Resource Summary Report

Generated by NIF on Aug 10, 2026

NGS-SNP

RRID:SCR_005182
Type: Tool

Proper Citation

NGS-SNP (RRID:SCR_005182)

Resource Information

URL: http://stothard.afns.ualberta.ca/downloads/NGS-SNP/

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Description: A collection of command-line scripts for providing rich annotations for SNPs identified by the sequencing of transcripts or whole genomes from organisms with reference sequences in Ensembl. Included among the annotations, several of which are not available from any existing SNP annotation tools, are the results of detailed comparisons with orthologous sequences. These comparisons allow, for example, SNPs to be sorted or filtered based on how drastically the SNP changes the score of a protein alignment. Other fields indicate the names of overlapping protein domains or features, and the conservation of both the SNP site and flanking regions. NCBI, Ensembl, and Uniprot IDs are provided for genes, transcripts, and proteins when applicable, along with Gene Ontology terms, a gene description, phenotypes linked to the gene, and an indication of whether the SNP is novel or known. A ?Model_Annotations? field provides several annotations obtained by transferring in silico the SNP to an orthologous gene, typically in a well-characterized species.

Abbreviations: NGS-SNP

Resource Type: software resource

Keywords: annotation, snp, sequencing, transcript, genome, reference sequence, indel, annotate, reference chromosome, reference transcript, gene, command-line

Resource Name: NGS-SNP

Resource ID: SCR_005182

Alternate IDs: OMICS_00177

Record Creation Time: 20220129T080228+0000

Record Last Update: 20260808T185834+0000

Ratings and Alerts

No rating or validation information has been found for NGS-SNP.

No alerts have been found for NGS-SNP.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 33 mentions in open access literature.

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

Li P, et al. (2026) Endonuclease-Assisted Selective Exponential Amplification for Ultrasensitive Enrichment and Detection of Low-Abundance Mutant Alleles in Lung Cancer. The Journal of molecular diagnostics : JMD.

Liu J, et al. (2025) Safety and Efficacy of KN046 in Combination with KN026 in Patients with Advanced HER2-Positive Breast Cancer: A Phase II Trial. Clinical cancer research : an official journal of the American Association for Cancer Research, 31(12), 2379.

Bolormaa S, et al. (2021) A conditional multi-trait sequence GWAS discovers pleiotropic candidate genes and variants for sheep wool, skin wrinkle and breech cover traits. Genetics, selection, evolution : GSE, 53(1), 58.

Buggiotti L, et al. (2021) Demographic History, Adaptation, and NRAP Convergent Evolution at Amino Acid Residue 100 in the World Northernmost Cattle from Siberia. Molecular biology and evolution, 38(8), 3093.

Sweet-Jones J, et al. (2021) Genotyping and Whole-Genome Resequencing of Welsh Sheep Breeds Reveal Candidate Genes and Variants for Adaptation to Local Environment and Socioeconomic Traits. Frontiers in genetics, 12, 612492.

Yuan Z, et al. (2021) Expression quantitative trait loci in sheep liver and muscle contribute to variations in meat traits. Genetics, selection, evolution : GSE, 53(1), 8.

Xiang R, et al. (2021) Genome-wide fine-mapping identifies pleiotropic and functional variants that predict many traits across global cattle populations. Nature communications, 12(1), 860.

Akanno EC, et al. (2018) Genome-wide association scan for heterotic quantitative trait loci in multi-breed and crossbred beef cattle. Genetics, selection, evolution : GSE, 50(1), 48.

Koufariotis L, et al. (2018) Sequencing the mosaic genome of Brahman cattle identifies historic and recent introgression including polled. Scientific reports, 8(1), 17761.

Xiang R, et al. (2018) Genome variants associated with RNA splicing variations in bovine are extensively shared between tissues. BMC genomics, 19(1), 521.

Agnew T, et al. (2018) A Wars2 Mutant Mouse Model Displays OXPHOS Deficiencies and Activation of Tissue-Specific Stress Response Pathways. Cell reports, 25(12), 3315.

Koufariotis LT, et al. (2018) Variance explained by whole genome sequence variants in coding and regulatory genome annotations for six dairy traits. BMC genomics, 19(1), 237.

Zhang C, et al. (2018) Genomic evaluation of feed efficiency component traits in Duroc pigs using 80K, 650K and whole-genome sequence variants. Genetics, selection, evolution : GSE, 50(1), 14.

van den Berg I, et al. (2017) Multi-breed genomic prediction using Bayes R with sequence data and dropping variants with a small effect. Genetics, selection, evolution : GSE, 49(1), 70.

Abo-Ismail MK, et al. (2017) Genome-wide association studies and genomic prediction of breeding values for calving performance and body conformation traits in Holstein cattle. Genetics, selection, evolution : GSE, 49(1), 82.

Wang M, et al. (2017) Putative enhancer sites in the bovine genome are enriched with variants affecting complex traits. Genetics, selection, evolution : GSE, 49(1), 56.

Miann?? J, et al. (2016) Correction of the auditory phenotype in C57BL/6N mice via CRISPR/Cas9-mediated homology directed repair. Genome medicine, 8(1), 16.

Potter PK, et al. (2016) Novel gene function revealed by mouse mutagenesis screens for models of age-related disease. Nature communications, 7, 12444.

Peveling-Oberhag J, et al. (2015) Whole exome sequencing of microdissected splenic marginal zone lymphoma: a study to discover novel tumor-specific mutations. BMC cancer, 15, 773.

Dorshorst B, et al. (2015) Dominant Red Coat Color in Holstein Cattle Is Associated with a Missense Mutation in the Coatomer Protein Complex, Subunit Alpha (COPA) Gene. PloS one, 10(6), e0128969.