Resource Summary Report

Generated by nidm-terms on Sep 23, 2026

RD-CONNECT

RRID:SCR_014985
Type: Tool

Proper Citation

RD-CONNECT (RRID:SCR_014985)

Resource Information

URL: http://rd-connect.eu/

Proper Citation: RD-CONNECT (RRID:SCR_014985)

Description: Integrated platform that serves as a central resource for rare disease research databases, registries, biobanks and clinical bioinformatics. The RD-CONNECT project utilizes researcher cooperation and data sharing to improve research methods and further scientific understanding of rare diseases.

Resource Type: data or information resource, portal

Keywords: data aggregate, integrated platform, interoperability, rare disease, central resource

Funding: European Union FP7 305444

Availability: The scientific community can contribute to this resource

Resource Name: RD-CONNECT

Resource ID: SCR_014985

Record Creation Time: 20220129T080323+0000

Record Last Update: 20260919T195604+0000

Ratings and Alerts

No rating or validation information has been found for RD-CONNECT.

No alerts have been found for RD-CONNECT.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 26 mentions in open access literature.

Listed below are recent publications. The full list is available at nidm-terms .

Jackson A, et al. (2026) Biallelic variants in RNU2-2 cause a remarkably frequent developmental and epileptic encephalopathy. Nature genetics, 58(4), 798.

Martins N, et al. (2026) Genome Sequencing of Undiagnosed European Patients Suspected of Hereditary Cancer: Diagnostic Yield and Identification of Candidate Causative Variants. JCO precision oncology, 10(4), e2500354.

Jackson A, et al. (2025) Analysis of R-loop forming regions identifies RNU2-2 and RNU5B-1 as neurodevelopmental disorder genes. Nature genetics, 57(6), 1362.

Osmond M, et al. (2025) One-Sided Matching Portal (OSMP): A Tool to Facilitate Rare Disease Patient Matchmaking. Human mutation, 2025, 5941599.

Núñez-Carpintero I, et al. (2024) Rare disease research workflow using multilayer networks elucidates the molecular determinants of severity in Congenital Myasthenic Syndromes. Nature communications, 15(1), 1227.

Hiz Kurul S, et al. (2022) High diagnostic rate of trio exome sequencing in consanguineous families with neurogenetic diseases. Brain : a journal of neurology, 145(4), 1507.

Zschüntzsch J, et al. (2022) The Evolution of Complex Muscle Cell In Vitro Models to Study Pathomechanisms and Drug Development of Neuromuscular Disease. Cells, 11(7).

Buphamalai P, et al. (2021) Network analysis reveals rare disease signatures across multiple levels of biological organization. Nature communications, 12(1), 6306.

Signorelli M, et al. (2021) Peripheral blood transcriptome profiling enables monitoring disease progression in dystrophic mice and patients. EMBO molecular medicine, 13(4), e13328.

Bonne G, et al. (2021) The Treatabolome, an emerging concept. Journal of neuromuscular diseases, 8(3), 337.

Te Paske IBAW, et al. (2021) A mosaic PIK3CA variant in a young adult with diffuse gastric cancer: case report. European journal of human genetics : EJHG, 29(9), 1354.

Harrow J, et al. (2021) ELIXIR: providing a sustainable infrastructure for life science data at European scale. Bioinformatics (Oxford, England), 37(16), 2506.

Bettio C, et al. (2021) The Italian National Registry for FSHD: an enhanced data integration and an analytics framework towards Smart Health Care and Precision Medicine for a rare disease. Orphanet journal of rare diseases, 16(1), 470.

Alves D, et al. (2021) Mapping, Infrastructure, and Data Analysis for the Brazilian Network of Rare Diseases: Protocol for the RARASnet Observational Cohort Study. JMIR research protocols, 10(1), e24826.

Perea-Romero I, et al. (2021) Genetic landscape of 6089 inherited retinal dystrophies affected cases in Spain and their therapeutic and extended epidemiological implications. Scientific reports, 11(1), 1526.

Perea-Romero I, et al. (2021) NGS and phenotypic ontology-based approaches increase the diagnostic yield in syndromic retinal diseases. Human genetics, 140(12), 1665.

Correia FB, et al. (2019) Handling Noise in Protein Interaction Networks. BioMed research international, 2019, 8984248.

Goisauf M, et al. (2019) Data in question: A survey of European biobank professionals on ethical, legal and societal challenges of biobank research. PloS one, 14(9), e0221496.

Boczonadi V, et al. (2018) Mitochondrial oxodicarboxylate carrier deficiency is associated with mitochondrial DNA depletion and spinal muscular atrophy-like disease. Genetics in medicine : official journal of the American College of Medical Genetics, 20(10), 1224.

Tagliafico E, et al. (2018) Workload measurement for molecular genetics laboratory: A survey study. PloS one, 13(11), e0206855.