Resource Summary Report

Generated by <u>NIF</u> on May 23, 2025

Residual Variation Intolerance Score (RVIS)

RRID:SCR_013850 Type: Tool

Proper Citation

Residual Variation Intolerance Score (RVIS) (RRID:SCR_013850)

Resource Information

URL: http://chgv.org/GenicIntolerance/

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Description: A gene-based score intended to help in the interpretation of human sequence data. The score is designed to rank genes in terms of whether they have more or less common functional genetic variation relative to the genome wide expectation given the amount of apparently neutral variation the gene has. A gene with a positive score has more common functional variation, and a gene with a negative score has less and is referred to as intolerant.

Abbreviations: RVIS

Synonyms: Residual Variation Intolerance Score

Resource Type: narrative resource, data or information resource, standard specification

Defining Citation: DOI:10.1371/journal.pgen.1003709

Keywords: gene, score, sequence, interpretation, rank, functional genetic variation

Funding: NIH Epi4K Sequencing ; Bioinformatics and Biostatistics Core U01NS077303

Resource Name: Residual Variation Intolerance Score (RVIS)

Resource ID: SCR_013850

License URLs: http://chgv.org/GenicIntolerance/terms.jsp

Record Creation Time: 20220129T080318+0000

Record Last Update: 20250523T055014+0000

Ratings and Alerts

No rating or validation information has been found for Residual Variation Intolerance Score (RVIS).

No alerts have been found for Residual Variation Intolerance Score (RVIS).

Data and Source Information

Source: <u>SciCrunch Registry</u>

Usage and Citation Metrics

We found 8 mentions in open access literature.

Listed below are recent publications. The full list is available at <u>NIF</u>.

Dimitrijevic S, et al. (2022) KCC2 rs2297201 Gene Polymorphism Might be a Predictive Genetic Marker of Febrile Seizures. ASN neuro, 14, 17590914221093257.

Samarakoon PS, et al. (2016) cnvScan: a CNV screening and annotation tool to improve the clinical utility of computational CNV prediction from exome sequencing data. BMC genomics, 17, 51.

Nicolaou N, et al. (2016) Prioritization and burden analysis of rare variants in 208 candidate genes suggest they do not play a major role in CAKUT. Kidney international, 89(2), 476.

Thomas RA, et al. (2016) Identification of genetic variants of LGI1 and RTN4R (NgR1) linked to schizophrenia that are defective in NgR1-LGI1 signaling. Molecular genetics & genomic medicine, 4(4), 447.

Bush WS, et al. (2016) Genetic variation among 82 pharmacogenes: The PGRNseq data from the eMERGE network. Clinical pharmacology and therapeutics, 100(2), 160.

Wang Q, et al. (2015) Increased co-expression of genes harboring the damaging de novo mutations in Chinese schizophrenic patients during prenatal development. Scientific reports, 5, 18209.

Jiang Y, et al. (2015) Incorporating Functional Information in Tests of Excess De Novo Mutational Load. American journal of human genetics, 97(2), 272.

, et al. (2014) De novo mutations in synaptic transmission genes including DNM1 cause epileptic encephalopathies. American journal of human genetics, 95(4), 360.