

Orphadata Products Data Catalogue

April 2026



OVERVIEW

This catalogue provides an overview of the different Orphadata Products datasets based on the Orphanet knowledge base.

We provide a range of pre-defined datasets, available for a one-year period with two updates in the year, in XML format, according to the [samples](#) and [dataset descriptions](#) (this document) available on the Orphadata Products site.

These datasets are available for public institutions, academics and patient organisations after signature of a Data Transfer Agreement. Other entities can [contact us](#) for pricing.

We also are now happy to offer a range of tailored datasets and services adapted to your use cases and end needs through Orphanet's non-profit spin-off, [Orphanet-AISBL](#). You can find more information on tailored solutions on the AISBL [website](#) and in the dedicated [tailored data and services catalogue](#).



The funds raised through the services and data we offer contribute to the sustainability of the Orphanet network so that we can continue to provide quality, curated data on rare diseases and orphan drugs to the community.

If you have any questions on our data, services or your specific use case, please get in [contact](#) so we can discuss your use case and discover what we can offer.



ORPHADATA PRE-DEFINED DATASETS WITH TWO UPDATES PER YEAR

Product 1 : Orphadata Science (no fee, no contract, CC BY 4.0 licence)



All users can access Orphadata Science data files in an XML/JSON format directly on the [Orphadata Science](#) site through the [CC BY 4.0 licence](#) (Open Science compatible licence). There is no fee, and no need to sign an agreement/contract to access Orphadata Science files, but the licence terms must be followed. These files can be used in conjunction with Orphadata Products files, thanks to the inclusion of an ORPHAcode for each and every data.

If you are interested in accessing Orphadata Science through an API, please [contact us](#).

Orphadata Science is an [ELIXIR Core Data Resource](#) and [Global Core Biodata Resource](#).

Product 2: Textual information

Price on request

Dataset with ORPHA code of the disease and name, abstract (about 250 words) in English, French, German, Italian, Portuguese, Spanish and Dutch.

Dataset with ORPHA code of the disease including url of external sources (review articles, emergency guidelines, clinical practice guidelines) providing textual information.

Dataset with ORPHA code of the disease including specific query to PubMed on the disease.

[Detailed description](#) and [samples](#) available.

Product 3: Patient organisations

Price on request

Dataset with name of patient organisation, country, geographical coverage (regional, national, European, international (beyond Europe)), website of patient organisation and ORPHA code of the associated disease and disease name.

Dataset with name of patient organisation network, country of the coordinator, geographical coverage (regional, national, European, international (beyond Europe)), website of the patient organisation network, list of each patient organisation participating in the network (name and website) and ORPHA code of the associated disease and disease name.

[Detailed description](#) and [samples](#) available.



Product 4: Expert centres

Price on request

Dataset with name of expert centres, status (officially designated, member of a European Reference Network), type of service provided (genetic counselling, medical management), type of public (children, adults, both), institution, city, country, and ORPHA code of the associated disease and disease name

Dataset with name of expert centre's network, status (European Reference Network), geographical coverage (regional, national, European, international (beyond Europe)), list of each expert centres participating in the network with city and country, and ORPHA code of the associated disease and disease name

[Detailed description](#) and [samples](#) available.

Product 5: Diagnostic tests & medical laboratories

Price on request

Dataset with name and acronym of the medical laboratory, name of the hosting institution of the laboratory, city and country, accreditation status of the laboratory and EQA participation, list of diagnostic tests provided by the laboratory.

Diagnostic tests are defined by the name of the test, the purpose, speciality, technique and objective of the test, preferred name and ORPHA code of diseases and genes tested, EQA participation for a specific test.

Please see Annex 1 for more information about the list of purposes, specialities, objectives and techniques referenced in our database.

[Detailed description](#) and [samples](#) available.

Product 6: Orphan drugs

Price on request

Dataset with ORPHA code of the diseases and names for which the substance is indicated, name of the product, chemical name, trade name, type of product, ATC code, status of the substance, orphan designation zone, link to PSO/EPAR, designation holder, MA holder, indication.

[Detailed description](#) and [samples](#) available.



Product 7: Research activities

Price on request

Dataset with name of the national research project, type of research project (see table), status of the research project (ongoing or terminated), start date and end date, name of the lab, institution, city, country, Website where the research project, stakeholders involved in the research projects, substances and/or genes involved/studied in the research project are described, if applicable, and ORPHA code of the associated disease and disease name

29 types of research projects

- Animal model creation / study
- In vitro functional study
- Biomarker development
- Induced pluripotent stem cells (iPS) creation / study
- Biorepositories development/creation
- Medical device / instrumentation development
- Biotechnology innovation Mutations search
- CRISPR-Cas9 study
- Natural history study
- Databases & Registries development/creation
- Observational clinical study
- Diagnostic tool/protocol development
- Ontology / bioinformatics study
- Drug repurposing
- Outcomes measures development
- Epidemiological study
- Pre-clinical cell therapy
- Gene expression profile
- Pre-clinical drug development / Drug delivery
- Gene search
- Pre-clinical gene therapy
- Genotype-phenotype correlation
- Pre-clinical vaccine development
- Health economics study
- Public health study (excluding health economics)
- Health sociology study
- Small molecule screening
- Human physiopathology study

Dataset with name of multinational research project, status (ongoing or terminated), institution and country of the coordinator, geographical coverage (European or international (beyond Europe)), website where the project stakeholders involved, substances and/or genes involved/studied in the research project is described, if applicable, list of each country participating in this multinational research project, and ORPHA code of the associated disease and disease name



Clinical trial activities

Dataset with name of the national clinical trial, status (ongoing or terminated) and recruitment, begin date and end date, hosting institution, city, country, website where the trial is described, name of the sponsor, phase of trial, type of trial (drug, protocol, gene therapy, cell therapy, vaccine, medical device), name of the substance/product/tradename involved, genes involved if applicable, and ORPHA code of the associated disease and disease name.

Dataset with name of multinational clinical trial, status (ongoing or terminated), institution and country of the coordinator, geographical coverage (European or international (beyond Europe)), Website where the trial is described, name of the sponsor, phase of trial, type of trial (drug, protocol, gene therapy, cell therapy, vaccine, medical device), name of the substance/product/tradename involved, genes involved if applicable, list of all countries participating in this multinational clinical trial with details on status, and ORPHA code of the associated disease and disease name.

Patient registries

Dataset with name of the patient registry, status (multicentric, funded by a member of IRDIRC, member of a European Reference Network), recruitment, start date and end date, URL of registry (website where the registry is described), country, geocoverage (regional, national, European, international (beyond Europe)), hosting institution and stakeholders involved, and ORPHA code of the associated disease and disease name

Dataset with name of the patient registry network, status (funded by a member of IRDIRC, member of a European Reference Network), country of the coordinator, geocoverage (regional, national, European, international (beyond Europe)), hosting institution and stakeholders involved, list of patient registries involved in the network with its whole dataset, and ORPHA code of the associated disease and disease name

Biobanks

Dataset with name of the biobank, status (multicentric, funded by a member of IRDIRC), begin date, URL of the biobank, country, hosting institution and stakeholders involved, genes involved/studied in the biobank if applicable, and ORPHA code of the associated disease and disease name

Dataset with name of biobank's network, status (multicentric, funded by a member of IRDIRC), country of the coordinator, geocoverage (regional, national, European, international (beyond Europe)), hosting institution and stakeholders involved, list of biobanks involved in the network with its whole dataset and ORPHA code of the associated disease and disease name.

[Detailed description](#) and [samples](#) available.



Annex 1 : Product 5 List of purposes, specialities, objectives and techniques in diagnostic tests

Purposes:

- Antenatal diagnosis
- Preimplantation diagnosis
- Postnatal diagnosis
- Presymptomatic diagnosis
- Pharmacogenetics
- Risk assessment
- Newborn screening
- Somatic genetics

Specialities:

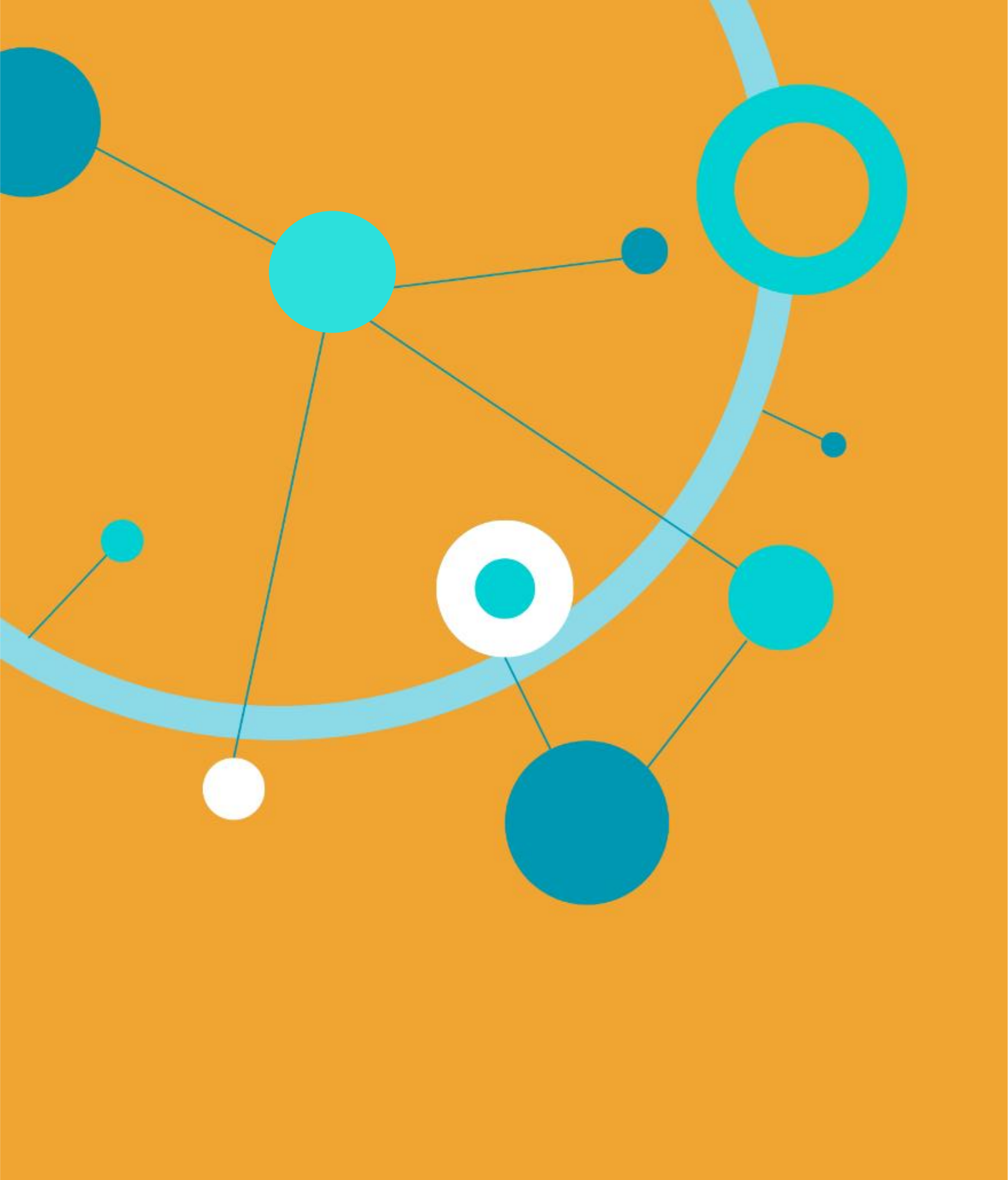
- Molecular genetics
- Cytogenetics
- Biochemical genetics
- Parasitology
- Bacteriology
- Virology
- Mycology
- Immunology
- Hematology
- Pathology
- Imaging
- Other

Objectives:

- Targeted mutation analysis
- Mutation scanning/screening and sequence analysis of selected exons
- Sequence analysis: entire coding region
- Uniparental disomy study
- Methylation analysis
- Deletion / Duplication analysis
- Detection of chromosome alterations large in size
- Detection of microdeletions/microduplications
- Chromosomal instability
- Analyte / Enzyme assay
- Protein expression

Techniques:

- Sanger sequencing
- NGS sequencing
- PCR based techniques
- MLPA based techniques
- Array based techniques
- Microsatellite analysis
- BS- Pyrosequencing
- FISH
- M-FISH / SKY
- Karyotyping
- Chromosomal instability
- Immunohistochemistry
- Western Blot



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https://www.orphadata.com/docs/Catalogue_Orphadata.pdf