

Resource Summary Report

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RD-CONNECT

RRID:SCR_014985

Type: Tool

Proper Citation

RD-CONNECT (RRID:SCR_014985)

Resource Information

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

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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: 20260803T040649+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

Usage and Citation Metrics

We found 22 mentions in open access literature.

Listed below are recent publications. The full list is available at [ASWG](#) .

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.

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.

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.

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.

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

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.

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.

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) 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.

Lochmüller H, et al. (2018) RD-Connect, NeurOmics and EURenOmics: collaborative European initiative for rare diseases. European journal of human genetics : EJHG, 26(6), 778.

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.

Bansagi B, et al. (2018) Multifocal demyelinating motor neuropathy and hamartoma syndrome associated with a de novo PTEN mutation. Neurology, 90(21), e1842.

Gainotti S, et al. (2018) The RD-Connect Registry & Biobank Finder: a tool for sharing aggregated data and metadata among rare disease researchers. European journal of human genetics : EJHG, 26(5), 631.

Wood L, et al. (2017) The UK Myotonic Dystrophy Patient Registry: facilitating and accelerating clinical research. Journal of neurology, 264(5), 979.