Resource Summary Report

Generated by dkNET on Apr 30, 2025

GeneTalk

RRID:SCR_005231
Type: Tool

Proper Citation

GeneTalk (RRID:SCR_005231)

Resource Information

URL: http://www.gene-talk.de

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Description: A web-based tool, knowledgebase and community for analysis and interpretation of human variant files. VCFs (Variant Call Formats) are preprocessed and annotated, you can filter them, access all databases and provide your expertise to the community by creating annotations.

Abbreviations: GeneTalk

Synonyms: GeneTalk - The Professional Network and Online Tool for Geneticists

Resource Type: data repository, service resource, database, narrative resource, community building portal, storage service resource, portal, data or information resource, blog

Defining Citation: PMID:22826540

Keywords: sequence variant, annotation, exome sequencing, genetic variant, gene, data sharing, bio.tools

Funding:

Availability: The community can contribute to this resource, Free, (during beta period)

Resource Name: GeneTalk

Resource ID: SCR_005231

Alternate IDs: OMICS_00270, biotools:genetalk

Alternate URLs: https://bio.tools/genetalk

Record Creation Time: 20220129T080229+0000

Record Last Update: 20250430T055337+0000

Ratings and Alerts

No rating or validation information has been found for GeneTalk.

No alerts have been found for GeneTalk.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 29 mentions in open access literature.

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

Chen B, et al. (2024) Predictive role of ctDNA in esophageal squamous cell carcinoma receiving definitive chemoradiotherapy combined with toripalimab. Nature communications, 15(1), 1919.

Zhao R, et al. (2023) Clonal dynamics and Stereo-seq resolve origin and phenotypic plasticity of adenosquamous carcinoma. NPJ precision oncology, 7(1), 80.

Zuo Y, et al. (2022) Genomic and epigenomic profiles distinguish pulmonary enteric adenocarcinoma from lung metastatic colorectal cancer. EBioMedicine, 82, 104165.

Panzer M, et al. (2022) Synonymous mutation in adenosine triphosphatase copper-transporting beta causes enhanced exon skipping in Wilson disease. Hepatology communications, 6(7), 1611.

Butscheidt S, et al. (2021) Relevant genetic variants are common in women with pregnancy and lactation-associated osteoporosis (PLO) and predispose to more severe clinical manifestations. Bone, 147, 115911.

Mandato C, et al. (2021) A ZFYVE19 gene mutation associated with neonatal cholestasis and cilia dysfunction: case report with a novel pathogenic variant. Orphanet journal of rare diseases, 16(1), 179.

Balashova MS, et al. (2020) The spectrum of pathogenic variants of the ATP7B gene in Wilson disease in the Russian Federation. Journal of trace elements in medicine and biology : organ of the Society for Minerals and Trace Elements (GMS), 59, 126420.

Rolvien T, et al. (2020) Whole-Exome Sequencing Identifies Novel Compound Heterozygous ZNF469 Mutations in Two Siblings with Mild Brittle Cornea Syndrome. Calcified tissue international, 107(3), 294.

Torrorey-Sawe R, et al. (2020) Pioneering Informed Consent for Return of Research Results to Breast Cancer Patients Facing Barriers to Implementation of Genomic Medicine: The Kenyan BRCA1/2 Testing Experience Using Whole Exome Sequencing. Frontiers in genetics, 11, 170.

Howaldt A, et al. (2019) Sclerosing bone dysplasias with hallmarks of dysosteosclerosis in four patients carrying mutations in SLC29A3 and TCIRG1. Bone, 120, 495.

Yang J, et al. (2018) A likely pathogenic variant putatively affecting splicing of PIGA identified in a multiple congenital anomalies hypotonia-seizures syndrome 2 (MCAHS2) family pedigree via whole-exome sequencing. Molecular genetics & genomic medicine, 6(5), 739.

Mrosk J, et al. (2018) Diagnostic strategies and genotype-phenotype correlation in a large Indian cohort of osteogenesis imperfecta. Bone, 110, 368.

Heinrich V, et al. (2017) A likelihood ratio-based method to predict exact pedigrees for complex families from next-generation sequencing data. Bioinformatics (Oxford, England), 33(1), 72.

Kuo DS, et al. (2017) Characterization of a variant of gap junction protein ?8 identified in a family with hereditary cataract. PloS one, 12(8), e0183438.

van der Merwe N, et al. (2017) Exome Sequencing in a Family with Luminal-Type Breast Cancer Underpinned by Variation in the Methylation Pathway. International journal of molecular sciences, 18(2).

Ehmke N, et al. (2017) De Novo Mutations in SLC25A24 Cause a Craniosynostosis Syndrome with Hypertrichosis, Progeroid Appearance, and Mitochondrial Dysfunction. American journal of human genetics, 101(5), 833.

Knaus A, et al. (2016) Rare Noncoding Mutations Extend the Mutational Spectrum in the PGAP3 Subtype of Hyperphosphatasia with Mental Retardation Syndrome. Human mutation, 37(8), 737.

Lindert U, et al. (2016) MBTPS2 mutations cause defective regulated intramembrane proteolysis in X-linked osteogenesis imperfecta. Nature communications, 7, 11920.

Lim EC, et al. (2015) Next-generation sequencing using a pre-designed gene panel for the molecular diagnosis of congenital disorders in pediatric patients. Human genomics, 9, 33.

Rymen D, et al. (2015) Key features and clinical variability of COG6-CDG. Molecular

genetics and metabolism, 116(3), 163.