

What is ClinVar?

An NCBI database of **variant interpretations**.

<https://www.ncbi.nlm.nih.gov/clinvar/>

Variants interpreted by

- Clinical laboratories
- Research groups
- Expert panels and more

Key statistics

- Over **273,000** interpreted variants
- From over **670** submitters

Search ClinVar by

- **Gene symbols**
HFE or BRCA1
- **HGVS expressions**
NM_000410.3:c.277G>C
- **rs numbers**
rs28934597
- **Protein changes**
p.Gly93Arg
- **Disease and phenotype**
Hemochromatosis
- **Submitting organization**
children's national medical center
- **PubMed ID or other citations**

Steps to submit your interpretation

1. **Go to ClinVar Submission Portal**
<https://submit.ncbi.nlm.nih.gov/clinvar/>
2. **Register your organization**
3. **Submit your data**
 - **single variant interpretation**
Use our single submission wizard
 - **Batch of variant interpretations**
Fill in our submission spreadsheet template, then upload your file. Learn more at:
<http://bit.ly/ncbi-clinvar-sub>

To learn more about ClinVar

- See our video tutorials http://bit.ly/clinvar_videos
- Read our online documents <http://bit.ly/ncbi-clinvar-1>, <http://bit.ly/ncbi-clinvar-2>

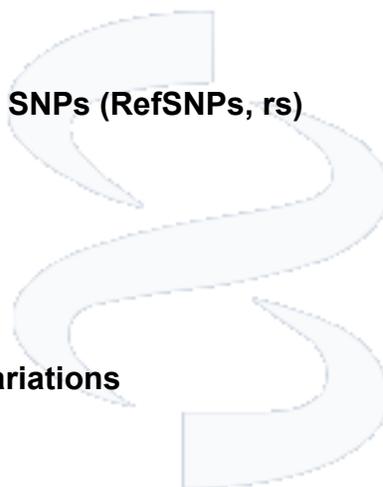
dbSNP from NCBI

An archive of **short nucleotide sequence variants** submitted by the public, with representation on sequences on which variants were defined, and on the current assemblies.

<https://www.ncbi.nlm.nih.gov/snp/>

dbSNP is **changing**

- New streamlined build system for establishing reference SNPs (RefSNPs, rs)
- New display to report on RefSNPs
- New content available in JSON format via FTP
- Consistent reporting on RefSNPs across products
- New internal representation of RefSNPs as objects
- New Variation Services for comparing and processing variations



Pre-Alpha release of new products

Files on FTP site

- VCF files for assemblies GRCh37 and CRCh38
ftp://ftp.ncbi.nlm.nih.gov/snp/.redesign/latest_release/VCF/
- Full set of RefSNPs in the JSON format
ftp://ftp.ncbi.nlm.nih.gov/snp/.redesign/latest_release/JSON/

Variation Services

- Web services for comparing, grouping and interconverting sequence variants
<https://www.ncbi.nlm.nih.gov/projects/variation/services/v0/>
- A BLOG post with additional details
<http://bit.ly/ncbi-spdi-1>

Spread the news

Please **share** this with your colleagues, particular those involved in managing the bioinformatics resources and sequence variation data for your group or institutions.

Address your questions or feedback to variation-services@ncbi.nlm.nih.gov