Scope and Access
The NCBI Short Genetic Variations database (dbSNP [1]), commonly known as dbSNP, catalogs short variations in nucleotide sequences for human. These variations include single nucleotide variations, short nucleotide insertions and deletions, short tandem repeats. Short Genetic Variations may be common, thus representing true polymorphisms, or they may be rare. Some rare human entries have additional information associated with them, including disease associations (from ClinVar [2]), genotype information and allele origin, as some variations are somatic rather than germline events.


Searching for and Displaying SNP Records
You can search for variations on the dbSNP homepage by typing a query term in the search box and clicking the Search button (A). You can also use the Advanced (B) page to create complex queries to produce more precise results. The search below, "hfe[gene] AND human[orgn]", retrieves variations mapped to the human HFE gene. You can use options in the Display settings popup (C) to change the number of records displayed or sort retrieved variations in a different order. You can further narrow down retrieved variations by selecting filters present in the left column (D), or save them to a local file using the Send to (E) option. Use links to separate displays to see gene-centric listings (GeneView, F), graphical presentation under the context of genome or mRNA sequences (via HGVS names, G), or gene-centric display in a genomic context (Varview, H). Using the “Find related data” portlet (I), you can retrieve related entries from other NCBI databases for the set of variations in the display.
The Reference SNP Cluster Report

The Reference SNP Cluster Report linked from rsID (rs1800730, shown in sections below and on p.3) provides details of a variation record. The report contains a summary of the allele (A), a link to the gene-centric display through the VarView icon (B, see p.4), mapping information in Human Genome Variation Society (HGVS) nomenclature (C), and minor allele frequencies (MAF, D) from various studies. The Integrated Maps table provides the genomic mapping details with the chromosomal coordinates (E) link to the same gene-centric display VarView icon provides. The magnifying glass (F) points to the 1000 Genomes Browser and provides genotyping details, if the variant this rsID represents is also called by that project.

For a summary of SNPs mapped to the gene, you can click the Go button (G) in the GeneView section to activate the SNP: GeneView display (p. 4).

The Gene Model(s) table below lists coordinates and changes on transcripts and proteins (H). The graphical panel (I) further below presents variants with various characteristics in different tracks (J) under the context of genome annotation. Individual variants are hyperlinked to provide additional details (K, activated upon hover).

The Submitter records table (L) lists alleles and flanking sequences from submitter SNPs (ssIDs) included in this reference SNP cluster. The ssIDs (M) link to submitter records with additional details.

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The Reference SNP Cluster Report (cont.)

The FASTA Sequence section shows the sequences flanking the variation. It uses the exemplar submitter record to represent the variation by presenting the sequence as the 5'- (A) and 3'- (B) flanking sequences, with the allele (C) in the middle. The Population Diversity section (D) sums up available genotype and allele frequency information for various populations from different studies. More genotype details are available through the 1000 Genomes browser link (E).

Other Ways to Access Data from dbSNP

The SNP database is fully integrated with the Entrez system, enabling the access of variation data through links present in records from other NCBI databases. For example, you can use the SNP: GeneView or Variation Viewer link (F) found in the Related information section of a Gene database record to see a summary list of variations mapped to that gene. You can project variations mapped to a segment of the RefSeq genomic or a mRNA record (with NT_, NG_, NW_ or NM_ accessions) by using the Customize view (G) menu in the upper right hand corner of the sequence record, simply check the SNPs checkbox and click Update View (H) to activate the selection.

dbSNP also integrates disease-related nucleotide variations that were reported in literature and cited in rsID format, collected by OMIM, or submitted to ClinVar. The table below is the Allelic Variant display for OMIM record 613609, which cites the rsIDs in the dbSNP column (I).
The SNP:GeneView Display

The SNP:GeneView display tabulates variations mapped to splicing variants of a particular gene. At the top, it lists all annotated splice variants (A) of the gene. The splice variant, whose variations are shown, is highlighted in yellow (B). The default setting shows only the non-clinical coding variations. Check “Clinical Source” and “in gene region” options, then click “Refresh” (C) to see the complete list. The table arranges mapped variants by their chromosomal coordinates (D) and color-codes them by their function: white for “in gene region” (E), orange for UTR (F), green for synonymous (G), red for non-synonymous (H), blue for frame-shift (I), purple for splicing site (J), and yellow for intron (K). The MAF (L) column lists the global minor allele frequencies from the 1000 Genomes project.

Variation Viewer

The GeneView display contains a link (M) to an interactive display in the Variation Viewer (N), which can display variation mapping and molecular consequences within the GRCh37 or GRCh38 context for more focused examination - by correlating a variation and its molecular consequences in the data table with its genomic context in the graphical display (O). Filters in the left hand column (not shown) are available to selectively display variants of interest.

More information on this tool is available online as a video tutorial, in a fact-sheet, as well as in the more detailed online help:

Variation Viewer factsheet
Online video tutorial
Variation Viewer help

https://www.youtube.com/watch?v=rnWZ9MFBwUM