



Orphanet Report Series

Rare Diseases collection

April 2024

Research Infrastructures useful to Rare Diseases

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Methodology and objectives

Research infrastructures are facilities, resources, computational tools and services useful for the scientific community to conduct research and foster innovation. The role of research infrastructures is continuously evolving and they are becoming more and more part of a connected ecosystem forming a unique resource for advanced research and interdisciplinary analysis of complex scientific problems, such as rare diseases.

This report provides the information collected by Orphanet so far, regarding the infrastructures that are relevant for fostering rare disease research at the international level. It has the aim to provide an at-hand short directory of research infrastructures, organized by categories, so to ease their identification by researchers. Some of these research infrastructures are specific for rare diseases (RD specific), while others are not but can be relevant for rare diseases.

Some of these infrastructures are recognized for their compliance with objective quality criteria by several initiatives, such as:



- International RD Research Consortium (IRDiRC) recognized resources



- ELIXIR Core Data Resources



- Human Variome Project Recommended Systems



- Global Core Biodata Resource

73 infrastructures have been identified by Orphanet and classified into 10 categories: Genomics deposition and analysis platform; Biobanking, biospecimens, cell lines, animal models, patient registries, pathogenic agents; Translational and clinical research; Chemical library, high-throughput drug screening and production/purification of molecules; In vivo, cellular and molecular imaging; Gene and cell therapy; Knowledge bases; Bioinformatics; Research tools; Structural biology and life science resources.

Each one of these resources will be listed below by specifying the more relevant information, such as the type of data, the mission, the coordinating country, the web link and, when it is the case, the “recognized resources” label.

In order to obtain more details on the data collected by Orphanet, we invite you to visit [Orphadata](#). This platform provides access to a number of massive datasets extracted from the Orphanet knowledge base, including datasets from our [catalogue of expert resources](#), upon signature of a Data Transfer Agreement or Service Contract.

Genomics deposition and analysis platform



Database of genomic variation and Phenotype in Humans using Ensembl Resources



- **Coordinating country:** United Kingdom
- **Data:** web-based phenotypic and genotypic patient database which incorporates a suite of tools
- **Mission:** aid the interpretation of genomic variants and enhance clinical diagnosis
- **RD specific:** No
- **Link:** <https://www.deciphergenomics.org/>
- Funded by IRDiRC member



Leiden Open (source) Variation Database



- **Coordinating Country:** Netherlands
- **Data:** gene-centered collection and display of DNA variants
- **Mission:** provide a flexible, freely available tool for genomic variant and phenotype collection, display and curation
- **RD specific:** No
- **Link:** <https://www.lovd.nl/3.0/home>



RD-Connect Genome-Phenome Analysis Platform



- **Coordinating country:** Spain
- **Data:** sequencing data from pseudonymized rare disease patients and family members
- **Mission:** identification of disease-causing mutations in rare disease patients and confirmation of diagnosis by finding matching cases submitted by other researchers around the world
- **RD specific:** Yes
- **Link:** <https://platform.rd-connect.eu/>
- Funded by IRDiRC member



EGA: European Genome-phenome Archive



- **Coordinating Country:** United Kingdom
- **Data:** permanent archiving and sharing of all types of personally identifiable genetic and phenotypic data resulting from biomedical research projects
- **Mission:** foster hosted data reuse, enable reproducibility, and accelerate biomedical and translational research
- **RD specific:** No
- **Link:** <https://ega-archive.org>



PhenomeCentral



- **Coordinating country:** Canada
- **Data:** phenotypic data (clinical symptoms and physical findings)
- **Mission:** allow clinicians to learn about the existence of cases similar to theirs, which may help to improve understanding of disorder manifestations and confirm underlying causes
- **RD specific:** Yes
- **Link:** <https://www.phenomecentral.org/About>
- **Funded by** IRDiRC member



CCG: **Cologne Center**
for **Genomics**

- **Coordinating Country:** Germany
- **Data:** sequencing, gene mapping, whole genome association studies in complex diseases, validation of copy number variations
- **Mission:** accelerate biological and medical research through the next generation sequencing
- **RD specific:** No
- **Link:** <https://ccg.uni-koeln.de/about-ccg>



WGGC: **West German**
Genome Center

- **Coordinating country:** Germany
- **Data:** gene sequencing
- **Mission:** advance Next Generation Sequencing technologies and applications
- **RD specific:** No
- **Link:** <https://wggc.de/>



CCGA: **Competence Centre**
for **Genomic Analysis Kiel**

- **Coordinating Country:** Germany
- **Data:** gene, transcriptome and microbiome sequencing
- **Mission:** strengthen the biological/medical infrastructure in Germany
- **RD specific:** No
- **Link:** <https://www.ccg.uni-kiel.de/>



DcGC: DRESDEN-concept
Genome Center

- **Coordinating country:** Germany
- **Data:** single cell applications, de novo genome sequencing
- **Mission:** shared technology resource that offers the infrastructure for a broad range of state-of-the-art genomic technologies
- **RD specific:** No
- **Link:** <https://genomecenter.tu-dresden.de/>



NCCT: NGS Competence
Center Tübingen

- **Coordinating Country:** Germany
- **Data:** gene sequencing
- **Mission:** strengthen the use of high-throughput sequencing methods in Germany
- **RD specific:** No
- **Link:** <https://portal.qbic.uni-tuebingen.de/portal/fr/web/ncct/our-center>

Genomics and Proteomics Core Facility



German Cancer Research
Center Genomics &
Proteomics Core Facility

- **Coordinating country:** Germany
- **Data:** genomics and proteomics
- **Mission:** provide access to sophisticated and expensive key technologies that are of critical relevance in biomedical science
- **RD specific:** No
- **Link:** <https://www.dkfz.de/gpcf/home>



SeqOIA

- **Coordinating Country:** France
- **Data:** gene sequencing
- **Mission:** allow equal access for all to genomic analyses so that cancer, rare disease, or common disease patients can benefit from the latest predictive and personalized medicine breakthroughs
- **RD specific:** No
- **Link:** <https://laboratoire-segoia.fr>



AURAGEN

- **Coordinating country:** France
- **Data:** gene sequencing
- **Mission:** provides access to high throughput sequencing to all actors involved in cancerology and management of rare diseases in the Auvergne Rhône-Alpes region
- **RD specific:** No
- **Link:** <https://www.auragen.fr>

Biobanking, biospecimens, cell lines, animal models, patient registries, pathogenic agents



EU RD Platform: European
Platform on Rare Disease
Registration

- **European Commission infrastructure**
- **Offices location:** Italy
- **Data:** patient registries
- **Mission:** facilitate access to data on RD patient registries, increase visibility and enable extended use and re-use of registries' data
- **RD specific:** Yes
- **Link:** <https://eu-rd-platform.jrc.ec.europa.eu/en>
- Funded by IRDiRC member



BBMRI-ERIC

Biobanking and Biomolecular
Resources Research Infrastructure
- European Research
Infrastructure Consortium

- **European infrastructure**
- **Offices location:** Austria
- **Data:** biobanks
- **Mission:** Facilitate access to collections of partner biobanks and biomolecular resources
- **RD specific:** No
- **Link:** <http://bbmri-eric.eu/>
- Funded by IRDiRC member



RaDiCo: Rare
Diseases Cohorts

- **Coordinating country:** France
- **Data:** anatomical, biochemical, molecular
- **Mission:** collection of extensive phenotype data in RD patients with a view to clinical and epidemiological research in liaison with translational and basic research
- **RD specific:** Yes
- **Link:** <http://www.radico.fr/>
- Funded by IRDiRC member



RADeep: Rare Anaemia
Disorders European
Epidemiological Platform

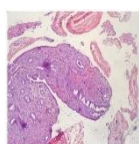
- **Coordinating Countries:** Spain, Belgium & Cyprus
- **Data:** patient registries, epidemiological
- **Mission:** map at the European level the diagnosis methods, demography, survival rate, treatments and main clinical features of RAD patients in order to facilitate research on RAD and development of new treatments and clinical trials
- **RD specific:** Yes
- **Link:** <https://www.radeepnetwork.eu/>



European Research Infrastructure
on Highly Pathogenic Agents

ERINHA: European Research
Infrastructure on Highly
Pathogenic Agents

- **Coordinating Country:** France
- **Data:** pathogens
- **Mission:** 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
- **RD specific:** No
- **Link:** <http://www.erinha.eu/>
- Funded by IRDiRC member



Cellosaurus - a knowledge resource
on cell lines



- **Coordinating Country:** Switzerland
- **Data:** cell lines
- **Mission:** Facilitate identification of cell lines, including iPS
- **RD specific:** No
- **Link:** <https://www.cellosaurus.org/>



hPSCreg - Human pluripotent
stem cell registry



- **Coordinating country:** Germany
- **Data:** cell lines
- **Mission:** offer a large and in-depth overview on the current status of human pluripotent stem cell research
- **RD specific:** No
- **Link:** <https://hpscereg.eu/>
- Funded by IRDiRC member



PHENOMIN: French
National Infrastructure for
Mouse Phenogenomics

- **Coordinating Country:** France
- **Data:** rodent models
- **Mission:** develop tools to advance basic research in mammals and better respond to the ethical demands of society on the use of animals in research and to improve the impact of preclinical studies on the development of therapies in humans
- **RD specific:** No
- **Link:** <http://www.phenomin.fr/>
- Funded by IRDiRC member



IMPC: International
Mouse Phenotyping
Consortium

- **Coordinating country:** United Kingdom
- **Data:** mouse genome
- **Mission:** create a comprehensive catalogue of mammalian gene function that is freely available for researchers and provide insights into the genetic basis of disease
- **RD specific:** No
- **Link:** <https://www.mousephenotype.org/>
- Funded by IRDiRC member



INFRAFRONTIER

- **Coordinating Country:** Germany
- **Data:** generation, phenotyping, archiving and distribution of model mammalian genomes.
- **Mission:** improve the understanding of gene function in human health and disease facilitating the production and use of mouse models
- **RD specific:** No
- **Link:** <https://www.infrafrontier.eu>

Translational and clinical research



EATRIS: European infrastructure for translational medicine

- **European infrastructure**
- **Offices location:** Netherlands
- **Data:** vast array of pre-clinical and clinical expertise and facilities to improve and optimize preclinical and early clinical development of drugs, vaccines and diagnostics
- **Mission:** support researchers in developing their biomedical discoveries into novel translational tools and interventions for better health outcomes for society
- **RD specific:** No
- **Link:** <http://www.eatris.eu/>



ECRIN: European Clinical Research Infrastructure Network

- **European infrastructure**
- **Offices location:** France
- **Data:** clinical trials
- **Mission:** facilitate researchers to set up and conduct multinational clinical trials in Europe
- **RD specific:** No
- **Link:** <https://ecrin.org/>



EORTC: European Organisation for Research and Treatment of Cancer

- **Coordinating country:** Belgium
- **Data:** translational and clinical research
- **Mission:** to coordinate and conduct international translational and clinical research to improve the standard of cancer treatment for patients and increase people's survival and quality of life
- **RD specific:** No
- **Link:** <http://www.eortc.org>



Care and Trial Site Registry

CTSR: Care and Trial Site Registry for neuromuscular and neurodegenerative diseases



- **Coordinating Country:** Germany
- **Data:** clinical trials
- **Mission:** help the pharmaceutical industry and clinical investigators select trial sites and identify potential partners for upcoming research projects
- **RD specific:** Yes
- **Link:** <https://ctsr.uniklinik-freiburg.de/>
- Funded by IRDiRC member



ERICA PROMs
Repository

- **Coordinating country:** Netherlands
- **Data:** Patient Reported Outcome Measures (PROMs)
- **Mission:** identify and centralize Clinical Assessment Outcomes questionnaires of relevance for rare diseases
- **RD specific:** Yes
- **Link:** <https://erica-rd.eu/work-packages/patient-centred-research/proms-repository/>
- Funded by IRDiRC member



Rare Diseases Clinical
Trials Toolbox



- **Coordinating country:** France
- **Data:** clinical trial toolbox
- **Mission:** help clinical trialists and R&D managers understand the regulations and requirements for conducting trials for RDs
- **RD specific:** Yes
- **Link:** <https://ecrin.org/rare-diseases-clinical-trials-toolbox>
- Funded by IRDiRC member



Innovation Management
Toolbox



- **Coordinating country:** France
- **Data:** reference library of resources in rare disease translational medicine
- **Mission:** provide investigators with self-help resources specific to their needs
- **RD specific:** Yes
- **Link:** <https://www.ejprarediseases.org/innovation-management-toolbox/>
- Funded by IRDiRC member



OrphanDev

- **Coordinating country:** France
- **Data:** clinical drug development
- **Mission:** support researchers, clinicians and the health industry actors in the development of drugs for rare diseases
- **RD specific:** Yes
- **Link:** <http://www.orphan-dev.org/>
- Funded by IRDiRC member

Toolkit For Patient-Focused Therapy Development

NCATS Toolkit



- **Coordinating Country:** United States
- **Data:** patient groups
- **Mission:** help patient groups become a driving force behind the development of therapies and/or a cure for RDs
- **RD specific:** Yes
- **Link:** <https://toolkit.ncats.nih.gov/>
- Funded by IRDiRC member



COMET: Core Outcome Measures for Clinical Trials

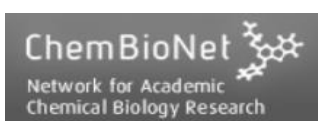
- **Coordinating Country:** United Kingdom
- **Data:** core outcome sets (COS)
- **Mission:** collate and stimulate relevant resources, both applied and methodological, to facilitate exchange of ideas and information, and to foster methodological research in clinical trials
- **RD specific:** No
- **Link:** <http://www.comet-initiative.org/>
- Funded by IRDiRC member

Chemical library, high-throughput drug screening and production/purification of molecules



EU-OPENSREEN ERIC -
European high-capacity
screening network

- **Coordinating country:** Germany
- **Data:** chemical compounds
- **Mission:** provide tools and expertise for advanced screening and medicinal chemistry technologies
- **RD specific:** No
- **Link:** <https://www.eu-openscreen.eu/>



ChemBioNet - Resource
Network Supporting
Academic Chemical Biology
Research

- **Coordinating Country:** Germany
- **Data:** chemical compounds
- **Mission:** support research projects for systematic usage of small molecules to explore biological systems
- **RD specific:** No
- **Link:** <http://www.chembionet.info/>



APTEEUS

- **Coordinating country:** France
- **Data:** chemical compounds
- **Mission:** respond quickly to the medical needs of patients by identifying candidates for repositioning and new avenues of treatment for diseases
- **RD specific:** No
- **Link:** <http://www.apteeus.fr/en/>



PCBIS: Platform of
Integrative Chemical Biology
of Strasbourg

- **Coordinating Country:** France
- **Data:** active molecules
- **Mission:** identify active molecules and give access to High Throughput Screening, chemical libraries and target libraries to academic and private laboratories
- **RD specific:** No
- **Link:** <http://www.pcbis.fr/en/>

In vivo, cellular and molecular imaging



Euro-BioImaging

- **Coordinating country:** Finland
- **Data:** imaging technologies and expertise
- **Mission:** provide researchers with imaging services that bridge biological and biomedical imaging and facilitate innovative and world-class research
- **RD specific:** No
- **Link:** <http://www.eurobioimaging.eu/>
- Funded by IRDiRC member



I-Stem: Institute for Stem cell
Therapy and Exploration of
Monogenic diseases

- **Coordinating country:** France
- **Data:** human pluripotent stem cells
- **Mission:** explore all the therapeutic potential of human pluripotent stem cells for applications in patients affected by rare diseases of genetic origin
- **RD specific:** Yes
- **Link:** <http://www.istem.eu/>
- Funded by IRDiRC member



Biotherapies Institute

- **Coordinating Country:** France
- **Data:** biotherapies
- **Mission:** unite the skills, expertise and resources, strengthen the complementarity of resources and better coordinate cross-functional projects, especially clinical development to accelerate the development of treatments against rare diseases
- **RD specific:** Yes
- **Link:** <http://www.institut-biotherapies.fr/>
- Funded by IRDiRC member



GENETHON

- **Coordinating country:** France
- **Data:** gene therapy
- **Mission:** 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
- **RD specific:** Yes
- **Link:** <http://www.genethon.fr/>
- Funded by IRDiRC member



CIDSTEM: Interdepartmental
Centre for Stem Cells and
Regenerative Medicine

- **Coordinating Country:** Italy
- **Data:** cell and gene therapy
- **Mission:** 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
- **RD specific:** No
- **Link:** <https://www.cidstem.unimore.it/>

Knowledge bases



EOSC: European Open
Science Cloud

- **European Commission infrastructure**
- **Data:** research data, methods, software, publications
- **Mission:** provide European researchers, innovators, companies and citizens with a federated and open multi-disciplinary environment where they can publish, find and reuse data, tools and services for research, innovation and educational purposes
- **RD specific:** No
- **Link:** <https://open-science-cloud.ec.europa.eu/>
- Funded by IRDiRC member



The EJP-RD Virtual
Platform

- **Coordinating country:** France
- **Data:** catalogues of resources, registries, biobanks, knowledge bases and tools compliant with agreed standards
- **Mission:** The Virtual Platform addresses the challenge of scattered, unconnected and incompatible resources, which pose a significant barrier to advancing treatments and knowledge about Rare Diseases, by allowing users to discover and search a range of interconnected Rare Disease resources.
- **RD specific:** Yes
- **Link:** <https://vp.ejprarediseases.org/>
- Funded by IRDiRC member

orphanet

Orphanet



- **Coordinating Country:** France
- **Data:** rare disease data
- **Mission:** provide high-quality information and data on rare diseases to improve the diagnosis, care and treatment of patients and to ensure equal access to knowledge for all stakeholders
- **RD specific:** Yes
- **Link:** <http://www.orpha.net/>
- Funded by IRDiRC member





Orphadata



- **Coordinating country:** France
- **Data:** rare disease data
- **Mission:** provide the scientific community with comprehensive, massive, re-usable and computable quality data sets related to rare diseases from the Orphanet knowledge base
- **RD specific:** Yes
- **Link:** <https://www.orphadata.com/>
- Funded by IRDiRC member



Wikipathways Rare Diseases

- **Coordinating Country:** Netherlands
- **Data:** biological pathway database
- **Mission:** facilitate the contribution and maintenance of pathway information by the biology community
- **RD specific:** Yes
- **Link:** <https://www.wikipathways.org/communities/rarediseases.html>
- Funded by IRDiRC member



IUPHAR/BPS

IUPHAR-Guide to Pharmacology

- **Coordinating country:** United Kingdom
- **Data:** drugs ligands and drugs
- **Mission:** provide a searchable database with quantitative information on drug targets and the prescription medicines and experimental drugs that act on them
- **RD specific:** No
- **Link:** <https://www.guidetopharmacology.org/>



UniProt: Universal Protein Resource



- **Coordinating Country:** United Kingdom
- **Data:** proteomics
- **Mission:** provide a comprehensive and freely accessible resource of protein sequence and functional information
- **RD specific:** No
- **Link:** <https://www.uniprot.org/>
- Funded by IRDiRC member





Nextprot

- **Coordinating country:** Switzerland
- **Data:** proteomics
- **Mission:** Provide a standardized vocabulary and a knowledge base on proteins
- **RD specific:** No
- **Link:** <https://www.nextprot.org/>



PRIDE Archive - **PR**oteomics
IDentifications Database



- **Coordinating Country:** United Kingdom
- **Data:** proteomics
- **Mission:** provide a standards compliant public data repository for mass spectrometry proteomics data to support reproducible research and enable the reuse of these data by other researchers
- **RD specific:** No
- **Link:** <https://www.ebi.ac.uk/pride/archive/>



MetaboLights



- **Coordinating Country:** United Kingdom
- **Data:** metabolomics
- **Mission:** Provide a cross-species and cross-technique database for metabolomics experiments and derived information
- **RD specific:** No
- **Link:** <https://www.ebi.ac.uk/metabolights/about>



BRENDA



- **Coordinating country:** Germany
- **Data:** enzyme functional data
- **Mission:** provide a collection of enzyme functional data available to the scientific community
- **RD specific:** No
- **Link:** <https://www.brenda-enzymes.org/introduction.php>





Rhea



- **Coordinating Country:** Switzerland
- **Data:** reaction participants
- **Mission:** Provide an expert-curated knowledgebase of chemical and transport reactions of biological interest
- **RD specific:** No
- **Link:** <https://www.rhea-db.org/>



ChEBI: **Chemical Entities of Biological Interest**



- **Coordinating Country:** United Kingdom
- **Data:** chemical entities
- **Mission:** provide a freely available dictionary and ontological classification of molecular entities focusing on “small” chemical compounds (atom, molecule, ion, ion pair, radical, radical ion, complex, conformer, etc.)
- **RD specific:** No
- **Link:** <https://www.ebi.ac.uk/chebi/aboutChebiForward.do>



ChEMBL



- **Coordinating Country:** United Kingdom
- **Data:** chemical compounds
- **Mission:** provide a database of bioactive molecules with drug-like properties. It brings together chemical, bioactivity and genomic data to aid the translation of genomic information into effective new drugs
- **RD specific:** No
- **Link:** <https://www.ebi.ac.uk/chembl/>



Ensembl



- **Coordinating Country:** United Kingdom
- **Data:** genome browser for vertebrate genomes
- **Mission:** support research in comparative genomics, evolution, sequence variation and transcriptional regulation through gene annotation, regulatory function prediction and alignments
- **RD specific:** No
- **Link:** <http://www.ensembl.org/index.html>





GenCC: The **Gene**
Curation **Coalition**

- **Coordinating country:** United States
- **Data:** genomics
- **Mission:** evaluate gene-disease validity with a willingness to share data publicly, to develop consistent terminology for gene curation activities and to facilitate the consistent assessment of genes that have been reported in association with disease
- **RD specific:** No
- **Link:** <https://thegencc.org/about.html>



Alliance of Genome
Resources

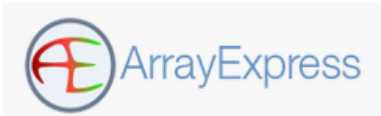
- **Coordinating Country:** United States
- **Data:** genomics
- **Mission:** develop and maintain sustainable genome information resources that facilitate the use of diverse model organisms in understanding the genetic and genomic basis of human biology, health and disease
- **RD specific:** No
- **Link:** <https://www.alliancegenome.org/>
- Funded by IRDiRC member



MARRVEL: **Model** organism
Aggregated Resources for Rare
Variant ExpLoration



- **Coordinating country:** United States
- **Data:** genomics
- **Mission:** facilitate the use of public genetic resources to prioritize rare human gene variants for study in model organisms
- **RD specific:** No
- **Link:** <https://marrvel.org/about>



ArrayExpress - Functional
Genomics Data



- **Coordinating Country:** United Kingdom
- **Data:** genomics
- **Mission:** store and provide data from high-throughput functional genomics experiments for reuse to the research community
- **RD specific:** No
- **Link:** <https://www.ebi.ac.uk/biostudies/arrayexpress>



GENEONTOLOGY

- **Coordinating country:** United States
- **Data:** genomics
- **Mission:** develop a comprehensive, computational model of biological systems, ranging from the molecular to the organism level, across the multiplicity of species in the tree of life
- **RD specific:** No
- **Link:** <http://geneontology.org/>
- Funded by IRDiRC member



Genome Aggregation Database

gnomAD

- **Coordinating Country:** United States
- **Data:** genomics
- **Mission:** aggregate and harmonize exome and genome sequencing data from a variety of large-scale sequencing projects and to make summary data available for the scientific community
- **RD specific:** No
- **Link:** <https://gnomad.broadinstitute.org/>
- Funded by IRDiRC member



HPO: Human
Phenotype Ontology



- **Coordinating Country:** United States
- **Data:** phenotype
- **Mission:** Provide a standardized vocabulary of phenotypic abnormalities encountered in human disease
- **RD specific:** Yes
- **Link:** <https://hpo.jax.org/>
- Funded by IRDiRC member

EMBL-EBI



EBI: European
Bioinformatics Institute

- **Coordinating country:** United Kingdom
- **Data:** genomics, proteins, small molecules, systems, ontologies and scientific literature
- **Mission:** help scientists realize the potential of big data in biology, exploiting complex information to make discoveries that benefit humankind
- **RD specific:** No
- **Link:** <http://www.ebi.ac.uk/>
- Funded by IRDiRC member



IFB: French Institute
of Bioinformatics

- **Coordinating Country:** France
- **Data:** genomics, proteins, small molecules, systems, ontologies
- **Mission:** 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
- **RD specific:** No
- **Link:** <https://www.france-bioinformatique.fr>
- Funded by IRDiRC member

Research tools



Bio.tools - Rare Diseases

- **Coordinating country:** Denmark
- **Data:** Rare disease resource catalogue
- **Mission:** provide the rare disease community with a meaningful and detailed registry of resources useful for rare disease research
- **RD specific:** Yes
- **Link:** <https://bio.tools/t?domain=rare-diseases>



Exomiser



- **Coordinating Country:** United States
- **Data:** genomic variants
- **Mission:** provide a program that functionally annotates variants from whole-exome sequencing data
- **RD specific:** No
- **Link:** <https://exomiser.monarchinitiative.org/exomiser/>
- Funded by IRDiRC member



Genomiser

- **Coordinating country:** United States
- **Data:** non-coding variants
- **Mission:** score the relevance of variation in the non-coding genome and associate regulatory variants to specific Mendelian diseases
- **RD specific:** No
- **Link:** <https://hpo.jax.org/tools/genomiser>
- Funded by IRDiRC member

LUMC Mutalyzer 3

LUMC Mutalyzer 3



- **Coordinating Country:** Netherlands
- **Data:** genomics
- **Mission:** to be of help when working with human genome variation society sequence variant nomenclature descriptions
- **RD specific:** No
- **Link:** <https://mutalyzer.nl/>
- Funded by IRDiRC member



PhenXToolkit - consensus
measures for Phenotypes
and eXposures

- **Coordinating country:** United States
- **Data:** measurement protocols intended for use in biomedical, epidemiological, clinical, translational, and genomic research
- **Mission:** facilitate cross-study analysis to potentially increasing the scientific impact of individual studies
- **RD specific:** No
- **Link:** <https://www.phenx.org/>
- Funded by IRDiRC member

Structural biology and life science resources



ELIXIR: European life sciences infrastructure for biological information

- **European infrastructure**
- **Offices location:** United Kingdom
- **Data:** databases, software tools, training materials and bioinformatics services
- **Mission:** Provide a registry of software and databases to form a single infrastructure that make it easier for scientists to find and share data, exchange expertise, and agree on best practices
- **RD specific:** No
- **Link:** <http://www.elixir-europe.org/page.php?page=home>
- Funded by IRDiRC member



Instruct-ERIC

- **Coordinating Country:** United Kingdom
- **Data:** structural biology
- **Mission:** promote innovation in biomedical science by making high-end technologies and methods in structural biology available to users
- **RD specific:** No
- **Link:** <https://instruct-eric.org/>
- Funded by IRDiRC member

Please note that all data presented in this report are available for download at <https://www.orphadata.com/>

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

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The correct form when quoting this document is:

« List of research infrastructures useful to rare diseases », Orphanet Report Series, Rare Diseases collection, April 2024
https://www.orpha.net/pdfs/orphacom/cahiers/docs/GB/Research_Infrastructures_for_rare_diseases.pdf

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