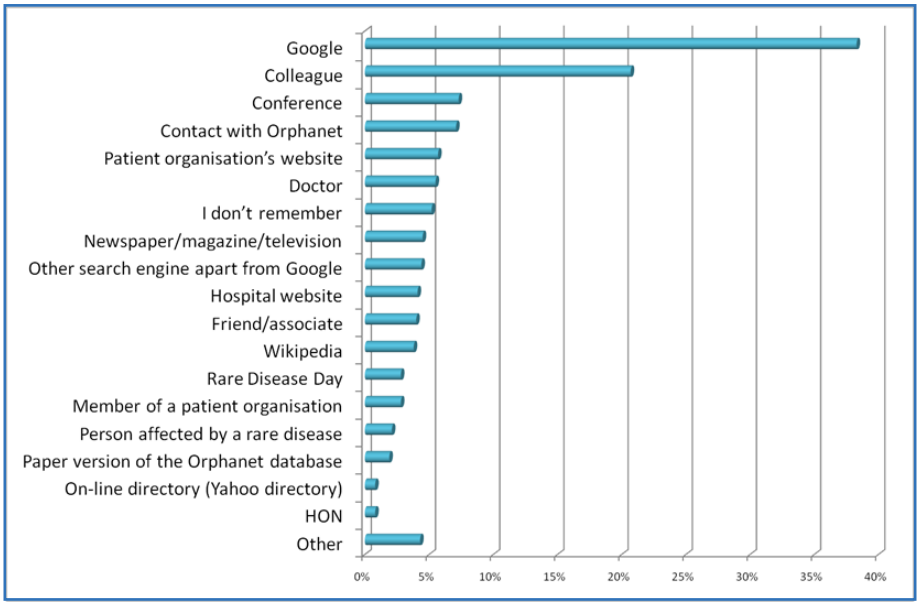
Here are the detailed results:

The site's users are :

- **Health professionals** **37.2%**
  - of which hospital specialists 16.0%
  - of which paramedicals 7.3%
  - of which independant doctors 3.3%
  - of which teachers/students 2.7%
  - of which biologists 2.7%
  - of which pharmacists 0.7%
- **Patient/relative or friend** **18.9%**
- **Researchers** **16.0%**
- **Field of education/communication** **9.9%**
- **Industry** **4.8%**
- **Health care managers/policy makers** **4.0%**
- **Other** **9.3%**



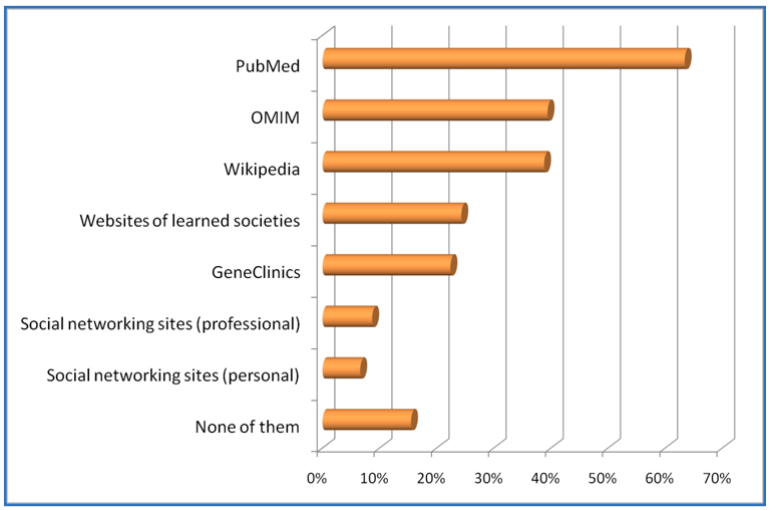
Users discover the site in the following ways (more than one choice possible):



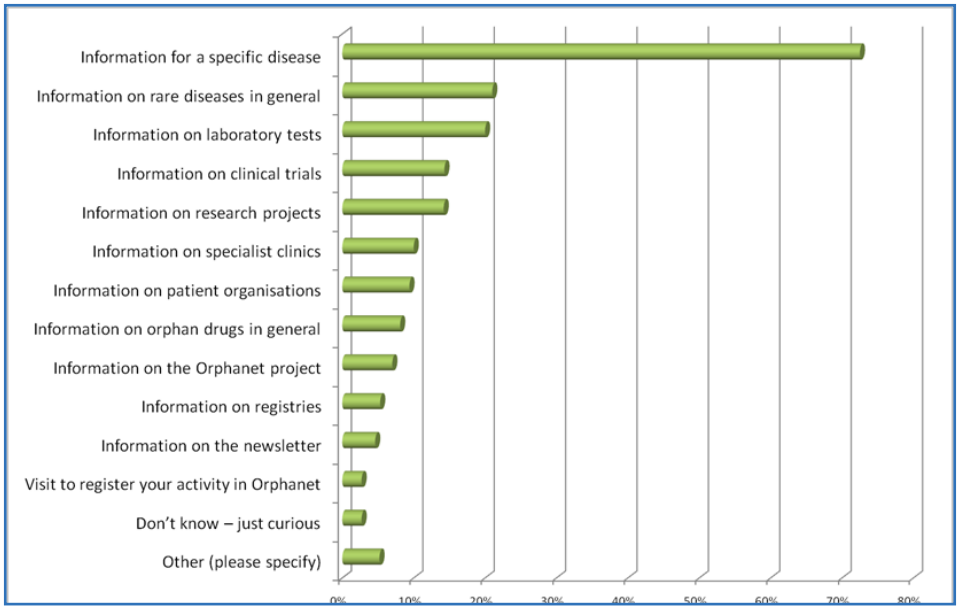
Frequency at which users visit the site:

- First visit 45 %
- More than twice a year 15%
- More than twice a month 22 %
- More than twice a week 18 %

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



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



Professionals' *opinion of usefulness of online services (percentage of users replying «very useful» and «useful»):*

	Very useful and useful
• List of diseases and classifications	90.5%
• Texts on diseases	87.0%
• Orphanet Report Series on epidemiology of Rare Diseases	59.0%
• Directory of medical laboratories	57.0%
• Directory of research projects	51.0%
• Directory of patient organisations	50.8%
• Directory of orphan drugs	50.8%
• Directory of clinical trials	49.3%
• OrphaNews newsletter	49.0%
• Directory of clinics	48.3%
• Search by sign facility	44.8%
• Orphanet Report Series on Orphan Drugs	44.3%
• Directory of registries	42.3%
• Emergency guidelines	40.5%

All users' opinion of usefulness of online services (percentage of users replying «very useful» and «useful»):

	Very useful and useful
• List of diseases and classifications	90.7%
• Texts on diseases	86.9%
• Orphanet Report Series on epidemiology of Rare Diseases	59.9%
• Directory of medical laboratories	56.8%
• Directory of orphan drugs	52.5%
• Directory of patient organisations	52.3%
• Directory of research projects	52.3%
• Directory of clinical trials	51.2%
• OrphaNews newsletter	49.9%
• Directory of clinics	49.7%
• Orphanet Report Series on Orphan Drugs	45.5%
• Search by sign facility	45.2%
• Directory of registries	43.9%
• Emergency guidelines	42.6%

Around 50% of the users who answered the online questionnaire left comments, criticism and suggestions to improve the site or words of thanks.

The audience of OrphaNews Europe, RDTF's newsletter

OrphaNews Europe is a bimonthly electronic newsletter in English, launched in June 2005. Currently more than 12,000 subscribers are registered.

2. The evolution of database content

The disease and genes database contains the following data:

- 7,842 diseases or groups of diseases and their synonyms, distributed as follows: 12,372 terms in French, 17,312 terms in English, 11,715 terms in German, 11,705 terms in Spanish, 11,674 terms in Italian, 10,198 terms in Portuguese.
- 2,132 diseases indexed with ICD10
- 3,198 diseases indexed with OMIM
- 2,496 diseases indexed with Pubmed
- 2,757 diseases indexed with prevalence data
- 2,836 diseases indexed with age of onset
- 2,892 diseases indexed with mode of inheritance
- 2,233 genes linked to 2,105 diseases
- 2,159 genes interfaced with UniProt KB
- 2,202 genes interfaced with OMIM
- 2,207 genes interfaced with Genatlas
- 2,233 genes interfaced with HGNC

The Orphanet encyclopaedia contains the following data:

- 2,592 summaries in French
- 2,687 summaries in English
- 2,221 summaries in German
- 2,339 summaries in Italian
- 1,026 summaries in Spanish
- 217 summaries in Portuguese
- 346 review articles in French
- 537 review articles in English
- 1,033 different addresses, generating 8,200 external links for 2,125 diseases
- 5 emergency guideline documents in English, and 21 emergency guideline documents in French

The directory of services in 38 European, and surrounding, countries contains the following data:

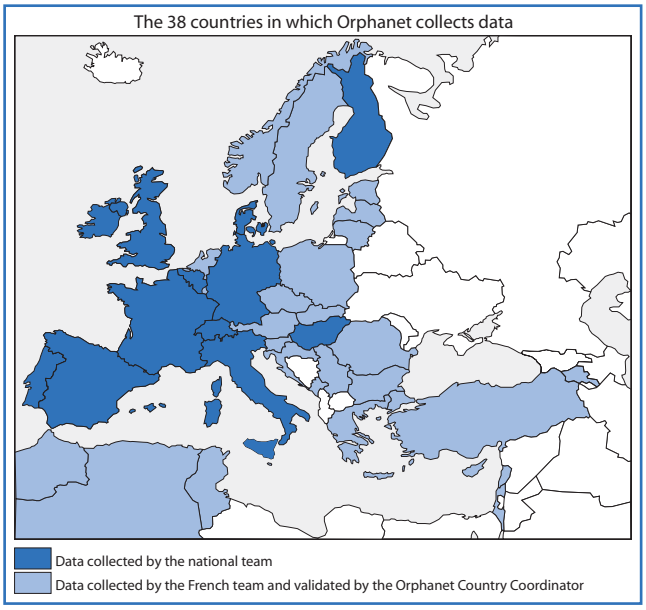
- 1,464 clinical laboratories dedicated to diagnosis
- 2,542 laboratories dedicated to research
- 21,349 medical laboratory tests linked to 2,175 diseases and 1,525 genes
- 4,770 research projects linked to 2,121 diseases
- 1,207 clinical trials for 210 diseases in 24 countries
- 1,032 registries and 743 networks
- 14,114 professionals are referenced in the database
- 4,236 specialised clinics
- 2,047 patient organisations

The 38 countries in which Orphanet collects data are the following:

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

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

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

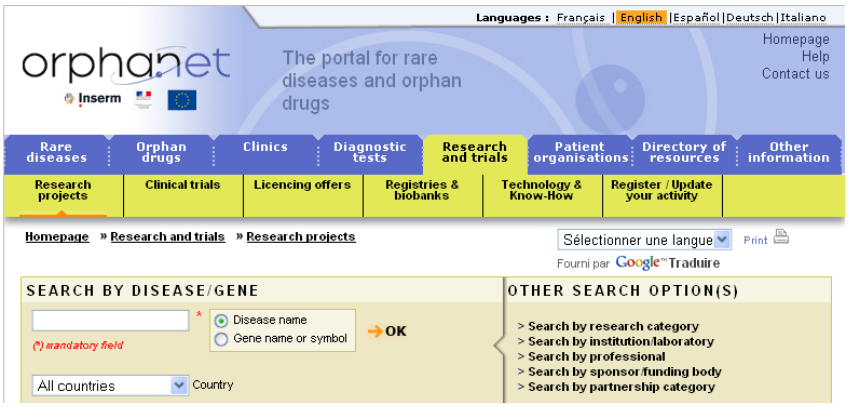


3. Evolution of products and services

Thanks to the French National Plan for Rare Diseases, and new partnerships, a new version of the site was developed and launched in April 2008, giving access to new products and services. It was not possible to put all the functionalities foreseen in the specifications in place in 2008. We have therefore continued these developments in 2009.

Functionalities of the new website developed in 2009

New functionalities were added to the "Research and trials" tab which has improved the user-friendliness of the tab and has made it easier to find the information looked for.



From now on, clinical trials and research projects are presented in two separate sub-tabs and a new sub-tab gives visitors information on registries. The search engine has also been optimised to allow for multi-criteria searches. As before, users can search by disease name, gene name or by type of project. Now users can also search by institution or laboratory, by professional, by sponsor or financing body, or by partnership category. This new development of Orphanet was achieved thanks to a grant from the European Commission (DG Research contract – RD PLATFORM HEALTH-F2-2008-201230).

New Orphanet Reports Series on orphan drugs

A new edition of the Orphanet Reports Series "List of Orphan Drugs in Europe" was published in 2009. In addition to listing the molecules which have obtained orphan drug designation in Europe, it also contains a list of drugs with an indication for a rare disease, but without prior orphan designation. Another new feature of the document is the inclusion of the detailed indication for which the products have received marketing authorisation from the European Medicines Agency (EMA).
The data is presented by trade name in alphabetical order, by date of MA in descending order, by ATC category, by ATC category or by MA holder. This document is the currently the only one of its kind to collect and synthesise this information, dispersed on the website of the European Medicines Agency.

Continuation of the Orphanet Emergency Guidelines

These guidelines are intended for pre-hospital emergency health care professionals (a dedicated section is included for their use) and for hospital emergency departments. These practical guidelines are devised in collaboration with reference centres and patient organisations, and are peer reviewed by emergency health care doctors from learned societies (SFMU, the French SAMU, SFP and SNFMI). These guidelines were launched in September 2007 and currently 9 guidelines are online and 21 others are in preparation. The initial 9 guidelines have been translated into four other languages (English, German, Italian, Spanish).

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

OJRD was indexed in Medline at the end of its first year of existence and was selected by Thompson Scientific after only two years in publication, which led to the OJRD receiving an impact factor of 3.14 in June 2009.

Currently, OJRD's production costs are covered by the Orphanet budget for solicited review articles. For spontaneously submitted articles, the authors cover the publication costs.

4. Communication

Communication documents

For the first time since the creation of Orphanet, this year we have designed, printed and distributed four types of 4 page leaflets in A4 format, in four colours, each aimed at a different target audience:

- A leaflet for all audiences on Orphanet as an information portal
- A leaflet for biologists on Orphanet as the source of information on biological tests for the diagnosis of rare diseases
- A leaflet for researchers and the 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 has been produced in 5 languages (French, English, German, Spanish and Italian).
The leaflets are freely available in the offices of the "Plateforme Maladies Rares" located at the Broussais Hospital in Paris. The leaflets have been distributed at 18 events in 2009 for a total of 860 leaflets in French, and 2790 leaflets in English. The distribution of leaflets in the other languages will start in 2010.

Invitations to give lectures at conferences in 2009

Orphanet participated in 40 conferences, of which 19 were in France and 21 abroad (Luxembourg, Italy, Spain, Germany, Belgium, Czech Republic, the United Kingdom, Austria, Romania, Turkey, Hungary and Taiwan). These presentations were mostly given at scientific conferences, in which Orphanet played the role of specialist in the field of rare diseases. These lectures dealt with medical and genetic approaches (15 lectures), public health care policy (15), the classification of diseases (5), orphan drugs (2).

In addition to this, 3 training seminars were run by Orphanet in 2009.

Booths at conferences in 2009

Orphanet had, as is customary, a booth at the annual meeting of the European Society of Human Genetics, which was held in Vienna (Austria), from 23 to 26 May 2009.

Organisation of symposia in 2009

- Symposium "Internet and rare diseases"

A forum bringing together representatives of over a hundred patient organizations was organised on 30 June 2009 around the theme "Medical data in a computerised world". This event was sponsored by the Groupama Foundataion. The event was, once again, a great success. The minutes of the event are available online on the Orphanet website.

5. The international influence of Orphanet

Collaboration with the European Commission

The director of Orphanet leads the European Commission Task Force on Rare Diseases and provides the scientific secretariat.

As such, Orphanet organised:

- A European workshop on the indexation of the Orphanet classification with SNOMED-CT: Paris, 11 March 2009
- A European workshop to discuss national initiatives taken in the field of rare diseases and to identify outcome indicators for policies put in place: Paris, 9-10 November 2009
- A European workshop to discuss research on rare diseases and in Europe, observed obstacles, the factors determining its processes, and areas to be developed: Paris, 3 December 2009

Collaboration with EPPOSI

The director of Orphanet is a board member of EPPOSI (European Platform of Patients' Organisations, Science and Industry) and, as such, was responsible for the organisation of a European workshop on rare disease registries: Brussels, 18-19 March 2009.

Collaboration with the WHO

The World Health Organisation (WHO) is currently revising the International Classification of Diseases (ICD): Orphanet collaborates with the WHO on this revision. This system aims to classify diseases, other connected health problems and external causes of disease and traumatisms in order to compile useful health information concerning deaths, diseases and traumatisms (mortality and morbidity). However, currently only 240 rare diseases are included in ICD-10 with their own code.

A group of rare disease experts, including Ségolène Aymé acting as chairperson, was established by the WHO in April 2007 in order to examine the status of these diseases in the classification system. Orphanet has been entrusted with the collection the information necessary in order to establish the alpha draft of the new classification of rare diseases. This classification will serve as a model for the entire revision of the ICD as rare diseases are present in all fields of medicine.

Orphanet has now completed the revision of three chapters of ICD-10: haematological and hematopoietic diseases and certain problems of the immune system; endocrine, nutritional and metabolic diseases; and metabolic diseases. These chapters are sent for revision to a wide panel of international experts. Orphanet also plans to revise each chapter of the classification in the following months. The new version of the ICD (ICD-11) should be adopted in 2014.

Scientific collaborations and partnerships with Industry

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

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

The provision of data to these parties is free for public institutions and charged for Industry and private companies. All situations require that Data Transfer Agreement be signed. Partnerships in the Industry are managed by Inserm Transfert.

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Communications officer
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Funding

1. Orphanet Funding

Orphanet was established in 1997 by the French Ministry of Health (Direction Générale de la Santé) and the INSERM (Institut National de la Santé et de la Recherche Médicale). Both agencies still fund the core project. The European Commission funds the encyclopaedia and the collection of data in European countries (since 2000 DG Public Health and Consumers Protection grants N°s S12.305098; S12.324970; SPC.2002269-2003220 and since 2004 DG Research grants N°s LSSM-CT-2004-503246; FP6-512148; LSHB-CT-2006-08933).

Other sponsors support some Orphanet services:

- In France: the "Association Française contre les Myopathies" sponsors OrphaNews France and OrphaNews Europe and the collection of data on clinical trials. "Les Entreprises du Médicament" (LEEM) sponsors the collection of data concerning orphan drugs and clinical trials. The "Fondation Groupama pour la Santé" sponsors the website services for patient organisations in France. "LFB Biomédicaments" sponsors the production and update of emergency guidelines and texts for the French encyclopaedia for general public. "Shire" sponsors the development of the Orphanet encyclopaedia. "Alexion" sponsors the translation of the emergency guidelines from French.
- In Germany: The "Land Niedersachsen" (Lower Saxony) sponsors part of the activities of the German Orphanet team.
- In Portugal: The "Fundação para a Ciência e a Tecnologia" sponsors part of the activities of the Portuguese Orphanet team.
- In Switzerland: The University Hospitals of Geneva, the 26 Swiss cantons and the Principality of Liechtenstein sponsors all the activities of the Swiss Orphanet team.
- In Spain: "CIBERER, Centro de Investigación Biomédica en Red de Enfermedades Raras" sponsors part of the activities of the Spanish Orphanet team.

Many other institutions from the participating countries contribute in kind to Orphanet.

2. National Funding or contribution in kind

Armenia
Center of Medical Genetics and Primary Health Care
Austria
Medical University Vienna
Belgium
University of Leuven
Bulgaria
Bulgarian Association for Promotion of Education and Science (BAPES)
Croatia
Zagreb University
Cyprus
Archbishop Makarios III Hospital

Czech Republic
Charles University Prague - 2nd School of Medicine (UCPRA-2SM) and UH Motol
Ministry of Health of Czech Republic
Grant Agency of Czech Republic - GACR
Denmark
Kennedy Center (KISOE)
Estonia
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