Catalogue of products 2018

One-year licence + monthly update on a private site at orphadata.org

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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 rhumatological 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: *No cost – via signature of a DTA**

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: *No cost – via signature of a DTA**

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: *No cost – via signature of a DTA**

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: *No cost – via signature of a DTA**

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: *No cost – via signature of a DTA**

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: *No cost – via signature of a DTA**

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
Animal model creation/study	economics)
Human physiopathology study	Natural history study
Biomarkers development	Drug repurposing
Pre-clinical gene therapy	Small molecule screening
Pre-clinical cell therapy	Biotechnology innovation
Pre-clinical drug development/drug delivery	Induced pluripotent stem cells (iPS)
Diagnostic tool/protocol development	creation/study
Pre-clinical vaccine development	Ontology/bioinformatics study
Medical device/instrument development	Outcomes measures 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: *No cost – via signature of a DTA**

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: *No cost – via signature of a DTA**

**DTA = Data Transfer Agreement*

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