Resource Summary Report

Generated by T1D on Aug 3, 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: National Institute of Health and Medical Research ;
Rennes ;
France ;
French Directorate General for Health ;
European Union

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: 20260803T040440+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 404 mentions in open access literature.

Listed below are recent publications. The full list is available at T1D .

Duque A, et al. (2025) An integrated approach for rare disease detection and classification in Spanish pediatric medical reports. Scientific reports, 15(1), 37973.

Alves D, et al. (2025) Promoting Comprehensive Care for People With Rare Diseases in a Tertiary Care Setting in Brazil: Protocol for a Mixed Methods Implementation Study. JMIR research protocols, 14, e68949.

Criado-Muriel MC, et al. (2025) Diagnostic yield of array-CGH in children with suspected rare disease. Jornal de pediatria, 101(6), 101450.

Bai X, et al. (2025) Quantifying coding integrity and reliability of ICD-11 MMS for rare disease registration: a case study of the Chinese rare disease catalogue. BMC medical informatics and decision making, 25(1), 440.

Aamer N, et al. (2025) Bind: large-scale biological interaction network discovery through knowledge graph-driven machine learning. Journal of translational medicine, 23(1), 856.

Dhombres F, et al. (2025) Revised orphanet nomenclature and classification for spina bifida and other spinal dysraphisms (SBoD). Orphanet journal of rare diseases, 20(1), 348.

Liu F, et al. (2025) MetaGP: A generative foundation model integrating electronic health records and multimodal imaging for addressing unmet clinical needs. Cell reports. Medicine, 6(4), 102056.

Li C, et al. (2025) Prevalence, genetic and clinical characteristics in first-degree relatives of patients with familial cerebral cavernous malformations in China. Stroke and vascular neurology, 10(1), 45.

Li MJ, et al. (2025) Spectrum and epidemiology of rare diseases in a Chinese natural population of 14.31 million residents, 2012-2023. Orphanet journal of rare diseases, 20(1), 410.

Akinbolade S, et al. (2025) Repurposed Medicines: A Scan of the Non-commercial Clinical Research Landscape. Pharmacology research & perspectives, 13(1), e70049.

Boccara O, et al. (2025) Diagnosis and management of superficial arteriovenous malformations: French healthcare network's recommendations. Orphanet journal of rare diseases, 20(1), 45.

Zucca S, et al. (2025) An AI-based approach driven by genotypes and phenotypes to uplift the diagnostic yield of genetic diseases. Human genetics, 144(2-3), 159.

Weber MT, et al. (2025) MedBot vs RealDoc: efficacy of large language modeling in physician-patient communication for rare diseases. Journal of the American Medical Informatics Association : JAMIA, 32(5), 775.

Braidotti S, et al. (2025) Pharmacological evaluation of drug therapies in Aicardi-Goutières syndrome: insights from patient-derived neural stem cells. Frontiers in pharmacology, 16, 1549183.

Michel É, et al. (2025) Rare diseases load through the study of a regional population. PLoS genetics, 21(10), e1011876.

Bougas N, et al. (2025) Employment and work ability in individuals living with rare diseases: a systematic literature review. Orphanet journal of rare diseases, 20(1), 193.

Rodriguez A, et al. (2025) Evaluating re-identification risks scores in publicly available clinical trial datasets: Insights and implications. Clinical trials (London, England), 22(6), 649.

Curado DDSP, et al. (2025) Drugs incorporated into the Brazilian Unified Health System: document analysis of methodological decisions in budget impact analyses between 2012 and 2024. Epidemiologia e servicos de saude : revista do Sistema Unico de Saude do Brasil, 34, e20250122.

Trampuž D, et al. (2025) International Survey on Phenylketonuria Newborn Screening. International journal of neonatal screening, 11(1).

Chen JH, et al. (2025) Evolutionarily new genes in humans with disease phenotypes reveal functional enrichment patterns shaped by adaptive innovation and sexual selection. Genome research, 35(3), 379.