Catalogue of products 2019

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

The price includes Inserm and Inserm-Transfert management fees.

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

INSERM US14 - Orphanet, Plateforme Maladies Rares, 96 rue Didot, 75014 Paris – France

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Email: data.orphanet@inserm.fr
**Product 2: Textual information: 16,000 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: 4,000 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: 6,000 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: 11,000 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: 11,000 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: 16,000 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

<table>
<thead>
<tr>
<th>Research project</th>
<th>Gene(s) search</th>
<th>Mutation(s) search</th>
<th>Genotype-phenotype correlation</th>
<th>In vitro functional study</th>
<th>Animal model creation/study</th>
<th>Human physiopathology study</th>
<th>Biomarkers development</th>
<th>Pre-clinical gene therapy</th>
<th>Pre-clinical cell therapy</th>
<th>Pre-clinical drug development/drug delivery</th>
<th>Diagnostic tool/protocol development</th>
<th>Pre-clinical vaccine development</th>
<th>Medical device/instrument development</th>
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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).
**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

**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

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

**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