

Resource Summary Report

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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: analysis service resource, data analysis service, production service resource, service resource

Defining Citation: [PMID:20676075](#)

Keywords: bio.tools

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: 20260821T042853+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 4781 mentions in open access literature.

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

Le DM, et al. (2026) Distinct Associations of GTF2I, TP53, and NOTCH1 Variants with Indolent and Aggressive Thymic Epithelial Tumors in Vietnamese Patients. *Genes*, 17(5).

Lipiński P, et al. (2026) Riboflavin treatment in L-2-hydroxyglutaric aciduria: report on a pediatric patient and literature review. *Journal of applied genetics*, 67(2), 443.

Chen Y, et al. (2026) Diagnosis and follow-up of a PCDH19 epilepsy patient. *Psychiatric genetics*, 36(1), 26.

Owring D, et al. (2026) Uncovering dual molecular diagnoses in families with complex phenotypes through structural and clinical studies of novel COL4A6 variants. *QJM : monthly journal of the Association of Physicians*, 119(3), 187.

Jiang Y, et al. (2026) Prenatal Exome Sequencing Analysis in Fetuses With Structural Anomalies: A Multicenter Prospective Cohort Study With Practical Implications. *Prenatal diagnosis*, 46(1), 46.

Krantz S, et al. (2026) Dissection of Gs and Hedgehog Signaling Cross-talk Reveals Therapeutic Opportunities to Target Hedgehog-Dependent Tumors. *Cancer research*, 86(4), 940.

Wang M, et al. (2026) Exploration of the Pathogenic Mechanism of the Factor XIII A Subunit in a Patient With Congenital Factor XIII Deficiency. *Molecular genetics & genomic medicine*, 14(1), e70193.

Ou S, et al. (2026) Clinical and Genetic Analysis of SMARCC2-Related Diseases in Three Chinese Patients. *Molecular genetics & genomic medicine*, 14(1), e70198.

Zhang YW, et al. (2026) A homozygous variant in HFM1 causes preimplantation embryo developmental arrest by disrupting zygotic genome activation. *Human reproduction (Oxford, England)*, 41(2), 296.

Jenni R, et al. (2026) Novel genetic variants identification and immune profiling in ataxia telangiectasia patients. *Journal of translational medicine*, 24(1).

Fatima S, et al. (2026) A novel protein truncating mutation of TTC8 causes Bardet-Biedl Syndrome (BBS) in a Pakistani family. *Genetics and molecular biology*, 49(1), e20250020.

Rey F, et al. (2026) Study of POLR3A variants in a family trio suggests mutation-specific pathogenetic mechanisms: insights from integrative OMIC approaches. *Cell communication and signaling : CCS*, 24(1).

Babur Güler G, et al. (2026) Phenotypic, Epidemiologic, and Imaging Features of Hypertrophic Cardiomyopathy: A Single-Center Experience. *Anatolian journal of cardiology*, 30(5), 321.

Kikuchi S, et al. (2026) Compound heterozygous CHAT gene mutations, a missense and a splice site variant, in two siblings with congenital myasthenic syndrome. *Scientific reports*, 16(1).

Yoon JG, et al. (2026) A predictive framework for stop-loss variants with C-terminal extensions. *Nucleic acids research*, 54(2).

Li JN, et al. (2026) Clinical features and genetic analysis of androgen receptor gene variants in 30 prepubertal patients with androgen insensitivity syndrome. *Asian journal of andrology*, 28(3), 335.

Aslam F, et al. (2026) Molecular characterization of recessively inherited ataxic and neuropathic disorders in consanguineous Pakistani families. *Scientific reports*, 16(1), 6529.

Santacruz Reyes MD, et al. (2026) Isobutyryl-coenzyme a dehydrogenase deficiency: disease, or non-disease? *Orphanet journal of rare diseases*, 21(1), 35.

Chaczka B, et al. (2026) Molecular Profiling of Polish Pediatric Patients with Epilepsy: A Single-Center Diagnostic Experience Using Next-Generation Sequencing. *Genes*, 17(2).

Zhu Y, et al. (2026) Short tandem repeat expansions in patients with neurodegenerative dementia. *EBioMedicine*, 125, 106190.