Resource Summary Report

Generated by dkNET on Apr 21, 2025

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: portal, data or information resource

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

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Record Creation Time: 20220129T080237+0000

Record Last Update: 20250421T053557+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 363 mentions in open access literature.

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

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.

Sun C, et al. (2025) Generating unseen diseases patient data using ontology enhanced generative adversarial networks. NPJ digital medicine, 8(1), 4.

Kishk A, et al. (2024) Metabolic models predict fotemustine and the combination of eflornithine/rifamycin and adapalene/cannabidiol for the treatment of gliomas. Briefings in bioinformatics, 25(3).

Domaradzki J, et al. (2024) "In God We Trust": An Exploratory Study of the Associations Between Religiosity and the Caregiving Experiences of Parents of Children with Rare Diseases in Poland. Journal of religion and health, 63(6), 4079.

Liu L, et al. (2024) The health-care utilization and economic burden in patients with genetic skeletal disorders. Orphanet journal of rare diseases, 19(1), 99.

Esteban-Medina M, et al. (2024) drexml: A command line tool and Python package for drug

repurposing. Computational and structural biotechnology journal, 23, 1129.

Berger A, et al. (2024) Pareto-principle in rare disease education: assessing the representation of common rare diseases in medical education and coding systems. Orphanet journal of rare diseases, 19(1), 340.

Lopez-de la Rosa A, et al. (2024) Clinical and genetic characterization of patients with eye diseases included in the Spanish Rare Diseases Patient Registry. Orphanet journal of rare diseases, 19(1), 234.

Fasshauer M, et al. (2024) Monogenic Inborn Errors of Immunity with impaired IgG response to polysaccharide antigens but normal IgG levels and normal IgG response to protein antigens. Frontiers in pediatrics, 12, 1386959.

Rao A, et al. (2024) Health-related quality of life in patients with diverse rare diseases: An online survey. Genetics in medicine open, 2, 101889.

Prignano F, et al. (2024) Epidemiology, Characteristics of Disease, and Unmet Needs of Patients with Generalized Pustular Psoriasis: A Large Italian Delphi Consensus. Dermatology (Basel, Switzerland), 240(3), 414.

Kim HH, et al. (2024) Explicable prioritization of genetic variants by integration of rule-based and machine learning algorithms for diagnosis of rare Mendelian disorders. Human genomics, 18(1), 28.

Iskrov G, et al. (2024) Are the European reference networks for rare diseases ready to embrace machine learning? A mixed-methods study. Orphanet journal of rare diseases, 19(1), 25.

Esteban-Medina M, et al. (2024) The mechanistic functional landscape of retinitis pigmentosa: a machine learning-driven approach to therapeutic target discovery. Journal of translational medicine, 22(1), 139.

Oliveira CC, et al. (2024) A systematic review of studies that estimated the burden of chronic non-communicable rare diseases using disability-adjusted life years. Orphanet journal of rare diseases, 19(1), 333.

Bradshaw MS, et al. (2024) Hypothesis generation for rare and undiagnosed diseases through clustering and classifying time-versioned biological ontologies. PloS one, 19(12), e0309205.

Lindell M, et al. (2024) Comparison of different radiographic methods to measure the slip angle in children with slipped capital femoral epiphysis (SCFE). Acta radiologica (Stockholm, Sweden : 1987), 65(9), 1109.

Zanardi A, et al. (2024) New orphan disease therapies from the proteome of industrial plasma processing waste- a treatment for aceruloplasminemia. Communications biology, 7(1), 140.

Herr K, et al. (2024) Estimating prevalence of rare genetic disease diagnoses using

electronic health records in a children's hospital. HGG advances, 5(4), 100341.