



Catalogue of products 2018

Price list for one-year licence + monthly update for commercial users
a private site at orphadata.org.

Public institutions, academics, and not-for-profits can access this data after
signature of a Data Transfer Agreement for no fee.

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Product 1: Inventory of rare diseases with annotations: No cost – free access

Rare Diseases and cross-references

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

Classifications of rare diseases (poly-hierarchy)

| Clinical classifications of rare diseases | |
|---|--|
| Rare cardiac diseases | Rare systemic and rheumatological diseases |
| Rare developmental anomalies during embryogenesis | Rare odontological diseases |
| Rare inborn errors of metabolism | Rare circulatory system diseases |
| Rare gastroenterological diseases | Rare bone diseases |
| Rare neurological diseases | Rare otorhinolaryngological diseases |
| Rare abdominal surgical diseases | Rare infertility |
| Rare hepatic diseases | Rare neoplastic diseases |
| Rare respiratory diseases | Rare infectious diseases |
| Rare urogenital diseases | Rare intoxications |
| Rare surgical thoracic diseases | Rare gynaecological and obstetric diseases |
| Rare skin diseases | Rare surgical maxillo-facial diseases |
| Rare renal diseases | Rare allergic diseases |
| Rare eye diseases | Rare teratologic diseases |
| Rare endocrine diseases | Rare cardiac malformations |
| Rare haematological diseases | Rare genetic diseases |
| Rare immunological diseases | Rare rheumatologic diseases of childhood |
| | Rare sucking/swallowing diseases |

Diseases with their associated genes

Table with Orpha number 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 number, 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.

Product 2: Textual information: 15,000 Euros

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

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

Table with Orpha number of the disease including specific query to PubMed on the disease.

Product 3: Patient organisations: 3,000 Euros

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

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

Product 4: Expert centres: 5,000 Euros

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

Product 5: Diagnostic tests & clinical laboratories: 10,000 Euros

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 a the name of the test, the purpose, speciality, technique and objective of the test, preferred name and Orpha number 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: 10,000 Euros

Table with Orpha number 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: 15,000 Euros

Table with Orpha number 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 | Epidemiological study |
| Mutation(s) search | Observational clinical study |
| Gene expression profile | Health sociology study |
| Genotype-phenotype correlation | Health economics study |
| In vitro functional study | Public health study (excluding health economics) |
| Animal model creation/study | Natural history study |
| Human physiopathology study | Drug repurposing |
| Biomarkers development | Small molecule screening |
| Pre-clinical gene therapy | Biotechnology innovation |
| Pre-clinical cell therapy | Induced pluripotent stem cells (iPS) creation/study |
| Pre-clinical drug development/drug delivery | Ontology/bioinformatics study |
| Diagnostic tool/protocol development | Outcomes measures development |
| Pre-clinical vaccine development | |
| Medical device/instrument development | |

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

Clinical trial activities

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

Disease Registries - Mutation registries/Databases

Table with Orpha number of the disease, name of the patient registry, URL of registry, country.

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

Table with Orpha number of the disease, Orphan number of the gene, name of mutation registry/database, URL of registry, country.

Biobanks

Table with Orpha number of the disease, name of the biobank, URL of the biobank, country.

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

Product 8: Rare diseases epidemiological data: 10,000 Euros

Table with preferred name and Orpha number 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 number of the diseases, their type of inheritance, interval average age of onset and age of death.

Product 9: All products: 65,000 Euros

Annex : 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