

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 [NIF](#) .

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).

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.

Wu G, et al. (2026) Pathogenicity and Functional Analysis of Multi-Variant Allele of RPE65 Causing Retinitis Pigmentosa. *Translational vision science & technology*, 15(2), 1.

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.

Ullah MF, et al. (2026) Whole-exome sequencing in obstructive coronary artery disease identifies rare and novel variants in cardiac arrhythmia and pulmonary arterial hypertension-associated genes. *Biomolecules & biomedicine*, 26(8), 1344.

Huang M, et al. (2026) WFS1-related isolated diabetes induced by a WFS1 missense mutation: focus on the isolated diabetes phenotype. *Orphanet journal of rare diseases*, 21(1).

Gao Y, et al. (2026) Clinical and Functional Characterization of Novel GALNT3 Mutations in a Chinese Child with Hyperphosphatemic Familial Tumoral Calcinosis. *International journal of molecular sciences*, 27(6).

Armengol-Alsina M, et al. (2026) Placental insufficiency markers to assess the risk of non-chromosomal genetic conditions in early-onset fetal growth restriction. *Ultrasound in obstetrics & gynecology : the official journal of the International Society of Ultrasound in Obstetrics and Gynecology*, 67(4), 492.

Zhou Z, et al. (2026) Somatic mosaicism in ALS and FTD identifies focal mutations associated with widespread degeneration. *Nature genetics*, 58(5), 1019.

Wei X, et al. (2026) A Novel CRYBB2 Splicing Mutation Is Associated With Lens Extracellular Matrix Remodeling and Vascular Alterations in Congenital Cataract. *Investigative ophthalmology & visual science*, 67(4), 36.

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

Babur Guler G, et al. (2026) Evaluation of artificial intelligence-based electrocardiogram analysis tools in patients with hypertrophic cardiomyopathy. *European heart journal. Digital health*, 7(2), ztag026.

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.

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

Su X, et al. (2026) Family-based genome-wide association study for asthma among Han Chinese children. *BMC pediatrics*, 26(1).

Wang PY, et al. (2026) Toward precision medicine in SCN3A variants-associated encephalopathies and epilepsy: optimizing genetic diagnosis and molecular subregional effects. *Frontiers in neurology*, 17, 1772239.

Sala E, et al. (2026) Intrafamilial phenotypic variability in hypophosphatasia: evidence from two families and the literature. *Frontiers in endocrinology*, 17, 1826307.

Ülker Üstebay D, et al. (2026) TRMT10A-Related Neurodevelopmental Disorder Without Metabolic Findings. *Human mutation*, 2026, 8058409.