

1. Publishable summary

RDPlatform was an international initiative bringing together organisations from 13 European countries. The project leader is Ségolène Aymé, and the coordinating team was composed of members of Orphanet Paris. Each of the thirteen participating countries was responsible for seeking and selecting the information at the national level. France carried out data collection for the remaining Orphanet countries (35 countries in total).

1. Objectives of the RDPlatform project and its potential impact

The RareDiseasePlatform was dedicated to developing a project-building platform to help researchers in the field of rare diseases (RDs) set up efficient, multidisciplinary teams to tackle RD research challenges. This project offered the opportunity for potential multinational teams to exchange ideas and strategies in order to structure future research proposals in the 27 EU member states.

The FP7 RDPlatform project was the continuation of the FP6 OrphanPlatform project, which delivered several products that have served as the basis for the development of RDPlatform. RDPlatform was conceived to fulfil the unmet needs of the European RD research community which were identified during the development of the previous European project.

The needs of the research community are diverse and include: finding a research team who developed an expert technological platform, finding a research team that has already created a desirable animal model or a collection of data and/or biological samples, discussing the clinical potential of a research project with clinicians who are more familiar with clinical research and development and constraints, as therapeutic developments are new in this field.

RDPlatform intends to facilitate the collaboration between academia and SMEs in order to convert scientific developments in the field of RD into diagnostic tools and therapies as quickly as possible. Over 30 million European citizens suffer from these severe and chronic diseases. They are entitled to be treated as efficiently as patients with more common diseases.

RDPlatform was structured around three central approaches

- Optimisation and integration of fragmented resources and data indispensable for research and development in the field of RDs and orphan medicinal products;
- Contribution to business intelligence activity for SMEs;
- Facilitation of all types of partnership: between academic teams, between academic teams and biotech companies, between two or more biotech companies, between biotech companies and big pharmaceutical companies.

RDPlatform was based on the experience acquired from:

- an EU-funded information service on RDs: Orphanet (www.orpha.net) which is the most visited information service in the field (20 000 daily users)
- an EU-funded project “Orphanplatform” (LSSM-CT-2004-503246) which is considered as a pilot of the current application
- an EU-funded project E-Rare which is an ERA-Net project on RDs (www.erare.eu)

2. Description of the work performed

i. Publicly available website for the RDPlatform project

We have constructed a publicly available website for the RDPlatform project (<http://www.rdplatform.org/>) where objectives and achievements of the RDPlatform project are explained. Information concerning the management structure and the members of the consortium is also available. A restricted member area is used to exchange documents between the members of the board of partners and the advisory board.

ii. Development of new Orphanet tools

A “Research & trials” area of the Orphanet website has been conceived to fulfil the unmet needs of the European RD research community, which were highlighted during the development of the previous European project. The new developments are accessible through the Orphanet website (<http://www.orpha.net/consor/cgi-bin/ResearchTrials.php?lng=EN>). A user guide for this new area of the website is available in the RDPlatform website (<http://www.rdplatform.org/>)

The “Research and trials” tab of the Orphanet website has been modified in order to collect additional information and to improve the search engine. A new multiple-criteria search engine allows users to find the specific information that they are interested in more easily.

We overhauled the orphan drugs section to improve and extend accessibility to information. We recalibrated the search engine to render data on Orphan Drugs more accessible from the website’s homepage. In addition to the existing options to search by drug, molecule, or by disease, we added four new sub-tabs that improve the visibility of information pertaining to orphan drugs, allowing users to search by a wider range of criteria. The results pages have also been redesigned to clearly separate substances from trade names, clarifying that trade names are used solely for products granted marketing authorisation, whereas substances with orphan designation status (prior to marketing authorisation) are referred to by the biochemical substance or molecule name. Finally, the orphan designation pages have been enlarged to include information on the sponsor, and the marketing authorisation pages now offer details on the MA holder. These new features are available in the five languages of the Orphanet website.

We also created functions to search for rare disease networks as networks are a growing phenomenon in the rare disease community, evolving from the recognition that collaboration and cooperation are crucial in order to combat scattered and limited resources, expertise, and patient populations in the field.

iii. Data collection and update

Each of the thirteen RDPlatform participating countries was responsible for collecting the information at the national level. France carries out data collection in France, and in the remaining Orphanet countries which are not partners of RDPlatform (22 countries).

The Orphanet Research & trials database contains more than **7200** entries to date -1 May 2011-, including 4337 research projects, 1720 clinical trials, 570 patient/product registries / 529 mutation registries / 93 biobanks.

A new Orphanet Report Series was published to present the rare disease networks in Europe. It is based on data extracted from the Orphanet database and captures the rare disease networks existing in Europe and surrounding countries. A network is defined as *“...a group of coordinated activities with financing or an official designation”*. This new Orphanet Report Series yields data on the rare disease research and clinical networks, providing statistics such as country distribution (number of networks for which a country is coordinator or participant), the diseases included in a particular network, geographical coverage of networks, distribution of networks by type of call, and funding mechanisms (including the EC DG-Research Framework Programmes, DG-Sanco, and E-Rare).

Another new Orphanet Report Series on patient registries in Europe was published. It offers data on patient registries and databases established in Europe and surrounding countries and which are open to collaboration with researchers. This work-in-progress cannot be considered exhaustive as data gathering is an ongoing process. However, the report offers a list of key registries organised by country, by type (regional, national, European or international), and by institution(s), as well as a list of network registries.

Finally, the way the collected data were disseminated was not foreseen at the beginning of the project: it was via personal requests from academic researchers and from Industry. Requests started to arrive in October 2008. Researchers wanted to receive our dataset of ongoing research projects, of clinical trials, of orphan drugs in development and on the market, together with our inventory of rare diseases and with our classifications. We had to define the terms of the possible transfer of data for research purpose. Standard Material Transfer Agreements (MTAs) were issued, one for academic teams, one for Industry teams.

We have signed 63 MTAs so far: 50 with academic teams and 13 with for-profit companies.

We received so many requests that we had to develop a new website to provide access to standard products at the address www.orphadata.org. The website was opened in June 2011 and received more than 1 000 visitors during the first month, without any publicity.

iv. Interactive Workshops of Experts on Rare Disease Research

Among the tasks of the partners of RDPlatform was the organisation of two workshops with top experts to analyse areas in need of collaborative research projects, based on an analysis of the current situation. The first workshop took place on 3 December 2009. The second one took place in Paris on 20 January 2011.

At the first workshop, panel discussions around the current status of the European research on rare diseases took place in the context of different presentations:

- The Orphanet team analysed the information collected and disseminated during the first half of the development of this project and produced a report intended to provide a clear view of what is currently funded at the national and international levels in Europe, in the field of rare diseases. The presentation of this report on 3 December 2009 served as the basis for discussion concerning the determinants of success for Therapy development in rare diseases.
- Prof. Busby, coordinator of a FP6-funded Network of Excellence presented a success example of European networks: the TreatNMD network.
- An overview of funding mechanisms, types of calls and areas to be promoted was given by Catherine Berens (Scientific Officer of the Medical and Public Health Research Unit of the EC DG Research), and by Sophie Koutouzov (General Secretary of the GIS Rare Diseases Institute and programme coordinator of the ERA-NET program on RD –E-RARE).

A final report, which included the main issues discussed that day was then produced and made publicly available by the end of January 2010 on the RDPlatform website.

A second round of revision of this report was started in the autumn of 2010, to prepare the final report planned to provide a State-of-the-Art of R&D in the field of rare diseases and orphan drugs but also an analysis of bottlenecks and a set of recommendations for the future.

A tentative list of 185 experts to be invited to contribute to this brain-storming exercise was established by considering that all project leaders of EC-funded research projects were the best possible ones. They received a letter of invitation in September, asking them if they were willing to review the documents and/or attend a workshop to be held in Paris. Those who replied positively received a formal invitation on 13 October 2010 to attend the workshop to be organised in January 2011.

A preparatory document was prepared by the coordinating team and sent to the 53 experts willing to review, whether they were able to attend the workshop or not, on 10 January. Written comments were expected within one month.

A final report on the State of the Art of R&D in Europe, in the field of rare diseases and orphan drugs was made publicly available in July 2011.

2. Project objectives

This project aimed at creating a set of tools intended to facilitate collaborations between academic teams, SMEs and even major companies, in the field of rare diseases (RDs). These tools were intended to contribute to building a community of stakeholders with the ultimate goal of speeding up RD research and development, and providing diagnostic tools and therapies as quickly as possible.

The specific objectives were:

- to identify expert groups in Europe, on-going funded research projects, technological platforms, databases and biobanks relevant to RD research and to release the information in a user-friendly manner on the existing Orphanet website;
- to identify, amongst the research projects funded at the MS level and at the EU level, those which are in need of partnership with other academic teams and/or which have a potential for market development and may benefit from a partnership with Industry;
- to release the information on partnership opportunities on the existing OrphanXchange website and adapt the website to meet the needs of all the types of partnerships identified so far;
- to develop partner search facilities based on the above mentioned databases and on an ad-hoc basis;
- to develop an electronic newsletter informing the community about newly posted partnership requests and business opportunities;
- to organise two workshops with top experts to analyse areas in need of collaborative research projects.