# Resource Summary Report

Generated by NIF on May 2, 2025

## QuantiSNP

RRID:SCR_013091
Type: Tool

### Proper Citation

QuantiSNP (RRID:SCR_013091)

### Resource Information

**URL:** https://sites.google.com/site/quantisnp/

**Proper Citation:** QuantiSNP (RRID:SCR_013091)

**Description:** THIS RESOURCE IS NO LONGER IN SERVICE, documented May 10, 2017. A pilot effort that has developed a centralized, web-based biospecimen locator that presents biospecimens collected and stored at participating Arizona hospitals and biospecimen banks, which are available for acquisition and use by researchers. Researchers may use this site to browse, search and request biospecimens to use in qualified studies. The development of the ABL was guided by the Arizona Biospecimen Consortium (ABC), a consortium of hospitals and medical centers in the Phoenix area, and is now being piloted by this Consortium under the direction of ABRC. You may browse by type (cells, fluid, molecular, tissue) or disease. Common data elements decided by the ABC Standards Committee, based on data elements on the National Cancer Institute''s (NCI''s) Common Biorepository Model (CBM), are displayed. These describe the minimum set of data elements that the NCI determined were most important for a researcher to see about a biospecimen. The ABL currently does not display information on whether or not clinical data is available to accompany the biospecimens. However, a requester has the ability to solicit clinical data in the request. Once a request is approved, the biospecimen provider will contact the requester to discuss the request (and the requester''s questions) before finalizing the invoice and shipment. The ABL is available to the public to browse. In order to request biospecimens from the ABL, the researcher will be required to submit the requested required information. Upon submission of the information, shipment of the requested biospecimen(s) will be dependent on the scientific and institutional review approval. Account required. Registration is open to everyone.Software to detect rare or de novo copy number alterations in normal DNA samples. Please note that QuantiSNP is no longer under active development.

**Abbreviations:** QuantiSNP

**Resource Type:** software resource

**Defining Citation:** PMID:17341461

**Keywords:** matlab, bio.tools

**Funding:**

**Availability:** THIS RESOURCE IS NO LONGER IN SERVICE

**Resource Name:** QuantiSNP

**Resource ID:** SCR_013091

**Alternate IDs:** biotools:quantisnp, OMICS_00730

**Alternate URLs:** https://bio.tools/quantisnp

**Record Creation Time:** 20220129T080314+0000

**Record Last Update:** 20250420T014632+0000

## Ratings and Alerts

No rating or validation information has been found for QuantiSNP.

No alerts have been found for QuantiSNP.

## Data and Source Information

**Source:** SciCrunch Registry

## Usage and Citation Metrics

We found 82 mentions in open access literature.

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

Huguet G, et al. (2024) Effects of gene dosage on cognitive ability: A function-based association study across brain and non-brain processes. Cell genomics, 4(12), 100721.

Halvorsen M, et al. (2024) A Burden of Rare Copy Number Variants in Obsessive-Compulsive Disorder. Research square.

Schmilovich Z, et al. (2024) Copy-number variants and polygenic risk for intelligence confer risk for autism spectrum disorder irrespective of their effects on cognitive ability. Frontiers in psychiatry, 15, 1369767.

Sha Z, et al. (2024) The copy number variant architecture of psychopathology and cognitive development in the ABCD® study. medRxiv : the preprint server for health sciences.

Ceroni F, et al. (2024) Deletion upstream of MAB21L2 highlights the importance of evolutionarily conserved non-coding sequences for eye development. Nature communications, 15(1), 9245.

Carlisle SG, et al. (2023) Rare Genomic Copy Number Variants Implicate New Candidate Genes for Bicuspid Aortic Valve. medRxiv : the preprint server for health sciences.

Fevga C, et al. (2023) PTPA variants and impaired PP2A activity in early-onset parkinsonism with intellectual disability. Brain : a journal of neurology, 146(4), 1496.

Kalinauskiene R, et al. (2023) A De Novo 8q22.2q22.3 Interstitial Microdeletion in a Girl with Developmental Delay and Congenital Defects. Medicina (Kaunas, Lithuania), 59(6).

Siavrien? E, et al. (2023) Molecular and Functional Characterisation of a Novel Intragenic 12q24.21 Deletion Resulting in MED13L Haploinsufficiency Syndrome. Medicina (Kaunas, Lithuania), 59(7).

Goh CJ, et al. (2023) Improving CNV Detection Performance in Microarray Data Using a Machine Learning-Based Approach. Diagnostics (Basel, Switzerland), 14(1).

Kopal J, et al. (2023) Rare CNVs and phenome-wide profiling highlight brain structural divergence and phenotypical convergence. Nature human behaviour, 7(6), 1001.

Tenney AP, et al. (2023) Noncoding variants alter GATA2 expression in rhombomere 4 motor neurons and cause dominant hereditary congenital facial paresis. Nature genetics, 55(7), 1149.

Kikas T, et al. (2023) Microdeletions and microduplications linked to severe congenital disorders in infertile men. Scientific reports, 13(1), 574.

Cato LD, et al. (2023) Genetic regulation of fetal hemoglobin across global populations. medRxiv : the preprint server for health sciences.

Alibutud R, et al. (2023) Structural Variations Contribute to the Genetic Etiology of Autism Spectrum Disorder and Language Impairments. International journal of molecular sciences, 24(17).

Chear S, et al. (2022) Lysosomal alterations and decreased electrophysiological activity in CLN3 disease patient-derived cortical neurons. Disease models & mechanisms, 15(12).

Tesfaye R, et al. (2022) Investigating the contributions of circadian pathway and insomnia

risk genes to autism and sleep disturbances. Translational psychiatry, 12(1), 424.

Balagué-Dobón L, et al. (2022) Fully exploiting SNP arrays: a systematic review on the tools to extract underlying genomic structure. Briefings in bioinformatics, 23(2).

Maihofer AX, et al. (2022) Rare copy number variation in posttraumatic stress disorder. Molecular psychiatry, 27(12), 5062.

Lavrichenko K, et al. (2021) SeeCiTe: a method to assess CNV calls from SNP arrays using trio data. Bioinformatics (Oxford, England), 37(13), 1876.