orphadata

orphadata.com

AGGREGATED RARE DISEASE DATA FROM ORPHANET

Reusable, predefined datasets, customisable services & APIs

ORPHADATA SCIENCE
ORPHANET SCIENTIFIC KNOWLEDGE BASE

Rare disease classifications, terminology alignments, epidemiology, gene-disease relationships, phenotype-disease relationships & functional consequences

GLOBAL CORE BIODATA RESOURCE

eliXir Core Data Resource

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ORPHANET NOMENCLATURE & TOOLS

ORPHA 716

All the files, tools and guidelines you need to implement the ORPHAcodes in health and research information systems

GLOBAL CORE BIODATA RESOURCE

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CATALOGUE OF EXPERT RESOURCES

Expert centres, research projects, clinical trials, patient registries, orphan drugs, patient organisations, diagnostic tests and laboratories, textual information, all curated with an ORPHAcode

ONTOLOGIES

Access the power of Orphanet through the Orphanet Rare Disease Ontology, and HPO-ORDO Ontological Module

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2024 Orphadata Services & Data Catalogue

OVERVIEW

This catalogue provides an overview of the different services offered by Orphadata based on the Orphanet knowledge base the rare disease expertise of the Orphanet and Orphadata teams.

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 available on the Orphadata website, and the [product description](product%20description). These datasets are available for public institutions, academics and patient organisations after signature of a Data Transfer Agreement. Other entities can contact data.orphanet@inserm.fr for pricing.

Thanks to feedback from our users, the Orphadata team is now happy to offer a range of tailored services adapted to your use cases and end needs. A few examples of how we can help you are given in section A of this catalogue. Please get in contact (data.orphanet@inserm.fr) so we can discuss your specific needs together and discover what we can offer.

The funds raised through the services we offer contribute to the sustainability of the Orphanet knowledge base so that we can continue to provide quality, curated data on rare diseases and orphan drugs to the community both through www.orpha.net and www.orphadata.com.

A) ORPHADATA SERVICES AND TAILORED DATASETS

You can get in contact with the Orphadata team data.orphanet@inserm.fr to discuss your specific rare disease-related use case and we will work with you to find a solution to best suit your needs. Below is a non-exhaustive list of services we can now provide to our end-users on a for-fee basis.

- Customised datasets (XML format or through an API)

- Customised data analysis interfaces

- Assistance in implementing and/or using Orphanet data

- Rare disease data expertise (curation/analysis/interpretation of data in the rare disease field)

- Data analysis reports

- Training modules

- Technical assistance for data integration

- Consultancy

B) ORPHADATA PRE-DEFINED DATASETS WITH TWO UPDATES PER YEAR

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

Rare diseases inventory with annotations

Rare Diseases and cross-references

List including preferred name, synonyms in English, French, German, Italian, Portuguese, Spanish, Dutch, Czech, or Polish, ORPHA code, type of entries, short definition of the disease. Entries are cross-referenced with ICD-10, ICD-11, OMIM, UMLS, MeSH, MedDRA and the alignments are characterized in order to indicate if the terms are perfectly equivalent (exact mapping) or not.

Clinical classifications of rare diseases (poly-hierarchy)

- Rare abdominal surgical diseases
- Rare allergic disease
- Rare bone diseases
- Rare cardiac diseases
- Rare cardiac malformations
- Rare circulatory system diseases
- Rare developmental anomalies during embryogenesis
- Rare diseases due to toxic effects
- Rare disorder without a determined diagnosis after full investigation
- Rare endocrine diseases
- Rare gastroenterological diseases
- Rare genetic diseases
- Rare gynaecological and obstetric diseases
- Rare haematological diseases
- Rare hepatic diseases
- Rare immunological diseases
- Rare inborn errors of metabolism

- Rare infectious diseases
- Rare infertility
- Rare neoplastic diseases
- Rare neurological diseases
- Rare odontological diseases
- Rare ophthalmic diseases
- Rare otorhinolaryngological diseases
- Rare renal diseases
- Rare respiratory diseases
- Rare skin diseases
- Rare surgical maxillo-facial diseases
- Rare surgical thoracic diseases
- Rare systemic and rheumatological diseases of childhood
- Rare systemic and rhumatological diseases
- Rare teratologic disorders
- Rare transplant-related diseases
- Rare urogenital diseases

Rare diseases with their associated genes
Table with ORPHA code of the disease linked to the associated genes, with a characterisation of the relationship between gene and disease (causative, modifier, susceptibility, or playing a role in the phenotype) and the kind of mutation germline or somatic. In addition, the table includes the name of the gene in English, its ORPHA code, chromosomal location, symbol and synonyms and cross-referenced with UniProtKB, HGNC, OMIM, Genatlas, ensembl, Reactome and IUPHAR-DB.

Phenotypes associated with rare diseases

Table with diseases listed in Orphanet annotated with HPO phenotypes. The alignment is characterised by frequency (obligatory, very frequent, frequent, occasional, very rare or excluded) and whether the annotated HPO term is a major diagnostic criterion or a pathognomonic sign of the rare disease.
Table with the source, the date and the validation status of the association between the rare disease and HPO terms.

Diseases with epidemiological data

Table with preferred name and ORPHA code of the diseases, groups of diseases or sub types: point prevalence, birth prevalence, lifelong prevalence and incidence, or the number of families reported together with their respective intervals per geographical area.
Table with preferred name and ORPHA code of the diseases, their type of inheritance, interval average age of onset.

Rare diseases and functional consequences

Table with diseases listed in Orphanet annotated with functional consequences or environmental factors leading to limitation of activity or restriction of participation.The alignment is characterised by frequency, temporality, degree of severity.

Product 2: Textual information   Price on request

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

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

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

Product 3: Patient organisations   Price on request

Table with ORPHA code of the disease, name of patient organisation, country, geographical coverage (regional, national, international).

Table with ORPHA code of the disease, name of patient organisation network, country of the coordinator, geographical coverage (regional, national, international).

Product 4: Expert centres   Price on request

Table with ORPHA code of the disease, name of expert centres, centre of expertise status, type of service provided (genetic counselling, disease management), type of public (children, adults), institution, type of institution (general hospital, private hospital, research institute, teaching hospital, university research centre, other), status of institution (private for profit, private not for profit, public for profit, public not for profit), city, country.

Table with ORPHA code of the disease, name of expert centre's network, country of the coordinator, geographical coverage (regional, national, international).

Product 5: Diagnostic tests & clinical laboratories   **Price on request**

Table with name and acronym of the laboratory, name and acronym of the hosting institution of the laboratory, speciality of the laboratory (molecular genetics, biochemistry, cytogenetics, immunology, haematology, virology, parasitology, bacteriology, pathology, address of the hosting institution (city and country), status of institution (private for profit, private not for profit, public for profit, public not for profit), 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.

Product 6: Orphan drugs   **Price on request**

Table with ORPHA code of the diseases for which the substance is indicated, name of the product, chemical name, trade name, type of product, INN, ATC code, status of the substance, orphan designation zone, link to PSO/EPAR, designation holder, MA holder, associated trials.

Product 7: Research activities   **Price on request**

Table with ORPHA code of the diseases, name of the research project, type of research project (see table), name of the lab, institution, type of institution (general hospital, private hospital, research institute, teaching hospital, university research centre, other), status of institution (private for profit, private not for profit, public for profit, public not for profit), city, country.

26 types of research projects

- Research project Gene(s) search
- Mutation(s) search
- Gene expression profile
- Genotype-phenotype correlation
- In vitro functional study
- Animal model creation/study
- Human physiopathology study
- Biomarkers development
- Pre-clinical gene therapy
- Pre-clinical cell therapy
- Pre-clinical drug development/drug delivery
- Diagnostic tool/protocol development
- Pre-clinical vaccine development
- Medical device/instrument development

- Epidemiological study
- Observational clinical study
- Health sociology study
- Health economics study
- Public health study (excluding health economics)
- Natural history study
- Drug repurposing
- Small molecule screening
- Biotechnology innovation
- Induced pluripotent stem cells (iPS) creation/study
- Ontology/bioinformatics study
- Outcomes measures development

Table with ORPHA code of the disease, name of research project's network, country of the coordinator, geographical coverage (regional, national, international).

Clinical trial activities

Table with ORPHA code of the diseases, name of the clinical trial, name of the sponsor, phase of trial, type of trial (drug, protocol, gene therapy, cell therapy, vaccine, medical device), name of the substance/product.

Table with ORPHA code of the disease, name of clinical trial network, country of the coordinator, geographical coverage (regional, national, international).

Patient Registries

Table with ORPHA code of the disease, name of the patient registry, URL of registry, country.

Table with ORPHA code of the disease, name of the patient registry network, country of the coordinator, geographical coverage (regional, national, international).

Biobanks

Table with ORPHA code of the disease, name of the biobank, URL of the biobank, country.

Table with ORPHA code of the disease, name of biobank's network, country of the coordinator, geographical coverage (regional, national, international).

Product 8: All products   Price on request

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