

Resource Summary Report

Generated by [RRID](#) on Apr 8, 2025

Genome Trax

RRID:SCR_001234

Type: Tool

Proper Citation

Genome Trax (RRID:SCR_001234)

Resource Information

URL: <http://www.biobase-international.com/product/genome-trax>

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Description: Service that provides a comprehensive compilation of variant knowledge that allows you to identify pathogenic variants in human whole genome or exome sequences. It makes it easy to upload a complete genome's worth of variations and identify the biologically relevant subset of known mutations, mutations that are novel and appear in a candidate disease genes, or mutations that are predicted to have a deleterious effect. The database includes a comprehensive collection of disease causing mutations from HGMD Professional, regulatory sites from TRANSFAC, and disease genes, drug targets and pathways from PROTEOME, as well as pharmacogenomic variants. It integrates the best public data-sets on somatic mutations, allele frequencies and clinical variants, in their most up-to-date version, for a total of more than 165 million annotations. It is possible to identify known pathogenic variants, remove harmless common variants, and obtain deleterious predictions for novel variants. With family data, it is possible to identify variants that are de novo, compound heterozygous only in the offspring. All of the results can be downloaded to Excel for further review. For core facilities and bioinformaticians, the complete underlying data is made available for download and easy integration into custom analysis pipelines. Genome Trax data is optimized to work with many other software packages, such as ANNOVARTM, CLC bio, Alamut, SimulConsult, and Cartagenia.

Abbreviations: Genome Trax

Synonyms: Genome Trax for Next Generation Sequencing

Resource Type: service resource

Keywords: next-generation sequencing, genome, exome, sequence, variation, mutation,

pathogenic, database, bio.tools, bio.tools

Funding:

Availability: Commercial, Subscription required

Resource Name: Genome Trax

Resource ID: SCR_001234

Alternate IDs: OMICS_02109, biotools:genome_trax

Alternate URLs: https://bio.tools/genome_trax

Record Creation Time: 20220129T080206+0000

Record Last Update: 20250214T182922+0000

Ratings and Alerts

No rating or validation information has been found for Genome Trax.

No alerts have been found for Genome Trax.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 2 mentions in open access literature.

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

Laššuthová P, et al. (2017) Confirmation of the GNB4 gene as causal for Charcot-Marie-Tooth disease by a novel de novo mutation in a Czech patient. *Neuromuscular disorders* : NMD, 27(1), 57.

Fuchsberger C, et al. (2016) The genetic architecture of type 2 diabetes. *Nature*, 536(7614), 41.