Resource Summary Report

Generated by Kravitz on Sep 10, 2026

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: 20260905T133200+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 Kravitz .

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).

Owrang 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.