Variation Resources

NCBI's variation resources offer human genomic variations, including common and rare SNV, other small-scale variations, large structural variations, and associated frequencies, including ALFA, a new aggregated frequency source based on data from millions of controlled-access research study subjects. Access through the web, APIs, and FTP downloads.

Visit now
bit.ly/3PDBN2D

Identify novel variants

Calibrate variant calling pipeline algorithms

Integrate dbSNP annotation with your data

Submit variants to share with the scientific community

dbSNP bit.ly/3RluBcJ

- Over 1.1 billion RefSNP(rs)
- Population Frequency for 95% of RefSNP; including common and rare variants
- Rich annotation reported on RefSeq *GRCH37* and *GRCH38* assemblies, mRNA, and protein
- VCF files for assemblies *GRCh37* and *GRCh38*
- Full set of RefSNPs in the JSON format
- Indexed Search (bit.ly/44ZK1GO)

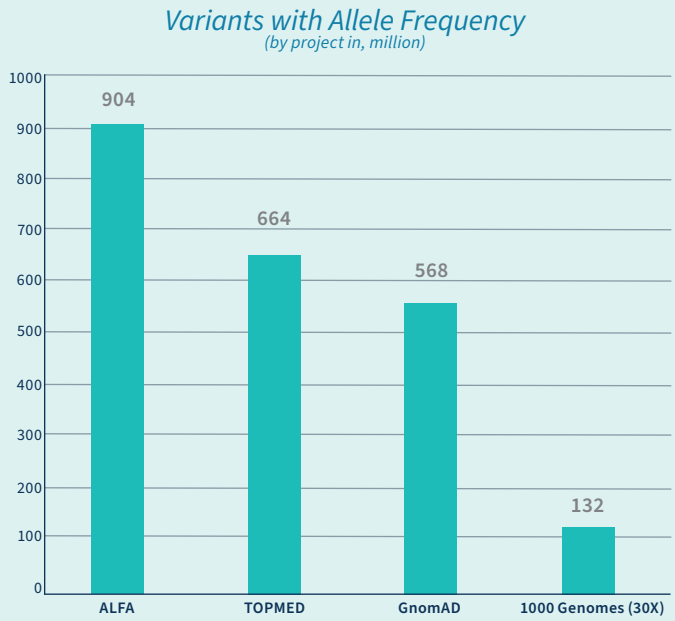
dbVar bit.ly/3sWdqUW

- 219 studies
- Clinically significant SV, Case-Control, and Curated
- Datasets (bit.ly/46j3qDp)
  - 7.7 million unique structural variants
  - 38 million submitted variant calls
- Updated monthly
- Population allele frequency
- Files are available in XML, GVF, VCF, BED, BEDPE, and TSV for assemblies *GRCh37* and *GRCH38*
- dbVar Tutorials and Datasets (bit.ly/2koP2Zg)
- Access full set of FTP (bit.ly/3RIRjfd) files

ALFA

- Latest Release 3 (August 2, 2023) included population frequency for 904 million variants from 204K subjects
- Access ALFA data along with other projects including 1000Genomes, GnomAD, and TOPMed

Learn about ALFA youtu.be/XM8HbSUNzfE



Variation Services

Web services for comparing, normalizing, annotating, and inter-converting variations

bit.ly/45TaYNJ

Variation Viewer

View, search, and navigate variations in genomic context. Review data or upload your own data

bit.ly/3EJSHq4

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Follow us on Twitter @ncbi_clinical

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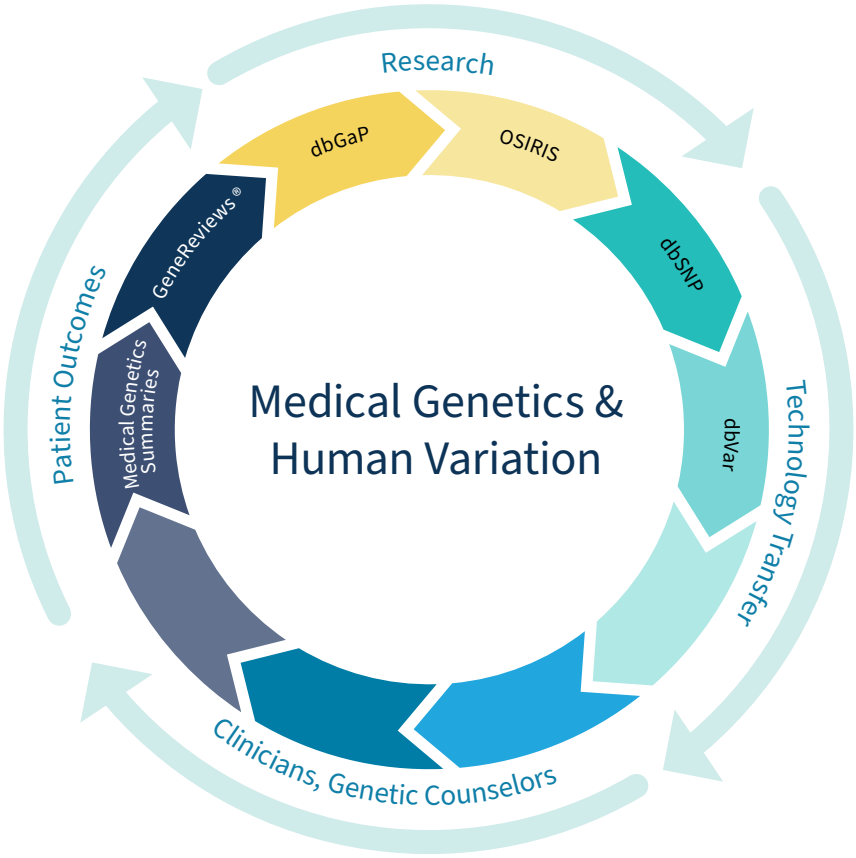
Visit us at ncbi.nlm.nih.gov/variation/

NCBI

The National Center for Biotechnology Information's medical genetics and human variation resources (MGV) help the genetics community advance the understanding of medical genetics and associated clinical applications.



ClinVar
user-submitted database for information about genomic variation and its relationship to human health.

dbGaP
database for genotype and phenotype research studies.

dbSNP and dbVar
databases of small and large genomic variants including both common variations and clinical mutations.

GTR®
provider-submitted database of clinical and research molecular, cytogenetic and biochemical genetic tests and supporting information.

MedGen
aggregates information from and provides access to authoritative medical genetics resources.

Medical Genetics Summaries and GeneReviews®
up-to-date, peer-reviewed, medically actionable summaries for heritable diseases and pharmacogenetics.

OSIRIS
open source short tandem repeat (STR) analysis tool for forensic, clinical and research use.

Variation Viewer
interactive browser for examination of nucleotide variants in a genomic context.

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