

Resource Summary Report

Generated by [ASWG](#) on Aug 3, 2026

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](#)

Availability: Free

Resource Name: MetaSKAT

Resource ID: SCR_003489

Alternate IDs: OMICS_00241

Record Creation Time: 20220129T080219+0000

Record Last Update: 20260801T190222+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: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 31 mentions in open access literature.

Listed below are recent publications. The full list is available at [ASWG](#) .

Cao J, et al. (2025) Integrating rare pathogenic variant prioritization with gene-based association analysis to identify novel genes and relevant multimodal traits for Alzheimer's disease. *Alzheimer's & dementia : the journal of the Alzheimer's Association*, 21(2), e14444.

Vuori N, et al. (2025) Exome and Genome Sequencing Reveals Novel Variants for Severe Diabetic Retinopathy in Type 1 Diabetes. *Investigative ophthalmology & visual science*, 66(13), 36.

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) Genetics of NLRP3 suggests lack of involvement and inefficient druggability in Parkinson's disease. *medRxiv : the preprint server for health sciences*.

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*.

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.

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.

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

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.

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

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.