Orphanet Report Series

Rare Diseases collection

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List of Research Infrastructures useful to Rare Diseases in Europe

By country of location

www.orpha.net

Inserm

La science pour la santé

From science to health

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AUSTRIA

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

BBMRI-ERIC is a distributed research infrastructure of biobanks and biomolecular resources. For its Member States, it provides expertise and services on a non-economic basis and facilitates access to collections of partner biobanks and biomolecular resources. BBMRI-ERIC is established for an unlimited period of time.

BBMRI-ERIC works towards connecting biobanks and researchers in Europe in order to facilitate the use of samples/data collected in Europe for the benefit of human health. It helps researchers to find the samples and data they need for their research through better utilisation of quality-defined biobanks and their samples/data in an ethically and legally compliant manner. BBMRI-ERIC helps biobanks with their visibility, quality development and usability and advise them onethical and legal questions such as the European regulation framework.

BBMRI offers quality management services, support with ethical, legal and societal issues, and a number of online tools and software solutions for biobankers and researchers.
http://www.bbmri-eric.eu/

BELGIUM

European Organisation for Research and Treatment of Cancer (EORTC)

The European Organisation for Research and Treatment of Cancer is an independent, non-profit cancer research organisation, whose mission is to coordinate and conduct international translational and clinical research to improve the standard of cancer treatment for patients. EORTC aims ultimately to increase people's survival and quality of life by testing new therapeutic strategies based on existing drugs, surgery and radiotherapy. We also help develop new drugs and approaches in partnership with the pharmaceutical industry and in patients' best interests.

http://www.eortc.org/

FRANCE

APTEEUS

APTEEUS is a Biotech specialized in drug discovery and repositionin in the field of rare monogenic diseases for which it is possible to measure the cause of the disease on cells of patients in culture. APTEEUS is currently particularly committed to hereditary metabolic diseases and genodermatoses. One of the objectives of APTEEUS is to identify new avenues of treatment for these diseases and less rare diseases. APTEEUS offers access to automated cell screening technology in partnership with patient organizations, research teams and pharmaceutical companies. Through use of primary cells directly from patients, APTEEUS is able to reproduce in vitro the physiopathology of a patient's disease and to measure the effect of specific molecules.
http://apteeus.fr/en/

Biotherapies Institute

The Biotherapies Institute unites the knowledge and expertise of three laboratories (Genethon, Institute of Myology, and I-Stem), initiated and supported by the AFM-Telethon with the objective 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.
www.institut-biotherapies.fr

SeqOIA

SeqOIA is one of of the two national high throughput sequencing platforms selected within the framework of the **Plan France médecine génomique 2025 (**the French initiative for genomic medicine - led by AVIESAN) set up to ensure that everyone has access to new technologies in genomic medicine in an equitable manner throughout the country. SeqOIA's mission is to allow equal access for all to genomic analyses so that cancer, rare disease, or common disease patients can benefit from the latest predictive and personalised medicine breakthroughs.
https://laboratoire-seqoia.fr
https://pfmg2025.aviesan.fr/le-plan/plateformes/

AURAGEN

AURAGEN is one of of the two national high throughput sequencing platforms selected within the framework of the **Plan France médecine génomique 2025 (**the French initiative for genomic medicine - led by AVIESAN) set up to ensure that everyone has access to new technologies in genomic medicine in an equitable manner throughout the country. AURAGEN provides access to high throughput sequencing to all actors involved in cancerology and management of rare diseases in the Auvergne Rhône-Alpes region.
https://www.auragen.fr

https://pfmg2025.aviesan.fr/le-plan/plateformes/

European Clinical Research Infrastructures Network (ECRIN)

The European Clinical Research Infrastructure Network (ECRIN) is a not-for-profit intergovernmental organisation that supports the conduct of multinational clinical trials in Europe. ECRIN acquired in 2013 the legal status of a European Research Infrastructure Consortium (ERIC). ECRIN provides tailored support and tools to facilitate multinational trials preparation and implementation and to overcome the various trial challenges (e.g. regulatory and ethical requirements, management and funding issues). Support areas include, among others, the preparation of applications for funding, protocol evaluation, trial management, and quality assurance. ECRIN focuses on independent trials across all disease areas and topics and its partners include clinical trial units or clinical research centres, academic institutions, investigators and sponsors, policymakers, patients and communities.
http://www.ecrin.org/

European Research Infrastructure on Highly Pathogenic Agents (ERINHA)

ERINHA-ASIBL (European Research Infrastructure on Highly Pathogenic Agents) is a pan-European Research Infrastructure dedicated to the study of High-consequence pathogens of Risk Group 4 (RG4). As a distributed Research infrastructure, it brings together leading European Bio-Safety Level 4 (BSL4) facilities and national research institutes with longstanding experience of research in this field. Such a coordinated approach is vital in a context marked by frequent globalization of infectious diseases with high risk for public health, society and economy.
The over-arching goal of ERINHA's Infrastructure research concept is to provide capacities to conduct projects which are broad in scope, ambition and require a range of capabilities inside and outside BSL4 facility that no single facility can provide on its own.
The access to the ERINHA is organized through its Central Coordinating Unit, hosted in Paris.
Since 2019 ERINHA is operational and provides access to its full range of services including the state-of- art high containment facilities.
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 - French Institute of Bioinformatics

The *Institut Français de Bioinformatique* (IFB, French Institute of Bioinformatics) is a national service infrastructure providing to life sciences and bioinformatics communities an access to vital research resources, project realisation support and the possibility to participate in ambitious national and international projects. Offererd services include data, software tools, training, support in biology research project, and access to an IT infrastructure dedicated to life sciences. The IFB federates 31 bioinformatics platforms affiliated to the main French research institutions.

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.

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IFB-core, in its capacity as the IFB hub, is the French national node of ELIXIR, the European hub in Hinxton (Cambridgeshire, UK).
https://www.france-bioinformatique.fr

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 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 and data 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: (1) an inventory of rare diseases and a classification of diseases elaborated using existing published expert classifications; (2) an encyclopaedia of rare diseases in English and French, progressively translated into Dutch, German, Italian, Spanish, Polish, Portuguese, and Czech; (3) an inventory of orphan drugs at all stages of development; (4) and 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 40 countries in the Orphanet Consortium which is managed by the INSERM in Paris. Orphanet datasets are available for research purposes at www.orphadata.org. Orphadata provides the scientific community with comprehensive, quality data sets related to rare diseases and orphan drugs from the Orphanet knowledge base, in reusable formats. Orphanet produces ORDO, the Orphanet Rare Diseases Ontology, and HOOM, the HPO-ORDO Ontological Module. Orphadata, part of the French node of ELIXIR, 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 is the National Infrastructure in Biology and Health granted in 2011 by the Investissements d'Avenir program. The objective of PHENOMIN is to develop and maintain according to the community's needs, innovative, standardized and parallel methodologies, functional analysis and comparative genetics for rodent models. PHENOMIN allows research teams to carry out integrated comparative functional analysis and to ensure a better translation of results to human. PHENOMIN currently aims (1) to develop or improve tools to advance basic research in mammals and better respond to the ethical demands of society on the use of animals in research (Welfare and Ethic), (2) to improve the impact of preclinical studies on the development of therapies in humans with new mouse models 3.0, and (3) to decipher the function of the mammalian genome. PHENOMIN aims at developing tools and technologies at the state of the art in the fields of genetic engineering, zootechny and phenotyping.
Phenomin is part of INFRAFRONTIER, European Research Infrastructure for the phenotyping and genotyping of mammal models, and of IMPC (International Mouse Phenotyping Consortium)
http://www.phenomin.fr/

PCBIS - Platform of Integrative Chemical Biology of Strasbourg

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/

Rare Disease Cohorts (RaDiCo) / French National Programme on Rare Disease Cohorts

RaDiCo program aims at endowing France with large epidemiological instruments, or "cohorts", in the field of rare diseases. These cohorts allow us to better understand health determinants, improve medical practices and public health policies, feed information to the French National Registry for Rare Diseases (BNDMR), and develop research programmes. The ultimate goal of RaDiCo is to improve the support to patients and their families. 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/

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 (Chemical Biology). 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.
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) and to the assessment as centres of expertise according to the EUCERD criteria. CTSR is an IRDiRC recognized resource.
https://ctsr.uniklinik-freiburg.de

Cologne Center for Genomics (CCG)

The center has a superior infrastructure for all kinds of sequencing, gene mapping and whole genome association studies in complex diseases as well as the validation of copy number variations. It offers various gene expression profiling platforms that may be used for high-throughput production and cross platform validations.
http://portal.ccg.uni-koeln.de/ccg/index.php

Competence Centre for Genomic Analysis (CCGA) Kiel

The CCGA is one of four DFG-funded Next Generation Sequencing (NGS) centres with the mission to strengthen the biological/medical infrastructure in Germany. Their sequencing services comprise short and long read sequencing technologies as well as bioinformatics support. The CCGA offers Illumina sequencing in different throughputs and read lengths on two NovaSeq6000, one HiSeq4000, one NextSeq500 and two MiSeqs. Long read sequencing can be done on the Pacific Biosciences Sequel system.
The CCGA offers a broad range of methods for RNA e.g. RNAseq, Single Cell transcriptomics or isoform sequencing as well as DNA e.g. whole genome sequencing, amplicon sequencing or exome sequencing.
https://www.ccga.uni-kiel.de/

DRESDEN-concept Genome Center (DcGC)

The DcGC constitutes a shared technology resource that offers the infrastructure for a broad range of state-of-the-art genomic technologies. It provides strong expertise in the areas of: (1) Single cell applications; (2) De novo genome sequencing and assemblies; and (3) A broad range of short read-based sequencing application. The DcGC is a joint sequencing center between the TU Dresden and the MPI-CBG, and one of four DFG-funded German competence centers for next generation sequencing (NGS-CC). These next generation sequencing (NGS) and associated technologies are available to researchers in the area of life sciences on the Dresden campus.
https://genomecenter.tu-dresden.de/

EU-OPENSCREEN

EU-OPENSCREEN ERIC is a non-profit research infrastructure, which operates on a global scale. **It is** funded by European countries (Czech Republic, Denmark, Finland, Latvia, Norway, Poland, Spain with Germany) and offer**s** access to academic high-throughput screening facilities and medicinal chemistry groups in these countries. Scientists from academia and industry can implement their screening projects at EU-OPENSCREEN´s partner laboratories using their general purpose diversity compound collection, the European Chemical Biology Library (ECBL), which is composed of 100.000 commercial compounds plus a growing number of compounds collected from academic chemistry groups. Hit optimisation can be done at their medicinal chemistry sites.
http://www.eu-openscreen.de/

Euro-BioImaging

Euro-BioImaging is the European *landmark* research infrastructure for biological and biomedical imaging as recognised by the European Strategy Forum on Research Infrastructures (ESFRI). Through Euro-BioImaging, life scientists can access imaging instruments, expertise, training opportunities and data management services that they might not find at their home institutions or among their collaboration partners. All scientists, regardless of their affiliation, area of expertise or field of activity can benefit from these pan-European open access services.
The fully distributed infrastructure of Euro-BioImaging is coordinated by a Hub and offers its services via 21 internationally imaging facilities called Nodes, which are located across 8 countries and the European Molecular Biology Laboratory, EMBL. The Euro-BioImaging Hub consists of Finland (Turku) as the headquarters, EMBL as the community-specific section for biological imaging and Italy (Torino) as the community-specific section for biomedical imaging.
http://www.eurobioimaging.eu

European Sequencing and Genotyping Infrastructure (ESGI)

On 1 February 2011, a consortium of leading European research institutes begun developing a European infrastructure for genome sequencing and genotyping. Led by the

Max-Planck-Institute for Molecular Genetics in Germany and funded by the European Commission, the European Sequencing and Genotyping Infrastructure (ESGI) brings together partners from Austria, France, Germany, Spain, Sweden and the United Kingdom. ESGI aims to solidify Europe's position as a world leader in genetics, genomics and systems biology research. The ESGI pools the efforts of leading European genomics and bioinformatics facilities to ensure that a larger scientific community can access new genomic technologies in an ethical way and use the latest analytic tools.
https://cnag.cat/projects/esgi

German Cancer Research Center Genomics & Proteomics Core Facility

The Genomics and Proteomics Core Facility (GPCF) at the Deutsches Krebsforschungszentrum (DKFZ) is a central research infrastructure providing access to sophisticated and expensive key technologies that are of critical relevance in biomedical science and which would otherwise not be accessible to the center's research groups. Its services are open to external users subject to available capacities.
https://www.dkfz.de/gpcf/home

Infrafrontier

INFRAFRONTIER is the European Research Infrastructure for the generation, phenotyping, archiving and distribution of model mammalian genomes. The INFRAFRONTIER Research Infrastructure 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 services 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 specialised 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/

NCCT - NGS Competence Center Tübingen

The NCCT (NGS Competence Center Tübingen) was conceptualized in 2017 and approved in early 2018, together with four other national centers by the DFG (German Research Foundation) to strengthen the use of high-throughput sequencing methods in Germany. The center offers service on the latest Illumina Next Generation Sequencer NovaSeq6000, Oxford Nanopore Technologies Sequencer PromethION and Pacific Bioscience Sequel.
https://ngs-kn.de/ngs-competence-center-tubingen-ncct/

WGGC - West German Genome Center

The West German Genome Center (WGGC) is one of four national Next Generation Sequencing (NGS) Competence Centers funded by the DFG, the national research council. The WGGC takes a leading role in harmonizing standards to serve the scientific community by providing NGS services. The WGGC is a collaborative network of universities and institutes located in the west of Germany and activities started in January 2019. The WGGC has three production sites where sequencing is performed, located in Cologne, Bonn and Düsseldorf and counts on additional expertise on NGS technologies from the universities in Aachen, Duisburg/ Essen, Saarbrücken as well as the DZNE, BfARM and MPIPZ. Centralized NGS services are complemented with existing decentralized NGS- and bioinformatics expertise of the WGGC partners. Such expertise covers most aspects of NGS research, encompassing capabilities in human and medical genetics as well as onco- and plant genomics.
https://wggc.de

ITALY

CIDSTEM - Interdepartmental Centre for Stem Cells and Regenerative Medicine

The Interdepartmental Center for Stem Cells and Regenerative Medicine (CIDSTEM) of the University of Modena and Reggio Emilia is based at the "Stefano Ferrari" Center for Regenerative Medicine. CIDSTEM belongs to the Life Sciences platform of the Emilia-Romagna High Technology Network and was created to bring industrial research supply and demand closer together. CIDSTEM 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

European Platform on Rare Disease Registration (EU RD Platform)

The main objective of the European Platform on Rare Disease Registration (EU RD Platform) is to cope with the enormous fragmentation of rare disease (RD) patients data contained in hundreds of registries across Europe. The Platform makes RD registries' data searchable and findable, thus increasing visibility for each registry, maximizing the value of each registry's information and enabling extended use and re-use of registries' data. This is ensured by the European RD Registry Infrastructure (ERDRI), which supports existing registries and the creation of new registries.
The EU RD Platform sets EU-level standards for RD data collection and data exchange and provides training on the use of the tools and services offered.

In addition to ERDRI, the EU RD Platform includes a data repository composed of the European RD Registry Data Warehouse (under preparation), the JRC-EUROCAT Central Registry and the JRC-SCPE Central Registry.

The EU RD Platform is open to all RD registries. Its final goal is to act as a knowledge generation centre benefiting healthcare providers including European Reference Networks, researchers, patients and policy-makers in the common effort to improve diagnosis and treatment for patients living with a rare disease.
https://eu-rd-platform.jrc.ec.europa.eu/

THE NETHERLANDS

EATRIS – European Infrastructure for Translational Medicine

The European Infrastructure for Translational Medicine (EATRIS) is a non-profit European Research Infrastructure Consortium (ERIC). EATRIS's organisational model is based on country membership. Institutions are selected within each country on the basis of their track record in public-private collaboration in the translational development and their multidisciplinary teams of leading academic experts, high-end research facilities, production laboratories and licenses. EATRIS provides researchers with guidance and support in the steps to be taken in translational research and developing new innovations (e.g. drug, vaccine or diagnostic development programme), and what expertise and technologies are suitable to perform these steps, by means of clinical, biological and technological expertise available within the infrastructure. Subsequently, EATRIS matches the need with the capabilities within the infrastructure. In this way, EATRIS facilitates collaboration among academics, physicians, and developers.
http://www.eatris.eu/

Leiden Open (source) Variation Database (LOVD)

The Leiden Open (source) Variation Database (LOVD) provides a flexible, freely available tool for genomic variant and phenotype collection, display and curation. LOVD allows both patient-centered and gene-centered views. LOVD is open source, released under the GPL license, and is actively being improved. On the server in Leiden, LOVD offers free hosting and support of LOVD-powered gene variant databases. To maintain a high quality of the data stored, LOVD connects with various resources, like HGNC, NCBI, EBI and Mutalyzer.
http://www.lovd.nl/3.0/home

SPAIN

RD Connect

RD-Connect began as a six-years project (2012 - 2018) funded by the European Commission, under the FP7 grant to create a unique infrastructure for rare disease research. In the collaborative work of partners in Europe and beyond, RD-Connect developed an integrated platform, which is now available free to use by scientists and clinicians in Europe and around the world. RD-connect is an IRDiRC Recognized recource.
RD-Connect developed the **RD-Connect Genome-Phenome Analysis Platform** is a user-friendly tool that lets researchers and clinicians analyse DNA sequencing data and link them to clinical information (i.e. description of symptoms of the patients). This helps to understand how genes determine the disease symptoms and their severity, make accurate genetic diagnosis and design better treatment.
RD-Connect has also developed several bioinformatic tools, integrated in the Platform to make the analysis of patient data and interpretation of DNA sequencing results easier.
To help researchers who deal with patients' clinical information and biosamples, RD-Connect has created the **Registry & Biobank Finder**, which allows them to find patients registries and biobanks dealing with their rare disease of interest. In addition, the **RD-Connect Sample Catalogue** helps them browse and find biosamples from rare disease patients, such as blood and DNA, which they might use and re-use for research.
To ensure protection of patient privacy without compromising research and drug discovery, the experts on ethics, legal and social issues engaged in RD-Connect provide guidance to researchers and work on the most appropriate models for data sharing
http://rd-connect.eu/

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

UNITED KINGDOM

Core Outcome Measures in Effectiveness Trials (COMET)

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/

DatabasE of genomiC varIation and Phenotype in Humans using Ensembl Resources (DECIPHER)

DECIPHER (DatabasE of genomiC varIation and Phenotype in Humans using Ensembl Resources) is an interactive web-based database which incorporates a suite of tools designed to aid the interpretation of genomic variants.
DECIPHER enhances clinical diagnosis by retrieving information from a variety of bioinformatics resources relevant to the variant found in the patient. The patient's variant is displayed in the context of both normal variation and pathogenic variation reported at that locus thereby facilitating interpretation.. DECIPHER is an IRDIRC recognized resource.
http://decipher.sanger.ac.uk/

European Bioinformatics Institute (EBI)

The European Bioinformatics Institute (EBI) is part of the European Molecular Biology Laboratory (EMBL), an international, innovative and interdisciplinary research organisation funded by 27 member states, 2 prospective member states and 2 associate member states.
EBI makes the world's public biological data freely available to the scientific community via a range of services and tools, perform basic research and provide professional training in bioinformatics. The EBI's databases and tools help scientists share data efficiently, perform complex queries and analyse the results in different ways. EBI's work supports researchers, who are wet-lab and computational biologists working in all areas of the life sciences, from biomedicine to biodiversity and agri-food research.
EBI's hands-on bioinformatics training help experimental biologists make the most of EBI's wide range of data resources. EBI also offers online training resources.
EMBL-EBI is a partner in several of Europe's emerging research infrastructures, including the ELIXIR infrastructure for biological information.
http://www.ebi.ac.uk

The European Genome-phenome Archive (EGA) is available at the European Bioinformatics Institute (EBI) and the Centre for Genomic Regulation (CRG). The 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 authorize 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. Studies and datasets can be browsed on the public website at the EBI or the CRG, providing information (metadata) about the aim, the experiments and the data used in the registered studies.
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 22 members and one Observer, bringing together over 220 research organisations. It was founded in December 2013 and began implementing its first scientific programme in 2014. It is currently implementing its second five-year scientific programme. The scientific and technical activities in ELIXIR are run by five Platforms and a number of Communities (formerly called 'Use Cases'). Platforms bring together experts to define the strategy and provide services in a particular area e.g. training, data. Communities 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 Phenotyping Consortium (IMPC) is an international effort by 19 research institutions to identify the function of every protein-coding gene in the mouse genome. The IMPC's mission is to create a comprehensive catalogue of mammalian gene function that is freely available for researchers. This data is obtained by systematically switching off or 'knocking out' each of the roughly 20,000 genes that make up the mouse genome. These knock out mice then undergo standardised physiological tests (phenotyping tests) across a range of biological systems in order to infer gene function. IMPC data can be used in a variety of ways, such as to investigate basic biology mechanisms that can lead to new therapeutic targets or to narrow down a suspected list of genes in patients. IMPC also aims to provide transformative insights into the genetic basis of disease that will impact upon clinical diagnosis and management and ultimately prevent, detect, diagnose and treat disease.
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 integrated 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/

UNITED STATES

HPO - the Human Phenotype Ontology

The Human Phenotype Ontology (HPO) provides a standardized vocabulary of phenotypic abnormalities encountered in human disease. Each term in the HPO describes a phenotypic abnormality. The HPO is currently being developed using the medical literature, Orphanet, DECIPHER, and OMIM. HPO currently contains over 13,000 terms and over 156,000 annotations to hereditary diseases. HPO provides an ontology of medically relevant phenotypes, disease-phenotype annotations, and the algorithms that operate on these to allow large-scale computational analysis of the human phenome. The HPO can be used to support differential diagnostics, translational research, and a number of applications in computational biology by providing the means to compute over the clinical phenotype. The HPO is being used for computational deep phenotyping and precision medicine as well as integration of clinical data into translational research. Although initially focused on rare, mainly Mendelian diseases, the focus of the HPO will be extended to other areas of medicine in coming years.
https://hpo.jax.org/app/

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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