Resource Summary Report

Generated by <u>NIF</u> on May 8, 2025

MetaSKAT

RRID:SCR_003489 Type: Tool

Proper Citation

MetaSKAT (RRID:SCR_003489)

Resource Information

URL: http://www.hsph.harvard.edu/skat/metaskat/

Proper Citation: MetaSKAT (RRID:SCR_003489)

Description: A R package for multiple marker meta-analysis.

Abbreviations: MetaSKAT

Resource Type: software resource

Defining Citation: PMID:23768515

Funding:

Availability: Free

Resource Name: MetaSKAT

Resource ID: SCR_003489

Alternate IDs: OMICS_00241

Record Creation Time: 20220129T080219+0000

Record Last Update: 20250420T014145+0000

Ratings and Alerts

No rating or validation information has been found for MetaSKAT.

No alerts have been found for MetaSKAT.

Data and Source Information

Source: <u>SciCrunch Registry</u>

Usage and Citation Metrics

We found 29 mentions in open access literature.

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

Senkevich K, et al. (2024) Lack of genetic evidence for NLRP3 inflammasome involvement in Parkinson's disease pathogenesis. NPJ Parkinson's disease, 10(1), 145.

Senkevich K, et al. (2024) Are rare heterozygous SYNJ1 variants associated with Parkinson's disease? NPJ Parkinson's disease, 10(1), 201.

Dorion MF, et al. (2024) MerTK is a mediator of alpha-synuclein fibril uptake by human microglia. Brain : a journal of neurology, 147(2), 427.

Haukka JK, et al. (2024) Whole-exome and whole-genome sequencing of 1064 individuals with type 1 diabetes reveals novel genes for diabetic kidney disease. Diabetologia, 67(11), 2494.

Senkevich K, et al. (2024) Are rare heterozygous SYNJ1 variants associated with Parkinson's disease? medRxiv : the preprint server for health sciences.

Antikainen AA, et al. (2024) Whole-genome sequencing identifies variants in ANK1, LRRN1, HAS1, and other genes and regulatory regions for stroke in type 1 diabetes. Scientific reports, 14(1), 13453.

Senkevich K, et al. (2023) GALC variants affect galactosylceramidase enzymatic activity and risk of Parkinson's disease. Brain : a journal of neurology, 146(5), 1859.

Senkevich K, et al. (2023) Association of rare variants in ARSA with Parkinson's disease. medRxiv : the preprint server for health sciences.

Senkevich K, et al. (2023) Genetics of NLRP3 suggests lack of involvement and inefficient druggability in Parkinson's disease. medRxiv : the preprint server for health sciences.

Jones-Tabah J, et al. (2023) The Parkinson's disease risk gene cathepsin B promotes fibrillar alpha-synuclein clearance, lysosomal function and glucocerebrosidase activity in dopaminergic neurons. bioRxiv : the preprint server for biology.

Neumann A, et al. (2022) Rare variants in IFFO1, DTNB, NLRC3 and SLC22A10 associate with Alzheimer's disease CSF profile of neuronal injury and inflammation. Molecular

psychiatry, 27(4), 1990.

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.

Delgado DA, et al. (2021) Rare, Protein-Altering Variants in AS3MT and Arsenic Metabolism Efficiency: A Multi-Population Association Study. Environmental health perspectives, 129(4), 47007.

Zhan L, et al. (2021) Rare variants in the endocytic pathway are associated with Alzheimer's disease, its related phenotypes, and functional consequences. PLoS genetics, 17(9), e1009772.

Hu S, et al. (2021) Whole exome sequencing analyses reveal gene-microbiota interactions in the context of IBD. Gut, 70(2), 285.

van Walree ES, et al. (2021) Germline variants in HEY2 functional domains lead to congenital heart defects and thoracic aortic aneurysms. Genetics in medicine : official journal of the American College of Medical Genetics, 23(1), 103.

Riveros-Mckay F, et al. (2020) The influence of rare variants in circulating metabolic biomarkers. PLoS genetics, 16(3), e1008605.

Marenne G, et al. (2020) Exome Sequencing Identifies Genes and Gene Sets Contributing to Severe Childhood Obesity, Linking PHIP Variants to Repressed POMC Transcription. Cell metabolism, 31(6), 1107.

Gaare JJ, et al. (2020) Meta-analysis of whole-exome sequencing data from two independent cohorts finds no evidence for rare variant enrichment in Parkinson disease associated loci. PloS one, 15(10), e0239824.

Ahluwalia TS, et al. (2019) A novel rare CUBN variant and three additional genes identified in Europeans with and without diabetes: results from an exome-wide association study of albuminuria. Diabetologia, 62(2), 292.