Resource Summary Report

Generated by RRID on Aug 15, 2026

LUMC Mutalyzer 3

RRID:SCR_026148
Type: Tool

Proper Citation

LUMC Mutalyzer 3 (RRID:SCR_026148)

Resource Information

URL: https://mutalyzer.nl/

Proper Citation: LUMC Mutalyzer 3 (RRID:SCR_026148)

Description: Software suite is designed to be of help when working with Human Genome Variation Society sequence variant nomenclature descriptions.

Synonyms: Mutalyzer 3

Resource Type: source code, software application, software resource

Keywords: HGVS variant descriptions, Human Genome Variation Society variant, HGVS, sequence variant,

Availability: Free, Available for download, Freely available

Resource Name: LUMC Mutalyzer 3

Resource ID: SCR_026148

License: MIT license

Record Creation Time: 20241210T053256+0000

Record Last Update: 20260815T042923+0000

Ratings and Alerts

No rating or validation information has been found for LUMC Mutalyzer 3.

No alerts have been found for LUMC Mutalyzer 3.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 78 mentions in open access literature.

Listed below are recent publications. The full list is available at RRID .

Calì F, et al. (2026) Potential Link Between a Disruptive CAPN6 Variant and Neurodevelopmental Disorders. International journal of molecular sciences, 27(3).

Zhang H, et al. (2026) Molecular mechanisms and therapeutic strategies for the recurrent F9 (c.520 + 13 A > G) variant in hemophilia B. Human genomics, 20(1), 33.

Resende KK, et al. (2026) Pre-eruptive Coronal Resorptions as a Clinical Feature of FAM83H-Related Amelogenesis Imperfecta: Insights from Two Brazilian Families. Calcified tissue international, 117(1).

Esmaeil Lashgarian H, et al. (2026) A Novel AP4M1 Variant in an Iranian Child with Spastic Paraplegia 50: A Case Report and Molecular Docking Approach. Iranian journal of medical sciences, 51(1), 70.

Fan S, et al. (2026) Identification and Functional Characterization of a Novel POU3F4 Frameshift Mutation in a Chinese Family. Life (Basel, Switzerland), 16(6).

Taha HB, et al. (2026) Prosaposin in CNS health and disease, metabolic stress and exercise adaptation. Journal of molecular medicine (Berlin, Germany), 104(1), 38.

Smaili F, et al. (2026) DYSF gene variant spectrum in Arab populations across eight countries: A systematic review. Biomolecules & biomedicine, 26(9), 1511.

Ma RX, et al. (2025) Case Report: Two children with factor XII deficiency caused by novel F12 compound heterozygous variants. Frontiers in pediatrics, 13, 1555426.

Heutinck PAT, et al. (2025) Diagnostic whole exome sequencing in presumably autosomal recessive inherited retinal dystrophies in an Iranian population. Scientific reports, 15(1), 23745.

Akhtar Z, et al. (2025) Mutations in retinal cyclic nucleotide-gated channels identified in familial cases of inherited retinal dystrophies from Pakistan. Molecular vision, 31, 206.

Cheerie D, et al. (2025) Consensus guidelines for assessing eligibility of pathogenic DNA variants for antisense oligonucleotide treatments. American journal of human genetics, 112(5), 975.

Genio E, et al. (2025) CTLA4 Alteration and Neurologic Manifestations: A New Family with Large Phenotypic Variability and Literature Review. Genes, 16(3).

de Melo MEC, et al. (2025) The Role of the AGPAT2 Gene in Adipose Tissue Biology and Congenital Generalized Lipodystrophy Pathophysiology. International journal of molecular sciences, 26(11).

Bouys L, et al. (2025) The mutational landscape of ARMC5 in Primary Bilateral Macronodular Adrenal Hyperplasia: an update. Orphanet journal of rare diseases, 20(1), 51.

Yan C, et al. (2025) Identification of a Novel EVC2 Variant in a Family with Non-Syndromic Tooth Agenesis and Its Potential Functional Implications. Genes, 16(11).

Kovermann P, et al. (2025) The severity of SLC1A2-associated neurodevelopmental disorders correlates with transporter dysfunction. EBioMedicine, 114, 105648.

Kwong A, et al. (2025) Germline BARD1 Mutation in High-Risk Chinese Breast and Ovarian Cancer Patients. Cancers, 17(15).

Arranz-Ledo M, et al. (2025) Genetic Features of Tumours Arising in the Context of Suspected Hereditary Cancer Syndromes with RAD50, RAD51C/D, and BRIP1 Germline Mutations, Results of NGS-Reanalysis of BRCA/MMR-Negative Families. Genes, 16(4).

De la Rosa SO, et al. (2025) MBOAT7 encephalopathy: Characterizing the neurology and epileptology. Epilepsia, 66(7), 2379.

Méndez-Vidal C, et al. (2025) A genomic strategy for precision medicine in rare diseases: integrating customized algorithms into clinical practice. Journal of translational medicine, 23(1), 86.