Resource Summary Report

Generated by PRECISE-TBI on Sep 8, 2026

Orphanet

RRID:SCR_006628
Type: Tool

Proper Citation

Orphanet (RRID:SCR_006628)

Resource Information

URL: http://www.orpha.net/

Proper Citation: Orphanet (RRID:SCR_006628)

Description: European website providing information about orphan drugs and rare diseases. It contains content both for physicians and for patients. Reference portal for rare diseases and orphan drugs to help improve diagnosis, care and treatment of patients with rare diseases.

Abbreviations: Orphanet

Resource Type: data or information resource, portal

Keywords: drug, clinical, diagnostic, test, rare, disease, molecule, gene, orphan, drug

Funding: European Union ;
French Directorate General for Health ;
National Institute of Health and Medical Research ;
Rennes ;
France

Availability: Free, Freely available

Resource Name: Orphanet

Resource ID: SCR_006628

Alternate IDs: nif-0000-21306, grid.458406.b, Wikidata: Q1515833

Alternate URLs: https://ror.org/03d3kf570

Record Creation Time: 20220129T080237+0000

Record Last Update: 20260905T133042+0000

Ratings and Alerts

No rating or validation information has been found for Orphanet.

No alerts have been found for Orphanet.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 474 mentions in open access literature.

Listed below are recent publications. The full list is available at PRECISE-TBI .

Li X, et al. (2026) Anesthesia management of a child with Melnick-Needles syndrome associated with retrognathia and significant tracheomalacia. BMC anesthesiology, 26(1).

Gaik C, et al. (2026) Evaluation and perspectives of the OrphanAnesthesia project - a survey among anesthesiologists in Germany. BMC anesthesiology, 26(1).

Groza T, et al. (2026) A systematic assessment of large language models' knowledge of rare diseases: How much do large language models know about rare disease? HGG advances, 7(1), 100558.

Qin X, et al. (2026) TPDdb: the comprehensive database of targeted protein degrader. Nucleic acids research, 54(D1), D1683.

Kang YM, et al. (2026) Awareness and knowledge gaps about pediatric rare disease among anesthesia practitioners in China: a survey-based study. BMC pediatrics, 26(1).

Rosenberger A, et al. (2026) Genes associated with genetic and rare lung diseases and the risk of lung cancer. BMC cancer, 26(1).

Wang H, et al. (2026) CRESTA: a comprehensive transcriptome atlas for cellular response to external stressors. Nucleic acids research, 54(D1), D1005.

Azarbonyad H, et al. (2026) Annotating and indexing scientific articles with rare diseases. Journal of biomedical semantics, 17(1), 3.

Demirkol A, et al. (2026) Diagnostic Role of Chromatic Full-Field Stimulus Test in Rod-Cone Versus Cone Dystrophies. Biomedicines, 14(2).

Ozaki Y, et al. (2026) Novel nonsense variant of KIF11 in a patient with MCLMR. Human genome variation, 13(1).

Prado HV, et al. (2026) Mapping Dental Care for Children and Adolescents With Rare Diseases: A Brazilian Multicentre Study. Community dentistry and oral epidemiology, 54(2), 163.

Marchi E, et al. (2026) Revisiting Minamata disease through computational phenotypic similarity analysis. PloS one, 21(2), e0342655.

Khandelwal MH, et al. (2026) Familial WT1-associated nephropathy - 46, XY Frasier syndrome and 46, XX steroid-resistant nephrotic syndrome in female siblings: A case report and review of literature. World journal of nephrology, 15(1), 110867.

Tumiene B, et al. (2026) Translating multi-omics into healthcare: requisites for scalable and equitable implementation. Human genomics, 20(1).

Reynolds G, et al. (2026) PPP1CB-Related Noonan Syndrome with Loose Anagen Hair: A Systematic Review. Genes, 17(6).

Yang P, et al. (2026) SeekPCMdb: knowledge on disease-associated protein-coding mutations. Nucleic acids research, 54(D1), D1646.

Zhou Y, et al. (2026) theRNA: a curated knowledgebase of functional RNA therapeutics spanning diverse modalities and disease applications. Nucleic acids research, 54(D1), D1672.

DeLuca M, et al. (2026) Medicines, Diseases, Indications, and Contraindications (MeDIC): a foundational resource to support drug repurposing. Nucleic acids research, 54(D1), D1477.

Hernandez-Beeftink T, et al. (2026) Contribution of dominant and recessive model effects to the genetic architecture of Idiopathic Pulmonary Fibrosis. medRxiv : the preprint server for health sciences.

Zhao W, et al. (2026) An agentic system for rare disease diagnosis with traceable reasoning. Nature, 651(8106), 775.