



# Orphadata Products Services & Data Catalogue

June 2025



## OVERVIEW

This catalogue provides an overview of the different services offered by Orphadata Products 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. 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](mailto: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](mailto: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](http://www.orpha.net) and [www.orphadata.com](http://www.orphadata.com).



## A) ORPHADATA SERVICES AND TAILORED DATASETS

You can get in contact with the Orphadata team [data.orphanet@inserm.fr](mailto: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, 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. These files can be used in conjunction with Orphadata Products files, thanks to the inclusion of an ORPHAcodes 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**

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



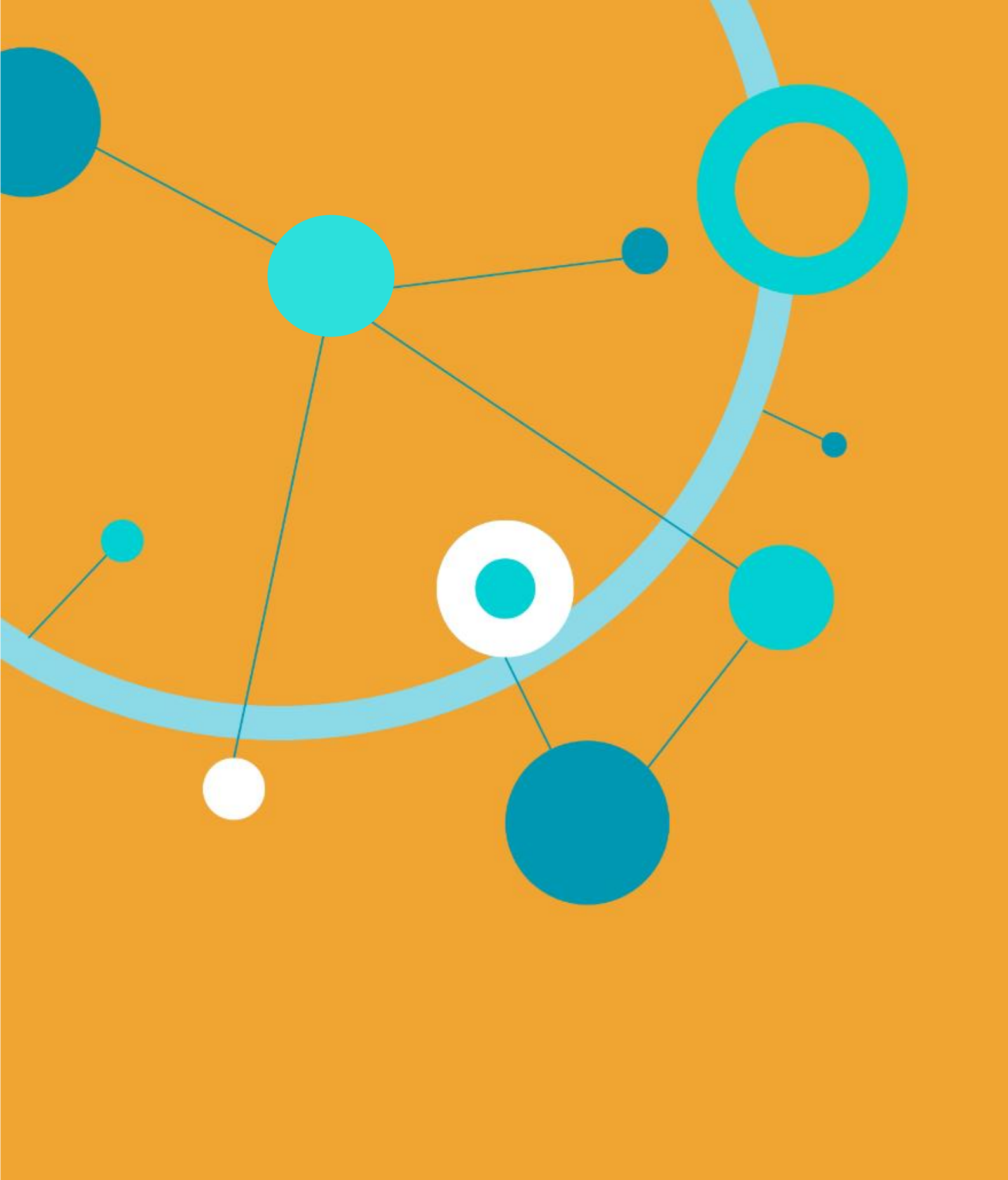
Objectives:

- 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





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[https://www.orphadata.com/docs/Catalogue\\_Orphadata.pdf](https://www.orphadata.com/docs/Catalogue_Orphadata.pdf)