Resource Summary Report

Generated by FDI Lab - SciCrunch.org on Apr 24, 2025

MutationTaster

RRID:SCR_010777
Type: Tool

Proper Citation

MutationTaster (RRID:SCR_010777)

Resource Information

URL: http://www.mutationtaster.org/

Proper Citation: MutationTaster (RRID:SCR_010777)

Description: Evaluates disease-causing potential of sequence alterations.

Abbreviations: MutationTaster

Resource Type: production service resource, data analysis service, analysis service resource, service resource

Defining Citation: PMID:20676075

Keywords: bio.tools

Funding:

Availability: Acknowledgement requested

Resource Name: MutationTaster

Resource ID: SCR_010777

Alternate IDs: biotools:mutation_taster, OMICS_00153

Alternate URLs: https://bio.tools/mutation_taster

Record Creation Time: 20220129T080300+0000

Record Last Update: 20250424T065113+0000

Ratings and Alerts

No rating or validation information has been found for MutationTaster.

No alerts have been found for MutationTaster.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 3892 mentions in open access literature.

Listed below are recent publications. The full list is available at FDI Lab - SciCrunch.org.

Heimer G, et al. (2025) Biallelic PIGM Coding Variant Causes Intractable Epilepsy and Intellectual Disability Without Thrombotic Events. Clinical genetics, 107(2), 179.

Yang J, et al. (2025) Manic Fringe promotes endothelial-to-mesenchymal transition mediated by the Notch signalling pathway during heart valve development. Journal of molecular medicine (Berlin, Germany), 103(1), 51.

Smith TB, et al. (2025) Bi-allelic variants in DAP3 result in reduced assembly of the mitoribosomal small subunit with altered apoptosis and a Perrault-syndrome-spectrum phenotype. American journal of human genetics, 112(1), 59.

Katsonis P, et al. (2025) Meta-EA: a gene-specific combination of available computational tools for predicting missense variant effects. Nature communications, 16(1), 159.

Albitar L, et al. (2025) Detection of exon2-MED12 mutations in uterine leiomyomas from Syrian patients. Scientific reports, 15(1), 452.

Bibi H, et al. (2025) Molecular and computational analysis of a novel pathogenic variant in emopamil-binding protein (EBP) involved in cholesterol biosynthetic pathway causing a rare male EBP disorder with neurologic defects (MEND syndrome). Molecular biology reports, 52(1), 101.

Alayoubi AM, et al. (2025) Zellweger syndrome; identification of mutations in PEX19 and PEX26 gene in Saudi families. Annals of medicine, 57(1), 2447400.

Bayam E, et al. (2025) Bi-allelic variants in WDR47 cause a complex neurodevelopmental syndrome. EMBO molecular medicine, 17(1), 129.

Yang L, et al. (2025) A novel de novo GABRA2 gene missense variant causing developmental epileptic encephalopathy in a Chinese patient. Annals of clinical and translational neurology, 12(1), 137.

Zhang S, et al. (2025) Exploratory analysis of a Novel RACK1 mutation and its potential role in epileptic seizures via Microglia activation. Journal of neuroinflammation, 22(1), 27.

El Fissi H, et al. (2025) Mucopolysaccharidoses types I and IIIA: Diagnosis and identification of novel polymorphisms associated with common mutations in Moroccan patients. Molecular genetics and metabolism reports, 42, 101186.

Fan X, et al. (2025) Genotype-phenotype correlations for 17 Chinese families with inherited retinal dystrophies due to homozygous variants. Scientific reports, 15(1), 3043.

Zin OA, et al. (2025) Genotype-Phenotype Correlations of Nance-Horan Syndrome in Male and Female Carriers of a Novel Variant. Genes, 16(1).

Dong J, et al. (2025) A Case Study Identified a New Mutation in the TTN Gene for Inherited Hypertrophic Cardiomyopathy. International journal of general medicine, 18, 447.

Yang Y, et al. (2025) A De Novo Frameshift Variant in SMC1A Causes Non-Classic Cornelia de Lange Syndrome With Epilepsy: A Case Report and Literature Review. Molecular genetics & genomic medicine, 13(1), e70058.

Singh S, et al. (2025) Biallelic variants in CCN2 underlie an autosomal recessive kyphomelic dysplasia. European journal of human genetics : EJHG, 33(1), 30.

Aref-Eshghi E, et al. (2025) Germline de novo alterations of RUNX1T1 in individuals with neurodevelopmental and congenital anomalies. HGG advances, 6(1), 100384.

Sallo FB, et al. (2025) Characterization of the Retinal Phenotype Using Multimodal Imaging in Novel Compound Heterozygote Variants of CYP2U1. Ophthalmology science, 5(1), 100618.

Zhou Z, et al. (2025) A novel homozygous mutation of CFAP300 identified in a Chinese patient with primary ciliary dyskinesia and infertility. Asian journal of andrology, 27(1), 113.

Shu Y, et al. (2025) Dias-Logan syndrome with a de novo p.Leu360Profs*212 heterozygous pathogenic variant of BCL11A in a Chinese patient: A case report. SAGE open medical case reports, 13, 2050313X251314069.