How to make use of RD data: The Orphanet contribution

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Inserm

La science pour la santé
From science to health

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I HAVE NO ACTUAL OR POTENTIAL CONFLICT OF INTEREST IN RELATION TO THIS PROGRAM/PRESENTATION

Making data usable: FAIR

- **F**
- F1. (meta)data are assigned a globally unique and eternally persistent identifier.
- F2. data are described with rich metadata.
- F3. (meta)data are registered or indexed in a searchable resource.
- F4. metadata specify the data identifier.

- **A**
- A1 (meta)data are retrievable by their identifier using a standardized communications protocol.
- A1.1 the protocol is open, free, and universally implementable.
- A1.2 the protocol allows for an authentication and authorization procedure, where necessary.
- A2 metadata are accessible, even when the data are no longer available.

- **I**
- I1. (meta)data use a formal, accessible, shared, and broadly applicable language for knowledge representation.
- I2. (meta)data use vocabularies that follow FAIR principles.
- I3. (meta)data include qualified references to other (meta)data.

- **R**
- R1. meta(data) have a plurality of accurate and relevant attributes.
- R1.1. (meta)data are released with a clear and accessible data usage license.
- R1.2. (meta)data are associated with their provenance.
- R1.3. (meta)data meet domain-relevant community standards.

Interoperability

Care
Health Information System (EHRs)

Research
Registries/Cohorts/ Biobanks

Orphanet central nomenclature

Genes

Genes:
- HGNC
- OMIM
- ensembl
- Genatlas

Proteins:
- UniProt

Pathways:
- Reactome

Targets & compounds:
- IUPHAR

Phenotypes

hpo

HOOM
HPO & ORDO Ontological Module

Disabilities

OD
ODrugs

Terminologies

- OMIM
- ICD10/11
- UMLS
- MedDRA
- SNOMED

orphanet

www.orpha.net

Orphanet RD nomenclature

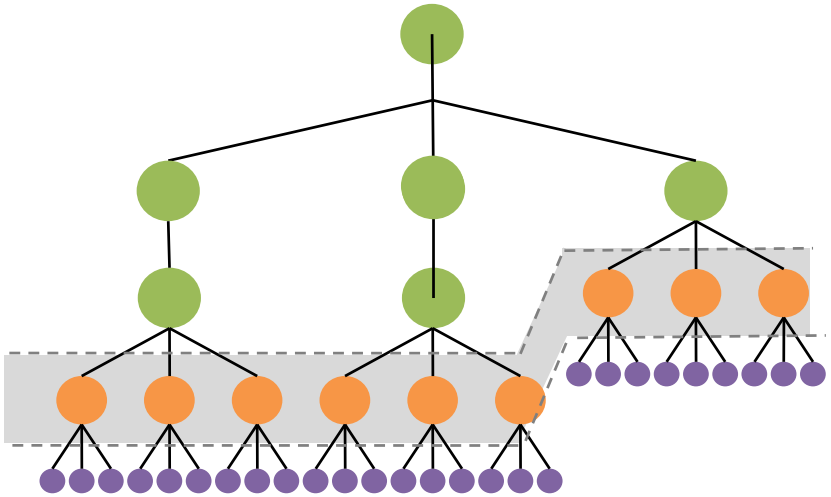
Around **6,000 Rare Diseases (amongst >9500**

ORPHA number	Preferred label	Synonyms
ORPHA:98672	Autosomal dominant optic atrophy	ADOA
ORPHA:893	WAGR syndrome	Del(11)(p13) Deletion 11p13 Monosomy 11p13 Wilms tumor-aniridia-genitourinary anomalies-intellectual disability syndrome
ORPHA:231169	Usher syndrome type 1	USH1

- The only nomenclature **specific for RD**
- **Unique, stable ORPHA number**
- **Definitions**
- 9 languages (Cz, En, Es, De, Fr, It, Nl, Pt, Pl)
- Peer-reviewed publications only (2 cases<RD<1/2000)

Logical structure

Complexity



Category

Group

Disorder: statistical reporting

- Disease, clinical syndrome, malformation syndrome, morphological anomaly, biological anomaly, particular clinical situation

Subtype:

- Etiological, clinical, histopathological

HOME FREE DATA ON REQUEST DATA ONTOLOGIES ABOUT TEAM LEGAL ISSUES CONTACT FAQ

ORPHADATA ONTOLOGIES POWERED BY orphanet

An ontology is a structured vocabulary that describes concepts within the same domain, capturing relationships between these concepts.

ORDO

The Orphanet Rare Disease Ontology (ORDO) was jointly developed by Orphanet and the European Bioinformatics Institute (EMBL-EBI) to provide a structured vocabulary for rare diseases, capturing relationships between diseases, genes and other relevant features, forming a useful resource for the computational analysis of rare diseases.

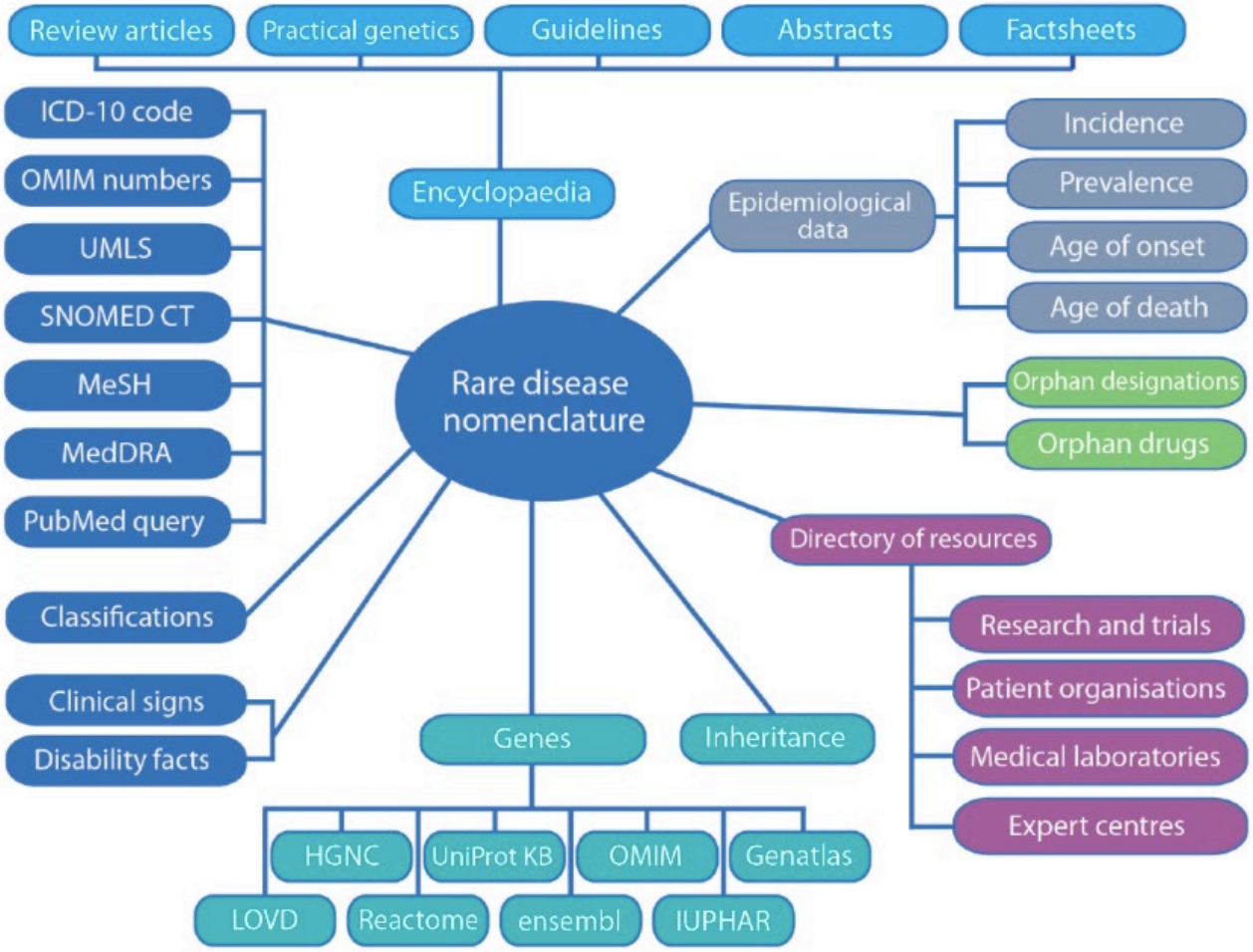
Sparql endpoint

Our freely available data sets can be queried using this SPARQL endpoint.

HOOM

The HPO – ORDO Ontological Module (HOOM) qualifies the annotations between a clinical entity (from ORDO) and phenotypic abnormalities from HPO (Human Phenotype Ontology) according to frequency and by integrating the notion of diagnostic criterion.

A comprehensive, computable, knowledge base for RD data



Making data usable and meaningful

- Adopt FAIR principles
- Adopt semantic standards
- Use available standardized knowledge resources to aggregate and interprete your data
- Interoperate with other data sources
- … and contribute to the improvement of the standards and knowledge resources

Orphanet's contribution to interoperability and usability of data

THANK YOU FOR YOUR ATTENTION