dbSNP: Database of Short Genetic Variations

National Library of Medicine National Center for Biotechnology Information

Catalog of short nucleotide changes for human genome and its annotated transcripts

Scope and Access

dbSNP

benign

other

Clinical Significance

pathogenicity

Validation Status

likely benign

pathogenic

risk factor

by-ALFA

by-cluster

by-frequency

Publication

PubMed Cited

PubMed Linked

Function Class

non coding transcript variant

intron

missense

Annotation

Global MAF

Custom range.

Show additional filters

somatic

Clear all

conflicting interpretations of

The NCBI Short Genetic Variation database [1], commonly known as dbSNP, catalogs short variations in human nucleotide sequences. Variations include single nucleotide variations, as well as insertions, deletions, and short tandem repeats less than 50 nucleotides in length. Short genetic variations may be common, thus representing true polymorphisms, or they may be rare. Some rare human entries have additional information including disease associations from NCBI's ClinVar database [2], genotype information and allele origin (germline / somatic).

You can access short nucleotide variation data from the dbSNP homepage www.ncbi.nlm.nih.gov/snp or the Entrez Programming Utilities API www.ncbi.nlm.nih.gov/books/NBK25501.

Download VCF and JSON files through FTP ftp.ncbi.nlm.nih.gov/snp/ or Aspera at www.ncbi.nlm.nih.gov/public/?snp/. The dbSNP API service, SPDI [3], is available at: api.ncbi.nlm.nih.gov/variation/v0/.

You can also access dbSNP and structural variation data from dbVar through the Variation Viewer www.ncbi.nlm.nih.gov/variation/view/.

Searching for and Displaying SNP Records

SNP

1.

clear

С

Search results

Items: 1 to 20 of 23

Variant type:

Chromosome:

Canonical SPDI:

Clinical significance:

Alleles:

Gene

Validated:

MAF:

HGVS

rs807209 [Homo sapiens]

Eunctional Consequence:

You can search for variations on the dbSNP homepage by typing query terms in the search box and clicking the Search button (A), or use the Advanced (B) page to create complex queries for more precise retrieval. For example, the query term HFE[gene] retrieves variations mapped to the human HFE gene. You can further refine the list with preset filters, such as "PubMed Cited" (C). Options in the Display Settings popup (D) allows you to change the number of records displayed and their sort order. The Send to dialog box (E) provides options to save retrieved records to a local file. The VarView (F) links to the Variation Viewer [4,5] where you can see the variant under the context of the genome annotation. The summary also provides allele frequencies from large population studies (G), including aggregated frequency from dbGaP subjects (ALFA). HGVS variant names (H) hyperlink to the graphical view on the Reference Sequence [6]. The Find related data feature (I) allows you to retrieve related entries from other NCBI databases for the set of variations selected.

HEE[gene]

Create alert Advanced

Display Settings: Summary, 20 per page, Sorted by SNP_ID

Filters activated: PubMed Cited. Clear all to show 4916 items.

SNV

benign

C>G,T [Show Flanks]

6:26092637 (GRCh38)

6:26092865 (GRCh37)

G=0.00219/62 (ALFA)

G=0.00006/1 (TOMMO)

G=0./0 (HapMap)

HFE (Varview), HFE-AS1 (Varview)

by frequency,by alfa,by cluster

NC_000006.12:g.26092637C>G, NC_0000

NC_000006.11:g.26092865C>G, NC_0000

NG 008720.2:q.10357C>G, NG 008720.2

NM_001406751.1:c.645C>G, NM_001406

NC 000006.12:26092636:C:G,NC 000006.12:26092636:C

...more

D

B

Page

Format

Summary

Ο5

O 10

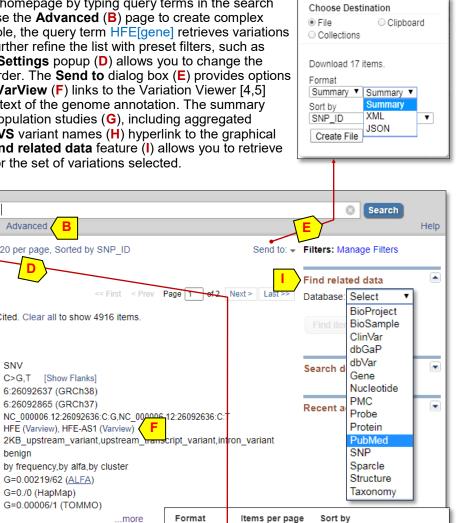
20

O 50

 $\bigcirc 100$

O 200

Next >



PubMed LitVar

Apply

O Default order

O Chromosome Base

SNP_ID

Position

The Reference SNP Report

The Reference SNP Report linked from a rsID (right & below) shows details of a variant record. The summary at the top (A) provides an overview of the variant. The allele change is relative to the plus strand of the chromosome. The report is also available in JSON format through the Download link (B). The Reference SNP report groups available details into categories (C) under separate tabs. The default Frequency tab (D) lists allele frequencies from major studies, such as ALFA from dbGaP samples and 1000 Genomes, broken down by subpopulation if available.

Sequence name

GRCh37.p13 chr 6

GRCh38.p14 chr 6

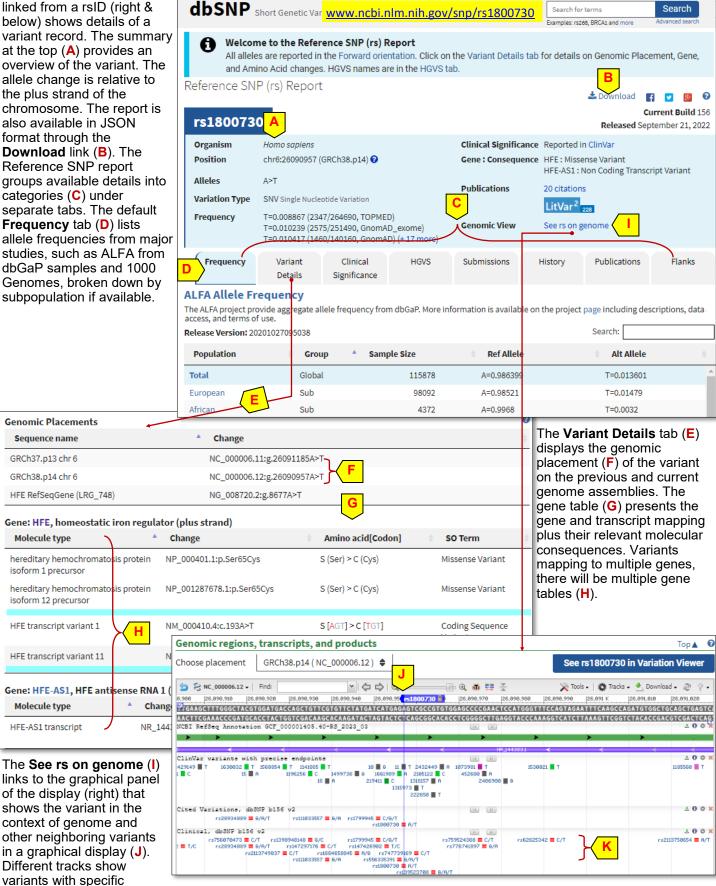
Molecule type

isoform 1 precursor

isoform 12 precursor

Molecule type

HFE-AS1 transcript



attributes, such with literature citations in the literature or with clinical significance from ClinVar (K).

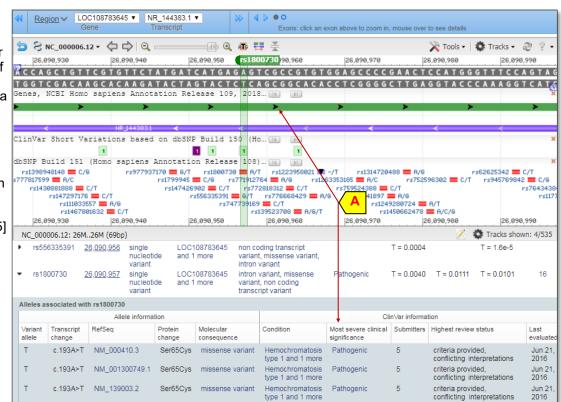
The Reference SNP Report (cont.) Other tabs in the new Reference SNP Report provide additional information.

Allele: T (allele ID: 15050)		Clinical		6	The Clinic	al Significance		
ClinVar Accession	Disease Names	Significance	🕴 Clinical Signi	Clinical Significance		tab (A) lists related clinical		
RCV00000028.12 H	emochromatosis type 1		Uncertain-Sign	Uncertain-Significance		assertions for the variant fro ClinVar, with links to ClinVar		
RCV000290779.11 H	ereditary hemochromatosis		Likely-Benign		records.			
ta d	zheimer disease,Familial porp Irda,Hemochromatosis type 1 Jabetes, susceptibility to, 7,Tra ait locus 2,Variegate porphyria	,Microvascular complication Insferrin serum level quantit		ificance		HGVS		
RCV000998547.11 n	v000998547.11 not provided		Benign-Likely-Benign					
RCV001328435.3 n	ot specified		Uncertain-Sign	ificance	Search:			
The HGVS tab (B) lists	Placement		▲ A=		≜ T			
HGVS names describing the variant using different	GRCh37.p13 chr 6		NC_000006.11:g.	NC_000006.11:g.26091185=		NC_000006.11:g.26091185A>T		
reference accessions.	GRCh38.p14 chr 6		NC_000006.12:g.	NC_000006.12:g.26090957=		NC_000006.12:g.26090957A>T		
Jse the table header to sort names or the search	hereditary hemochromatosis protein isoform 1 precursor		NP_000401.1:p.S	NP_000401.1:p.Ser65=		NP_000401.1:p.Ser65Cys		
box (C) to filter the list.	hereditary hemochromatosis protein isoform 12 precursor		NP_001287678.1	NP_001287678.1:p.Ser65=		NP_001287678.1:p.Ser65Cys		
	hereditary hemochromatosis protein isoform 13 precursor		NP_001371093.1	NP_001371093.1:p.Ser65=		NP_001371093.1:p.Ser65Cys		
Submissions D The	hereditary hemochromatos	is protein isoform 3 precursor	NP_620572.1:p.S	er65=	NP_620572	.1:p.Ser65Cys		
1 SubSNP, 19 Frequency, 5 ClinVa	r submissions	Search	(ssion tab (I	D) lists submitted		
No Submitter	Submis	sion ID 🕴 D	ate (Build) 🕴	entries	from large	projects or		
60 1000G_HIGH_COVERAGE	ss526779	ss5267790513 Oct 13, 2022 (156)						
64 1000G_HIGH_COVERAGE	se5552202280 E Oct 12 2022 (156) Th				he History tab (F) tracks changes			
						s other rsIDs that		
72 1000Genomes		t 12, 2018 (152)	variants rs115372583 and					
73 1000Genomes_30x	NC_0000	t 13, 2022 (156)		4888 (G) were merged into				
45 ACPOP	ss373330		13, 2019 (153)	rs1800	730.			
34 AFFY	ss298535		ov 08, 2017 (151)					
90 ALFA	NC_0000	06.12 - 26090957 Ap	or 26, 2021 (155) 🚽]			
			Searc	h:				
Associated ID		History Updated (I	Build)		·			
rs28934888 May 25, 2008 (130)						History		
rs115372583		Oct 26, 2010 (133)				F		
dded to this RefSNP Cluster:			Searc	h:				
					Source			
Submission IDs		Observation SPDI	Canonical SPDI	÷	RSIDs			
ss160462894, ss410868034, ss49188198	1, ss1592256975	NC_000006.10:261991	63:A:T NC_000006.12:26	090956:A:T	(self)	Publications		
31165441, 17368896, 12371682, 820886	1, 40652, 7745290, <mark>4</mark> 723249, 7707	7902, NC_000006.11:260911	34:A:T NC_000006.12:26	090956:A:T	(self)	н		
bSNP connects Referen		0 citations for rs1800730						
and biomedical literature text-mining. The Reference						Search:		
displays relevant citations	under the	PMID 🕴 Title		Author	🕴 Year 🗸	Journal		
Publications tab (H). Use		34120733 Whole-body R2* map iron storage organs: re	Clinical radiology					
PubMed" button (I) to retr citations in PubMed and e		Genetic test for the prescription of diets in support of Naureen Z			Z et al. 2020 Acta bio-medica			
abstracts for more information	ation. Some of	physical activity. Hemochromatosis: Hereditary hemochromatosis and						
these citations may also h available from PubMed C		HFE gene.	Katsarou MS et al. 2019 Vitamins and hormones					
		View All in PubMed						

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Variation Viewer

The Variation Viewer provides an interactive display of the variant under the context of annotation of the selected genome assembly. It also provides a table of variants in the display with molecular and clinical consequences (**A**). Facet filters in the left column (not shown) allows you to filter for variants with particular characteristics. More information on this tool is available online [4, 5]



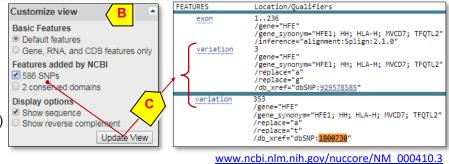
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Other Ways to Access dbSNP Data

The dbSNP database is fully integrated with the Entrez system that provides links to SNP records from other NCBI databases. For example, you can show variations mapped to a RefSeq mRNA record (with NM_ accessions) by using the **Customize view (B)** menu in the upper right hand corner of the sequence record, simply check the SNPs checkbox and click

dbSNP also includes variations reported in literature, collected by OMIM or submitted to ClinVar. For example, the OMIM <u>Allelic</u> <u>Variants</u> table links to dbSNP entries (**D**).

Update View (C).



613609 HFE GENE; HFE Allelic Variants (11 Selected Examples) : All cline Derivations									
Number 4	Phenotype	Mutation 🖕	dbSNP	ExAC	ClinVar				
.0001	HEMOCHROMATOSIS, TYPE 1 PORPHYRIA CUTANEA TARDA, SUSCEPTIBILITY TO, INCLUDED PORPHYRIA VARIEGATA, SUSCEPTIBILITY TO, INCLUDED HEMOCHROMATOSIS, JUVENILE, DIGENIC, INCLUDED ALZHEIMER DISEASE, SUSCEPTIBILITY TO, INCLUDED TRANSFERRIN SERUM LEVEL QUANTITATIVE TRAIT LOCUS 2, INCLUDED MICROVASCULAR COMPLICATIONS OF DIABETES, SUSCEPTIBILITY TO, 7, INCLUDED	HFE, CYS282TYR	[<u>rs1800562]</u>	-	[RCV000210820]				
.0002	HEMOCHROMATOSIS, TYPE 1 MICROVASCULAR COMPLICATIONS OF DIABETES, SUSCEPTIBILITY TO, 7, INCLUDED	HFE, HIS63ASP	[rs1799945]	[rs1799945]	[RCV00000027]				
.0003	HEMOCHROMATOSIS, TYPE 1	HFE, SER65CYS	[rs1800730]	-	[RCV000290779]				
.0004	HFE INTRONIC POLYMORPHISM	HFE, 5569G-A	[rs1800758]	[rs1800758]	[RCV00000031]				
.0005	HFE POLYMORPHISM	HFE, VAL53MET	[rs28934889]	-	[RCV00000032]				
.0006	HFE POLYMORPHISM	HFE, VAL59MET	[rs111033557]	-	[RCV00000033]				
.0007	HEMOCHROMATOSIS, TYPE 1	HFE, GLN127HIS	[rs28934595]	-	[RCV00000034]				
.0008	HEMOCHROMATOSIS, TYPE 1	HFE, ARG330MET	[rs111033558]	-	[RCV00000035]				

References

- 1. The Database of Short Genetic Variation (dbSNP). Kitts A, et. al. In The NCBI Handbook [Internet], 2nd ed.
- 2. ClinVar: improving access to variant interpretations and supporting evidence. Landrum MJ, et al. <u>Nucleic Acids Res.</u> 2018 Jan 4;46(D1):D1062-D1067.
- 3. New Web Services for Comparing and Grouping Sequence Variants. go.usa.gov/xUeKT.
- 4. Variation Viewer factsheet. ftp.ncbi.nih.gov/pub/factsheets/Factsheet Variation Viewer.pdf
- 5. Variation Viewer video tutorial. www.youtube.com/watch?v=rnWZ9MFBwUM
- 6. Graphical Sequence Viewer factsheet. ftp.ncbi.nih.gov/pub/factsheets/Factsheet_Graphical_SV.pdf