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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 (Biobanking and Biomolecular Resources Research Infrastructure)

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.eu

BELGIUM

CHD wiki

The CHD wiki bioinformatics tool will combine the sequence data and expression data generated by the human and mouse studies with morphology and literature to prioritize genes and generate hypotheses. The Wiki, which will contain existing and new genetic and environmental knowledge on heart development, will be an aid for many groups working on cardiovascular development and a novel means of disseminating our findings.

http://homes.esat.kuleuven.be/~bioiuser/chdwiki/index.php/Main_Page

EuroGentest

EuroGentest is a project funded by the European Commission to harmonize the process of genetic testing, from sampling to counseling, across Europe. The ultimate goal is to ensure that all aspects of genetic testing are of high quality thereby providing accurate and reliable results for the benefit of the patients. It contributes to the continuous improvement and implementation of new technologies in genetic testing.

www.eurogentest.org

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/about-us/aims-mission

FRANCE

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 ERINHA infrastructure will provide 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) will promote the harmonization of biosafety and biosecurity procedures, will develop standards for the management of biological resources, diagnosis of group 4 pathogens, and training of BSL4 labs users.

http://www.erinha.eu/

French National Rare Disease Database (BNDMR)

The French National Rare Disease Database aims at structuring a national database containing clinical and, subsequently, in all likelihood, biological and therapeutic data on rare disease based on results collected in the centres of expertise and in the disease registries. It is ultimately destined to be interfaced with specific cohort monitoring data (in particular the RADICO) cohort and with other medical and administrative databases. A minimum set of data to be gathered has been decided in cooperation with the reference centres, structured into sectors, INSERM and INVS, taking into account the recommendations issued at the European level. Data collection about rare diseases is done using the Orphanet nomenclature.

http://www.isyrare.fr/isy-rare/

Mouse Clinical Institute - MCI

The Mouse Clinical Institute is a research infrastructure of excellence for translational research and functional genomics. Founded in 2002 by Pierre Chambon, operated by Inserm, CNRS and the University of Strasbourg and supervised by GIE-CERBM, it provides a comprehensive set of specialised services to academic and industrial users and is a major player in the European post-genomics era programs. The close interaction with the IGBMC strongly contributes to the development and design of new techniques and functional assays. The ICS combines the capacity of generating mutant mice on a large scale with a high-throughput and comprehensive phenotypic analysis of the animals. The Institute's phenotyping platforms are adapted for the study of genetically engineered mouse models and genetic reference populations but can also be used for preclinical studies including the validation of therapeutic targets as well as pharmacological and toxicological studies in the mouse.

http://www.ics-mci.fr/en/about-ics/institute/

Orphanet

Orphanet is the reference portal for information on rare diseases (which include all mendelian diseases) and orphan drugs, for all audiences. Orphanet's aim is to help improve the diagnosis, care and treatment of patients with rare diseases. It offers a range of freely accessible services: An inventory of rare diseases and a classification of diseases elaborated using existing published expert classifications; a encyclopaedia of rare diseases in English and French, progressively translated into German, Italian, Spanish and Portuguese; an inventory of orphan drugs at all stages of development; a directory of expert resources, providing information on expert clinics, medical laboratories, on-going research projects, clinical trials, registries, biobanks and patient organizations in the field of rare diseases, in each of the 38 countries in Orphanet's consortium which is managed by the INSERM in Paris. The Orphanet dataset is freely available for research purpose at www.orphadata.org

http://www.orpha.net

PHENOMIN: French National Infrastructure for Mouse Phenogenomics

The French National Infrastructure for Mouse Phenogenomics constitutes a unique distributed resource for the creation, the care, the phenotyping, the distribution and archiving of animal models for academics and private corporations.

http://www.phenomin.fr/

Platform Mutations Initiative

The Platform Mutations Initiative is a platform of high throughput sequencing for the identification of mutations, settled at Genoscope. In 2011 five other national public and private high throughput sequencing platforms have joined this action, leading to the rise of a network of six platforms of high expertise and capacity. The network aims at meeting the growing needs of the scientific community to use this new technology for the identification of genes involved in human diseases, with a particular focus on monogenic diseases. The goal of the open call for proposals is to support research projects aimed at identifying – by the use of high throughput exome sequencing and/or region specific sequencing - genes involved in rare diseases whose molecular basis remains unknown.

http://www.cns.fr/spip/High-throughput-sequencing-and.html

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 (RAre DIsease COhorts)

The RADICO Project is to form rare disease patient cohorts (in the epidemiological sense of the term). The project, which is expected to include over 250,000 patients in France, is premised on the creation of a RD platform designed to unite existing strengths on the field: 131 reference centres, 501 skills centres, molecular diagnosis laboratories, CRBs in connection with research laboratories, technological platforms and patient associations. This platform, which will in particular be responsible for collecting the data necessary for epidemiological and public health studies, is expected to stimulate the emergence of clinical and translational research programmes in the field of rare diseases, all the while anticipating future needs, such as the integration of data from "omics" approaches and systems biology.

http://radico.isyrare.fr/

GERMANY

chILD better together – European Management Platform for Childhood Interstitial Lung Disease

The chILD EU aims to better understand the natural course, risk factors, treatments and reasons for the development of childhood interstitial lung disease (chILD). It will collect and analyse details of symptoms and quality of life, clinical data and also biological material in a Register and Biobank. In the long run, this Register will serve the improved understanding of the disease and will lead to the development of new and effective approaches to treatment.

http://www.klinikum.uni-muenchen.de/Child-EU/en/index.html

EUMINAfab

EUMINAfab is a European Research Infrastructure offering open access to state of the art of multimaterial micro and nanotechnologies. By combining scientific expertise with technological capabilities, EUMINAfab provides innovative and efficient solutions to your challenges in the area of micro and nanofabrication of functional structures and devices out of a knowledge-based multimaterials' repertoire.

http://www.euminafab.eu/

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

Euro-BioImaging is a pan-European infrastructure project whose mission is to build a distributed imaging infrastructure across Europe that will provide open access to innovative biological and medical imaging technologies for European researchers. The infrastructure established by Euro-BioImaging will consist of a set of geographically distributed but strongly interlinked imaging facilities (Euro-BioImaging Nodes), which will be selected among the leading European imaging facilities based on an independent evaluation process.

http://www.eurobioimaging.eu/

European Sequencing and Genotyping Infrastructure (ESGI)

ESGI, the European Sequencing and Genotyping Infrastructure, aims at the integration of world class high-throughput sequencing and genotyping facilities. The project in particular provides access to sequencing and genotyping technologies as well as data analysis methodologies to the scientific community, for a broad range of genetic and systems biology studies using well-phenotyped samples, for example those derived from standardized European biobanks and animal facilities.

http://www.esgi-infrastructure.eu/

Human Phenotype Ontology (HPO)

The Human Phenotype Ontology aims to provide a standardized vocabulary of phenotypic abnormalities encountered in human disease. The HPO was initially developed using information from Online Mendelian Inheritance in Man (OMIM), which is a hugely important data resource in the field of human genetics and beyond. The HPO is currently being developed using information from OMIM and the medical literature and contains approximately 10,000 terms. Over 50,000 annotations to hereditary diseases are available for download or can be browsed using the PhenExplorer. The HPO is now being developed in collaboration with members of the OBO Foundry (Open Biological and Biomedical Ontologies), and logical definitions for HPO terms are being developed using PATO and a number of other ontologies including the FMA, GO, ChEBI, and MPATH. The HPO can be used for clinical diagnostics in human genetics (Phenomizer), bioinformatics research on the relationships between human phenotypic abnormalities and cellular and biochemical networks, for mapping between human and model organism phenotypes, and for providing a standardized vocabulary for clinical databases, among many other things. There exists a webpage for every HPO-term. The HPO project encourages input from the medical and genetics community with regards to the ontology itself and to clinical annotations.

http://www.human-phenotype-ontology.org/index.php/hpo_home.html

Infrafrontier

Infrafrontier, the European infrastructure for phenotyping and archiving of model mammalian genomes provide the facilities and resources for the phenotyping of medically relevant mouse models, and for the archiving and dissemination of those models. Mice indeed constitute a model system to understand the molecular basis of health and disease in humans, due in particular to the high (> 95 %) similarity of genes with humans.

https://www.infrafrontier.eu/

IRELAND

European Surveillance of Congenital Anomalies (Eurocat)

EUROCAT is a network of population-based registries for the epidemiological surveillance of congenital anomalies, covering 1.7 million births in 21 countries of Europe.

http://www.eurocat-network.eu/

QualityNano

QualityNano is a Research Infrastructure for nanosafety assessment. This four year project integrates 28 top European analytical & experimental facilities in nanotechnology, medicine and natural sciences with the goal of developing and implementing best practice and quality in all aspects of nanosafety assessment.

http://www.qualitynano.eu/

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/en/home.html

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/SitePages/home.aspx

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.

http://umcecaruca01.extern.umcn.nl:8080/ecaruca

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

Prime-xs

PRIME-XS provides European Union funded access to an infrastructure of state-of-the-art proteomics technology to the biological and biomedical research community in Europe.

http://www.primexs.eu/

SPAIN

Spanish Rare Diseases Registries Research Network (SpainRDR)

The SpainRDR is a project financed by the Institute of Health Carlos III (ISCIII). It aims to build the National Rare Diseases Registry in Spain based on the input of two different strategies: patient registries addressed to patient outcome research and population-based registries addressed to epidemiologic research and social and health systems planning.

https://spainrdr.isciii.es/en/Pages/default.aspx

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.

http://www.incf.org/resources/data-space

UNITED-KINGDOM

BioMedBridges

BioMedBridges is a joint effort of ten biomedical sciences research infrastructures on the ESFRI roadmap. Together, the project partners will develop the shared e-infrastructure—the technical bridges—to allow data integration in the biological, medical, translational and clinical domains and thus strengthen biomedical resources in Europe.

http://www.biomedbridges.eu

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/about/overview

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.

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 project 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://www.cosmos-fp7.eu/

The project **Impact** for Improving protein annotation and coordination through technology contributes to the further development and enhancement of the 'InterPro' database of predictive protein signatures, i.e. entities that are used to recognize a particular domain or protein family, and its other contributing databases.

http://www.ebi.ac.uk/impact

ELIXIR

ELIXIR aims at building an effective data infrastructure for biological information that will encompass hundreds of existing biological databases in Europe, ranging from major core datasets – such as the vertebrate genomes in Ensemble – to very small specialist collections overseen on a part-time basis by individual researchers. ELIXIR can be considered a networking effort between national nodes with the addition of a major U.K.-funded new hub, with each of these facilities operating according to national rules. Collaboration Agreements will set out the exact services each node will provide and the relevant KPIs are expected to be developed out of these collaboration agreements.

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

GEN2PHEN

The GEN2PHEN project aims to unify human and model organism genetic variation databases towards increasingly holistic views into Genotype-To-Phenotype (G2P) data, and to link this system into other biomedical knowledge sources via genome browser functionality. The project establishes the technological building-blocks needed for the evolution of today's diverse G2P databases into a future seamless G2P biomedical knowledge environment, by the projects end. This will consist of a European-centred but globally-networked hierarchy of bioinformatics GRID-linked databases, tools and standards, all tied into the Ensembl genome browser.

http://www.gen2phen.org/about-gen2phen/project-summary-and-objectives

Instruct

Instruct, the Integrated Structural Biology Infrastructure, consists of distributed centres for structural biology. All centres maintain a set of core technologies (e.g. protein production, NMR, crystallography, microscopy) accessed by users to obtain multi-scale structural data.

http://www.structuralbiology.eu/

RD-CONNECT

RD-Connect will develop a critical mass for harmonisation and provide a strong impetus for a global trial-ready infrastructure ready to support the IRDiRC goals for diagnostics and therapies for RD in close collaboration with the successful A/B projects. It will build on and transform the current state-of-the-art across databases, registries, biobanks, bioinformatics, and ethical considerations to develop a quality-assured and comprehensive integrated hub/platform in which complete clinical profiles are combined with -omics data and sample availability for RD research. The integrated, user-friendly RD-Connect platform, built on efficient informatics concepts already implemented in international research infrastructures for large-scale data management, will provide access to federated databases/registries, biobank catalogues, harmonised -omics profiles, and cutting-edge bioinformatics tools for data analysis.

http://rd-connect.eu/

SLING: Serving Life-science Information for the Next Generation

SLING makes available a comprehensive range of databases and services from its five Partners: the European Molecular Biology Laboratory European Bioinformatics Institute (EMBL-EBI), the Swiss Institute of Bioinformatics (SIB), the BRENDA database at the Technische Universität Braunschweig (TUBS), the European Patent Office (EPO), and Enzymeta GmbH.

http://www.sling-fp7.org/

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