

OrphanPlatform



Contract n°: **LSSM-CT-2004-503246**

Project acronym : **OrphanPlatform**

Instrument: **Specific Support Action**

Project title: **A European Platform of Integrated Information Services for the coordination of rare disease research in Europe, with various stakeholders from research, SMEs and patient organisations and the coordination of early clinical trials**

Thematic priority: **LSH-2002-2.1.1-12.**

first annual activity report

Period covered: from 1 April 2004 to 31 March 2005

Date of preparation: 15 may 2005

Start date of project: 1 April 2005

Duration: 2 years

Project coordinator: **Ségolène AYME**

Organisation name **INSERM**

Draft 2

Executive summary

Project objectives

The project aims at developing information tools to address in a comprehensive and integrated approach the set of factors that currently affects research on rare diseases and its coordination.

The specific objectives are: (1) to develop an information service, freely accessible on Internet, dedicated to research activities in the field of rare diseases and orphan medicinal products, including a database of research projects, funded at MS level and at the EU level, and a database of collections and research networks. (2) to develop services aiming at speeding up the enrolment of patients in clinical research. (3) to develop a database of research projects with development potential, to help scientists and Industry establish the necessary partnerships. (4) to organise a workshop with all stakeholders to discuss known bottlenecks and find solutions.

The project aims at establishing the platform of services in 11 European countries in the pilot phase in order to propose an extension to the 25 European countries in 2006.

Ultimately, the goal is to convert scientific developments in the field of rare diseases into diagnostic tools and therapies as quickly as possible.

Two websites will be launched to support these activities: www.orphanplatform.org to serve as a management tool between partners; and www.orphanXchange.org to serve as a tool to facilitate partnership between researchers and Industry. All the collected information on on-going research activities and all the new services developed during the course of this contract will be available on www.orpha.net

Contractors involved

This project is based on input from the following (1) an EU funded information network: Orphanet (www.orpha.net) (2) A European platform of Patients organization, Science and Industry (EPPOSI) which actively supports partnering activities. (3) two umbrella organizations of patient support groups (Eurordis and VSOP) involved in supporting research and regulatory activities. (4) three umbrella organisations of Industry: Emerging Biopharmaceutical Enterprises (EFPIA), EuropaBio, LEEM. (5) 11 Academic institutions (INSERM (France), University College of Cork (Ireland), Istituto Casa Sollievo Della Soffenza (Italy), Universita Pompeu Fabra (Spain), Victoria University of Manchester (UK), Katholieke Universiteit Leuven (Belgium), Academic Medical Center Amsterdam (Netherlands), University of Turku (Finland), Instituto de Genetica Medica Jacinto Magalhaes (Portugal), Medizinische Hochschule Hannover (Germany), Medical University of Vienna (Austria).

Coordinators contact details

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Work performed

1- The first sub project was to extend the Orphanet directory of services in order to provide accurate information on on-going research activities at the Member states level and at the EU level, in a format easy to use for researchers, clinicians, Industry, patients, administrative bodies and any decision-maker.

This specific objective was composed of two subprojects: (1) the extension of the Orphanet database to accommodate this new type of information and (2) the collection of data in participating countries.

All the developments were performed during the first months. The database was ready for data injection by September 04.

The lists of research funding agencies and the sources to identify research projects in the Orphanet partner countries have been established. Collection of data in all participating countries is mainly performed through a common questionnaire that has been translated into all National languages. Collection of projects published on the DG SanCo and the DG Research websites was performed by the coordinating team and specific information sent to corresponding country partners.

2- The second subproject was to develop a new facility to allow patients to find out whether there is a clinical research project on their disease, which they could participate in.

This facility has been developed according to plans.

Only patients can register, or parents and legal representatives of a person suffering from a rare disease; family and doctors can pass on information to patients (an information letter is available and may be downloaded from the website).

It is be an **opt-in** service

- Patients can access the list of clinical trials regarding their disease. If there is no on-going trial, the patient can decide to leave his contact details in order to be informed about any new project.
- If there is an on-going trial, the patient can access the protocol to check whether he seems eligible. He is advised to discuss with his physician about the possibility to apply for enrolment.
- The patients who leave their contact details can access, modify or suppress their data at any moment.
- They will be systematically asked to re-confirm their willingness to remain in the database, once a year.

The service to allow patients to register as volunteers for participating in clinical trials was launched in French and English on 6 January 2005. Translation of the service into German, Spanish, Portuguese and Italian has been done as required by the OrphanPlatform contract for the end of the first year: however, the service is not yet on-line in these languages as the service requires more administration than previously thought. We are currently discussing the possibility that OrphanPlatform partners could manage this service in their national language after having obtained permission to manage the data from their legal authorities.

3- The third sub project was to identify, among all the research projects listed in the database, the research projects which reach a stage at which they can be carried out by an industrial company or form the basis for a start-up company, and be developed into a commercial product or technique. These projects were be made accessible and visible on the OrphanXchange website, a

separate website from Orphanet but with functional links. The two projects would share part of the Orphanet database but the front ends would be different. The necessity to have a separate website came from the fact that the OrphanXchange website will be password protected when the Orphanet website is fully accessible without any registration.

This subproject had two components: (1) the establishment of a website and of a database of research projects seeking partnership with Industry, (2) the collection of research projects to fill in the database. The website opened in June 2004. It is accessible at : www.orphanxchange.org

The research projects included in the database are coming from three sources: (1) a selection of projects of potential industrial interest listed in Orphanet; (2) potential orphan designations identified through mail surveys sent to European clinicians; (3) projects collected by departments of technology transfer of research institutions.

4- The fourth sub project was to organise a partnering workshop during the second year of the contract. There is a strong view that additional action needs to be taken to energise and expedite the development of therapies and suitable diagnostics for rare diseases. A workshop is needed to exchange views and explore options for facilitating the development of therapies and identify research projects in advanced stages. It will follow the scheme of previous partnering workshops organised by EPPOSI: Brussels (2000); Paris (2001), Rome (2002), The Hague (2003), Berlin (2004), but it will focus on Orphanplatform main concepts: coordination of research activities and research funding in Europe; improvement of partnering activities; improvement of clinical trials in the field of rare diseases. The total number of participants is 100 to 150 of which 50 will be invited, the other ones having to register. The registration is free of charge for scientists and patients.

The workshop will be advertised by a mailing to the previous participants to EPPOSI workshops, by putting the information of the EPPOSI website, the Orphanet website and the EURORDIS website, and by putting the information in OrphaNews and in EURORDIS newsletter.

The EPPOSI workshops are problem-solving oriented workshops to identify bottlenecks and solutions at all stages of research and therapy development. They are also partnering workshop between patients, industry, researchers and venture capital.

During the first year we established the organising committee of the workshop and held two organising committee meetings. We identified London as the place. Information about the workshop can be obtained from www.orphanplatform.org

Results achieved so far

1- During the first year, over **400** research projects and clinical trials have been entered into the Orphanet database. 1600 additional research projects need still to be collected and entered into the database before the end of the contract.

2- The service to allow patients to register as volunteers has registered **457** patients so far, who are suffering from **230** distinct diseases. These patients are from **11** (Austria, Belgium, Denmark, France, Germany, Italy, Luxembourg, Spain, Switzerland, United Kingdom, and Sweden). Even if patients were able to register from distinct countries in Europe, it appears obvious that English language may act as a strong barrier to registration. Optimisation of the service would thus probably be reached with multi-language options. Service into French is already in place. Translation into German, Spanish, Italian and Portuguese has been performed but is not yet accessible as we have to identify who can manage the service into these languages as all the process cannot be automated.

3- OrphanXchange, the database of research projects seeking partnership with Industry has received 1580 requests. 70% of the requests are performed by registered users of which 50% comes from the private pharmaceutical sector, the major targeted public for the database.

The number of registered users is **130** (50% Pharma-Biotech-Venture Capital-Consulting, 36% Public research/Healthcare, 7% patient support groups, and 7% other). The number of registered visiting countries: **21** (Austria, Belgium, Bulgaria, Canada, Denmark, Estonia, Finland, France, Germany, Ireland, Italy, Japan, Malta, Netherlands, Pakistan, Portugal, Sweden, Switzerland, United Kingdom, United States).

The visits by registered users have induced 47 contact requests of which 29 were accepted by the researcher. A survey of the outcome of these requests is planned for the second semester of 2005.

4- The planned workshop is attracting a lot of interest, as judged through the number of industrial sponsorship already collected to support the event. It will be organised in collaboration with the FDA, the NIH, the EMEA and American and European patients organisations, and will be co-sponsored by the British Department of Health as well as by Industry. The programme is available on-line at www.epposi.org

Expected end results

We plan to deliver on time all the proposed services. The next step will be to extend these services to all European countries with the support of an other contract. In addition to the current country partners, we have identified possible new partners in the following additional countries so far: Bulgaria, Cyprus, Czech Republic, Denmark, Estonia, Greece, Hungary, Lebanon, Morocco, Poland, Romania, Slovakia, Slovenia, Sweden, Switzerland, Tunisia. Discussions are in progress in Israel, Turkey and Norway.

Intention for use and impact

All the services developed through this action will be freely accessible to all stake holders.

OrphanPlatform is particularly relevant in four areas:

1. The project addresses the needs of diseases specific research networks

Through bridging the upstream needs for data availability with the downstream issues faced by clinical researchers, the project aims at building synergies with, and acting as a facilitator for other important research and development projects in the fields of rare disorders, genomic and post genomic, gene and cell therapies.

2. The project addresses the needs of EMEA and industry

The project is of course directly relevant to the activities of the Committee for Orphan Medicinal Products and of the Committee for Proprietary Medicinal Products, including its Scientific Review Group to provide protocol assistance and its safety, efficacy and quality Working Parties. The project is also expected to have direct benefits for European industry and particularly to small and medium enterprises (SMEs), which account for 80% of OMP applications submitted to EMEA. The platform of services at the centre of the project will provide these industry partners with cost-effective services and solutions that are not yet available. The platform will also steer innovation capacity for new therapies and will reduce the current competitiveness gap of EU industry versus US.

3. The project will benefit to paediatric drugs and cancer treatments development

This represents another major public health objective for the EU on which the European Commission envisages specific regulatory and research initiatives. 80% of rare diseases appear at an early age and are directly responsible for 25% of the mortality in childhood. Experience gathered in the US and in Europe also shows that around 40 % of Orphan Medicinal Products are new and innovative products for cancer and leukaemia treatment.

4. The project is an archetype of the need for action at European level

Not only the project could not be developed at the level of Member States acting independently, but non-integrated national actions are now adding to obstacles faced by clinical researchers and industry. The specificity of the small number of patients, scarce professional competences and fragmented resources are defining the relevance of this structuring project at the Community level.

Section 1 - Project objectives and major achievements during the reporting period

The project aims at developing information tools to address in a comprehensive and integrated approach the set of factors that currently affects research on rare diseases and its coordination.

The specific objectives are: (1) to develop an information service, freely accessible on Internet, dedicated to research activities in the field of rare diseases and orphan medicinal products, including a database of research projects, funded at MS level and at the EU level, and a database of collections and research networks. (2) to develop services aiming at speeding up the enrolment of patients in clinical research. (3) to develop a database of research projects with development potential, to help scientists and Industry establish the necessary partnerships. (4) to organise a workshop with all stakeholders to discuss known bottlenecks and find solutions.

The project aims at establishing the platform of services in 11 European countries in the pilot phase in order to propose an extension to all European countries in 2006 years time.

Ultimately, the goal is to convert scientific developments in the field of rare diseases into diagnostic tools and therapies as quickly as possible.

Specific objective 1: The Orphanet database extension to research activities

The project was to extend the Orphanet directory of services in order to provide accurate information on on-going research activities at the Member states level and at the EU level, in a format easy to use for researchers, clinicians, Industry, patients, administrative bodies and any decision-maker.

This specific objective was composed of two subprojects: (1) the extension of the Orphanet database to accommodate this new type of information and (2) the collection of data in participating countries.

Extension of the database

Orphanet is a relational database, which is implemented on a Unix server. This database is at the SQL standard and is run via SYBASE ASE 12.0. The query interface is written in html for the static pages and in PHP for the dynamic pages. The updating interface is written in Java. Access is restricted to the local area network. All accesses to the database are authenticated. Only authorised persons can access the database. Authorisation is delivered to the information scientists in charge of the data entry.

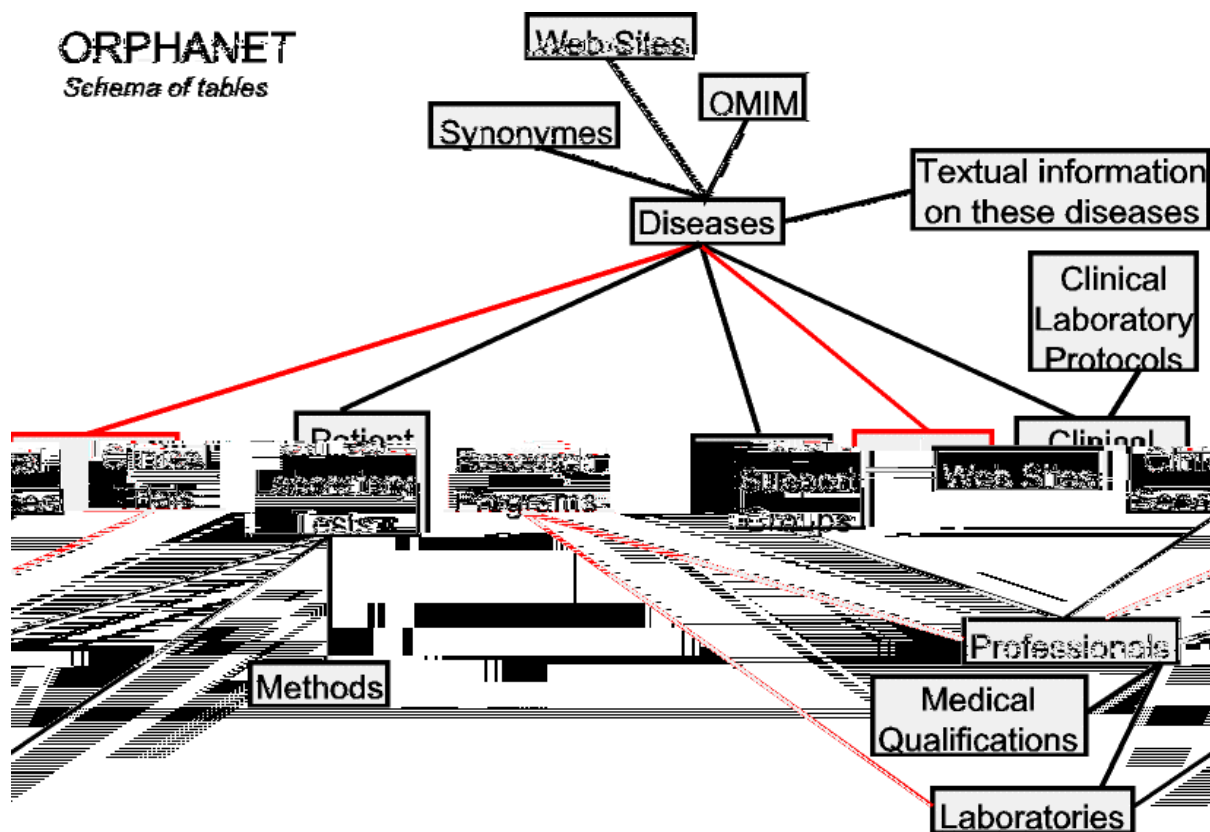
The developments have included the creation of the following new tables:

- a table of research projects defined as any project from basic science up to social science, which is directly relevant to improve the knowledge about 1 or more rare diseases or to contribute to better prevention, diagnostic, or treatment of these rare diseases (for instance the development of a diagnostic kit or an orphan drug). The database structure includes the title of the research project, the principal investigator details and the protocol as an optional item, necessary if there is a call for participation to research (see recruitment of volunteers). Each project is linked to 1 or more rare diseases.

- a table of registries/ database of patients/cohorts defined as any collection of clinical data systematically organised to be suitable for further analysis with the aim to improve our knowledge of the clinical course of rare diseases. The table contains the title of the data collection, the principle investigator details and the protocol as an optional item, necessary if there is a call for participation to research (see recruitment of volunteers). Each project is linked to 1 or more rare diseases. So far we have collected data on registries/cohorts/data collections.
- a table of networks of professionals organised around a rare disease or a group of diseases defined as any group of professionals sharing a common goal in research and sharing data and protocols. The table contains the title of the network, the principle investigator details and the protocol as an optional item, necessary if there is a call for participation to research (see enrolment of volunteers). Each network is linked to the relevant diseases.

General design of the database

The general design of the relational database is given below. The existing tables are in black. The new tables are in red.



All the developments were performed during the first months. The database was ready for data injection by September 04.

Collection of research projects at the MS level:

The lists of research funding agencies and the sources to identify research projects in the Orphanet partner countries have been established for all countries (see Appendix 1) except 3: Belgium, Finland and The Netherlands. In Belgium and Finland our partners have not yet started collecting data on research activities. In the Netherlands the partner was not able to work as he had to move to UK at the beginning of this contract. The new partner will do the job in the remaining time hopefully. This change will not impact on the end result as Pr Martina Cornel, the new partner, has the capacity to achieve the supervision of the data collection in her country.

The OrphanPlatform participants have started to establish effective partnerships with the main funding agencies as well as the main research foundations to collect research projects. This step is still in progress in some countries. Once the projects are identified, the researchers are contacted.

Collection of data in all participating countries is mainly performed through a common questionnaire that has been translated into all National languages. The questionnaires are given in Appendix 2.

Clinical trials are collected through a specific questionnaire giving information on the title of the clinical research, the diseases covered by the study, the sponsor and investigator contacts, the inclusion and exclusion criteria, the time period covered by the study, the phase of the trial and the drug used in the trial if applicable.

If specific data has already been collected through collaborations established with institutions and funding agencies, the questionnaires are filled in before being sent to principal investigators. We experience a much higher level of returned questionnaires, in these cases.

The data are entered into the Orphanet database both in the national language and in English for the six languages available on the website: English, French, German, Italian, Portuguese, and Spanish. The data from Finland and the Netherlands are entered only in English.

Collection of research projects at the EU level:

Collection of projects published on the DG SanCo and the DG Research websites was performed by the coordinating team and specific information sent to corresponding country partners.

Projects funded by DG SanCo in the field of rare diseases:

- 10 projects identified
- 3 projects listed in the Orphanet Database
- 6 projects pending agreement from EU project leaders
- 1 project already ended.

Projects funded by DG Research in the field of rare diseases:

- 13 projects identified in FP5
- 6 projects listed in the Orphanet Database
- 3 projects pending agreement from EU project leaders
- 3 projects already ended.

- 18 projects identified in FP6
- 3 projects listed in the Orphanet Database
- 15 projects pending agreement from EU project leaders

It appears necessary to confirm all data collected on the European Commission website before entering research projects into the Orphanet database because a large majority of them are not updated. It is also necessary to validate with project leaders all the rare diseases covered by specific research projects.

Volume of data already collected

The goal of the OrphanPlatform contract is to reference 2000 research projects at the end of the contract, ie after 2 years.

During the first year, over 400 research projects and clinical trials have been entered into the Orphanet database. 1600 additional research projects need still to be collected and entered into the database before the end of the contract.

The data collection started only less than 6 month ago. Funding was sent to partners at the end of June. Probably due to the summer break, most local administration did not attribute the funding to our partners before September. Hiring an information scientist could not start without knowing when the money would be available. This explains that some of them were hired in October or November.

In the Netherlands, the data collection has not started yet as our partner moved to UK at the beginning of the contract and informed us only recently that he wanted to be replaced. We have identified a new partner but the official change of partner is not yet in place as the change requires an amendment to the contract. In UK the local hosting institution was particularly slow in establishing the frame to use the funding. The data collection only started at the beginning of 2005. In Belgium our partner could not hire a proper collaborator and will, hopefully, do all the work during the second year.

The distribution of research projects between participating countries is given in Appendix 3. In March 2005, the total number of research projects is 1988 and the total number of clinical studies is 131.

Country name	Number research projects	Number Clinical trials
Austria	10	0
Belgium	4	0
Finland	18	0
France	1429	99
Germany	90	7
Ireland	5	0
Italy	257	9
Netherlands	0	0
Portugal	21	0
Spain	153	16
United Kingdom	1	0

Several other projects have been collected but are still being validated before being entered into the database.

The main problem identified by the participants is to get complete information from researchers, despite numerous requests using different methods, including phone calls.

However we are confident that the data collection will be effective during the second year of the contract as contacts with main funding agencies for medical research have been developed and all information scientists in participating countries have been trained. The information scientist of the Belgium team has also been trained and is now supervised by the French team.

For the collection of data about trials, we will benefit from the new regulation stating that trials must be registered in a public registry at or before the onset of patient enrolment. Although that does not guarantee that the data will be publicly available, it ensures the fact that National regulatory agencies will be aware of the on-going trials on their territory, which was not the case up to now in most countries. We are contacting all industrial companies developing orphan drugs in order to encourage them to register clinical trials in Orphanet and we are encouraging our partners to establish a partnership with their regulatory agency.

In addition, main scientific journals have recently stated that they will require, as a condition for publication from 1 July 05, registration in a public trials registry. Orphanet, being the registry for rare diseases in Europe, will develop a strategy to benefit from this proposition.

Specific objective 2: The Eclor project: Enrolment of volunteers for research

To meet the needs of researchers and the wishes of the patients, we planned to develop a new facility to allow patients to find out whether there is a clinical research project on their disease, which they could participate in. A pilot project had been funded by the French Ministry of Research and had received an approval (n° 820077) from the C.N.I.L (National Committee on Informatics and Liberty) in France. That sub-project is a complement to the extension of the Orphanet database to clinical trials. This subproject is supposed to accelerate the enrolment of patients in clinical research projects.

The technical solutions, which have been selected to secure the nominative data, are the following:

- the identifier of a person is a combination of its first and last name and of a password. The password is freely selected by the person and needs to be complex enough, with at least 6 characters including 2 letters and 1 number.
- the flux of data between the web server and the service user are encrypted. The 128 bits encrypting process is secured by a SSL protocol with the Apache server. The applicative server is behind a firewall, which is filtering all the ports of the UNIX server. The access to the database is done through the applicative server or through a strong authentication. To access the server, it is necessary to use a SSH tunnel (protocol SSH2) using a public and a private key which are protected by a pass phrase.

The project was to extend the service which is currently available on the French Orphanet website to all participating countries after legal permission is obtained country by country, and translation made in the 6 languages of the project.

Description of the Eclor service

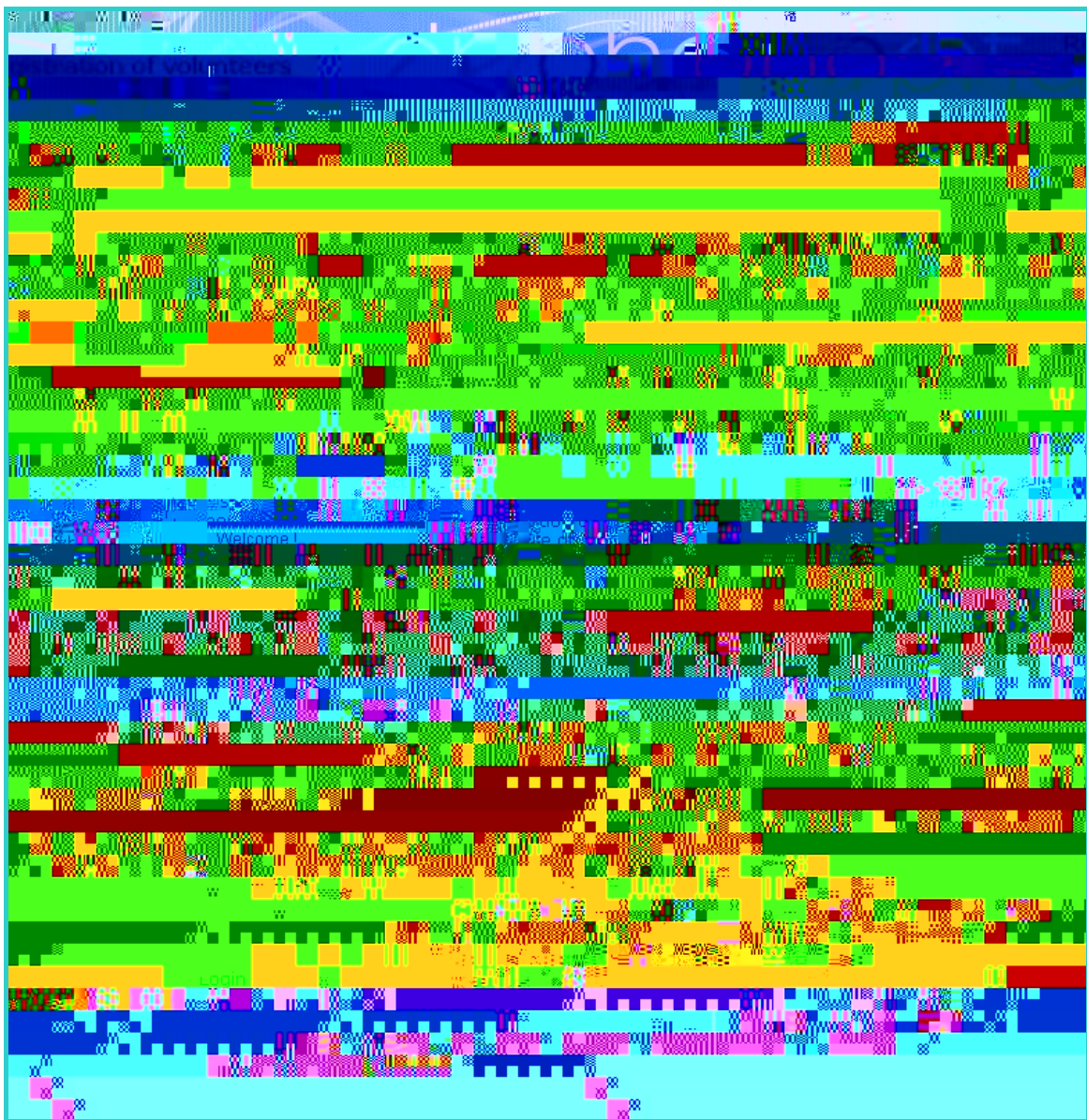
This facility has been developed according to the plans.

Only patients can register, or parents and legal representatives of a person suffering from a rare disease; family and doctors can pass on information to patients (an information letter is available and may be downloaded from the website).

It is be an **opt-in** service

- Patients can access the list of clinical trials regarding their disease. If there is no on-going trial, the patient can decide to leave his contact details in order to be informed about any new project.

- If there is an on-going trial, the patient can access the protocol to check whether he seems eligible. He is advised to discuss with his physician about the possibility to apply for enrolment.
- The patients who leave their contact details can access, modify or suppress their data at any moment.
- They are systematically asked to re-confirm their willingness to remain in the database, once a year.
- The list of potential volunteers is not accessible on line and will not be provided to researchers or companies. It is only used by the Coordinating team to inform volunteers when there is a new project in the database which is relevant for them. It is up to them to contact the principal investigator to be included in the trial. The sponsors can be informed, on request, of the number of registered volunteers.



The search for a disease is as in Orphanet. A list of diseases corresponding to the search is proposed. The patient may choose the disease he/she is interested in. If the patient does not find in the proposed list the corresponding disease, he/she can describe the disease (symptoms/clinical signs) and register.



To register, the mandatory fields are: email address, Name, First name, City, Country and date of birth. The postal address is optional.

orphanet Registration of volunteers

[Orphanet home](#)

Participating in clinical research [Print](#)

Clinical studies

Welcome !
Patient
Parents
Doctor
Family or friend
Contact us

Orphanet
Welcome !

If you wish to be informed via e-mail of the launch of a new clinical study that may concern the disease you are interested in, please fill in the form.
You can, at anytime, access, delete or amend your data.
We will contact you on a yearly basis in order for you to update or delete your data.
In keeping with Article 27 of the French law 'Informatique et libertés' n° 78-17, the data you submitted are and will remain confidential, and will not be disclosed to any third parties.

The following fields are mandatory :

Name of the disease you did not find

Symptoms/clinical signs of the disease

REGISTRATION FORM FOR THE INFORMATION NETWORK OF CLINICAL STUDIES RECRUITING VOLUNTEERS

The following fields are mandatory :

I accept the terms and conditions defined above.
Thank you for ticking this box if you agree.

*Login

*Password

*Type new password again

*Letters and digits only
from 6 to 8 letters or digits only.

Your email address

Civility

Name

Firstname

City

Country

Date of birth

The following fields are optional :

Post Code

Address

Registration allows users:

- to access, modify or suppress their data at any moment
- to register a child for the same disease
- to access the list of clinical trials regarding their disease
- to access the protocol of the clinical research. User is advised to discuss with his/her physician about the possibility to apply for enrolment.

A request for confirmation of willingness to remain in the database will be systematically sent by email once a year. One reminder will be sent at 365 + 30 days before deletion of the account. This update function is automatically performed by the Eclor application.

Achievements regarding the Eclor service

The service was launched in French and English on 6 January 2005.

Translation of the service into German, Spanish, Portuguese and Italian has been done as required by the OrphanPlatform contract for the end of the first year: however, the service is not yet on-line in these languages as the service requires more administration than previously thought. The reason is that many patients register without knowing exactly what is the name of their disease. This obliges us to start discussing through e-mails to find out what could be the right diagnosis. This activity is unexpectedly time-consuming. We are currently discussing the possibility that OrphanPlatform partners could manage this service in their national language but up to now the straightforward solution is to open the service in the 6 languages but to manage the communication with patients in only 2 of them: French and English, the only languages spoken by the Paris team.

Communication about Eclor

All OrphanPlatform participants were informed by email of the new version of the Eclor service. They were asked to transmit the information to colleagues.

In addition, the launch of the Eclor service was advertised in OrphaNews France, the Orphanet newsletter in French.

Since March 2005, new clinical studies entered in the Orphanet Database are listed in OrphaNews with a link to the Eclor service. This information has resulted in a great increase in the number of patients registering to the service.

In June the service will be advertised in OrphaNews Europe as well, the newsletter of the rare diseases task force, which will increase a lot the visibility of the service.

Administration of the Eclor service

The list of patients who register as volunteers is not accessible on line except by a few nominative members of the team.

The persons in charge of this service have:

- to follow up registrations to check the correct spelling of the email address entered into the database. It is necessary to contact by regular mail if problems are detected.
- to follow up the diseases entered into the database to adapt entries if necessary. This action is performed in agreement with users. Many contacts may be necessary.
- to answer additional questions sent through Internet about the disease, about the state of art of research in the field, about the likelihood that a trial will start soon....These unsolicited questions are unavoidable when a communication interface is in place.

The time allocated to these actions was underestimated in the OrphanPlatform contract. Indeed, some patients do not select specific rare diseases but prefer instead to describe their condition. This point has to be discussed with the EU partners and solutions have to be identified:

- We can either share the management of registration of the Eclor service with the EU partners (this could facilitate the follow up of messages written in different languages)

- or we keep only French and English as languages for communication with the administrator (in that case the administration can be performed by the coordination team using the currently available tool) but the web pages are in the 6 languages.

The Eclor service is greatly appreciated by patients who register every day since opening. It is also of great interest for physicians who contact Orphanet when they are facing difficulties to

Specific objective 3: the OrphanXchange project: from development to the marketing of diagnostic tools and therapeutic products in the field of rare diseases

The project was to identify, among all the research projects listed in the database, the research projects, which reach a stage at which they can be carried out by an industrial company or form the basis for a start-up company, and be developed into a commercial product or technique. These projects were made accessible and visible on the OrphanXchange website, a separate website from Orphanet but with functional links. The two projects would share part of the Orphanet database but the front ends would be different. The necessity to have a separate website came from the fact that the OrphanXchange website will be password protected.

This subproject had two components: (1) the establishment of a website and of a database of research projects seeking partnership with Industry, (2) the collection of research projects to fill in the database

Description of www.orphanxchange.org

The website and the database were developed according to plans. The website opened officially on **June 2004**.

Access to general information and search for projects in the OrphanXchange website are free of charge and possible without registration.

The query for projects is made by selection of the type of health product and the type of rare disease.

Types of health product are:

Therapeutic molecule	Diagnostic tool
Vaccine	Nutritional supplement
Gene therapy	Cell Therapy
Medical device	Other




Types of rare diseases are:

- Name of rare disease as in Orphanet
- MeSH disease category

The number of results and the titles of the identified projects are listed.

The user may register to orphanXchange without charge in order:

- to save search results into a personalised area
- to access full description of the projects by an automated request to the researcher
- to manage potential partnerships
- to be informed of new data releases in the database

	Services
OXC Home Page Search database Submit a project	
	
General Information About OrphanXchange About Rare Diseases About Orphan Drugs OrphaNews	
	
Contact us	

Register as a user

Registration is required:

- ◆ to access full description of projects matching your query through an automated request to the researcher
- ◆ to be informed of new data releases in the OrphanXchange database

Account Type User

Login *

Password must contain at least 4 characters and 2 digits:

Password *

Password again *

Civility *

Title

Firstname *

Lastname *

Email *

Affiliation

Company / Institute profile

Company / Institute status

Company / Institute *

Address *

Post Code 1

City *

Post Code 2

Country *

The registration allows the administrator:

- to identify users in order to better serve them
- to follow up the potential partnerships and thus to measure the impact of action

The OrphanXchange service has been approved by the C.N.I.L. (Commission Nationale Informatique et Liberté) n° 898336 BO 2004-51.

Selection of projects for OrphanXchange

The first source of information is a selection of projects of potential industrial interest listed in Orphanet. To identify new projects among those collected by Orphanet teams, the willingness of researchers to develop partnerships with industrial companies and the stage of development of their projects are identified in the questionnaire sent to investigators.

All investigators responsible for projects matching OrphanXchange criteria are contacted.

A first contact is made by email with general information on the OrphanXchange project. A reminder is sent few months later.

We do not insist if investigators are not interested; indeed, the service we propose must meet their needs.

We had conducted a survey in 2003 to identify molecules already marketed for an other indication and that could be of interest for orphan indications. A questionnaire was sent by email to 1541 health care professionals referenced in Orphanet France. 226 orphan indications including 107 molecules were identified. Researchers/physicians were contacted in order to investigate their willingness to display information on the website. In addition, agreement of industrial companies was necessary before online publication of information related to drugs (Appendix 4).

From this survey, 65 molecules were referenced in OrphanXchange. They constitute potential orphan designations.

On 24 March 2005, a second survey was conducted and an online questionnaire was made available for health care professionals and patient organisations referenced in Orphanet. The questionnaire was available in 3 languages French, English and Spanish and was sent to 2115 professionals. The preliminary results revealed more than 100 potential orphan indications. Once precise identification of molecules has been done, we will follow the same strategy as before to include molecules into the OrphanXchange database.

Departments of technology transfer from research organisations and universities constitute also a source to identify products with potential interest for treatment and diagnosis in the field of rare diseases.

Partnerships have already been developed with French departments: **Inserm, CNRS, CEA, Institut Pasteur, Institut Curie** and **universities Paris V, VI and VII**. Development of partnerships with similar departments in Europe is in progress.

Projects identified through INSERM, CEA and Institut Pasteur are already online.

Communication about the OrphanXchange project

The website <http://www.orphanxchange.org> was launched on June 15th, 2004. A press conference was held on the same day at the Plateforme Maladies Rares in the presence of Pierre Le Sourd, president of the French pharmaceutical companies association, Christian Brechot, Director of INSERM, Alain Fischer, Director of GIS Institut des Maladies Rares, Jacques Bernard, representative of the French rare diseases alliance, and Ségolène Aymé, director of Orphanet. The press release (see Appendix 5) has been translated into English by Orphanet and sent to EFPIA(EBE), EPPOSI, EuropaBio, Eurordis, European Commission, EMEA, FDA and to 20 Orphanet partners in Europe. The press release was then translated into 15 national languages and sent to the national media and national competent authorities by our partners.

In France,

- 3 Press Agency news releases - June 15th 2004 (AFP, APM et AEF)
- 1 News Brief in national daily newspaper (Le Parisien - June 16th 2004)
- 5 articles in national medical/specialised journals (Le Quotidien du Médecin - June 17th 2004, Le généraliste and Impact Médecine - June 18th 2004, Le Concours Médical - June 30 2004, Scrip August 13th 2004)
- Newsletter Pharmaclient (June 2004)
- Bulletin International d'Information (June-July 2004)
- Yahoo News (June 15 2004)
- La Croix (June 16th 2004)
- Nouvel Obs.com (June 16th 2004)
- Samaritains Handicap France website (June 16th 2004)
- L'usine nouvelle Biotech Info (July 2004)
- ivs-info.com, Biotechs diffusion letter (July 7 2004)
- Destination santé.com (July 8 2004)

We have no data on the impact in the media of other countries.

New entries in the OrphanXchange database

The registered users were all informed by email of new entries into the database on July 16th 2004, on October 25th 2004 and on March 25th 2005.

The total number of visits and the total number of pages seen the week before and the week after the announcement measured the impact of these calls:

	Visits	Pages
First call	+ 40 %	+ 63 %
Second call	+ 45 %	+ 117 %
Third call	+ 6 %	+ 71 %

The two first calls clearly indicate an increase in the numbers of total visits and an increase in the number of pages seen by the users.

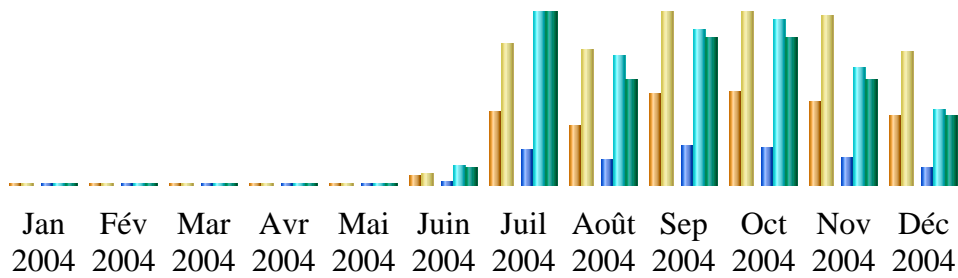
As indicated in the table, the last call was not as efficient as the two first calls for the number of visits. However, the increase of accessed pages clearly demonstrated high interest for searching projects in OrphanXchange.

Statistics as of 15 May 2005:

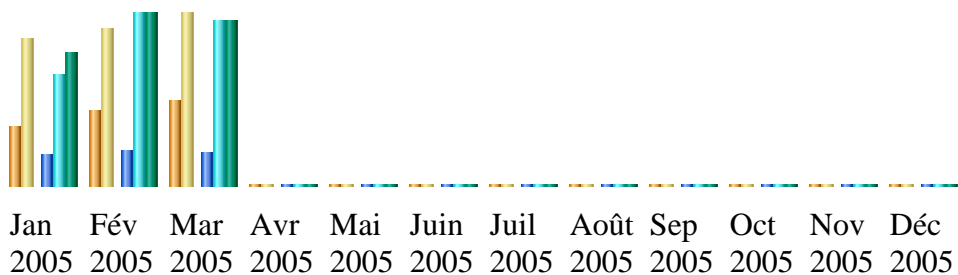
Number of projects in the database: **125**

Number of registered users: **130** (50% Pharma-Biotech-Venture Capital-Consulting, 36% Public research/Healthcare, 7% patient support groups, and 7% other).

Number of registered visiting countries: **21** (Austria, Belgium, Bulgaria, Canada, Denmark, Estonia, Finland, France, Germany, Ireland, Italy, Japan, Malta, Netherlands, Pakistan, Portugal, Sweden, Switzerland, United Kingdom, United States).



Mois	Visiteurs différents	Visites	Pages	Hits	Bande passante
Juin 2004	35	41	169	1125	9.03 Mo
Juil 2004	262	509	2161	10866	90.40 Mo
Août 2004	216	490	1514	7843	52.39 Mo
Sep 2004	330	622	2378	9302	73.57 Mo
Oct 2004	333	649	2214	9901	73.47 Mo
Nov 2004	302	612	1646	7069	52.43 Mo
Déc 2004	248	480	1035	4573	34.77 Mo
Total	1726	3403	11117	50679	386.06 Mo



Mois	Visiteurs différents	Visites	Pages	Hits	Bande passante
Jan 2005	198	494	1260	4577	37.47 Mo
Feb 2005	251	526	1457	7418	50.99 Mo
Mar 2005	282	604	1383	6793	46.52 Mo

These data reflect the global activity of the website but not the number of requests performed either by registered users or unregistered users: Since the launch of the OrphanXchange website, registered users have performed over **1100 requests**. The total number of requests is over **1580** if anonymous requests are included.

Overall, the data show that 70% of the requests are performed by registered users of which 50% comes from the private pharmaceutical sector, the major targeted public for the database.

Impact of the OrphanXchange project on potential partnership development

Researchers must reply to the request from the registered user wishing to access the detailed description of the project of interest for him/her. They are simply informed by email with a functional link to easily respond to the request.

Number of total contact requests: 47

Number of positive answers to contact requests: 29

Number of pending answers to contact requests: 18

Probably because of the great number of emails posted to researchers, their answers could take some time. The administrator of the website has then to email again or even to call researchers to make sure the message arrived correctly.

The administrator usually proposes to reply to the request on behalf of researchers and to keep them informed.

In order to refine and improve the content of the OrphanXchange database to make it more relevant to users, we plan to conduct a survey in June 2005 as we will reach the one-year milestone for the database.

We will propose to enlarge possibilities of partnership development through OrphanXchange. Indeed, after one year, it appears obvious that OrphanXchange could be the place for all kinds of collaboration/partnerships.

The other types of partnership are:

Academia-Academia: search for an academic partner to set up a collaboration at national or international level.

Industry-Industry: SME searching for a partner to develop a project.

This development will require the modification of the information collected on research projects as it will be necessary to ask the researchers whether they are interested by a collaboration.

Specific objective 4: Partnering workshop

There is a strong view that additional action needs to be taken to energise and expedite the development of therapies and suitable diagnostics for rare diseases.

A workshop is supposed to be organised to exchange views and explore options for facilitating the development of therapies and identify research projects in advanced stages. It will follow the scheme of previous partnering workshops organised by EPPOSI: Brussels (2000); Paris (2001), Rome (2002), The Hague (2003), Berlin (2004), but it will focus on Orphanplatform main concepts: coordination of research activities and research funding in Europe; improvement of partnering activities; improvement of clinical trials in the field of rare diseases.

The total number of participants is 100 to 150 of which 50 will be invited, the other ones having to register. The registration is free of charge for scientists and patients.

The workshop will be advertised by a mailing to the previous participants to EPPOSI workshops, by putting the information of the EPPOSI website, the Orphanet website and the EURORDIS website, and by putting the information in OrphaNews Europe and in EURORDIS newsletter which are sent to 5,000 stakeholders in Europe.

The EPPOSI workshops are problem-solving oriented workshops to identify bottlenecks and solutions at all stages of research and therapy development. They are also partnering workshop between patients, industry, researchers and venture capital.

During the first year we established the organising committee of the workshop and held two organising committee meetings. We identified London as the place.

The pre-conference paper is as follows:

SIXTH EPPOSI CONFERENCE ON RARE DISEASE THERAPY DEVELOPMENT AND PARTNERING

“People with Rare Diseases – No Longer Alone in the World”

Topic:

Encouraging research, facilitating development, and securing take up of innovation and treatments for rare diseases through a supportive regulatory framework and functional partnering to ensure equal access to treatments for all.

London, 25-27 October 2005

Pre-conference paper

About EPPOSI

The European Platform for Patients’ Organisations, Science and Industry (EPPOSI) is an EU patient-led partnership between patient organisations, industry and academic science and clinicians, founded in 1994 to discuss and influence policies in human healthcare in Europe based on joint views by its stakeholders.

EPPOSI is officially registered as a not-for-profit International Society under Belgian law. It is managed by a Board of Directors, consisting of EPPOSI members representing patient organisations, academia and industry. The Chair is always a patient representative.

EPPOSI’s mission is achieved through a range of activities including meetings, debates and workshops between stakeholder representatives. For the past five years, EPPOSI has organised an annual therapy-development partnering workshop: in Brussels in 2000, Paris in 2001, Rome in 2002, The Hague in 2003 and Berlin in 2004. This year EPPOSI is organising an international conference to be held in London under the UK EU presidency, with the support of the UK Department of Health and Department of Trade & Industry.

The aim of EPPOSI events is to provide a forum for discussion between political decision makers, healthcare providers, patient representatives and industry from several regions of the world, on the research and development of innovative health products and health services and their impact on the quality of life of patients. They seek to open up debate in order to evaluate how, and if, specific conclusions and recommendations can be reached, whether a basis for consensus is achievable and, if so, what this consensus will be.

The London conference in context

The EU orphan medicinal products regulation entered into force in April 2000 with unanimous approval from the European Parliament. As of April 2005, 254 orphan designations have been granted by the European Commission and 20 orphan medicinal products have been given marketing authorisation in the EU. Stakeholders agree that this positive start is a source of hope for patients and their families, but much remains to be done to ensure that more products reach

those that need them. The “orphan drug” regulation is currently being re-assessed after its first 5 years of existence. This is an ideal time to compare the situation in Europe with that in other regions of the world, such as the USA, where similar regulation has been in place for a longer period of time. It is why this conference is organised in partnership with NIH and FDA.

It is also time to look at the R&D process as a whole, to identify bottlenecks and possible solutions and to benchmark the initiatives that have shown to be efficient in providing an appropriate framework for the development and provision of innovative therapies for rare diseases. With the European Commission’s next Research Framework and Public Health programmes both under discussion this year, this conference will provide an excellent opportunity to discuss the impact of EC support for basic research and the development of information systems in the field of rare diseases. The USA has also supported several important initiatives to boost R&D in the field of rare diseases and the results can usefully be compared with the EU approach.

Patients’ organisations are holding a major European conference in Luxembourg in June 2005 to present their case. This event is organised by Eurordis. Their views will be an important basis for discussion at the London EPPOSI conference.

Topics to be debated during the conference will include priority setting in research, facilitating development and the process of technology transfer, comparing diagnostic approaches in Europe and the USA, facilitating clinical trials and encouraging transatlantic collaboration in the field, harmonisation of reference frames, facilitating access to innovative therapies, and care provision for rare diseases in the context of political and economic globalisation.

These same topics are currently the subject of active debate amongst stakeholders, sometimes with very conflicting views being expressed. During the conference, participants will be asked to actively contribute to an action plan of key recommendations addressed to specific stakeholders, which will be debated in the final session. By providing such a forum for open and interactive debate, the 2005 EPPOSI conference will give all stakeholders the opportunity to impact on the future of patients with rare diseases through international cooperation.

Section 2- Workpackages progress

WP 1 Management

WP1 is targeted at organising the scientific and financial management of the project by scheduling events and milestones, by creating information tools for the partners and by launching a website where all information will be available for the partners to achieve their tasks. All the management activities are performed by participant 1 INSERM.

Every thing has been done according to schedule, except for the consortium agreement, which has not been drafted by lack of time. This does not impact on the project.

A kick-off meeting has been organised on 5 April 2004. The standing committees met during the meeting.

The Orphanplatform website has been opened at month 3 (www.orphanplatform.org) (deliverable 1), requiring 1 person-month.

All the documents used for the kick-off meeting are available on the contract website in the private area. We also post on the website the statistics. The activity of each partner is monitored monthly and reported on the website. This activity required 2 person-month.

A press conference has been organised when the orphanXchange website has been launched (deliverable 14)

The main difficulties we faced were the delay of the INSERM administration to transfer the funding to all partners and the slowness of the administration of the partners to provide the partners with their budget. We roughly lost 5-6 months, which is a lot for a 2-year project.

In addition two partners could not deliver anything during the first year: The Dutch partner resigned from his position to go to UK at the beginning of the contract. It took one year to identify an other Dutch team accepting to serve. The Belgian partner has not taken his job seriously and has not hired someone capable to deliver what was expected. This will be corrected by having someone full time during the second year to perform all the tasks of the two years.

WP1 List of deliverables

Deliverable 1: OrphanPlatform website done on month 2 according to schedule

Deliverable 12: authorisation of competent authority for Eclor: obtained in due time

Deliverable 13: Final plan for using and disseminating the knowledge. This is due by the end of the contract.

Deliverable 14: Report on raising public awareness. This is due by the end of the contract, but we have already communicated actively on the project (press conference on June 04; see enclosed press release).

WP2 Database of research activities

WP2 is targeted at creating a database of research activities in the Austria, Belgium, Finland, France, Germany, Ireland, Italy, Netherlands, Portugal, Spain, Switzerland, United-Kingdom, including all research projects, which are funded through a peer-review process at the national level or at the EU level. We were supposed to design the structure of the relational database to be build up and the type of screens to be developed, both to query the database and to update it; to validate the concepts with the scientific advisory committee of Orphanet and with the sub-project participants and to do the necessary development f36611patinh

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WP4 OrphanXchange database of projects at a near to the market stage

WP4 was targeted at developing OrphanXchange, a relational database and a web site of research projects at a near to the market stage of development.

This has been achieved according to schedule by participant 1 INSERM. It took 6 person-month. The OrphanXchange website has been launched on 15 June 2004 (deliverable 4).

We have not faced any problem.

WP4 List of deliverables

Deliverable 4: OrphanXchange a website and a database of near to the market research projects. It was delivered at month 2.

WP5 Collection of data on research activities

WP5 was targeted at collecting data on research activities in Austria, Belgium, Finland, France, Germany, Ireland, Italy, Netherlands, Portugal, Spain, Switzerland, United-Kingdom, to fill in the database created by WP2.

The partnership with funding agencies has been established in most countries except 2 (Belgium and Netherlands). The questionnaires to collect the data have been translated and adapted to the country specificities by all partners.

The data collection was supposed to start at month 2 and to continue during all the project. In fact it started much later (average 6 months) for administrative reasons, but it is now going on smoothly in most countries. As already said, 3 countries are late but will catch up hopefully.

Coherence and consistency is checked at the central level, once a week.

The data collection requested the following person-month:

Partner 3 UCC: 6 person-month as scheduled

Partner 5 CSS: 6 person-month as scheduled

Partner 7 UNIMAN: 1 person-month when 6 were scheduled. It is planned that the team will catch-up during the second year

Partner 9 KU Leuven: 0 person-month when 6 were scheduled. It is planned that the team will catch-up during the second year

Partner 15 U Turku: 6 person-month as scheduled

Partner 23 MHH: 0 person-month as scheduled. In fact someone was employed to do the job and did it but we discovered when preparing the financial report that this person was not eligible to be paid through a EU grant (internal rules of the German university). The financial report is at 0 for expenses when in fact the work has been done. It is planned that the team will spent the 12 person-month during the second year

Partner 29 MUW: 6 person-month as scheduled

WP5 List of deliverables

Deliverable 5: collection of research projects in participating countries. This is in progress with a 6 month delay. It is expected that the partners will catch up during the second year. One fourth of the projects have already been collected.

WP6 Collection of data for OrphanXchange

WP6 was targeted at collecting data to fill in the database created for OrphanXchange, the relational database of research projects at a near to the market stage of development (deliverable 6).

The data collection was done by participant 1 INSERM. It is progressing well. The number of research projects in the database was expected to be 150 at the end of the contract. We have already collected 125 projects at the end of the first year. This data collection was done thanks to resources outside this contract.

WP6 List of deliverables

Deliverable 6: collection of projects to be put in OrphanXchange. This has been done according to schedule and the number of projects already collected is higher than expected.

WP7 Partnering workshop

WP7 was targeted at organising a partnering workshop based on the OrphanPlatform experience to further define the needs of the stakeholders and the possible solutions. The targeted participants are the scientists currently involved in clinical research, the emerging biotech companies, and the key-players from patients support groups.

The workshop is already well designed; ahead of schedule. The organisation of this workshop is supposed to take place during the second year. The work already done was done outside of this contract.

WP7 List of deliverables

Deliverable 7: workshop on rare disease research specificities: from bottlenecks to solutions. This workshop is now planned for October 05 (month 18 instead of 20). It will be organised in London. The organisation is ahead of schedule.

Deliverable 8: final report: this will be issued after the workshop. Arrangements are in place to produce it.

Section 3 – Consortium management

The consortium is managed by three bodies:

- 1- The *scientific advisory committee* is composed of key actors in the field of rare disease research and orphan drug development. It includes 1 observer from the EMEA (Jordi Llinares), 3 COMP members representing 3 countries involved in the project (Spain: Josep Torrent-Farnell; France: Dr Emmanuel Heron; Belgium: Dr André Lhoir), and 4 representatives of National governmental organisations in charge of coordinating research funding in the field of rare diseases (Spain: Instituto de Salud Carlos III (Manuel Posada); Netherlands: Dutch steering Committee on Orphan Medicinal Products (Sonja van Weely); France: Institut des maladies Rares (Pascale Borensztein); Germany: PT-DLR which is a non-governmental, non-profit organisation specialised in research management and funding (Ralph Schuster); 1 patient representative: Yann Le Cam (CEO Eurordis) who is co-chairing this committee with Josep Torrent-Farnell, Chairperson of the Committee for Orphan Medicinal Products.

Its mission is to participate in the definition of the goals and tasks of the projects; to assess the quality of the products and the relevance of the outcomes for the Rare Disease Community; to identify all legal, social and ethical issues related to the project; to advise the executive board on any relevant matter. It met at the kick-off meeting and commented actively on the plans. Its main recommendation was to identify the Companies having obtained an orphan designation as the main source of information about clinical trials. This recommendation is at the origin of a new planned development of the database for 2006 in the field of orphan drugs. Up to now we included in Orphanet all orphan drugs with a market authorisation. The plan is to register also all orphan designations in order to follow up the development from the designation phase up to the availability on national markets. The clinical trial phase will be one stage of this development.

- 2- The *executive board* is composed of the leaders of the sub-projects and of one representative of each participating organisation not yet represented in the scientific advisory committee: Eurordis (Fabrizia Bignami), Europabio (Patrick Squiban) and Epposi (Cees Smit). Their task is to supervise the implementation of the subprojects and to take decisions when ever necessary. The executive board met at the kick-off meeting in conjunction with the scientific advisory committee.

In fact they met jointly with the scientific advisory committee as we were at the beginning of the project with no executive questions to be debated.

- 3- Each sub-project is under the responsibility of a *sub-project leader*. The participants of the sub-projects 1 and 2 are the national partners in charge of collecting the information in their country. The participants of sub-project 3 are representatives of the SMEs and of patients' organisations. The participants of sub-project 4 are the EPPOSI board members. The sub-project leaders have met at the kick-off meeting.

The plans, ways to achieve the goals, potential bottlenecks and solutions were extensively discussed.

The contractors contributed up to now as planned except for 3 of them:

- The UK partner (Dian Donnai) could not start working until almost January 05 due to the administrative delay at the UK level. The team is now well in place and working hard to catch up.
- The Dutch partner (Raoul Hennekam) resigned from his position in Amsterdam to move to London. He took sometime to decide whether he was keeping his responsibility or not for orphanplatform. He finally decided to quit. We have now identified a new partner (Martina Cornel). She will be in a position to work as soon as the amendment to the contract is in place. She is expected to be able to deliver on time.
- The Belgian partner (Jean-Pierre Fryns) did not hire the proper professional to do the data collection as planned. This person will be closely supervised during the second year to catch up.

There is no change in the project timetable.

Section 5 – Plan for using and disseminating Knowledge

1. Exploitable knowledge and its use

Two databases and 1 service were established during the first year: New tables in Orphanet to

In France,

- 3 Press Agency news releases - June 15th 2004 (AFP, APM et AEF)
- 1 News Brief in national daily newspaper (Le Parisien - June 16th 2004)
- 5 articles in national medical/specialised journals (Le Quotidien du Médecin - June 17th 2004, Le généraliste and Impact Médecine - June 18th 2004, Le Concours Médical - June 30 2004, Scrip August 13th 2004)
- Newsletter Pharmaclient (June 2004)
- Bulletin International d'Information (June-July 2004)
- Yahoo News (June 15 2004)
- La Croix (June 16th 2004)
- Nouvel Obs.com (June 16th 2004)
- Samaritains Handicap France website (June 16th 2004)
- L'usine nouvelle Biotech Info (July 2004)
- ivs-info.com, Biotechs diffusion letter (July 7 2004)
- Destination santé.com (July 8 2004)

We have no data on the impact in the media of other countries.

The Eclor service was advertised in OrphaNews France, the newsletter in French which is sent to 3,000 readers electronically every 2 weeks. It will be advertised in OrphaNews Europe when this newsletter will be launched.

The project leader of OrphanPlatform was invited to give a lecture in several meetings:

- "Identifier de nouvelles indications de molécules déjà sur le marché: le projet orphanXchange"
Colloque du GIS Maladies Rares "Développer la thérapeutique des maladies rares: stratégies de recherche de molécules d'intérêt", Paris, 7 juin 2004

- "OrphanPlatform: services for the development of orphan drugs"
EFPIA/EBE workshop on orphandrugs. Brussels, 11 June 2004

- "What epidemiological data are lacking in orphan drugs development and what are the possible solutions?"
International conference on pharmacoepidemiology and therapeutic risk management
Bordeaux, 25 August 2004

- « Are orphan drugs priority medicines for Europe ?
Drug Information Association European meeting, Lisbon, 8 March 2005

Appendix 1
Major research project funding agencies
and sources of information to collect research projects
on rare diseases

FRANCE

Ministère de la Santé : Délégation à la recherche clinique de l'APHP

Carré historique de l'hôpital Saint-Louis
1, avenue Claude Vellefaux 75010 Paris
Tel : 01 44 84 17 65

Ministère de la recherche

PHRC
Ségolène Aymé
INSERM ORPHANET

Inserm , BIR : Banque d'informations sur les recherches

Catherine Trecourt..... 101 Rue de Tolbiac 75013 Paris Tel : 01 44 23 61 25

E-mail : trecourt@tolbiac.inserm.fr

GIS - Institut des maladies rares

Pascale Borensztein
Plateforme Maladies Rares
Hôpital Broussais
102 rue Didot, 75014 Paris
pborensz@infobiogen.fr

CNRS :

emmanuelle.wollman@cnrs-dir.fr

CEA

www.cea.fr/

INRA :

www.inra.fr/

INRIA

www.inria.fr/

Institut Pasteur :

<http://www.pasteur.fr/>

Institut Curie :

<http://www.curie.fr/>

Main patient associations

AFM :

<http://www.afm-france.org/>

Anne D'Andon
adandon@afm.genethon.fr

Vaincre la Mucoviscidose

181 rue de Tolbiac
75013 Paris
Stéphane Mazur
smazur@vaincrelamuco.org

Ligue contre le cancer :
www.ligue-cancer.asso.fr/

ARC : Association pour la Recherche sur le Cancer

Pr FRIDMAN Hervé
ARC 94803 VILLEJUIF CEDEX

FNLCC :
www.fnclcc.fr

BELGIUM

Public institutions

FWO VL Fond de la recherche scientifique en Flandre
FNRS Fond de la recherche scientifique en Wallonie
IWT Institut de la recherche technologique en Flandre
IRSIA Institut de la recherche technologique pour la Wallonie
Ministère de la Recherche au travers des pôles universitaires

Private institutions

Association Belge de l'étude du Cancer (ABEC-BVSK)
Ligue Flamande contre le Cancer
Association Belge de Lutte contre la Mucoviscidose
Fond Forton (au sein de la Fondation Roi Baudouin)

ITALY

TELETHON

Italian Ministry of Health

CNR (the National Research Council)

Scientific Institutes for Research, and Research Hospital (Istituti di Ricovero e Cura a Carattere Scientifico)

ISS - Istituto Superiore di Sanità

Regional Health Authorities

UNITED KINGDOM

ARC

Arthritis Research Campaign
Copeman House
St Mary's Court
St Mary's Gate
Chesterfield
Derbyshire
S41 7TD
United Kingdom
Tel: 0870 850 5000 or +44 (0)1246 558033
Fax: +44 (0)1246 558007
www.arc.org.uk

BBSRC

Polaris House
North Star Avenue
Swindon
SN2 1UH
United Kingdom
+44 (01793) 413349
grants.awards@bbsrc.ac.uk

Cancer Research UK

P.O. Box 123
Lincoln's Inn Fields
London WC2A 3PX
tel. 020 7242 0200
fax. 020 7269 3100
<http://science.cancerresearchuk.org/>

Department of Health

The Department of Health
Richmond House
79 Whitehall
London SW1A 2NL
0207 210 4850
www.dh.gov.uk

MRC

Medical Research Council,
20 Park Crescent,
London
W1B 1AL
UK
Tel: +44 (0)20 7636 5422
Fax: +44 (0)20 7436 6179
corporate@headoffice.mrc.ac.uk

New Life (Birth Defects Foundation)

[BDF Newlife](#)
[BDF Centre,](#)
[Hemlock Business Park,](#)

Hemlock Way,
Cannock,
Staffordshire, WS11 7GF
01543 468888
www.bdfcharity.co.uk

The Royal Society
6-9 Carlton House Terrace
London
SW1Y 5AG
tel: +44 (0)20 7451 2500
fax: +44 (0)20 7930 2170
info@royalsoc.ac.uk
<http://www.royalsoc.ac.uk>

WELLCOME TRUST
215 Euston Road
London
NW1 2BE
UK
tel:+44 (0)20 7611 8888
email:contact@wellcome.ac.uk
www.wellcome.ac.uk

Also, many other medical research charities which may fund rare disease research can be found at www.amrc.org.uk.

PORTUGAL

Foundation for Science and Technology (FCT), Ministry of Science -
Fundação para a Ciência e Tecnologia
Av. D. Carlos I, 126
1249-074 Lisboa
PORTUGAL
<http://www.fct.mces.pt>

Agency for Innovation, Ministry of Science/Economics -
Agência da Inovação
Edifício IDIT - Rua do IDIT - Espargo
4520-102 S.M.Feira
Tel: 256 330 820
Fax: 256 332 891
<http://www.adi.pt>

Gulbenkian Foundation (private) -
Fundação Calouste Gulbenkian
Av. de Berna, 45A
1067-001 Lisboa
Telf: 21 782 3000
Fax: 21 782 3021

www.gulbenkian.pt
info@gulbenkian.pt

There are occasionally other private foundations that inject money into research, but these ones (namely FCT and Gulbenkian) fund the majority of the projects existing in state and academic labs.

AUSTRIA

Bundesministerium für Gesundheit und Frauen

Referat I/A/1b
A-1030 Wien, Radetzkystraße 2
01/71100-0
johannes.wohlfahrt@bmgf.gv.at

Medizinisch-wissenschaftlicher Fonds des Bürgermeisters der Bundeshauptstadt Wien

Büro der Geschäftsgruppe Gesundheit und Soziales, Rathaus, Stiege 5, 2. Stock
A-1082 Wien
01/4000/81235

Dachverband der österreichischen Kinder-Krebs-Hilfe

A-1090 Wien, Borschkegasse 1/7
01/4028899
dachverband@kinderkrebshilfe.at

Fonds zur Förderung der wissenschaftlichen Forschung (FWF)

A-1040 Wien, Weyringergasse 35
01/5056740-0
office@fwf.ac.at

Jubiläumsfonds der österreichischen Nationalbank

A-1090 Wien, Otto-Wagner-Platz 3
01/40420/7100
romana.rebec@oenb.co.at

Medizinische Universität Wien – Hans-und-Blanca-Moser-Stiftung

A-1090 Wien, Spitalgasse 23, BT 88
01/40160/10001
karin.tentulin-wawra@meduniwien.ac.at

Magistrat der Stadt Wien – Stiftung zur Förderung der Bekämpfung der Tuberkulose und anderer Lungenerkrankungen

Magistratsabteilung 15, Referat I/3, Tuberkulosebekämpfung
A-1010 Wien, Neutorgasse 18
01/53114/87658

Österreichische Forschungsförderungsgesellschaft mbH (FFG)

A-1015 Wien, Kärntnerstraße 21-23
01/5124584
office@fff.co.at

Österreichische Gesellschaft für Chemotherapie

A-1090 Wien, Alserstraße 4

01/405138320

Österreichische Gesellschaft für Orthopädie und orthopädische Chirurgie

A-1090 Wien, Alserstraße 4

01/40513/8320

Österreichische Gesellschaft für Rheumatologie und Rehabilitation

A-1121 Wien, Dr.-Boehringer-Gasse 5-11

01/8039880

office@rheuma2000.at

Österreichischer Herzfonds

A-1090 Wien, Währinger Straße 15/16

01/4059155

office@herzfonds.at

Uni Venture

(Universitäts-Spin-Off-Fonds)

A-1010 Wien, Seitzergasse 2-4

01/53453/22381

Universität Innsbruck

(Forschungsprojekte mit Industriebezug)

A-6020 Innsbruck, Christoph-Probst-Platz

0512/507/3967

i-med-qm@uibk.ac.at

IRELAND

Public funding-

- Health Research Board.** (www.hrb.ie)
- Science Foundation Ireland** (www.sfi.ie)
- Enterprise Ireland** (www.enterprise-ireland.com)

Private Funding/ Support Groups

- Fighting Blindness Ireland**
(c/o Michael Griffith, 1 Chrishchurch hall, Highstreet, Dublin 8.)
- Irish Cancer Society**
(5 Northumberland Road, Dublin 4.)
- Irish Heart Foundation**
(4 Clyde Road, Ballsbridge, Dublin 4.)
- Cystic Fibrosis Association of Ireland**
(CF House, 24 Lower Rathmines Road, Dublin 6.)
- DEBRA Ireland**
(10 Burgh Quay, Dublin 2.)

FINLAND

Aaltosen Säätiö
Jalmari ja Rauha Ahokkaan Säätiö
Allergiatutkimussäätiö
Alzheimer-säätiö
Doris och Holger Bergenheims Stiftelse för medicinsk Forskning
Diabetestutkimussäätiö
Mary och Georg Ehrnrooths stiftelse
Epilepsiatutkimussäätiö
Farmoksen tutkimus- ja Tiedesäätiö
Finska läkaresällskapet r.f.
Folkhälsans forskningsstiftelse - Kansanterveyden tutkimussäätiö
Fysiologisten hoitomuotojen tutkimussäätiö
Biomedicum Helsinki -säätiö
Instrumentariumin Tiedesäätiö
Invalidisäätiö
Itsenäisyyden Juhlavuoden Lastenrahaston Säätiö
Yrjö Jahnssonin säätiö
Sigrid Juseliuksen Säätiö
Väinö ja Hilikka Kiltin Säätiö
Kirurgian Edistämissäätiö
Väinö ja Laina Kiven Säätiö
Korvatautien tutkimussäätiö
Aarne Koskelon säätiö
Kuopion Yliopistosäätiö
Maaherra Wilho Kytän säätiö
Urho Känkäsän Säätiö
Reino Lahtikarin Säätiö
Lastenpsykiatrian tutkimussäätiö
Lastentautien tutkimussäätiö
Kyllikki ja Uolevi Lehikoisen Säätiö
Leiraksen tutkimussäätiö
Lihastautien Tutkimussäätiö
Lääketutkimussäätiö
Mehiläisen Tutkimussäätiö
Minervastiftelsen - Minervasäätiö
Mjöbolsta stiftelse för medicinsk forskning
Moikoisten Syöväntutkimussäätiö
Ida Montinin Säätiö
Munuaissäätiö
Neurologiasäätiö
Nummelan parantolan lääketietellinen tutkimussäätiö
Paavo Nurmen Säätiö
Ellen ja Artturi Nyysösen säätiö
Orion-yhtymän Tutkimussäätiö
Kurt och Doris Palanders Stiftelse för Medicinsk Forskning
Pharmacalin stipendi- ja tutkimussäätiö
Kaarina ja Erkki Piippolan säätiö
Psykiatrian Tutkimussäätiö
Reumatautien tutkimussäätiö
Rinnekodin tutkimussäätiö

Salonoja-säätiö
Silmä- ja kudospankkisäätiö
Silmäsäätiö
Stiftelsen Alma och K.A. Snellman Säätiö
Päivikki ja Sakari Sohlbergin Säätiö
Maija Sulamaan säätiö
Suomen Hammaslääketieteen Säätiö
Suomen Kulttuurirahasto

GERMANY

1. DFG (Deutsche Forschungsgemeinschaft)

Deutsche Forschungsgemeinschaft
Kennedyallee 40
53175 Bonn
Telefon: 0228/885-1
Telefax: 0228/885-2777
E-Mail: postmaster@dfg.de
URL: <http://www.dfg.de>
<http://www.dfg.de/en/index.html>

2. BMBF (Bundesministerium für Bildung und Forschung)

Federal Ministry of Education and Research
Bonn office
Heinemannstr. 2
53175 Bonn
Telephone: 01888/57- 0
Fax: 01888/57- 83601

Federal Ministry of Education and Research

Berlin office
Hannoversche Straße 28-30
10115 Berlin
Telephone: 01888/57- 0
Fax: 01888/57- 83601

E-Mail: information@bmbf.bund.de
<http://www.bmbf.de/en/>

3. Bundesministerium für Gesundheit und Soziale Sicherung (BMGS)

Postfach 500,
53108 Bonn,
Fax: 0180 - 51 51 511
<http://www.bmgs.bund.de/deu/gra/service/email/index.php>

4. Max Planck Society

Hofgartenstr. 8
80539 München

Max Planck Society
P.O. Box 10 10 62
80084 München

Tel: +49 (89) 2108 - 0
Fax: +49 (89) 2108 - 1111
<http://www.mpg.de/english/portal/index.html>

5. VW-Stiftung (VolkswagenStiftung)

VolkswagenStiftung
Kastanienallee 35
30519 Hannover
Germany
Phone: +49 (0)511 8381-0
Fax: +49 (0)511 8381-344
E-mail: mail@volkswagenstiftung.de
<http://www.volkswagen-stiftung.de/english.html>

6. Deutsche Krebshilfe

Deutsche Krebshilfe e.V.
Thomas-Mann-Str. 40
53111 Bonn
Postfach 1467, 53004 Bonn

Telefon: 02 28/7 29 90-0
Fax: 02 28/7 29 90-11
E-Mail: deutsche@krebshilfe.de
<http://www.krebshilfe.de/>

7. Wilhelm-Sander-Stiftung

Goethestraße 74
80336 München
Telefon: 089 / 544 1870
Telefax: 089 / 544 18720
e-Mail: info@sanst.de
<http://www.sanst.de/index.html>

8. Gemeinnützige Hertie-Stiftung (Hertie Foundation)

Gemeinnützige Hertie-Stiftung

Grüneburgweg 105
60323 Frankfurt am Main

Telefon: +49 (69) 660 756 -0
Telefax: +49 (69) 660 756 -999 Berliner Büro
Oranienburgerstr. 13/14
10178 Berlin
<http://www.ghst.de/en/index.php>

9. Deutsche José Carreras Leukämie-Stiftung e.V.

Arcisstraße 61
80801 München
Tel.: 089/272904-0
Fax.: 089/272904-44
<http://www.carreras-stiftung.de/>

10. Christiane Herzog Stiftung

Geißstraße 4
70173 Stuttgart
Tel. 0711 / 24 63 46
Fax 0711 / 24 26 31
<http://www.christianeherzogstiftung.de/>

11. Bertelsmann Stiftung

Carl-Bertelsmann-Str. 256
33311 Gütersloh
Tel: + 49 5241-810
Fax:+ 49 5241-81681502
E-Mail: info@bertelsmann-stiftung.de
Web: <http://www.bertelsmann-stiftung.de/>
<http://en.bertelsmann-stiftung.de/index.html>

12. Friedrich- Ebert- Stiftung

Godesberger Allee 149
D-53175 Bonn
Tel. +49(0)228/883-0
Fax +49(0)228/883-396
Friedrich-Ebert-Stiftung
Hiroshimastraße 17
D-10785 Berlin
Tel. +49(0)30/26935-6
Fax +49(0)30/26935-850

<http://www.fes.de/indexnav.html>

<http://www.fes.de/indexkontakt.html>

13. Hans-Böckler-Stiftung

Hans-Böckler-Straße 39
40476 Düsseldorf
Telefon: +49 211 7778 0
Telefax: +49 211 7778 120
E-Mail: zentrale@boeckler.de
<http://www.boeckler.de/>

14. Konrad-Adenauer-Stiftung e.V

Rathausallee 12
53757 Sankt Augustin
Tel.: (0 22 41) 24 6-0
Fax: (0 22 41) 24 6-5 91
E-Mail: zentrale@kas.de

http://www.kas.de/1_webseite.html

http://www.kas.de/1642_webseite.html

13) Deutsche Diabetes-Stiftung

Geschäftsstelle
Am Klopferspitz 19
82152 Martinsried / München

Telefon: (0 89) 579 579 - 0
Fax: (0 89) 579 579 - 19

E-Mail: info@diabetesstiftung.de
Internet: <http://www.diabetesstiftung.de>

14) Helmholtz-Gemeinschaft

http://www.helmholtz.de/en/General_Information/Service/Impressum.html

15) Alexander von Humboldt-Stiftung

<http://www.avh.de/en/index.htm>

16) Arbeitsgemeinschaft industrieller Forschungsvereinigungen "Otto von Guericke" e.V.

Bayenthalgürtel 23
50968 Köln
Telefon 0221 37680-0
Telefax 0221 37680-27

E-Mail: info@aif.de

17) Geschäftsstelle Berlin

Tschaikowskistraße 49
13156 Berlin
Telefon 030 48163-3
Telefax 030 48163-401/402
E-Mail: gsb@aif.de
<http://www.aif.de/>

17) Fritz Thyssen Stiftung

Am Römerturm 3
50667 Köln
Kontaktdaten
Telefon: 0221 – 27 74 96-0
Telefax: 0221 – 27 74 96-29
<http://www.fritz-thyssen-stiftung.de/>

SPAIN

PUBLIC LEVEL

- Plan Nacional de Investigación Científica, Desarrollo e Innovación Tecnológica 2004-07, Ministerio de Educación y Ciencia (http://wwwn.mec.es/ciencia/plan_idi/)
- Fondo de Investigación Sanitaria, Instituto de Salud Carlos III, Ministerio de Sanidad y Consumo (<http://www.isciii.es/fis/>)
- Pla de Recerca i Innovació 2005-2008 , Departament d'Universitats, Recerca i Societat de la Informació, Generalitat de Catalunya (<http://www10.gencat.net/dursi/>)
- Agencia de Gestió d'Ajuts Universitaris i de Recerca (<http://agaur.gencat.net/>)
- Plan Regional de Investigación Científica e Innovación Tecnológica de la Comunidad de Madrid, Dirección General de Universidades e Investigación, Consejería de Educación, Comunidad de Madrid (<http://www.madrid.org/ceducacion/investigacion/index.htm>)
- Plan Andaluz de Investigación, Consejería de Innovación, Ciencia y Empresa, Junta de Andalucía (<http://www.juntadeandalucia.es/innovacioncienciayempresa/>)
- Plan Autonómico de Investigación, Desarrollo y Transferencia de Conocimientos de Aragón , Departamento de Ciencia, Tecnología y Universidad, Gobierno de Aragón (<http://www.aragoninvestiga.org/>)
- Plan Tecnológico de Navarra, Gobierno de Navarra (<http://www.plantecnologico.com/>)
- Plan de Ciencia, Tecnología e Innovación, Gobierno del Principado de Asturias (<http://www.pctiasturias.com/>)
- Cantabria
- Plan Riojano de I+D+I, Gobierno de La Rioja (<http://www.larioja.org/i+d+i/index.htm>)
- Plan Regional de Investigación, Desarrollo Tecnológico e Innovación de Extremadura, Junta de Extremadura, (<http://www.juntaex.es/consejerias/idt/dgidi/IIpri-infr/indice.htm>)
- Plan Galego de Investigación, Desenvolvemento e Innovación Tecnolóxica, Xunta de Galicia (<http://www.sxid.org/>)

- Estrategia Regional de Investigación Científica, Desarrollo Tecnológico e Innovación (I+D+i), Junta de Castilla y León (http://www.jcyl.es/jcyl-client/jcyl/cee/ade/tkContent?idContent=24415&locale=es_ES&textOnly=false)
- Castilla – La Mancha Innovación (<http://www.clminnovacion.com/>)
- Convocatoria de ayudas para la realización de Proyectos de Investigación Básica y/o Aplicada, Humanidades y Ciencias Sociales y ayudas a Grupos de Alto Rendimiento, Departamento de Educación, Universidades e Investigación, Gobierno Vasco, (http://www.hezkuntza.ejgv.euskadi.net/r43-2493/es/contenidos/informacion/dib4/es_2035/proyectos_c.html)
- Programa de Investigación y Desarrollo Tecnológico, Conselleria d'Empresa, Universitat i Ciència, Generalitat Valenciana (<http://www.impiva.es/>)

FOUNDATIONS

- Fundació Marató TV3 (<http://www.tvcatalunya.com/marato/>)
- Fundació La Caixa (<http://www.fundacio.lacaixa.es>)
- Fundación Caja Madrid (<http://www.fundacioncajamadrid.org/>)
- Fundación Ramón Areces (<http://www.fundacionareces.es/>)
- Real Patronato sobre Discapacidad (<http://www.rpd.es/>)
- Fundación Séneca , Región de Murcia (<http://www.f-seneca.org/>)
- Fundació Ibit, Illes Balears Innovació Tecnològica (<http://www.ibit.org/>)

Appendix 2 Questionnaires to collect information on research activities

Return this form by:

Fax :

Post :

E-Mail :

RESEARCH PROJECT ON RARE DISEASES*

* A disease is considered to be rare when it affects less than 1 person in 2,000.

Type of project			
Cartography/Cloning of the responsible gene (s)	<input type="checkbox"/>	Development of a new diagnostic tool	<input type="checkbox"/>
Search for mutations in the responsible gene (s)	<input type="checkbox"/>	Development of a new diagnostic protocol	<input type="checkbox"/>
Gene Expression	<input type="checkbox"/>	Development of a medical device	<input type="checkbox"/>
Genotype-phenotype correlation	<input type="checkbox"/>	Clinical trial for a new drug	<input type="checkbox"/>
Physiopathology of the disease	<input type="checkbox"/>	Clinical trial for a new protocol	<input type="checkbox"/>
Natural history of the disease	<input type="checkbox"/>	Epidemiological Study	<input type="checkbox"/>
Animal model of the disease	<input type="checkbox"/>	Registry	<input type="checkbox"/>
Gene therapy / Cell therapy	<input type="checkbox"/>	Research in health sociology	<input type="checkbox"/>
Preclinical development of a new drug	<input type="checkbox"/>	Research in health economy	<input type="checkbox"/>

Other aspects of the project

Patients' recruitment needed for the study: Yes No

Scheduled duration of the project:

Do you belong to a network: National European International

Name of the network:

Are you the project leader of the network? Yes No

If Clinical trial, name of the sponsor:

Title of the research project

In local language:

In English:

Names of the rare disease(s) concerned by the project

Person responsible for the project

Gender M F Last name: First name:
 Title: Dr Prof Middle initial:

Professional degree: PhD MD PhD - MD other

E-mail address:

Additional information only for Orphanet team; not for online publication:

Phone number (direct line or mobile)

Personal e-mail :

Name of the laboratory or service

Address:

Phone number of the **secretary**: Fax:

Website URL:

Status: Public Independent / Private Private non for profit

Setting: Research setting Clinical setting

Affiliation :

Department/ Institution (if applicable):

Acronym :

Full name :

Director's name :

Hosting institution's name:

Funding information

Grant(s) from public institutions:
 Names of institutions

Grant(s) from private companies:
 Names of companies

Grant(s) from charities:
 Names of charities

Partnerships

Are you interested in developing collaborations / partnerships?

with academic researchers: Yes No
 with biotech/industry Yes No

Protocol summary of the project

If you want the protocol of your project to be available on Orphanet, please send your text by e-mail.

INFORMED CONSENT TO BE RETURNED WITH SIGNATURE

According to the electronic data protection law, your consent is required before diffusion of any nominative information. You have all rights to access, opposition to, or correction of these data. Phone numbers and e-mail addresses may be not available to the public. You need only to contact the director of the Orphanet database, Dr Ségolène Aymé. The file composed from this information is declared at the French electronic data protection Committee.

I agree to the terms and conditions for publication of this information on the ORPHANET database website.

Date:

Signature:

Return this form by:

Fax :

Post :

E-Mail :

CLINICAL TRIAL ON RARE DISEASES*

* A disease is considered to be rare when it affects less than 1 person in 2,000.

Title of the trial:

In local language:

In English:

Name of the rare disease(s) concerned by the trial:

Sponsor of the clinical study:

Name:

Address:

Phone number of the **secretary**:

Fax number:

Contact person:

Gender: M F
Title: Dr Prof

Last name:

First Name:
Middle initial:

Additional information only for Orphanet team; not for online publication:

Phone number (direct line or mobile) :

Personal e-mail :

Principal investigator

Gender: M F Last name: First name:
Title: Dr Prof Middle initial:
Professional degree: PhD MD PhD- MD other
E-mail address:

Additional information only for Orphanet team; not for online publication:
Phone number (direct line or mobile) :
Personal e-mail

Name of the service:

Address:

Phone number of the **secretary**: Fax number:

Website URL:

Status: Public institution Independent / Private Private non for profit
Setting : Research setting Clinical setting

Affiliation:

Department/ Institution (if applicable):

Acronym :
Full name :
Director's name :
Hosting institution's name:

Other investigator (if applicable)

Gender: M F Surname: First name:
Title: Dr Prof
E-mail address:

Key aspects of the trial

Date of the start of the trial: Duration scheduled:
Patients' recruitment needed for the study: Yes No
Recruiting period: Expected number of individuals to be included:
Phases of the trial: Phase 1 Phase 2 Phase 3 Phase 4
Single centre trial National multi-centre trial International multi-centre trial
Participating countries if international trial:

Inclusion criteria:

Exclusion criteria:

Name of the drug(s)* or product(s) concerned by the trial:

* Indicate: Speciality ®, ICD or name of the substance or code name, pharmaceutical form, dosage, ATC code

According to the electronic data protection law, your consent is required before diffusion of any nominative information. You have all rights to access, opposition to, or correction of these data. Phone numbers and e-mail addresses may be not available to the public. You need only to contact the director of the Orphanet database, Dr Ségolène Aymé. The file composed from this information is declared at the French electronic data protection Committee.

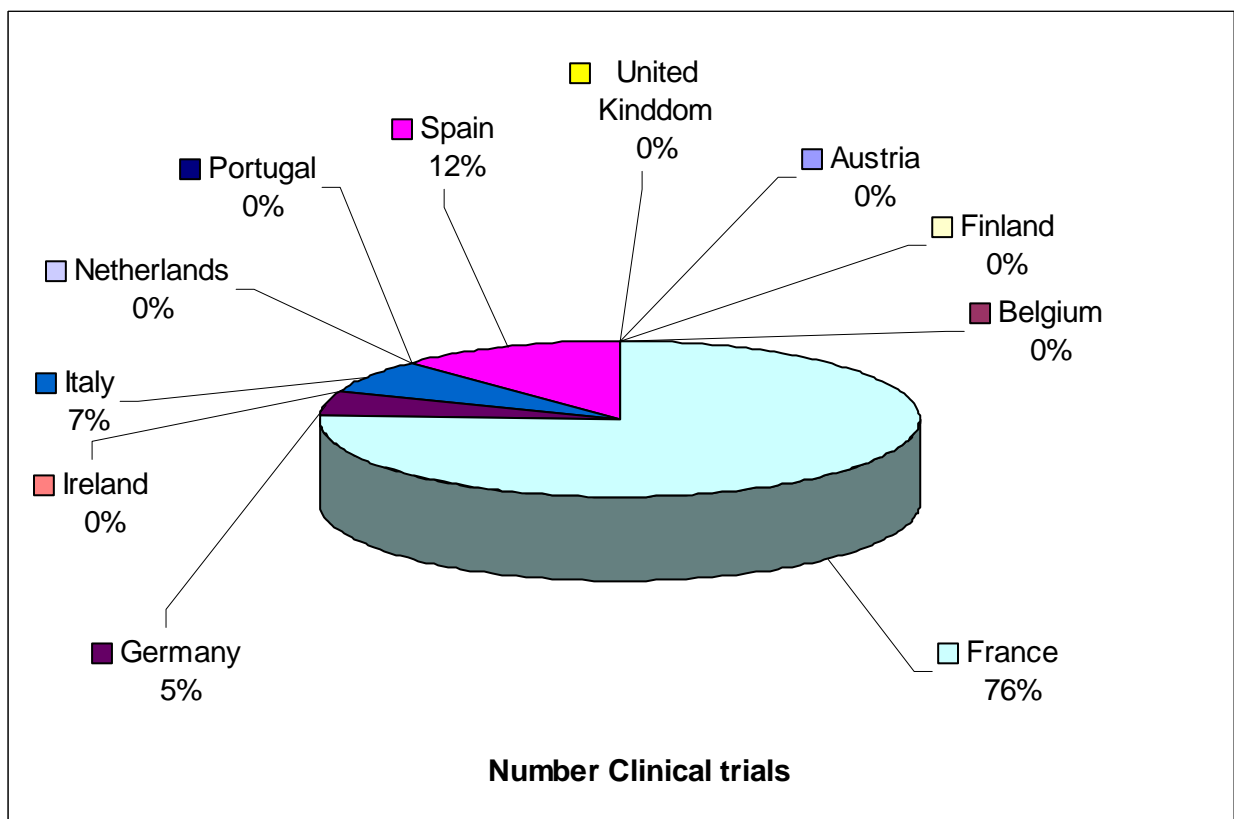
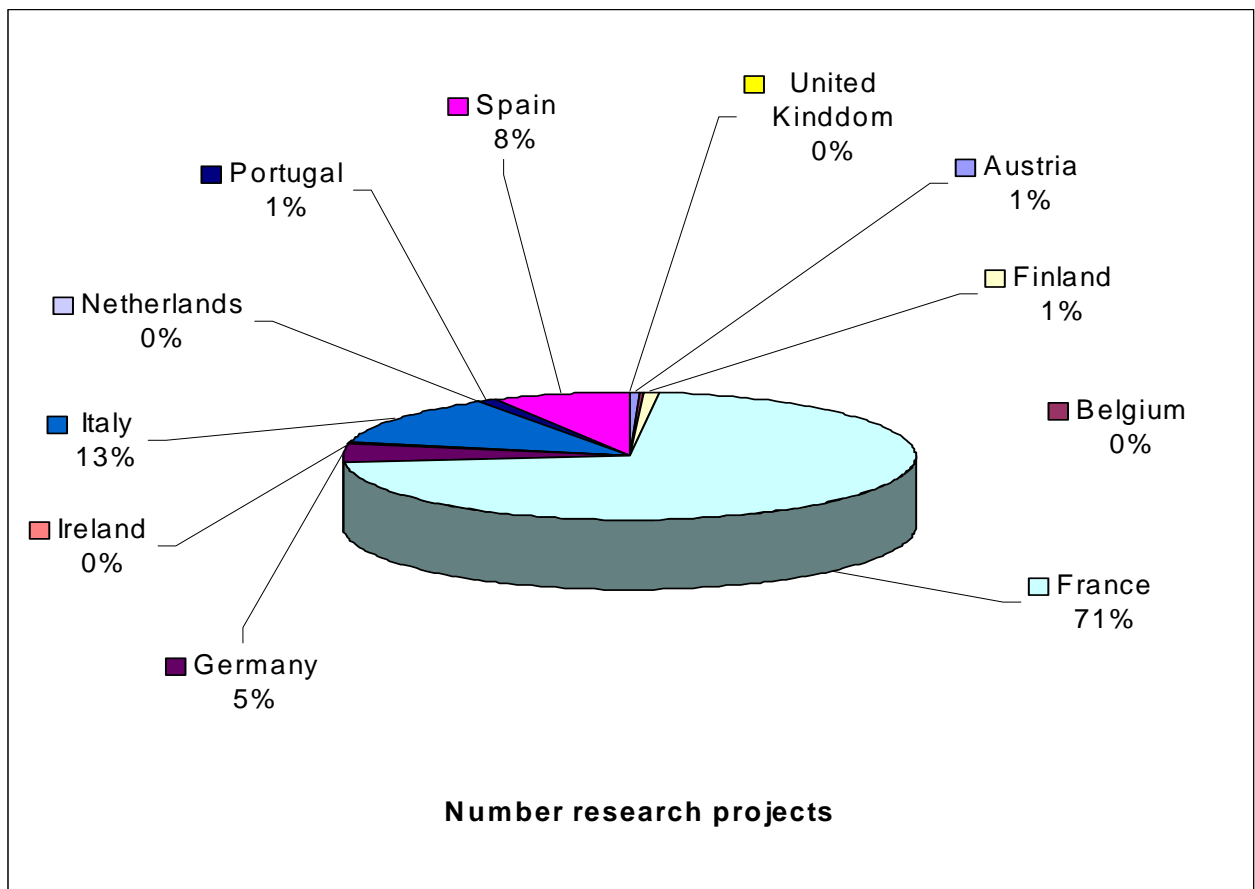
INFORMED CONSENT TO BE RETURNED WITH SIGNATURE

I agree to the terms and conditions for publication of this information on the ORPHANET database website.

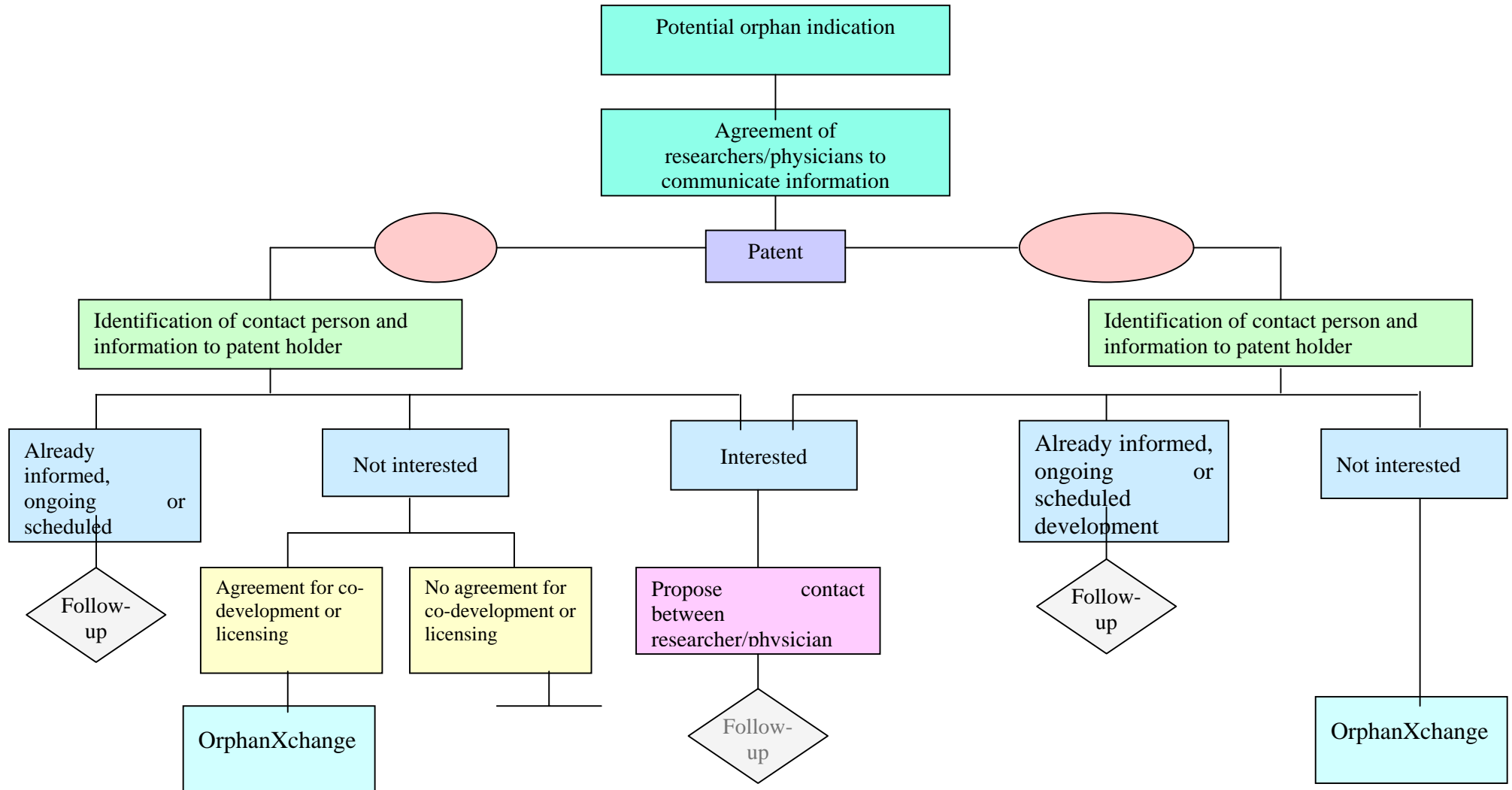
Date:

Signature:

Appendix 3



Orphan indications: off label use





LSSM-CT-2004-503246

Press release

July 2004

**Launch of "OrphanXchange" and "Erditi":
Two European pioneering initiatives to boost
the development of therapies for rare diseases**

After a two-year joint effort, the French Alliance for Rare Diseases, the French Institute for Rare Diseases Research, the French Pharmaceutical Companies Association (LEEM), the European database of rare diseases Orphanet and Inserm (National Institute of Health and Medical Research) announce the creation of two major initiatives.

These are an important step as co-operation is vital to discovery, development and registration of treatments for rare diseases or "Orphan Diseases" as they are generally known. Rare diseases affect 20 million European citizens. Solutions are difficult to discover and scientific breakthroughs are needed. By promoting partnerships between the different organisations, whether public or private, the chances of success are increased.

- **OrphanXchange** (www.orphanxchange.org) seeks to promote partnerships between academic research projects and private companies with the aim of developing diagnostic solutions and "orphan" drugs. This exchange of information is facilitated via OrphanXchange's database website. Therapy development projects can result from academic research alone or may involve compounds that are already marketed for other indications, and which may, in turn, also be used in the treatment of rare diseases. The OrphanXchange database already contains projects for possible orphan drug indications involving over 50 rare diseases. The OrphanXchange website is free of charge and accessible to everyone. Access to the detailed description of projects requires registration. This site is powered by the European database of rare diseases, Orphanet (www.orpha.net). OrphanXchange, a program developed within an Inserm department, is supported by the European Commission's DG Research Framework 6 Programme and the LEEM (French Pharmaceutical Companies Association).

- **Erditi** (European Rare Diseases Therapeutic Initiative) offers European academic researchers access to existing pharmaceutical compounds for further therapeutic investigation in the field of rare disease. Such compounds, that have been or are being developed by pharmaceutical companies in more common diseases, may thus also be developed for rarer, or "orphan" indications. To date, four major pharmaceutical companies, i.e., Aventis, Glaxosmithkline, Roche and Servier, and about 10 European academic research institutions are already ERDITI's partners. Erditi is coordinated by the French Institute for Rare Diseases Research, with the support of LEEM Research a

The aim of these two unique programs is to reinforce the existing operations between patients' associations, pharmaceutical companies, academic research and healthcare professionals. As part of the LEEM working group on rare diseases, these programs develop new avenues of research and cooperation between all parties involved in **fighting** rare diseases. Today such a fight is realistic: an **increasing number** of rare diseases, including genetic disorders, can now be treated, **either** by cell therapy, gene therapy, replacement enzymes or conventional pharmacological therapies. Major advances in biotechnology research raise the hope of better-targeted and more effective treatments. Therapies have been under development in Europe for the last three years as a result of the 1999 passing of a European orphan drug regulation aimed at providing incentives for pharmaceutical companies. Over 300 applications have been submitted to the European Medicines Agency and 14 drugs have been granted Marketing Authorization (MA).

The French Alliance for Rare diseases, a full participant in the **thought** process that led to the origin of these two promising initiatives, expects much from the cooperation of public and private researchers. It is absolutely **essential** that all participants fully engage in this endeavor.

Rare diseases

- ° A disease is referred to as rare, or "orphan", when it affects less than five persons in 10,000.
- ° Rare diseases affect over 20 million persons in Europe
- ° Rare diseases are serious, chronic, progressive disorders, which often are life threatening.
- ° There are about 6,000 - 7,000 known rare diseases.
- ° About 80% of rare diseases are of genetic origin.
- ° Three out of four rare diseases are pediatric diseases resulting in severe disabilities.

OrphanXchange: www.orphanxchange.org
Contact Valérie Thibaudeau - Phone: +33 1 56 53 81 55
vthibaud@orpha.net

Erditi: www.erditi.org
Contact Claire Roussel - Phone: +33 1 58 14 22 84
croussel@infobiogen.fr

LEEM press information: Contact Stéphanie Fleurot - Phone.: +33 (0)1 45 03 88 38
e-mail: sfleurot@leem.org Agnès Garcia Gibot
Press and Publications information: Contact Anne Brossard and Catherine Gros – Phone.: +33 (0)1 46 99 69 69