

Resource Summary Report

Generated by T1D on Aug 2, 2026

PLINK/SEQ

RRID:SCR_013193

Type: Tool

Proper Citation

PLINK/SEQ (RRID:SCR_013193)

Resource Information

URL: <https://atgu.mgh.harvard.edu/plinkseq/>

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Description: An open-source C/C++ library for working with human genetic variation data. The specific focus is to provide a platform for analytic tool development for variation data from large-scale resequencing projects, particularly whole-exome and whole-genome studies. However, the library could in principle be applied to other types of genetic studies, including whole-genome association studies of common SNPs. (entry from Genetic Analysis Software)

Resource Type: software resource, software application, software library, software toolkit

Keywords: gene, genetic, genomic, c/c++, r, macos, linux, bio.tools

Availability: Open unspecified license

Resource Name: PLINK/SEQ

Resource ID: SCR_013193

Alternate IDs: nlx_154213, biotools:plink-seq

Alternate URLs: <https://bio.tools/plink-seq>

Record Creation Time: 20220129T080314+0000

Record Last Update: 20260801T190750+0000

Ratings and Alerts

No rating or validation information has been found for PLINK/SEQ.

No alerts have been found for PLINK/SEQ.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 77 mentions in open access literature.

Listed below are recent publications. The full list is available at [T1D](#) .

Ishorst N, et al. (2025) Role of ZFHX4 in orofacial clefting based on human genetic data and zebrafish models. European journal of human genetics : EJHG, 33(5), 595.

Lokki AI, et al. (2025) Understanding rare genetic variants within the terminal pathway of complement system in preeclampsia. Genes and immunity, 26(1), 22.

Bánfai Z, et al. (2024) Analysis of Gyimes Csango population samples on a high-resolution genome-wide basis. BMC genomics, 25(1), 942.

Lokki A, et al. (2024) Rare variants in genes coding for components of the terminal pathway of the complement system in preeclampsia. Research square.

Fernandez TV, et al. (2023) Primary complex motor stereotypies are associated with de novo damaging DNA coding mutations that identify KDM5B as a risk gene. PloS one, 18(10), e0291978.

Bánfai Z, et al. (2023) Characterization of Danube Swabian population samples on a high-resolution genome-wide basis. BMC genomics, 24(1), 9.

Spena S, et al. (2022) Genetic variants at the chromosomal region 2q21.3 underlying inhibitor development in patients with severe haemophilia A. Haemophilia : the official journal of the World Federation of Hemophilia, 28(2), 270.

Shin JJ, et al. (2021) Clinical, Radiographic, and Genetic Analyses in a Population-Based Cohort of Adult Spinal Deformity in the Older Population. Neurospine, 18(3), 608.

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

Mikhaylova AV, et al. (2021) Whole-genome sequencing in diverse subjects identifies genetic correlates of leukocyte traits: The NHLBI TOPMed program. American journal of human genetics, 108(10), 1836.

Mahdi H, et al. (2021) Genomic analyses of high-grade neuroendocrine gynecological malignancies reveal a unique mutational landscape and therapeutic vulnerabilities. Molecular oncology, 15(12), 3545.

Bis-Brewer DM, et al. (2020) Assessing non-Mendelian inheritance in inherited axonopathies. Genetics in medicine : official journal of the American College of Medical Genetics, 22(12), 2114.

Serra EG, et al. (2020) Somatic mosaicism and common genetic variation contribute to the risk of very-early-onset inflammatory bowel disease. *Nature communications*, 11(1), 995.

Bobbili DR, et al. (2020) Excess of singleton loss-of-function variants in Parkinson's disease contributes to genetic risk. *Journal of medical genetics*, 57(9), 617.

Nguyen TH, et al. (2020) mTADA is a framework for identifying risk genes from de novo mutations in multiple traits. *Nature communications*, 11(1), 2929.

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.

Xu C, et al. (2020) Medium-coverage DNA sequencing in the design of the genetic association study. *European journal of human genetics : EJHG*, 28(10), 1459.

Crooks L, et al. (2020) Identification of single nucleotide variants in the Moroccan population by whole-genome sequencing. *BMC genetics*, 21(1), 111.

Witten A, et al. (2020) ADAMTS12, a new candidate gene for pediatric stroke. *PloS one*, 15(8), e0237928.

Monroe TO, et al. (2020) PCM1 is necessary for focal ciliary integrity and is a candidate for severe schizophrenia. *Nature communications*, 11(1), 5903.