Resource Summary Report

Generated by SPARC SAWG on Sep 14, 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: 20260912T200206+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 SPARC SAWG .

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).

Lipi?ski 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.

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.

Krantz S, et al. (2026) Dissection of G?s 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.

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).

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