



# dbSNP: Database for Short Genetic Variations

Catalog of nucleotide changes for human and other model organisms

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

National Center for Biotechnology Information • National Library of Medicine • National Institutes of Health • Department of Health and Human Services

## Scope and access

The NCBI Short Genetic Variations database, commonly known as dbSNP, catalogs short variations in nucleotide sequences from a wide range of organisms. These variations include single nucleotide variations, short nucleotide insertions and deletions, short tandem repeats and microsatellites. 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, genotype information and allele origin, as some variations are somatic rather than germline events.



Short nucleotide variation data can be accessed via the SNP homepage or EUtils API:

[www.ncbi.nlm.nih.gov/snp](http://www.ncbi.nlm.nih.gov/snp) or [www.ncbi.nlm.nih.gov/projects/SNP/SNPeutils.htm](http://www.ncbi.nlm.nih.gov/projects/SNP/SNPeutils.htm)

VCF files and database bcp files are available for download through FTP or Aspera client at:

[ftp.ncbi.nlm.nih.gov/snp/](http://ftp.ncbi.nlm.nih.gov/snp/) or [www.ncbi.nlm.nih.gov/public/?snp/organisms/](http://www.ncbi.nlm.nih.gov/public/?snp/organisms/)

SNP data can also be accessed interactively through the new Variation Viewer:

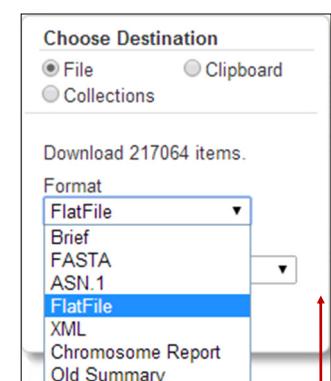
[www.ncbi.nlm.nih.gov/variation/view/](http://www.ncbi.nlm.nih.gov/variation/view/)

For help documentation, SNP database FAQs are available at:

[www.ncbi.nlm.nih.gov/books/NBK3848/](http://www.ncbi.nlm.nih.gov/books/NBK3848/)

For information on how to find known SNPs in a sequence, refer to this handout:

[ftp.ncbi.nlm.nih.gov/pub/factsheets/HowTo\\_Finding\\_SNP\\_by\\_BLAST.pdf](http://ftp.ncbi.nlm.nih.gov/pub/factsheets/HowTo_Finding_SNP_by_BLAST.pdf)



## 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 show variations in other formats, or sort retrieved variations in a different order. You can further narrow down retrieved variations by selecting filters listed 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 (**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” portet (**I**), you can retrieve related entries from other NCBI databases for the set of variations in the display.

**D** Organism: Homo sapiens  
Variation Class: in del, mmp, snp  
Clinical Significance: benign, likely benign, other, pathogenic  
Annotation: Cited in PubMed, OMIM, PubMed, nucleotide, protein, structure  
Function Class: 3'utr, 5' splice site, 5'utr, coding synonymous frame shift, intron, missense, nonsense, stop gained  
Global MAF: Custom range...  
Validation Status: by-1000 Genomes, by-2bit-2allele, by-cluster

**A** SNP: **human[orgn] AND HFE[gene]**  
Save search Advanced

**B**

**C** Display Settings:  Summary, 20 per page, Sorted by SNP\_ID

**E**

**F** [HFE \(GeneView\)](#)

**G** [HGVS](#)

**H** [Varview](#)

**I** Find related data  
Database: Select  
Find items  
Search details  
"Homo sapiens" [Organism] AND HFE [gene]  
Search See more...

**D** Results: 1 to 20 of 841

1. rs1799945 [Homo sapiens]

Chromosome: 6:2609051  
Gene: HFE (GeneView)  
Functional Consequence: intron variant, missense, nc transcript variant  
Allele Origin: G(germline)/C(germline)  
Clinical significance: other  
Validated: by 1000G, by cluster, by frequency, by hapmap  
Global MAF: G=0.0731/366  
HGVS:  
NC\_000006.11:g.26091179C>G, NC\_000006.12:g.26090951C>G,  
NG\_008720.2:g.8671C>G, NM\_000410.3:c.187C>G, NM\_001300749.1:c.187C>G,  
NM\_139003.2:c.187C>G, NM\_139004.2:c.187C>G, NM\_139006.2:c.187C>G,  
NM\_139007.2:c.77-363C>G, NM\_139008.2:c.77-1734C>G, NM\_139009.2:c.118C>G,  
NM\_139010.2:c.77-1734C>G, NM\_139011.2:c.77-2168C>G, NP\_000401.1:p.His63Asp,  
NP\_001287678.1:p.His63Asp, NP\_620572.1:p.His63Asp, NP\_620573.1:p.His63Asp,  
NP\_620575.1:p.His63Asp, NP\_620578.1:p.His40Asp, XM\_005249040.1:c.187C>G,  
XM\_011514543.1:c.187C>G, XM\_011514544.1:c.187C>G,  
XP\_005249097.1:p.His63Asp, XP\_011512845.1:p.His63Asp,  
XP\_011512846.1:p.His63Asp, XR\_241893.1:n.309C>G, XR\_241893.2:n.309C>G,  
XR\_241894.1:n.434C>G

2. rs1800562 [Homo sapiens]

Chromosome: 6:26092913  
Gene: HFE (GeneView)  
Functional Consequence: intron variant, missense, nc transcript variant  
Allele Origin: G(germline)/A(germline)  
Clinical significance: Pathogenic  
Validated: by 1000G, by cluster, by frequency, by hapmap  
Global MAF: A=0.0126/63  
HGVS:  
NC\_000006.11:g.26093141G>A, NC\_000006.12:g.26093141G>A, NG\_008720.2:g.10633G>A, NM\_000410.3:c.845G>A, NM\_001300749.1:c.845G>A,

**D** Format:  Summary,  Graphic Summary,  FASTA,  FlatFile,  Chromosome Report,  Old Summary,  dbSNP Batch Report  
Items per page: 5, 10, 20, 50, 100, 200  
Sort by: Default order, Organism, SNP\_ID, Success Rate, Heterozygosity, Chromosome Base Position  
**E** Apply

## 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**), mapping information in Human Genome Variation Society (HGVS) nomenclature (**B**), and a link to a new gene-centric display (via the VarView icon **C**, see pg.4). Its integrated map table (**D**) details the genome mapping information with the chromosomal coordinates (**E**) link to the same gene-centric display as the VarView icon. The magnifying glass (**F**) points to the 1000 Genomes Browser and provides genotyping details if the rsID is also reported by that project.

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

The Gene Model(s) table below lists transcript and protein coordinates and changes (**H**). The graphical presentation (**I**) further below presents variations with different characteristics in different tracks (**J**) under the context of annotated genome assembly. Individual variations are hyperlinked to provide additional details in the popup (not shown).

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

Reference SNP (refSNP) Cluster Report: rs1800730																																																																										
** With Pathogenic allele **																																																																										
<b>RefSNP</b> Organism: human ( <i>Homo sapiens</i> ) Molecule Type: Genomic Created/Updated in build: 89/144 Map to Genome Build: <a href="#">107</a> / <a href="#">Weight</a> <b>Validation Status:</b> Citation: <a href="#">PubMed</a>				<b>Allele</b> <b>A</b> <b>B</b> <b>Variation Class:</b> SNV: single nucleotide variation <b>RefSNP Alleles:</b> A/T (FWD) <b>Allele Origin:</b> A:germline T:germline <b>Ancestral Allele:</b> A <b>Variation Viewer:</b> <b>Clinical Significance:</b> With Pathogenic allele [ <a href="#">ClinVar</a> ] <b>MAF/MINorAlleleCount:</b> T=0.0040/20 <b>MAF Source:</b> 1000 Genomes				<b>HGVS Names</b> NC_00006.11:g.26091185A>T NC_00006.12:g.2609057A>T NG_008720.2:g.8677A>T NM_00410.3:c.193A>T NM_001300749.1:c.193A>T NM_139003.2:c.193A>T NM_139004.2:c.193A>T NM_139006.2:c.193A>T NM_139007.2:c.77-357A>T NM_139008.2:c.77-357A>T NM_139009.2:c.124A>T																																																																		
<b>Integrated Maps (Hint: click on 'E' Pos' to see variant in the new NCBI variation viewer)</b> <b>D</b>																																																																										
Assembly	Annotation Release	Chr	Chr Pos	Contig	Contig Pos	SNP to Chr	Contig allele	Contig to Chr	Neighbor SNP	Map Method																																																																
GRCh38.p2	<a href="#">107</a>	6	<a href="#">26090957</a>		<a href="#">NT_007592.16</a>	<a href="#">26030957</a>	Fwd	A	Fwd	<a href="#">view</a>	mapup																																																															
GRCh37.p13	<a href="#">105</a>	6	<a href="#">26091185</a>		<a href="#">NT_007592.15</a>	<a href="#">26031185</a>	Fwd	A	Fwd	<a href="#">view</a>	blast																																																															
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<input type="checkbox"/> View more variation on this gene (click to hide). <input checked="" type="checkbox"/> Clinical Source: <input type="radio"/> in gene region <input checked="" type="radio"/> cSNP <input type="radio"/> has frequency <input type="radio"/> double hit <a href="#">Go</a> <b>G</b>																																																																										
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Submitter records for this RefSNP Cluster <b>K</b>												
The submission ss24431742 has the longest flanking sequence of all cluster members and was used to instantiate sequence for rs1800730 during BLAST analysis for the current build.												
NCBI Assay ID	Handle Submitter ID	Validation Status	ss to rs Orientation /Strand	Alleles	5' Near Seq 30 bp		3' Near Seq 30 bp		Entry Date	Update Date	Build Added	Molecule Type
ss2420855	<a href="#">HGBASEISNP000007282</a>	fwd/T	A/T	gctgttcgttctatgtatcatgag	gtcgccgttgaggccccgaactcc				11/07/00	10/10/03 89		Genomic
ss68969473	PERLEGEN PGP04777602	fwd/	A/T	gaccaggctttgttttatgtatcatgatggatgtccgttgaggccccgaactccatgg	01/30/07 08/14/07 127							Genomic
ss160462894	ILLUMINA HumanOmni-Quad v1-0_B_rs1800730...	fwd/	A/T	gaccaggctttgttttatgtatcatgatggatgtccgttgaggccccgaactccatgg	08/04/09 10/20/09 131							Genomic
ss233370756	1000GENOMES pilot_1_CEU_2975385_chr6_261	fwd/	A/T	gaccaggctttgttttatgtatcatgatggatgtccgttgaggccccgaactccatgg	05/01/10 05/01/10 132							Genomic
ss24431742	[2010_April_001_075_HFE_235200_0003...]	fwd/	A/T	gaccaggctttgttttatgtatcatgatggatgtccgttgaggccccgaactccatgg	06/16/10 06/16/10 132							Genomic
ss28828927	Omm-CURATED-RECORDS 6335	fwd/	A/T	gaccaggctttgttttatgtatcatgatggatgtccgttgaggccccgaactccatgg	12/21/10 12/21/10 133							Genomic
ss342203118	NHLBI-ESP ESP2500-chr6-26091185	fwd/	A/T	gaccaggctttgttttatgtatcatgatggatgtccgttgaggccccgaactccatgg	03/25/11 03/26/11 134							Genomic
ss410868034	ILLUMINA Cardio-Metabo_Chip_A_chr6_26199164...	fwd/	A/T	gaccaggctttgttttatgtatcatgatggatgtccgttgaggccccgaactccatgg	06/07/11 06/07/11 135							Genomic
ss48106736	ILLUMINA HumanOmni-Quad v1-0_rs1800730-128...	fwd/	A/T	gaccaggctttgttttatgtatcatgatggatgtccgttgaggccccgaactccatgg	01/30/12/01/12/137							Genomic
ss490921024	1000GENOMES 20110521_exome_428822_chr6_260...	fwd/	A/T	gaccaggctttgttttatgtatcatgatggatgtccgttgaggccccgaactccatgg	02/10/12/02/21/12/137							Genomic
ss491378666	EXOME_CHIP nonsyn_94892_chr_6_26091185	fwd/	A/T	gaccaggctttgttttatgtatcatgatggatgtccgttgaggccccgaactccatgg	03/05/12/03/05/12/137							Genomic
ss491881981	CLINSEQ_SNPNISNV-chr6-26199164	fwd/	A/T	gctgttcgttctatgtatcatgag	gtccgcgttgaggccccgaactcc				03/06/12/03/13/12/137			Genomic

## 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. Additional links may be available in the NCBI Resource Links section (D) if available.

The Population Diversity section (E) 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 (p. 2, E).

**Fasta sequence (Legend)**

```
>gnl|dbSNP|rs1800730|allelePos=251|totalLen=501|taxid=9606|snpclass=1|alleles='A/T'|mol=Ge
CAGGACTGCA ACTCACCCCT CACAAAATGA GGACCAAGACA CAGCTGATGG TATGAGTTGA TGCGAGGTGTG
TGGAGCCTCA ACATCTGCT CCCCTCTAC TACACATGGT TAAGGCCCTGT TGCTCTGTCT CCAGGTCAC
ACTCTCTGCA CTACCTCTTC ATGGGTCCTC CAGACCCAGGA CCTTGGTCTT TCCTTGTGAA AAGCTTGGG
CTACGT GACCGACTGT TCGTGTCTA TGATCATGAG
W C GTCGCCCTT GGAGCCCCGA ACTCCATGGG TTTCAGTAG AATTCAGAC CAGATGTGGC TGCGAGTGAG
TCAGACTCTG AAAGGGTGGG ATCACATGTT CACTGTGAC TTCTGGACTA TTATGGAAAA TCACAACAC
AGCAAGGGTA TGTGGAGAGG GGGCTCACC TTCCCTGAGGT TGTCAAGACT TTTCATCTT TCATGCATCT
TGAAGGAAAC AGCTGGAAGT GTGGTCTT GTGGGAGCGAG
```

**NCBI Resource Links**

Submitter-Referenced	dbSNP Blast Analysis	UniGene Cluster ID	3D structure mapping	OMIM
GenBank Z92910		233325	NP_000401 NP_620572 NP_620573 NP_620575 NP_620578	235200_0003 613609_0003

**Population Diversity (in rs orientation)**

ss#	Population	Individual Group	Chrom.	Sample Cnt.	Source	Genotype Detail			Alleles		
						A/A	A/T	T/T	HWP	A	T
ss233370756_pilot_1_CEU_low_coverage_panel				120	AF				0.967	0.033	
ss342203118_ESP_Cohort_Populations				4552	GF	0.978	0.022		0.655	0.989	0.011
ss491881981_CSAgent				1323	GF	0.983	0.017		0.992	0.009	

## 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 link (G) 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 (H) menu in the upper right hand corner of the sequence record, simply check the SNPs checkbox and click Update View (I) to update the display.

**Customize view**

- Basic Features
  - Default features
  - Gene, RNA, and CDS features only
- Features added by NCBI
  - 65 SNPs
- Display options
  - Show sequence
  - Show reverse complement

**Update View**

**Related information**

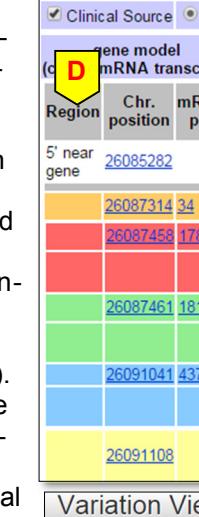
- Order cDNA clone
- 3D structures
- BioAssay, by Gene target
- BioProjects
- Books
- CCDS
- Conserved Domains
- dbVar
- Full text in PMC
- Genome
- GEO Profiles
- GTR
- HomoloGene
- Map Viewer
- Nucleotide
- OMIM
- Probe
- Protein
- PubChem Compound
- PubChem Substance
- PubMed
- PubMed (GeneRIF)
- PubMed (OMIM)
- RefSeq Proteins
- RefSeq RNAs
- RefSeqGene
- SNP
- SNP: GeneView
- SNP: Genotype
- SNP: VarView
- Taxonomy
- UniGene
- UniSTS

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 (J) display for OMIM record 613609, which cites the rsIDs in the dbSNP column (K).

613609				
HFE GENE; HFE				
Allelic Variants (11 Selected Examples):				
Number	Phenotype	Mutation	dbSNP	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	-	✓
.0002	HEMOCHROMATOSIS, TYPE 1 MICROVASCULAR COMPLICATIONS OF DIABETES, SUSCEPTIBILITY TO, 7, INCLUDED	HFE, HIS63ASP	[rs1799945]	✓
.0003	HEMOCHROMATOSIS, TYPE 1	HFE, SER65CY5	[rs1800730]	✓
.0004	HFE INTRONIC POLYMORPHISM	HFE, 5569G-A	-	✓
.0005	HFE POLYMORPHISM	HFE, VAL53MET	[rs28934889]	✓
.0006	HFE POLYMORPHISM	HFE, VAL59MET	[rs111033557]	✓
.0007	HEMOCHROMATOSIS, TYPE 1	HFE, GLN127HIS	[rs28934595]	✓
.0008	HEMOCHROMATOSIS, TYPE 1	HFE, ARG330MET	[rs111033558]	✓
.0009	HEMOCHROMATOSIS, TYPE 1	HFE, ILE105THR	[rs28934596]	✓
.0010	HEMOCHROMATOSIS, TYPE 1	HFE, GLY93ARG	[rs28934597]	✓
.0011	HEMOCHROMATOSIS, TYPE 1	HFE, GLN283PRO	[rs111033563]	✓

## The SNP:GeneView display

The SNP:GeneView display tabulates variations mapped to splice variants of a particular gene. The top of the page lists all annotated splice variants (**A**), and highlights the splice variant, for which nucleotide variations are shown in yellow (**B**). The default setting displays only the non-clinical coding variations. For the complete list, click “Refresh” after checking “Clinical Source” and “in gene region” options (**C**). This table arranges these variants by their sequential orders on the genome (**D**) and color-codes them by their function: white for “in gene region” (**E**), orange for UTR (**F**), red for non-synonymous (**G**), green for synonymous (**H**), blue for frame-shift (**I**), and yellow for intronic (**J**). Red, green, and blue are for the coding region. The MAF (**K**) column lists the global minor allele frequencies computed from 1000 Genomes data available.



Gene model (cDNA mRNA transcript):		Cont...	
Region	Chr. position	mRNA pos	dbSNP ID
5' near gene	<a href="#">26085282</a>		rs728...
	<a href="#">26087314</a>	34	rs626...
	<a href="#">26087458</a>	178	rs149...
	<a href="#">26087461</a>	181	rs114...
	<a href="#">26091041</a>	437	rs773...
	<a href="#">26091108</a>		rs207...

**Variation Viewer**

- ▶ Pick Assembly
- ▼ Search

## Variation Viewer

The GeneView display contains a link (**L**) to a new interactive display generated by the Variation Viewer (**M**), 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 (**N**). Filters (not shown) are available to selectively display variants of interest. More information on this tool is available online as a video tutorial, in a factsheet, or in the more detailed online help documentation.

**View display**

display tabulates variants of a particular gene. The page lists all annotations (A), and highlights which nucleotide variants follow (B). The default is non-clinical coding. To see complete list, click "Clinical Source" options (C). This table

Gene Model (mRNA alignment) information from genome sequence											
Total gene model (contig mRNA transcript):					12	Contig	Contig Label	List SNP			
mRNA	transcript	protein	mRNA orientation	Contig	Contig Label						
NM_000410_3	plus strand	NP_000401_1	forward	NT_007592_16	GRCh38.p2	< currently show			B		
XM_011514544_1	plus strand	XP_011512846_1	forward	NT_007592_16	GRCh38.p2	<a href="#">View SNP on GeneModel</a>					
XM_011514543_1	plus strand	XP_011512845_1	forward	NT_007592_16	GRCh38.p2	<a href="#">View SNP on GeneModel</a>					
NM_139011_2	plus strand	NP_620580_1	forward	NT_007592_16	GRCh38.p2	<a href="#">View SNP on GeneModel</a>					
NM_139010_2	plus strand	NP_620579_1	forward	NT_007592_16	GRCh38.p2	<a href="#">View SNP on GeneModel</a>					
NM_139009_2	plus strand	NP_620578_1	forward	NT_007592_16	GRCh38.p2	<a href="#">View SNP on GeneModel</a>					
NM_139009_2	plus strand	NP_620577_1	forward	NT_007592_16	GRCh38.p2	<a href="#">View SNP on GeneModel</a>					

**D** Clinical Source **E** in gene region **F** cSNP **G** has frequency **H** double hit **I** refresh **J**

Gene model (mRNA transcript): Contig Label Contig mRNA protein mRNA orientation transcript SNP count																
Region	Chr. position	mRNA pos	dbSNP rs# cluster id	Heterozygosity	Validation	MAF	Allele origin	3D	Clinically Associated	Clinical Significance	Function	dbSNP allele	Protein residue	Codon pos	Amino acid pos	PubMed
5' near gene	<a href="#">26085282</a>	<a href="#">rs72834669</a>	0.040			0.0202					5' near gene					<b>E</b>
	<a href="#">26087314</a>	<a href="#">34</a>	<a href="#">rs52625316</a>	0.000		0.0002					5' UTR	C				<b>F</b>
	<a href="#">26087458</a>	<a href="#">178</a>	<a href="#">rs149342416</a>	0.002						missense	C	Ser [S]	3	6	<b>G</b>	
										contig reference	G	Arg [R]	3	6		
	<a href="#">26087461</a>	<a href="#">181</a>	<a href="#">rs114758821</a>	0.003		0.0014					synonymous	A	Pro [P]	3	7	<b>H</b>
										contig reference	G	Pro [P]	3	7		
	<a href="#">26091041</a>	<a href="#">437</a>	<a href="#">rs773296212</a>	0.000						frame shift	-	Gly [G]	1	9	<b>I</b>	
										contig reference	G	Trp [W]	1	9		
	<a href="#">26091108</a>	<a href="#">rs2071303</a>	0.489			0.4267				Likely benign	intron					<b>J</b>

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## Factsheet for Variation Viewer: Online video tutorial: