

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.

dbSNP Short Genetic Variations www.ncbi.nlm.nih.gov/snp/rs1800730 Search for terms Search

Welcome to the Reference SNP (rs) Report
All alleles are reported in the Forward orientation. Click on the Variant Details tab for details on Genomic Placement, Gene, and Amino Acid changes. HGVS names are in the HGVS tab.

Reference SNP (rs) Report

rs1800730 (A)

Organism: *Homo sapiens* Clinical Significance: Reported in ClinVar
Position: chr6:26090957 (GRCh38.p14) Gene : Consequence: HFE : Missense Variant
Alleles: A>T HFE-AS1 : Non Coding Transcript Variant
Variation Type: SNV Single Nucleotide Variation Publications: 20 citations
Frequency: T=0.008867 (2347/264690, TOPMED) LitVar² 228
T=0.010239 (2575/251490, GnomAD_exome) Genomic View: See rs on genome (I)
T=0.010417 (1460/140160, GnomAD) (+ 17 more)

Download (B)

Current Build 156
Released September 21, 2022

Frequency (D) Variant Details Clinical Significance HGVS Submissions History Publications Flanks

ALFA Allele Frequency
The ALFA project provide aggregate allele frequency from dbGaP. More information is available on the project page including descriptions, data access, and terms of use.
Release Version: 20201027095038

Population	Group	Sample Size	Ref Allele	Alt Allele
Total	Global	115878	A=0.986399	T=0.013601
European	Sub	98092	A=0.98521	T=0.01479
African	Sub	4372	A=0.9968	T=0.0032

Genomic Placements

Sequence name	Change
GRCh37.p13 chr6	NC_000006.11:g.26091185A>T
GRCh38.p14 chr6	NC_000006.12:g.26090957A>T (F)
HFE RefSeqGene (LRG_748)	NG_008720.2:g.8677A>T (G)

Gene: HFE, homeostatic iron regulator (plus strand)

Molecule type	Change	Amino acid[Codon]	SO Term
hereditary hemochromatosis protein isoform 1 precursor	NP_000401.1:p.Ser65Cys	S (Ser) > C (Cys)	Missense Variant
hereditary hemochromatosis protein isoform 12 precursor	NP_001287678.1:p.Ser65Cys	S (Ser) > C (Cys)	Missense Variant
HFE transcript variant 1	NM_000410.4:c.193A>T	S [AGT] > C [TGT]	Coding Sequence (H)
HFE transcript variant 11			

Gene: HFE-AS1, HFE antisense RNA 1 (minus strand)

Molecule type	Change
HFE-AS1 transcript	NR_144083.1

The Variant Details tab (E) displays the genomic placement (F) of the variant on the previous and current genome assemblies. The gene table (G) presents the gene and transcript mapping plus their relevant molecular consequences. Variants mapping to multiple genes, there will be multiple gene tables (H).

The See rs on genome (I) links to the graphical panel of the display (right) that shows the variant in the context of genome and other neighboring variants in a graphical display (J). Different tracks show variants with specific attributes, such with literature citations in the literature or with clinical significance from ClinVar (K).

Genomic regions, transcripts, and products

Choose placement: GRCh38.p14 (NC_000006.12) See rs1800730 in Variation Viewer

NC_000006.12 | Find: | Tools | Tracks | Download

AACTTCGAAACCCGATGCACCTACTGGTCGCAAGCACAAGATACTAGTACTCAGCGGCACACCTCGGGCTTGAAGTACCCAAAGGTCATCTTAAAGTTCGGTCTACACCGCTGACTCAG

NCBI RefSeq Annotation GCF_000001405.40-RS_2029_03

ClinVar variants with precise endpoints

429149	T	1630832	T	1568054	T	1141805	T	10	0	11	T	2432449	R	1873981	T	1508021	T	1105500	T
	C	15	R	1196256	C	1499738	G	1661989	R	2105122	C	452680	R	2486900	G				
								219411	C	1316157	R								
										1315973	T								
												222650	T						

Cited Variations, dbSNP b156 v2

rs28934889	G/R/T	rs11033557	G/R	rs1799945	C/G/T	rs1090730	R/T
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Clinical, dbSNP b156 v2

rs756070473	C/T	rs1390948140	G/C	rs1799945	C/G/T	rs759524380	C/T	rs62625342	C/T	rs213750654	R/T
rs28934889	G/R/T	rs147297176	C/T	rs147426902	T/C	rs776741897	G/R				
		rs2113749037	C/T	rs1084650845	R/G	rs747729169	C/T				
		rs11033557	G/R	rs1080730	R/T	rs556335291	R/T				
				rs1090730	R/T						
				rs139523708	G/R/T						

The Reference SNP Report (cont.)

Other tabs in the new Reference SNP Report provide additional information.

Allele: T (allele ID: 15050)

ClinVar Accession	Disease Names	Clinical Significance
RCV000000028.12	Hemochromatosis type 1	Uncertain-Significance
RCV000290779.11	Hereditary hemochromatosis	Likely-Benign
RCV000764641.2	Alzheimer disease,Familial porphyria cutanea tarda,Hemochromatosis type 1,Microvascular complications of diabetes, susceptibility to, 7,Transferrin serum level quantitative trait locus 2,Variegate porphyria	Uncertain-Significance
RCV000998547.11	not provided	Benign-Likely-Benign
RCV001328435.3	not specified	Uncertain-Significance

The **Clinical Significance** tab (A) lists related clinical assertions for the variant from ClinVar, with links to ClinVar records.

The **HGVS** tab (B) lists HGVS names describing the variant using different reference accessions. Use the table header to sort names or the search box (C) to filter the list.

Placement	A=	T
GRCh37.p13 chr 6	NC_000006.11:g.26091185=	NC_000006.11:g.26091185A>T
GRCh38.p14 chr 6	NC_000006.12:g.26090957=	NC_000006.12:g.26090957A>T
hereditary hemochromatosis protein isoform 1 precursor	NP_000401.1:p.Ser65=	NP_000401.1:p.Ser65Cys
hereditary hemochromatosis protein isoform 12 precursor	NP_001287678.1:p.Ser65=	NP_001287678.1:p.Ser65Cys
hereditary hemochromatosis protein isoform 13 precursor	NP_001371093.1:p.Ser65=	NP_001371093.1:p.Ser65Cys
hereditary hemochromatosis protein isoform 3 precursor	NP_620572.1:p.Ser65=	NP_620572.1:p.Ser65Cys

Submissions (D)

71 SubSNP, 19 Frequency, 5 ClinVar submissions

No	Submitter	Submission ID	Date (Build)
60	1000G_HIGH_COVERAGE	ss5267790513	Oct 13, 2022 (156)
64	1000G_HIGH_COVERAGE	ss5553392389	Oct 13, 2022 (156)
72	1000Genomes	NC_000006.11 - 26091185	Oct 12, 2018 (152)
73	1000Genomes_30x	NC_000006.12 - 26090957	Oct 13, 2022 (156)
45	ACPOP	ss3733306376	Jul 13, 2019 (153)
34	AFFY	ss2985356717	Nov 08, 2017 (151)
90	ALFA	NC_000006.12 - 26090957	Apr 26, 2021 (155)

Submission tab (D) lists submitted entries from large projects or individual submitters (E).

The **History** tab (F) tracks changes of the cluster and lists other rsIDs that were merged with the variant. Here, variants rs115372583 and rs28934888 (G) were merged into rs1800730.

Associated ID | **History Updated (Build)**

rs28934888	May 25, 2008 (130)
rs115372583	Oct 26, 2010 (133)

Added to this RefSNP Cluster:

Submission IDs	Observation SPDI	Canonical SPDI	Source RSIDs
ss160462894, ss410868034, ss491881981, ss1592256975	NC_000006.10:26199163:A:T	NC_000006.12:26090956:A:T	(self)
31165441, 17368896, 12371682, 8208864, 40652, 7745290, 4723249, 7707902,	NC_000006.11:26091184:A:T	NC_000006.12:26090956:A:T	(self)

The **History** tab (F) tracks changes of the cluster and lists other rsIDs that were merged with the variant. Here, variants rs115372583 and rs28934888 (G) were merged into rs1800730.

dbSNP connects Reference SNP variants and biomedical literature citations through text-mining. The Reference SNP Report displays relevant citations under the **Publications** tab (H). Use the "View All in PubMed" button (I) to retrieve the list of citations in PubMed and examine their abstracts for more information. Some of these citations may also have free full-text available from PubMed Central (PMC).

20 citations for rs1800730

PMID	Title	Author	Year	Journal
34120733	Whole-body R2* mapping to quantify tissue iron in iron storage organs: reference values and a genotype.	Kromrey ML et al.	2021	Clinical radiology
33170161	Genetic test for the prescription of diets in support of physical activity.	Naureen Z et al.	2020	Acta bio-medica
30798813	Hemochromatosis: Hereditary hemochromatosis and HFE gene.	Katsarou MS et al.	2019	Vitamins and hormones

View All in PubMed (I)

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]

The screenshot shows the NCBI Variation Viewer interface. At the top, the region is identified as LOC108783645 on chromosome 16 (16p11.2). The sequence viewer shows the genomic context with a green bar indicating the variant rs1800730. Below the sequence, a table lists variants with their clinical consequences. A yellow box labeled 'A' highlights the variant rs1800730, which is a single nucleotide variant (G/T) with a pathogenic clinical consequence (missense variant, non coding transcript variant) and a p-value of 0.0004.

Variant	Transcript	RefSeq	Protein change	Molecular consequence	Condition	Most severe clinical significance	Submitters	Highest review status	Last evaluated
T	c.193A>T	NM_000410.3	Ser65Cys	missense variant	Hemochromatosis type 1 and 1 more	Pathogenic	5	criteria provided, conflicting interpretations	Jun 21, 2016
T	c.193A>T	NM_001300749.1	Ser65Cys	missense variant	Hemochromatosis type 1 and 1 more	Pathogenic	5	criteria provided, conflicting interpretations	Jun 21, 2016
T	c.193A>T	NM_139003.2	Ser65Cys	missense variant	Hemochromatosis type 1 and 1 more	Pathogenic	5	criteria provided, conflicting interpretations	Jun 21, 2016

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 **Update View** (C).

The screenshot shows the 'Customize view' menu and the 'FEATURES' panel. The 'Customize view' menu has a yellow box labeled 'B' next to it. The 'FEATURES' panel shows a list of features for the HFE gene, with a yellow box labeled 'C' next to the 'variation' feature.

www.ncbi.nlm.nih.gov/nuccore/NM_000410.3

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

The screenshot shows the OMIM Allelic Variants table for HFE. The table shows a list of variants with their dbSNP IDs and clinical consequences. A yellow box labeled 'D' highlights the dbSNP ID rs1800562.

Number	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	[rs1800562]	-	[RCV000210820...]
.0002	HEMOCHROMATOSIS, TYPE 1 MICROVASCULAR COMPLICATIONS OF DIABETES, SUSCEPTIBILITY TO, 7, INCLUDED	HFE, HIS63ASP	[rs1799945]	[rs1799945]	[RCV000000027...]
.0003	HEMOCHROMATOSIS, TYPE 1	HFE, SER65CYS	[rs1800730]	-	[RCV000290779...]
.0004	HFE INTRONIC POLYMORPHISM	HFE, 5569G-A	[rs1800758]	[rs1800758]	[RCV000000031]
.0005	HFE POLYMORPHISM	HFE, VAL53MET	[rs28934889]	-	[RCV000000032]
.0006	HFE POLYMORPHISM	HFE, VAL59MET	[rs111033557]	-	[RCV000000033]
.0007	HEMOCHROMATOSIS, TYPE 1	HFE, GLN127HIS	[rs28934595]	-	[RCV000000034]
.0008	HEMOCHROMATOSIS, TYPE 1	HFE, ARG330MET	[rs111033558]	-	[RCV000000035]

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. [Nucleic Acids Res. 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