Orphanet Report Series

Rare Diseases collection

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List of Research Infrastructures useful to Rare Diseases in Europe

By country of location

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List of research infrastructures useful to rare diseases in Europe

AUSTRIA

BBMRI-ERIC (Biobanking and Biomolecular Resources Research Infrastructure – European Research Infrastructure Consortium)

BBMRI is a distributed research infrastructure and has been on the ESFRI Roadmap since 2006. It is mostly a networking and standardization effort, whose stated mission as an ERIC is to increase efficiency and excellence in European biomedical research in an ethically and legally compliant way, and to promote standard operating procedures and international best practices on a variety of pre-existing national bio-banks. The National nodes are not part of the ERIC. The relation to the central entity is achieved via membership on the Management Board, thus national bio-banks have only an indirect link to the ERIC. This is reflected in the Partner Charter with the national nodes.

http://www.bbmri-eric.eu/

BELGIUM

European Organisation for Research and Treatment of Cancer (EORTC)

The European Organisation for Research and Treatment of Cancer aims to develop, conduct, coordinate, and stimulate translational and clinical research in Europe to improve the management of cancer and related problems by increasing survival but also patient quality of life. The EORTC contributes to the development of new drugs and other innovative approaches in partnership with the pharmaceutical industry. This is accomplished mainly by conducting large, multicenter, prospective, randomized, phase III clinical trials. In this way, the EORTC facilitates the passage of experimental discoveries into state of the art treatments. EORTC Headquarters, a unique pan European clinical research infrastructure, is based in Brussels, Belgium, from where its various activities are coordinated and run.

http://www.eortc.org/

FRANCE

APTEEUS: engaging patients in discovery

APTEEUS is a clinical stage company addressing monogenetic disorders. Focusing on the molecular causes of diseases, APTEEUS dedicates its strong experience to the discovery and the development of Disease Modifying Therapies.

http://apteeus.fr/en/

European Clinical Research Infrastructures Network (ECRIN)

The European Clinical Research Infrastructures Network is a sustainable, not-for-profit infrastructure supporting multinational clinical research projects in Europe. Multinational clinical research is hampered by the fragmentation of health and legislative systems in Europe. ECRIN provides information, consulting and services to investigators and sponsors in the preparation and in the conduct of multinational

clinical studies, for any category of clinical research and in any disease area. This is particularly relevant for investigator- initiated clinical trials and for clinical research on rare diseases where international cooperation is a key success factor. ECRIN is based on the connection of coordinating centres for national networks of clinical research centres and clinical trials units, able to provide support and services to multinational clinical research. Relevant tools for clinical researchers involved in multinational clinical trials are available on the website.
http://www.ecrin.org/

European Research Infrastructure on Highly Pathogenic Agents (ERINHA)

The **ERINHA2** project aims at building a pan-European research infrastructure to reinforce the European coordination and capacities for the study and the surveillance of highly pathogenic micro-organisms. The ERINHA infrastructure provides open access to state-of-the-art BSL-4 facilities for the European scientific community to enhance basic and finalised research activities. The European Research Infrastructure on Highly Pathogenic Agents (ERINHA) promotes the harmonization of biosafety and biosecurity procedures, develops standards for the management of biological resources, diagnosis of group 4 pathogens, and training of BSL4 labs users.
http://www.erinha.eu/

GENETHON

Genethon's mission is to design gene therapy products for rare diseases, and to ensure their pre-clinical and clinical development in order to provide patients with access to these innovative treatments. To meet this challenge, Genethon has acquired the technical and human resources needed to accelerate the medical application of scientific discoveries arising from fundamental research. The translational research programs are supported by a first-rate technological platform and nearly 200 people (researchers, Pharm.Ds, M.Ds, Ph.Ds, engineers and technicians,…) who have joined forces in order to develop these innovative treatments.
http://www.genethon.fr/

IFB

The Institut Français de Bioinformatique (IFB, French Institute of Bioinformatics) is a national service infrastructure in bioinformatics that was created following the call for proposals, "National Infrastuctures in Biology and Health", of the "Investments for the Future" initiative (ANR-11-INBS-0013). This project gathers together the bioinformatics platforms of the main French research organizations, CNRS, INRA, INRIA, CEA and INSERM, as well as CIRAD, the Pasteur and Curie Institutes, and the French universities.
IFB-Core acts as an interface between the bioinformatics community, the life science community that uses the services provided by IFB, and the representatives of the main French scientific research organizations. IFB-core, in its capacity as the IFB hub, is also in charge of maintaining an effective relationship with ELIXIR, the European hub in Hinxton (Cambridgeshire, UK).
https://www.france-bioinformatique.fr/en

I-STEM

Created in 2005 through collaboration between Inserm – Institut National Health and Medical Research – and AFM-Telethon – French Association against Myopathies – I-

Stem is the largest French laboratory research and development dedicated to human pluripotent stem cells, embryonic origin or obtained by reprogramming gene. I-Stem is part of the Biotherapy Institute for Rare Diseases, which includes so far the four centers of research and development funded directly by the AFM Telethon. The specific vocation of I-Stem is to explore all the therapeutic potential of human pluripotent stem cells for applications in patients affected by rare diseases of genetic origin. In this context, two major areas of research are developed. The first one is cell therapy, which aims to replace lost or diseased cells to other cells with the same characteristics, produced in the laboratory from pluripotent stem cells. The second area is pharmacology based on automated screening of large libraries of compounds with therapeutic potential, following modeling of molecular mechanisms associated with diseases, as revealed by the study of pluripotent stem cells from affected donors. I-Stem teams are currently working on a dozen genetic diseases that affect different organs. The Institute also hosts every year many researchers interested in other diseases and provides training and technology support.

http://www.istem.eu/

Orphanet and Orphadata

Orphanet, http://www.orphanet.net, is a unique resource, gathering and improving knowledge on rare diseases so as to improve the diagnosis, care and treatment of patients with rare diseases. Orphanet aims to provide high-quality information on rare diseases, and ensure equal access to knowledge for all stakeholders. Orphanet maintains the Orphanet rare disease nomenclature (ORPHA number/ ORPHA codes), essential in improving the visibility of rare diseases in health and research information systems. Orphanet offers a range of freely accessible services: an inventory of rare diseases and a classification of diseases elaborated using existing published expert classifications; an encyclopaedia of rare diseases in English and French, progressively translated into Dutch, German, Italian, Spanish, Polish and Portuguese; an inventory of orphan drugs at all stages of development; a directory of expert resources, providing information on expert clinics, medical laboratories, ongoing research projects, clinical trials, registries, biobanks and patient organizations in the field of rare diseases, in each of the 34 countries in Orphanet's network and 5 contact points which are managed by the INSERM in Paris. Orphanet datasets are available for research purposes at www.orphadata.org. Orphanet produces ORDO, the Orphanet Rare Diseases Ontology, and HOOM, the HPO-ORDO Ontological Module. Orphadata is an Elixir Core Data Resource. Orphanet and ORDO are IRDIRC Recognized Resources.

http://www.orpha.net ; www.orphadata.org

OrphanDev

OrphanDev is an academic platform specialized in supporting researchers, clinicians and the health industry actors in the development of drugs for rare diseases. This multidisciplinary team supports all projects from private or public sector, throughout the crucial stages of drug development. It provides its scientific and regulatory expertise in Orphan Designation and Protocol Assistance Applications; its logistical and methodological support for clinical trials in rare diseases with a specificity in the patient's recruitment strategy; and its experience in national and European calls for projects. This set of services and tools allows the acceleration of the development of

therapies for rare diseases.
http://www.orphan-dev.org/

PHENOMIN: French National Infrastructure for Mouse Phenogenomics

PHENOMIN aims at developing tools and technologies at the state of the art in the fields of genetic engineering, zootechny and phenotyping. To achieve this, PHENOMIN:

- Participates to different National and International Research programs to improve scientific community knowledge and opportunities.
- Is involved in different scientific networks and consortia to be aware of the last innovations and trends.
- Develops new tools on its own and to address its customers' projects with the best and most appropriate technological options.

http://www.phenomin.fr/

FRANCE GENOMIQUE

France Genomique brings together and mutualises the resources of the most efficient French genomics and bioinformatics platforms. Its ambition is to maintain French research at the highest level of competitiveness and performance in the production and analysis of genomic data. It offers the public and private scientific community the highest level of expertise and skills, as well as project support.

http://www.france-genomique.org/?lang=en

Platform of Integrative Chemical Biology of Strasbourg (PCBIS)

The Platform of Integrative Chemical Biology of Strasbourg (PCBIS) gives access to High Throughput Screening (HTS), chemical libraries and target libraries to academic and private laboratories. The platform evaluates the ADME properties of active molecules.

http://www.pcbis.fr/

RADICO / French National Programme on Rare Disease Cohorts (RAre Disease COhorts)

RaDiCo program aims at endowing France with large epidemiological instruments or "cohorts" in the field of rare diseases. These cohorts are designed to improve the understanding of health determinants of rare diseases and ultimately, to the development of new therapeutic strategies. RaDiCo is coordinated by the French Institute on Health and Medical Research (Inserm). It is funded by the French Health Ministry in the framework of the so-called "Investissements d'Avenir" funding scheme. Based in Paris, RaDiCo offers a clinical research and services platform for the translational research on rare diseases.

http://radico.fr/

BIOTHERAPIES INSTITUTE

The Biotherapies Institute unites the knowledge and expertise of four laboratories, initiated and supported by the AFM-Telethon, world leaders of biotherapies for rare diseases. The objective is to accelerate the development of treatments against rare diseases. The mission of the Biotherapies Institute is to unite the skills, expertise and resources, strengthen the complementarity of resources and better coordinate cross-

functional projects, especially clinical development. 650 researchers, engineers and experts in project management, clinical development, regulatory affairs, technology transfer and 25 000 m2 of laboratory space in Paris, Evry and Nantes: the Biotherapies Institute is a unique and exceptional force for translational research, from fundamental to making treatments available for patients.

http://www.institut-biotherapies.fr/

GERMANY

ChemBioNet: resource network supporting academic chemical biology research

The ChemBioNet was initiated by biologists and chemists from academia who realized the need for interdisciplinary open access platforms to support research projects for systematic usage of small molecules to explore biological systems. This initiative wants to provide chemists with bioprofiles for their unique synthetic molecules and biologists developing unique assay systems, with access to high throughput technologies to identify compounds useful for dosage dependent, temporally or locally controlled interference with biological functions. In summary a novel discipline termed Chemical Biology.

http://www.chembionet.info

Care and Trial Site Registry for neuromuscular and neurodegenerative diseases (CTSR)

The aim of the Care and Trial Site Registry (CTSR) is to help the pharmaceutical industry and clinical investigators select trial sites as well as to help to identify potential partners for upcoming research projects.

The **CTSR** provides information relevant

- to clinical studies (such as personnel, facilities and patient population)
- to the assessment as centres of expertise according to the EUCERD criteria

https://ctsr.uniklinik-freiburg.de

EU-OPENSCREEN

EU-OPENSCREEN, the European infrastructure of open screening platforms for chemical biology provides access to bioactive small molecules. A large collection of diverse compounds, representing the chemical knowledge in Europe, is available for many fields of the life sciences, e.g. human and veterinary medicine, systems biology, biotechnology, agriculture, nutrition. It also offers access to services in all of chemical biology: high-throughput screening, chemical synthesis for hit-optimisation, bio-profiling and in vivo studies, a central database, and training.

http://www.eu-openscreen.de/

Euro-BioImaging

The European Research Infrastructure for Imaging Technologies in Biological and Biomedical Sciences (Euro-BioImaging, EuBI or EuBI ERIC) provides open physical user access to a broad range of state-of-the-art technologies in biological and biomedical imaging for life scientists. In addition, EuBI will offer image data support and training for infrastructure users and providers. The EuBI consists of a set of 29 geographically distributed Node Candidates (specialised imaging facilities) that can grant access to scientists from all European countries and beyond. Currently, researchers can apply to use some of 36 imaging technologies offered through Euro-BioImaging.

http://www.eurobioimaging.eu

Infrafrontier

Infrafrontier, the European infrastructure for phenotyping and archiving of model mammalian genomes provides access to first-class tools and data for biomedical research, and thereby contributes to improving the understanding of gene function in human health and disease using the mouse model. The core service of Infrafrontier comprise the systemic phenotyping of mouse mutants in the participating mouse clinics, and the archiving and distribution of mouse mutant lines by the European Mouse Mutant Archive (EMMA). In addition, Infrafrontier provides specialized services such as the generation of germ-free mice (axenic service) and training in state of the art cryopreservation and phenotyping technologies. https://www.infrafrontier.eu/

West German Genome Center (CCG)

The Cologne Center for Genomics has been selected as the major production site of the newly founded West German Genome Center. Through the accompanying funding provided by the DEG, the CCG will achieve a superior level of capacity, throughput and cost effectiveness in next generation sequencing (NGS)

http://portal.ccg.uni-koeln.de/ccg/index.php

ITALY

Interdepartmental Centre for Stem Cells and Regenerative Medicine (CIDSTEM)

The Interdepartmental Centre for Stem Cells and Regenerative Medicine aims to provide patients with effective therapeutic solutions for severe diseases for which regenerative medicine is either the only or the best therapeutic chance, through ethical and scientific excellence. The GMP facility is devoted to the preparation of epithelial grafts, destined to clinical application in cell therapy and gene therapy and to the development of phase I/II clinical trials based on different types of ATMP, also on external commission.

http://www.cidstem.unimore.it/chi_siamo.html

European Platform on Rare Disease Registration (EU RD Platform)

The EU Rare Disease Platform aims to provide researchers, healthcare providers, patients and policy-markers with a consistent instrument to improve knowledge, diagnosis and treatment of rare diseases. It will make registries' data searchable at EU level and will standardize data collection and data exchange; which will increase the value of each registry and its registration.

https://eu-rd-platform.jrc.ec.europa.eu/

THE NETHERLANDS

EATRIS

EATRIS aims to provide access to a top-academic high-end infrastructure and related academic services for research, translational research expertise and large patient cohorts. EATRIS is expected to facilitate the efficient translation of novel biomedical targets into the development of innovative preventive, diagnostic and therapeutic products, including their early stage clinical evaluation in a coordinated effort to address the "innovation gap" that afflicts both industry and academic science

worldwide.

http://www.eatris.eu/

ECARUCA

ECARUCA is a European-based database, that collects cytogenetic and clinical data of rare chromosomal aberrations from (cyto)genetic centres in Europe and the rest of the world. Many ECARUCA accountholders are also member of the European Cytogeneticists Association (ECA). During recent years there has been an enormous improvement in diagnostic techniques, enabling cytogeneticists to find more and smaller chromosomal aberrations. However, accurate clinical knowledge about rare chromosome disorders is frequently lacking, as a considerable percentage of cases remains unpublished. The resulting gap in clinical knowledge is in sharp contrast with the increasing demand from parents and physicians for reliable information on the disorder of their child or patient.

https://omictools.com/ecaruca-tool

LOVD: Leiden Open (source) Variation Database

LOVD aims to provide a flexible, freely available tool for Gene-centered collection and display of DNA variations. LOVD 3.0 extends this idea to also provide patient-centered data storage and storage of NGS data, even of variants outside of genes. To maintain a high quality of the data stored, LOVD connects with various resources, like HGNC, NCBI, EBI and Mutalyzer. The LOVD team is located in Leiden, The Netherland.

http://www.lovd.nl/3.0/home

SPAIN

SEFALer: Laboratory Animals Phenotyping Network

SEFALer is a service coordinated by CIBERER through several of its research groups, unprecedented in Spain and specialising in phenotyping animal models with specific applications for rare diseases. The SEFALer is a network technology platform which research groups and services (SEFALer units) are joining to carry out work on phenotyping laboratory animals, particularly genetically modified mice. The SEFALer platform is permanently open to having new research groups join.

http://www.ciberer.es/en/platforms/sefaler

RD Connect

RD connect was established in November 2012 as a 6-year project financed by the European Commission under an FP7 grant. That multidisciplinary consortium developed an integrated platform of tools and resources to improve data analysis, sharing and linkage in rare disease research. In May 2017, the consortium decided to continue its work beyond the end of the FP7 funding period (October 2018) as the RD-Connect Community. RD-connect is an IRDiRC Recognized recource. RD-Connect's scientific coordination is moved from Newcastle University to CNAG-CRG in Barcelona. RD-Connect's objectives are:

- Harmonisation and development of common standards for databases and patient registries for rare diseases by collaborating internationally to implement common registry infrastructure and data elements across a federated system.
- Harmonisation and development of common standards and a common catalogue for rare disease biobanks that collect and provide standardised, quality-controlled

biomaterials for translational research.

- Development of a suite of clinical bioinformatics tools, including data mining and knowledge discovery tools for analysis and integration of molecular and clinical data to discover new disease genes, pathways and therapeutic targets.
- Development of an integrated platform to host the processed data from the research projects Neuromics, EURenOmics and future IRDiRC projects.
- Development of best ethical practices for balancing patient-related interests associated with rare disease research using databases/registries, biobanks and omics databases, engaging with relevant stakeholders, including patient organisations, clinical and research networks, legislators and policymakers and the pharmaceutical industry.
- Development of a proposal for an expedient regulatory framework for linking of medical and personal data related to rare disease on a European and global level.
- Ensuring access to project results and broad and global impact in science, diagnostics and translational research including industrial collaborations.

http://rd-connect.eu/

SWEDEN

INCF-DataSpace

The purpose of the INCF Dataspace is to enable collaboration between researchers through the sharing of neuroscience data, text, images, sounds, movies, models and simulations.

https://www.incf.org/network

UNITED-KINGDOM

COMET (Core Outcome Measures in Effectiveness Trials)

The COMET Initiative brings together people interested in the development and application of agreed standardised sets of outcomes, known as 'core outcome sets'. These sets represent the minimum that should be measured and reported in all clinical trials of a specific condition, and are also suitable for use in clinical audit or research other than randomised trials. The existence or use of a core outcome set does not imply that outcomes in a particular trial should be restricted to those in the relevant core outcome set. Rather, there is an expectation that the core outcomes will be collected and reported, making it easier for the results of trials to be compared, contrasted and combined as appropriate; while researchers continue to explore other outcomes as well. COMET aims to collate and stimulate relevant resources, both applied and methodological, to facilitate exchange of ideas and information, and to foster methodological research in this area.

http://www.comet-initiative.org/

DECIPHER (Database of Chromosomal Imbalance and Phenotype in Humans Using Ensembl Resources)

DECIPHER is an interactive web-based database which incorporates a suite of tools designed to aid the interpretation of submicroscopic chromosomal imbalance. DECIPHER enhances clinical diagnosis by retrieving information from a variety of bioinformatics resources relevant to the imbalance found in the patient. Known and predicted genes within an aberration are listed in the DECIPHER patient report,

common copy-number changes in healthy populations are displayed and genes of recognized clinical importance are highlighted. DECIPHER is an IRDIRC recognized resource.
http://decipher.sanger.ac.uk/

Diagnostic Mutation Database (DMuDB)

The Diagnostic Mutation Database is a secure repository of clinical quality variant data collected from diagnostic genetics laboratories. Access to DMuDB is available by annual laboratory subscription, and must be for diagnostic purposes only. DMuDB is accessed through a secure website; the data can also be accessed through a graphical browser, which is the preferred method for viewing data and also allows access to other mutation databases. Access to DMuDB is not permitted for any purpose other than the investigation and interpretation of patient results in order to provide a diagnosis. Researchers or healthcare professionals wishing to query the database and obtain data that may be held there send a request, which will be passed on to laboratories that have submitted relevant data. It may be possible for data to be shared for research and other non-diagnostic purposes but this must be done with express permission of the submitting laboratory and with data transfer agreements in place. The web site is a private site operated and maintained by Certus Technology. Access to the site is restricted. The DMuDB is located in Manchester, United-Kingdom.
https://secure.dmudb.net/ngrl-rep/Home.do

EBI (European Bioinformatics Institute)

The EBI is part of EMBL, Europe's flagship laboratory for the life sciences. EMBL-EBI provides freely available data from life science experiments covering the full spectrum of molecular biology. While they are best known for their provision of bioinformatics services, about 20% of the institute is devoted to basic research. An extensive training program helps researchers in academia and industry to make the most of the incredible amount of data being produced every day in life science experiments. The EBI is a non- profit, intergovernmental organisation funded by EMBL member states. The 500 staff from 43 nationalities work on the Wellcome Trust Genome Campus in Hinxton, Cambridge in the United Kingdom.

http://www.ebi.ac.uk

The EBI coordinates several projects which are very relevant for RD: The project **Cosmos**, Coordination of Standards in Metabolomics, brings together European data providers to set and promote community standards that will make it easier to disseminate metabolomics data through life science e-infrastructures.

http://cosmos-fp7.eu/

The European Genome-Phenome Archive (EGA) is designed to be a repository for all types of sequence and genotype experiments, including case-control, population, and family studies. EGA provides a service for the permanent archiving and distribution of personally identifiable genetic and phenotypic data resulting from biomedical research projects. Data at EGA was collected from individuals whose consent agreements authorise data release only for specific research use to bona fide researchers. Strict protocols govern how information is managed, stored and distributed by the EGA project.
https://www.ebi.ac.uk/ega/home

ELIXIR

ELIXIR is an intergovernmental organisation that brings together life science resources from across Europe. These resources include databases, software tools, training materials, cloud storage and supercomputers. The goal of ELIXIR is to coordinate these resources so that they form a single infrastructure. This infrastructure makes it easier for scientists to find and share data, exchange expertise, and agree on best practices. Ultimately, it will help them gain new insights into how living organisms work. ELIXIR includes 23 member states and over 220 research organisations. It was founded in 2014, and is currently implementing its first five-year scientific programme. The scientific and technical activities in ELIXIR are run by five Platforms (Data, Tools, Interoperability, Compute and Training) and four Use Cases (Human Data, Rare Disease, Marine Metagenomics and Plant Sciences). Platforms bring together experts to define the strategy and provide services in a particular area e.g. training, data. Use Cases bring together scientists who work in a specific domain (e.g. Plant Sciences). These scientists develop services targeted for their own domains. They also give feedback on the services the Platforms provide, to make sure that the services solve real world problems.

http://www.elixir-europe.org/

International Mouse Phenotype Consortium (IMPC)

The International Mouse Phenotype Consortium is a research infrastructure of excellence for translational research and functional genomics.
After completing the mouse genome sequence, an international consortium was developed, the International Knock-out Mouse Consortium (IKMC) to systematically generate mutant ES cells for every gene in the mouse genome (20,000 plus genes).
The IMPC builds on the efforts of International Knock-out Mouse Consortium (IKMC) to produce knockout mice and carry out high-throughput phenotyping of each line in order to determine the function of every gene in the mouse genome. These mice are preserved in repositories and made available to the scientific community representing a valuable resource for basic scientific research as well as generating new models for human diseases.
https://www.mousephenotype.org

Instruct-ERIC

Instruct-ERIC is a pan-European distributed research infrastructure making high-end technologies and methods in structural biology available to users. Instruct-ERIC promotes innovation in biomedical science and operates on a non-economic basis within the scope of the ERIC Regulation.

Instruct-ERIC provides open access to cutting edge structural biology, specifically supporting research that uses intergrated approaches and technologies. It operates with the following principles: a) scientific excellence is our priority in the services we provide and the research we support; b) transparency, equality and legality is the cornerstone of our operational model.

https://instruct-eric.eu/

Please note that all data presented in this report are available for download at www.orphadata.org

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

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