Programme of community action on rare diseases

Contract 2003220

ORPHANET 4:
Final Implementation Report

Start Date: 01 April 2004
Duration: 36 Months
Project Leader: Dr Ségolène Aymé, INSERM, Paris, France
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Summary

Objective

The project’s objective was to offer reliable, up-to-date, relevant information on rare diseases and orphan drugs to health care professionals, scientists, health authorities, patients and their relatives, the media and the community at large on an existing open-access website, Orphanet. The information provided on rare diseases is comprised of the name, general description, prevalence rate in the community, synonyms, symptoms, causes, epidemiological data, preventive measures, standard treatments (e.g. orphan drugs), clinical trials, diagnostic laboratories, specialised consultations, research projects and additional resources for most rare diseases. This information has been made available through two products: an Encyclopaedia and a Directory of Services both available at the project’s website, www.orpha.net.

Project Goals

The project goals included an increase in number of the Orphanet website users, an increase in the number of Encyclopaedia entries and an increase of data in the Directory of Services. At the beginning of this contract, 1 April 2004, the Orphanet website was accessed by an average of 4,000 independent users per day from all over Europe. At that time, the Encyclopaedia of rare diseases was available both in English and French and translation into German, Italian, Spanish and Portuguese was projected. The Encyclopaedia covered 990 diseases with abstracts in French and in English for all and a review article for 445 of them, either in French or in English. The goal was to cover 1,200 diseases in English by 2006. In April 2004, the Directory of Services contained information about clinical laboratories and specialised consultations in Austria, Belgium, France, Germany, Italy, Portugal, Spain and Switzerland. The goal was to expand data collection to include clinical trials and research projects in these countries. Another goal was also to expand data collection to include diagnostic laboratories, specialised clinics, research projects and clinical trials in Bulgaria, Cyprus, Denmark, Estonia, Finland, Greece, Hungary, Lithuania, Ireland, Netherlands, Romania, United Kingdom. The success of this project was to be measured both in terms of the increase in volume of information published on the website and in terms of number of users of the website.

Achievements

The volume of users of the website has dramatically increased. At the end of March 2007, the average number of visits per day was approximately 22,000 from over 160 countries. Two-thirds of these users are professionals, and one-third is patients and families. The percentage of satisfied users is 99.2 % among Francophone users and 96.1 % among Anglophone users. Orphanet is now the most highly accessed website in the world in the field of rare diseases.

The Encyclopaedia has since expanded as planned. As of March 2007, it contained 2,072 summaries in English, 2,191 summaries in French, 1,822 summaries in Italian, 609 summaries in Spanish, 1,512 summaries in German, 228 summaries in Portuguese, 270 review articles in
French and 436 review articles in English. The data collection of services is continually in progress in all participating countries. As of March 2007 the Directory of Services included data concerning: 1,185 clinical laboratories, 2,613 specialised clinics, 4,073 research projects, 668 clinical trials, 1,680 support groups, and 9,245 professionals in all participating countries. In conclusion, the project has developed according to plan and even surpassed initial expectations in the number of website users, the number of summaries published in the Encyclopaedia, and the data collected in the Directory of Services. Through the establishment of a network of European partners, the consolidation of scarce and scattered rare disease information and resources has addressed a great unmet need of the rare disease community.

A. Detailed Description Activities

1. Objective

The project’s objective was to offer reliable, up-to-date, relevant information on rare diseases and orphan drugs to health care professionals, scientists, health authorities, patients and their relatives, the media and the community at large on an existing open-access website, Orphanet. Through the establishment of a network of European partners and their exchange of rare disease information, the consolidation of scarce and scattered rare disease information and resources on the Orphanet database has addressed a great unmet need of the rare disease community. The information provided on the Orphanet database is comprised of the name, general description, prevalence rate in the community, synonyms, symptoms, causes, epidemiological data, preventive measures, standard treatments (e.g. orphan drugs), clinical trials, diagnostic laboratories, specialised consultations, research projects and additional resources for most rare diseases. This information has been made widely available on the European level through two products: an Encyclopaedia and a Directory of Services, both available on the project’s website, www.orpha.net.

2. Activities foreseen

The foreseen activities were conducted over a period of 36 months from 1 April 2004 to 31 March 2007. The expected results, presented by year in Table 1, describe the expected increase in website users, development of the Encyclopaedia, development of the Directory of Services, and remaining project management tasks.
Table 1. Expected results

<table>
<thead>
<tr>
<th></th>
<th>Year 1</th>
<th>Year 2</th>
<th>Year 3</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Website Users</strong></td>
<td>• Increase in the average number of users per day up to average of 5,000</td>
<td>• Increase in the average number of users per day up to average of 6,000</td>
<td>• Increase in the average number of users per day up to average of 7,000</td>
</tr>
<tr>
<td><strong>Encyclopaedia</strong></td>
<td>• Publication of 1,000 expert written summaries in English</td>
<td>• Publication of 1,100 expert written summaries in English</td>
<td>• Publication of 1,200 expert written summaries in English</td>
</tr>
<tr>
<td></td>
<td>• Publication of expert written review articles in English on 350 diseases</td>
<td>• Publication of expert written review articles in English on 450 diseases</td>
<td>• Publication of expert written review articles in English on 550 diseases</td>
</tr>
<tr>
<td></td>
<td>• Publication of short summaries in English on 200 disease written in-house</td>
<td>• Publication of short summaries in English on 400 disease written in-house</td>
<td>• Publication of short summaries in English on 600 disease written in-house</td>
</tr>
<tr>
<td><strong>Directory of Services</strong></td>
<td>• Data collection and publication of clinical trials in Austria, Belgium, France, Germany, Italy, Portugal, Spain, and Switzerland on website</td>
<td>• Data collection and publication of diagnostics laboratories and specialised clinics in Bulgaria, Cyprus, Denmark, Estonia, Ireland, Lithuania, Finland, Greece, Hungary, Netherlands, Romania, United Kingdom on website</td>
<td>• Data collection and publication of clinical trials in Bulgaria, Cyprus, Denmark, Estonia, Ireland, Lithuania, Finland, Greece, Hungary, Netherlands, Romania, United Kingdom on website</td>
</tr>
<tr>
<td><strong>Management</strong></td>
<td>• Establishment of National Scientific Advisory Board committees</td>
<td>• Executive Board Meeting</td>
<td>• Executive Board Meeting</td>
</tr>
<tr>
<td></td>
<td>• 4 day training session for each new partner</td>
<td>• Update of all information</td>
<td>• Update of all information</td>
</tr>
<tr>
<td></td>
<td>• Executive Board Meeting</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>• Update of all information</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

3. Achieved Activities

a) User Statistics
As of the end of March 2007, and according to Google statistics, the number of Orphanet visitors per average weekday is 22,000, from over 160 countries. The increase is three times much larger than expected. Orphanet is now the most accessed website in the world in the field of rare diseases.

b) Encyclopaedia
As of the end of March 2007, the Orphanet Encyclopaedia contained 2,072 summaries in English, 2,191 summaries in French, 1,822 summaries in Italian, 609 summaries in Spanish, 1,512 summaries in German, 228 summaries in Portuguese, 270 review articles in French and 436 review articles in English. We have produced more summaries in English than planned (1200 written by experts and 600 written in-house) and a bit less review articles than expected (550).
This was based on a decision to reallocate resources toward the production of summaries which are the key products sought by end users. Translation of summaries has also progressed.

c) Directory of Services
The collection of data for the Directory of Services has taken place in all the participating countries excepted Lithuania and Bulgaria where their respective governments had not yet signed the memorandum on public health required necessary for funding.
The increase in the volume of data collected and published on the website since the beginning of the contract is as follows:

<table>
<thead>
<tr>
<th></th>
<th>April 2004</th>
<th>March 2007</th>
</tr>
</thead>
<tbody>
<tr>
<td>Clinical laboratories</td>
<td>693</td>
<td>1,185</td>
</tr>
<tr>
<td>Specialised clinics</td>
<td>1,136</td>
<td>2,613</td>
</tr>
<tr>
<td>Research projects</td>
<td>1,580</td>
<td>4,073</td>
</tr>
<tr>
<td>Clinical trials</td>
<td>107</td>
<td>668</td>
</tr>
<tr>
<td>Support groups</td>
<td>766</td>
<td>1,680</td>
</tr>
<tr>
<td>Professionals</td>
<td>4,148</td>
<td>9,245</td>
</tr>
<tr>
<td>Total</td>
<td>8,430</td>
<td>19,464</td>
</tr>
</tbody>
</table>

The total amount of data has, therefore, increased by 2.3 times during the duration of the contract. Detailed statistics are available on the OrphanPlatform website, www.orphanplatform.org.

d) Updates
The Orphanet Encyclopaedia is currently updated every three years. The database of services is updated yearly. During Year 3, we developed an online system allowing professionals to directly update their activity. Currently, they receive a copy of all the data which are linked to them once a year and can modify them if necessary. This new system has increased the response rate up to 80%.

e) Scientific Advisory Boards
The list of the members of the National Scientific Advisory Boards can be found on the web page of each country accessible from the Orphanet home page (click on "about Orphanet"). Every participating country except Bulgaria and Lithuania has established a National Scientific Advisory Board. In the two latter countries, the effective participation in Orphanet activities could not start as their respective governments had not yet signed the memorandum on public health necessary for funding.

f) Meetings
Four meetings of the Executive Board (April 2004, April 2005, November 2005, April 2006), one in addition to the three foreseen, have taken place in Paris. These meetings were organized outside this contract which did not provide financial support for them. Four-day-long training sessions were organized by the central team in Paris for each partner during the 3 year period of the contract.
B. Manpower for the execution of activities

1. Manpower in France
Orphanet was run by a dedicated Inserm team in Paris.
The manpower dedicated to management activities was 330 days of a director + 295 days of an administrative manager + 330 days of a secretary.
The informatic aspects of the project required 660 days of a database expert and 640 days of a network/system expert.
The production of the Encyclopaedia required 1,653 days of an editor.
The database of diseases required 155 days of information scientists.
The collection of data on services in France required 1,155 days of an information scientist.
The supervision of the data collection, training and quality control required 495 days of a data manager.

2. Manpower in Estonia
The collection of data on services required 64 days of the coordinator, 34 days of a secretary and 562 days of an information scientist.

3. Manpower in Cyprus
The collection of data on services required 330 days of the coordinator, 330 days of an information scientist.

4. Manpower in Portugal
The collection of data on services required 73 days of the coordinator and 785 days of an information scientist.

5. Manpower in Germany
The collection of data on services required 468 days of an information scientist.

6. Manpower in the Netherlands
The collection of data on services required 5 days of the coordinator and 581 days of an information scientist.

7. Manpower in Denmark
The collection of data on services required 9 days of the coordinator and 218 days of an information scientist.

8. Manpower in Greece
The collection of data on services required 330 days of an information scientist.

9. Manpower in Hungary
The collection of data on services required 124 days of the coordinator and 468 days of an information scientist.
10. Manpower in Spain
The collection of data on services required 348 days of the coordinator and 1,291 days of an information scientist.

11. Manpower in Italy
The coordination required 40 days of an MD. The Encyclopaedia in Italian required 546 days of an editor. The collection of data on services required 779 days of an information scientist.

12. Manpower in United Kingdom
The collection of data on services required 547 days of an information scientist.

13. Manpower in Romania
The collection of data on services required 330 days of an information scientist.

C. Distribution of Work

1. Coordinating Team (Orphanet France)

The coordination of the project was managed by the coordinating team, Orphanet France, located in department SC11 of Inserm (the French National Institute of Health and Medical Research). The team was in charge of coordinating network activities, all hardware and software aspects of the project, the database of rare diseases and the production of the Encyclopaedia.

The coordination activities included the organisation of the partners’ meetings and of the training sessions, the supervision of the distant teams in charge of collecting national data, the quality control of the data about services in the participating countries, the production of data statistics for the follow-up of the project and their release on the Orphanplatform website.

The hardware and software aspects of the project included the management of the Unix server (over 20,000 connections per day and 800 transactions per day to update the data) and the development of new tools to collect and update information. Until December 2005, the database was hosted by the Orphanet Sun Server in the Infobiogen Centre located in Evry, suburb of Paris. Due to the closure of Infobiogen, the server was transferred to the department of informatics of the University Paris IV La Sorbonne. As we were not satisfied with the service in terms of security of the server, we established a partnership with the Informatics Department of Inserm and transferred the Orphanet server to the Inserm Central Informatics Facility in Villejuif, in February 2007. The service is now guaranteed 7 days per week, 24 hours per day. The main change in the methodology that took place during Year 3 is the development of an online system to allow professionals to update their activity. Currently they receive a copy of all data linked to them once a year and can modify them if necessary. This development facilitates the updating and the collection of new data. Due to this new methodology we had a 80% response rate this year, much higher that in previous years.
The coordinating team was also in charge of the database of rare diseases which includes the names of the disease and synonyms, prevalence rate in the community, symptoms, epidemiological data, OMIM and ICD codes. The list was updated through a permanent survey of the medical and scientific literature. It was also in charge of producing the Encyclopaedia by contacting the editorial board members to ask them to nominate authors, approaching the nominated authors, checking the quality of the manuscript and the conformity with the Orphanet format, sending the manuscripts to editors for review, making the final changes according to the editor’s requests, entering the texts into the database, managing the updating process. Moreover, the coordinating team is also responsible for collecting, validating and entering data on clinical trials, diagnostic laboratories, specialised consultations, research projects, support groups and additional sources of information in France.

2. Partner Activities

The establishment of a Directory of Services can only be achieved by consolidation of data collected at the MS level as the identification of the expert resources requires a very good knowledge of the national research and health care networks. Each country coordinator is a national expert – an essential element in establishing a scientific committee at the MS level. All national coordinators are located in high profile institutions which can provide the adequate environment for the information scientists to work, in terms of documentation, secretarial facilities and access to the network.

Each national partner is responsible for collecting, validating and entering data on clinical trials, diagnostic laboratories, specialised consultations, research projects, support groups and additional sources of information in their respective countries. Each partner has the choice either to be provided with the computer system tools to access and update the database from its own premises or to send the data to the central team for data processing. Currently the countries which are fully responsible for their data entry include: Belgium, Germany, Italy, The Netherlands, Portugal, Spain, Switzerland, UK. This process is illustrated on figure 1.

![Figure 1](image-url)
3. Management boards

The project is run by three committees:
   a) The management board, composed of country coordinators and chaired by the scientific coordinator. It meets once a year and decides on the quality charter, the evolution of the database, the budget, and the dissemination of information.
   b) The national scientific advisory boards, composed of national experts from all relevant fields of medicine. It is in charge of advising the executive board on all relevant matters and of validating the data before it is released. Members are nominated by country coordinators. The organisation of the work and formal meeting of this committee is decided at the national level.
   c) The editorial board of the Encyclopaedia composed of 83 experts in charge of nominating authors of review articles. All work is done electronically with no formal meetings (Annex 1).

D. Achievement of objectives

1. Evaluation of results

a) User Statistics

User statistics are available on the Orphanet website on a daily basis. As the reputation of Orphanet was already well established at the beginning of this contract, the increase in the average number of users is significantly larger than expected. At the end of this contract, an average of more than 22,000 independent users visited the Orphanet website daily (figure 2). Orphanet is now the most accessed website in the world in its category.

![User Statistics 2004-2006](Figure 2)
The website is visited by users from over 160 countries as illustrated in the map below, extracted from Google analytics (Figure 3).

![Synthèse géographique](image)

**Figure 3**

b) Encyclopaedia

As of March 2007, the Orphanet Encyclopaedia contained 2,072 summaries in English, 2,191 summaries in French, 1,822 summaries in Italian, 609 summaries in Spanish, 1,512 summaries in German, 228 summaries in Portuguese, 270 review articles in French and 436 review articles in English. Far more summaries and a bit fewer review articles have been produced than originally planned. This was based on a decision to reallocate resources toward the production of summaries which are the key products that end users are seeking. The number of articles and summaries by year is illustrated on Figure 4.
Diseases Covered in Encyclopedia

Review Articles

Summaries

* note that 2004 review article statistics include review article in French and English together
c) Directory of Services

As of March 2007 the Directory of Services included data concerning: 1,185 clinical laboratories, 2,613 specialised clinics, 4,073 research projects, 668 clinical trials, 1,680 support groups, and 9,245 professionals in Austria, Belgium, France, Germany, Italy, Portugal, Spain, Switzerland, Cyprus, Denmark, Estonia, Finland, Greece, Hungary, Ireland, the Netherlands, Romania, and the United Kingdom. The data collection of services could not start in Lithuania or Bulgaria, as their respective governments had not yet signed a memorandum on public health necessary for funding.
d) Establishment of the Orphanet Journal of Rare Diseases

In order to motivate authors to contribute and to publish their solicited review articles, an electronic open-access journal published by to BioMedCentral, “Orphanet Journal of Rare Diseases”, was established. This development provides an opportunity for the articles to be indexed by Medline and to have an official impact factor (with a usual delay of two years) increasing the visibility of the articles. The journal was launched in March 2006. Approximately 25% of the published articles are classified by the National Library of Medicine as “highly accessed”, suggesting that the right decision was made. The only negative consequence as a result of this decision has been an increase in the cost of publishing, as BioMedCentral charges for the publication. The users of the Orphanet website continue to access the articles directly from the website as in the past.

e) Establishment of a partnership with the European Journal of Human Genetics

Orphanet has also signed an agreement with the European Journal of Human Genetics (EJHG) to co-publish the Orphanet articles which have an important genetic content. The review articles are published on the Orphanet website and in the EJHG with the Orphanet logo. This development was also intended to motivate authors who are better referenced through this well established journal.
2. Satisfaction of users

An online survey was performed in April 2006 to better understand the needs of end users and assess their satisfaction. The questionnaire was systematically proposed to all visitors who had the option of refusing to complete it before entering the website. The questionnaire was proposed to all visitors until 1,200 were completed.

The results of the survey are as follows:

<table>
<thead>
<tr>
<th>Website users</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Health professionals</td>
<td>55.8 %</td>
</tr>
<tr>
<td>Patients and families</td>
<td>29.8 %</td>
</tr>
<tr>
<td>Others</td>
<td>14.4 %</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Referral to Website</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Search engine</td>
<td>51.6 %</td>
</tr>
<tr>
<td>Physician</td>
<td>7.6 %</td>
</tr>
<tr>
<td>Colleague</td>
<td>6.6%</td>
</tr>
<tr>
<td>Hospital website</td>
<td>5.2%</td>
</tr>
<tr>
<td>Expert clinics</td>
<td>2.2%</td>
</tr>
<tr>
<td>Other</td>
<td>19.9%</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Frequency of visits</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>First visit</td>
<td>49.3 %</td>
</tr>
<tr>
<td>More than twice a year</td>
<td>17.8 %</td>
</tr>
<tr>
<td>More than twice a month</td>
<td>22.7 %</td>
</tr>
<tr>
<td>More than twice a week</td>
<td>10.2 %</td>
</tr>
</tbody>
</table>
### Type of information sought

<table>
<thead>
<tr>
<th>Type of Information Sought</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Specific disease</td>
<td>51.6%</td>
</tr>
<tr>
<td>Patient organisation</td>
<td>7.6%</td>
</tr>
<tr>
<td>Clinical laboratory</td>
<td>6.6%</td>
</tr>
<tr>
<td>Research project</td>
<td>5.2%</td>
</tr>
<tr>
<td>Expert clinics</td>
<td>2.2%</td>
</tr>
<tr>
<td>Clinical trial</td>
<td>19.9%</td>
</tr>
</tbody>
</table>

### Satisfaction with the information

<table>
<thead>
<tr>
<th>Satisfaction Level</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Totally satisfied</td>
<td>74.9%</td>
</tr>
<tr>
<td>Partially satisfied</td>
<td>23.9%</td>
</tr>
<tr>
<td>Not satisfied</td>
<td>1.2%</td>
</tr>
</tbody>
</table>

### 3. Communication

Orphanet accomplishments were presented through the following lectures, publications, presentations and media:

**Estonian team:**

- **Communications:**
  - Õunap K.: Presentation about the Orphanet project. Tartu University Hospital Annual Conference, Tartu
  - Metspalu A.: Presentation about the Orphanet project. Children hospital, May 2004

- **Press, Media:**
  - Eesti Arst, Introduction about the Orphanet project, July 2006
Cyprian team:
Communications:
• Minister of Health: “ORPHANET network and our involvement in Orphanet-Cyprus”. WHO ‘International day for health’, April 2005

Danish team:
Communications:
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4. Conclusion

The Orphanet project has developed according to plan and even surpassed initial expectations despite a difficult first year during which many new partners had to be trained, supervised, and supported without the required financial resources.

The audience of the website has increased much more rapidly than expected. Orphanet now ranks very well in search engines making it the most frequently visited site in the world in its category. Through the establishment of a network of European partners, the consolidation of scarce and scattered rare disease information and resources has addressed a great unmet need of the rare disease community.

The challenges of the future are to expand the network to all European countries and to other surrounding countries. The Orphanet network will include soon Croatia, Czech Republic, Latvia, Lebanon, Luxembourg, Malta, Morocco, Norway, Poland, Serbia, Montenegro, Slovakia, Slovenia, Sweden, Tunisia, and Turkey which will be partners of the next contract. The current version of the database is entirely available in English and French, and partially available in Italian, German, Spanish and Portuguese. Funding for translation was not available.

Collecting information on expert services in MS provided an opportunity to confront the peculiarities of the health care systems and the heterogeneity of the national approaches toward rare diseases. The Orphanet database content was used for the analysis of the situation of centres of reference in participating countries. This analysis was published as Rare Diseases Task Force reports in September 2005 and December 2006. These documents are accessible from the EU health portal. An analysis of the situation regarding genetic testing is ongoing in collaboration with EuroGenTest, an EU funded network of excellence. In addition to this collaboration, Orphanet was requested to become partner of several other EU funded FP6 projects: Evi-Genoret, CliniGene, EMIL, PHGEN and Treat-NMD and serve as a foundation for another FP6 project: OrphanPlatform. At the International level, a formal collaboration has been established with GeneTest (NIH), SwissProt (Switzerland) and the World Health Organization for the coding of rare diseases. All these collaborations demonstrate that Orphanet is now considered as the database of reference for rare diseases at the international level. This was possible thanks to the long-term commitment of the French public institutions (Ministry of Health and Inserm). The current contract contributed to funding the expansion of the data collection in new countries and to fund the development of the Encyclopaedia in English. The sustainability of Orphanet must now be considered.
Current network of Orphanet partners
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