

Resource Summary Report

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Orphanet Rare Disease Ontology

RRID:SCR_010402

Type: Tool

Proper Citation

Orphanet Rare Disease Ontology (RRID:SCR_010402)

Resource Information

URL: <http://purl.bioontology.org/ontology/ORDO>

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Description: Ontology to provide a structured vocabulary for rare diseases capturing relationships between diseases, genes and other relevant features which will form a useful resource for the computational analysis of rare diseases. It derived from the Orphanet database (<http://www.orpha.net>) , a multilingual database dedicated to rare diseases populated from literature and validated by international experts. It integrates a nosology (classification of rare diseases), relationships (gene-disease relations, epidemiological data) and connections with other terminologies (MeSH, SNOMED CT, UMLS, MedDRA), databases (OMIM, UniProtKB, HGNC, ensembl, Reactome, IUPHAR, Geantlas) or classifications (ICD10). The ontology will be maintained by Orphanet and further populated with new data. Orphanet classifications can be browsed in the OLS view. The Orphanet Rare Disease Ontology is updated monthly and follows the OBO guidelines on deprecation of terms. It constitutes the official ontology of rare diseases produced and maintained by Orphanet (INSERM, US14).

Abbreviations: ORDO

Resource Type: ontology, data or information resource, controlled vocabulary

Keywords: owl

Related Condition: Rare disease

Resource Name: Orphanet Rare Disease Ontology

Resource ID: SCR_010402

Alternate IDs: nlx_157540

Record Creation Time: 20220129T080258+0000

Record Last Update: 20260805T044240+0000

Ratings and Alerts

No rating or validation information has been found for Orphanet Rare Disease Ontology.

No alerts have been found for Orphanet Rare Disease Ontology.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We have not found any literature mentions for this resource.