Resource Summary Report

Generated by dkNET on May 5, 2025

KGGSeq

RRID:SCR_005311
Type: Tool

Proper Citation

KGGSeq (RRID:SCR_005311)

Resource Information

URL: http://statgenpro.psychiatry.hku.hk/limx/kggseq/

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Description: A biological Knowledge-based mining platform for Genomic and Genetic studies using Sequence data. The software platform, constituted of bioinformatics and statistical genetics functions, makes use of valuable biologic resources and knowledge for sequencing-based genetic mapping of variants / genes responsible for human diseases / traits. It facilitates geneticists to fish for the genetic determinants of human diseases / traits in the big sea of DNA sequences. KGGSeq has paid attention to downstream analysis of genetic mapping. The framework was implemented to filter and prioritize genetic variants from whole exome sequencing data.

Abbreviations: KGGSeq

Synonyms: KGGSeq: A biological Knowledge-based mining platform for Genomic and Genetic studies using Sequence data

Resource Type: software resource

Defining Citation: PMID:22241780

Keywords: genomic, genetic, sequence, mutation, exome sequencing, disease, gene, variant, bio.tools

Related Condition: Monogenic disorder, Cancer

Funding:

Resource Name: KGGSeq

Resource ID: SCR_005311

Alternate IDs: biotools:kggseq, OMICS_02260

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

Record Creation Time: 20220129T080229+0000

Record Last Update: 20250420T014248+0000

Ratings and Alerts

No rating or validation information has been found for KGGSeq.

No alerts have been found for KGGSeq.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 53 mentions in open access literature.

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

Amin AK, et al. (2024) 11p13 microduplication: a differential diagnosis of Silver-Russell syndrome? Molecular cytogenetics, 17(1), 5.

Venger K, et al. (2023) Unusual phenotypes in patients with a pathogenic germline variant in DICER1. Familial cancer, 22(4), 475.

Loose S, et al. (2023) Peripheral temperature dysregulation associated with functionally altered NaV1.8 channels. Pflugers Archiv : European journal of physiology, 475(11), 1343.

Bilo L, et al. (2023) Molecular characterisation of 36 multilocus imprinting disturbance (MLID) patients: a comprehensive approach. Clinical epigenetics, 15(1), 35.

Chui MMC, et al. (2023) Evaluating High-Confidence Genes in Conotruncal Cardiac Defects by Gene Burden Analyses. Journal of the American Heart Association, 12(4), e028226.

Jiang L, et al. (2022) Deviation from baseline mutation burden provides powerful and robust rare-variants association test for complex diseases. Nucleic acids research, 50(6), e34.

Xu Y, et al. (2022) Adult human kidney organoids originate from CD24+ cells and represent an advanced model for adult polycystic kidney disease. Nature genetics, 54(11), 1690.

Tang CSM, et al. (2022) Sequencing of a Chinese tetralogy of Fallot cohort reveals clustering mutations in myogenic heart progenitors. JCI insight, 7(2).

Pagliari MT, et al. (2021) Role of ADAMTS13, VWF and F8 genes in deep vein thrombosis. PloS one, 16(10), e0258675.

Kim YJ, et al. (2021) The burden of rare damaging variants in hereditary atypical parkinsonism genes is increased in patients with Parkinson's disease. Neurobiology of aging, 100, 118.e5.

Papa R, et al. (2021) Type I interferon activation in RAS-associated autoimmune leukoproliferative disease (RALD). Clinical immunology (Orlando, Fla.), 231, 108837.

Amanat S, et al. (2021) Burden of rare variants in synaptic genes in patients with severe tinnitus: An exome based extreme phenotype study. EBioMedicine, 66, 103309.

Li M, et al. (2021) A host-based whole genome sequencing study reveals novel risk loci associated with severity of influenza A(H1N1)pdm09 infection. Emerging microbes & infections, 10(1), 123.

Di Fede E, et al. (2021) Expanding the phenotype associated to KMT2A variants: overlapping clinical signs between Wiedemann-Steiner and Rubinstein-Taybi syndromes. European journal of human genetics : EJHG, 29(1), 88.

Meyer R, et al. (2021) One test for all: whole exome sequencing significantly improves the diagnostic yield in growth retarded patients referred for molecular testing for Silver-Russell syndrome. Orphanet journal of rare diseases, 16(1), 42.

Marx D, et al. (2021) Atypical focal segmental glomerulosclerosis associated with a new PODXL nonsense variant. Molecular genetics & genomic medicine, 9(5), e1658.

Lam WY, et al. (2021) Identification of a wide spectrum of ciliary gene mutations in nonsyndromic biliary atresia patients implicates ciliary dysfunction as a novel disease mechanism. EBioMedicine, 71, 103530.

Kim JJ, et al. (2020) No genetic evidence for involvement of alcohol dehydrogenase genes in risk for Parkinson's disease. Neurobiology of aging, 87, 140.e19.

Jiang H, et al. (2020) Exome sequencing analysis identifies frequent oligogenic involvement and FLNB variants in adolescent idiopathic scoliosis. Journal of medical genetics, 57(6), 405.

Castro CN, et al. (2020) NCKAP1L defects lead to a novel syndrome combining immunodeficiency, lymphoproliferation, and hyperinflammation. The Journal of experimental medicine, 217(12).