Resource Summary Report

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Genetic and Rare Diseases Information Center

RRID:SCR_008695
Type: Tool

Proper Citation

Genetic and Rare Diseases Information Center (RRID:SCR_008695)

Resource Information

URL: http://rarediseases.info.nih.gov/GARD/Default.aspx

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Description: Genetic and Rare Diseases Information Center (GARD) is a collaborative effort of two agencies of the National Institutes of Health, The Office of Rare Diseases Research (ORDR) and the National Human Genome Research Institute (NHGRI) to help people find useful information about genetic conditions and rare diseases. GARD provides timely access to experienced information specialists who can furnish current and accurate information about genetic and rare diseases. So far, GARD has responded to 27,635 inquiries on about 7,147 rare and genetic diseases. Requests come not only from patients and their families, but also from physicians, nurses and other health-care professionals. GARD also has proved useful to genetic counselors, occupational and physical therapists, social workers, and teachers who work with people with a genetic or rare disease. Even scientists who are studying a genetic or rare disease and who need information for their research have contacted GARD, as have people who are taking part in a clinical study. Community leaders looking to help people find resources for those with genetic or rare diseases and advocacy groups who want up-to-date disease information for their members have contacted GARD. And members of the media who are writing stories about genetic or rare diseases have found the information GARD has on hand useful, accurate and complete. GARD has information on: :- What is known about a genetic or rare disease. :- What research studies are being conducted. :- What genetic testing and genetic services are available. :- Which advocacy groups to contact for a specific genetic or rare disease. :- What has been written recently about a genetic or rare disease in medical journals. GARD information specialists get their information from: :- NIH resources. :- Medical textbooks. :- Journal articles. :- Web sites. :- Advocacy groups, and their literature and services. :- Medical databases.

Abbreviations: GARD

Synonyms: Genetic Rare Diseases Information Center

Resource Type: data or information resource, disease-related portal, topical portal, portal

Keywords: genetic, disease, information, genome, human, rare disease, health, physician, counselor, gene, journal, medical

Funding: Office of Rare Diseases Research ;
NHGRI

Resource Name: Genetic and Rare Diseases Information Center

Resource ID: SCR_008695

Alternate IDs: nif-0000-37627

Record Creation Time: 20220129T080248+0000

Record Last Update: 20250430T055620+0000

Ratings and Alerts

No rating or validation information has been found for Genetic and Rare Diseases Information Center.

No alerts have been found for Genetic and Rare Diseases Information Center.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 13 mentions in open access literature.

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

Delaye L, et al. (2024) Untangling the Evolution of the Receptor-Binding Motif of SARS-CoV-2. Journal of molecular evolution, 92(3), 329.

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.

Lucaci AG, et al. (2024) The evolution of mammalian Rem2: unraveling the impact of purifying selection and coevolution on protein function, and implications for human disorders.

Frontiers in bioinformatics, 4, 1381540.

Park JY, et al. (2023) Automating Rey Complex Figure Test scoring using a deep learning-based approach: a potential large-scale screening tool for cognitive decline. Alzheimer's research & therapy, 15(1), 145.

Lang D, et al. (2022) The Emergence and Dynamics of Tick-Borne Encephalitis Virus in a New Endemic Region in Southern Germany. Microorganisms, 10(11).

Sattar S, et al. (2022) Phenotypic characterization and genome analysis of a novel Salmonella Typhimurium phage having unique tail fiber genes. Scientific reports, 12(1), 5732.

Lewin TD, et al. (2021) Dynamic Molecular Evolution of Mammalian Homeobox Genes: Duplication, Loss, Divergence and Gene Conversion Sculpt PRD Class Repertoires. Journal of molecular evolution, 89(6), 396.

Toshkhujaev S, et al. (2020) Classification of Alzheimer's Disease and Mild Cognitive Impairment Based on Cortical and Subcortical Features from MRI T1 Brain Images Utilizing Four Different Types of Datasets. Journal of healthcare engineering, 2020, 3743171.

Vázquez-Rosas-Landa M, et al. (2020) Population genomics of Vibrionaceae isolated from an endangered oasis reveals local adaptation after an environmental perturbation. BMC genomics, 21(1), 418.

Zhu Q, et al. (2020) An integrative knowledge graph for rare diseases, derived from the Genetic and Rare Diseases Information Center (GARD). Journal of biomedical semantics, 11(1), 13.

Kotov AA, et al. (2016) Phylogeography of the Chydorus sphaericus Group (Cladocera: Chydoridae) in the Northern Palearctic. PloS one, 11(12), e0168711.

Gross R, et al. (2014) Quasispecies in population of compositional assemblies. BMC evolutionary biology, 14, 265.

Bello G, et al. (2011) The Use of Bioinformatics for Studying HIV Evolutionary and Epidemiological History in South America. AIDS research and treatment, 2011, 154945.