

ONLINE GENOME DATABASES

AÜTF D1M1, 2019

Tools

[All tools](#)

BioMart >

Export custom datasets from Ensembl with this data-mining tool

BLAST/BLAT >

Search our genomes for your DNA or protein sequence

Variant Effect Predictor >

Analyse your own variants and predict the functional consequences of known and unknown variants

Search

All species for [input] Go

e.g. BRCA2 or rat 5:62797383-63627669 or rs699 or coronary heart disease

All genomes

-- Select a species --

- View full list of all Ensembl species
Edit your favourites

Favourite genomes

Human GRCh38.p12

Switching GRCh37?

Mouse GRCh38.p6

Zebrafish GRCz11

Ensembl is a genome browser for vertebrate genomes that supports research in comparative genomics, evolution, sequence variation and transcriptional regulation.

Ensembl Release 94 (October 2018)

- New fish: 38 new and updated fish genomes
GENCODE update 29 for human and M19 for mouse
Additional pathogenicity predictors for missense variants
New transcription factor binding motifs from SELEX
Gene trees using HMMs

More release news on our blog

Other news from our blog

- 06 Nov 2018: Job: Regulation Project Leader
06 Nov 2018: Job: Bioinformatician - comparative genomics
26 Oct 2018: Cool stuff the VEP can do: splice site variant annotation

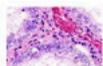
Compare genes across species



Find SNPs and other variants for my gene

Genetic sequence alignment showing SNPs and variants.

Gene expression in different tissues



Retrieve gene sequence

ATCGATCCCGGGTGGG
GGGCTGTGGCCGAGC
GGGCTCTGCTCCGCTT
AAGGACAGATTTGTGAM
CACCTCTGGAAGCGGTTT
GCAATTCAGGCTGGG

Find a Data Display



Use my own data in Ensembl



EMBL-EBI Ensembl creates, integrates and distributes reference datasets and analysis tools that enable genomics. We are based at EMBL-EBI and our software and data are freely available. Our acknowledgements page includes a list of current and previous funding bodies. How to cite Ensembl in your own publications.



Ensembl's latest human genome "assembly"

Insan genomunda arama penceresi

Ensembl BLAST/BLAT | VEP | Tools | BioMart | Downloads | Help & Docs | Blog

Human (GRCh38.p12) ▾

Search Human (*Homo sapiens*)

Search all categories ▾ Search Human... Go

e.g. **BRCA2** or **17:63992802-64038237** or **rs1333049** or **osteoarthritis**

Genome assembly: GRCh38.p12 (GCA_000001405.27)

- More information and statistics
- Download DNA sequence (FASTA)
- Convert your data to GRCh38 coordinates
- Display your data in Ensembl

Other assemblies

GRCh37 Full Feb 2014 archive with BLAST, VEP and BioMart Go

Gene annotation

What can I find? Protein-coding and non-coding genes, splice variants, cDNA and protein sequences, non-coding RNAs.

- More about this genebuild
- Download FASTA files for genes, cDNAs, ncRNA, proteins
- Download GTF or GFF3 files for genes, cDNAs, ncRNA, proteins
- Update your old Ensembl IDs

Comparative genomics

What can I find? Homologues, gene trees, and whole genome alignments across multiple species.

- More about comparative analysis
- Download alignments (EMF)

Regulation

What can I find? DNA methylation, transcription factor binding sites, histone modifications, and regulatory features such as enhancers and repressors, and microarray annotations.

- More about the Ensembl regulatory build and microarray annotation
- Experimental data sources
- Download all regulatory features (GFF)

Variation

What can I find? Short sequence variants and longer structural variants; disease and other phenotypes

- More about variation in Ensembl
- Download all variants (GVF)
- Variant Effect Predictor **Ve!P**

View karyotype

Example region

Example gene tree

Example regulatory feature

ENCODE data in Ensembl

Example transcript

Example variant

Example phenotype

Example structural variant

GRCh38.p13 statistics

Human chromosomes

GRCh38.p12 statistics



Human assembly and gene annotation

Assembly

This site provides a data set based on the December 2013 *Homo sapiens* high coverage assembly GRCh38 from the [Genome Reference Consortium](#). This assembly is used by UCSC to create their hg38 database. The data set consists of gene models built from the genewise alignments of the human proteome as well as from alignments of human cDNAs using the cDNA2genome model of exonerate.

This release of the assembly has the following properties:

- contig length total 3.4 Gb.
- chromosome length total 3.1 Gb (excluding haplotypes).

It also includes 261 alt loci scaffolds, mainly in the LRC/KIR complex on chromosome 19 (35 alternate sequence representations) and the [MHC region on chromosome 6](#) (7 alternate sequence representations).

 [Watch a video on YouTube](#) about patches and haplotypes in the Human genome.

Patches

As the GRC maintains and improves the assembly, patches are being introduced. Currently, assembly patches are of two types:

- Novel patch: new sequences that add alternative sequence at a loci and will remain as haplotypes in the next major assembly release by GRC
- Fix patch: sequences that correct the reference sequence and will replace the given region of the reference assembly at the next major assembly release by GRC.

The genome assembly represented here corresponds to GenBank Assembly ID [GCA_000001405.27](#)

Other assemblies

Gene annotation

The Ensembl human gene annotations have been updated using Ensembl's automatic annotation pipeline. The updated annotation incorporates new protein and cDNA sequences which have become publicly available since the last GRCh38 genebuild (December 2013).

In the current release, we continue to display a joint gene set based on the merge between the automatic annotation from Ensembl and the manually curated annotation from Havana. See the statistics table, right, for the corresponding GENCODE version number. The Consensus Coding Sequence (CCDS) identifiers have also been mapped to the annotations. More information about the [CCDS project](#).

Updated manual annotation from Havana is merged into the Ensembl annotation every release. Transcripts from the two annotation sources are merged if they share the same internal exon-intron boundaries (i.e. have identical splicing pattern) with slight differences in the terminal exons allowed. Importantly, all Havana transcripts are included in the final Ensembl/Havana merged (GENCODE) gene set.

- [Detailed information on genebuild](#) (PDF)

Neanderthal genome

A preliminary assembly of the Neanderthal (*Homo sapiens neanderthalensis*) genome is available via the [Neanderthal Genome Browser](#), an Ensembl-powered project based at the Max Planck Institute.

More information

General information about this species can be found in [Wikipedia](#).

Statistics

Summary

Assembly	GRCh38.p13 (Genome Reference Consortium Human Build 38), INSDC Assembly GCA_000001405.28 , Dec 2013
Base Pairs	4,537,931,177
Golden Path Length	3,096,649,726
Annotation provider	Ensembl
Annotation method	Full genebuild
Genebuild started	Jan 2014
Genebuild released	Jul 2014
Genebuild last updated/patched	Jun 2019
Database version	98.38
Gencode version	GENCODE 32

Gene counts (Primary assembly)

Coding genes	20,444 (incl 667 readthrough)
Non coding genes	23,949
Small non coding genes	4,871
Long non coding genes	16,857 (incl 304 readthrough)
Misc non coding genes	2,221
Pseudogenes	15,214 (incl 8 readthrough)
Gene transcripts	227,530

Gene counts (Alternative sequence)

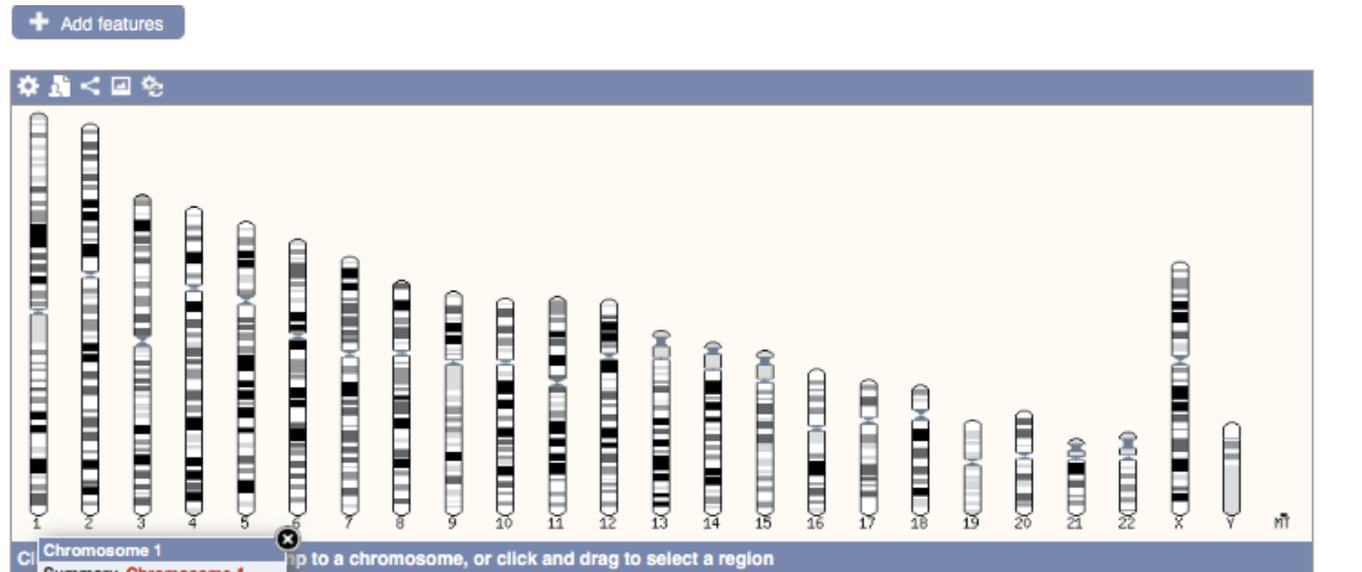
Coding genes	3,054 (incl 44 readthrough)
Non coding genes	1,555
Small non coding genes	297
Long non coding genes	1,071 (incl 45 readthrough)
Misc non coding genes	187
Pseudogenes	1,798
Gene transcripts	21,636

Other

Genscan gene predictions	51,756
Short Variants	665,834,144
Structural variants	6,013,113

İnsan karyotip görüntüsü

+ Add features



Chromosome 1

Summary [Chromosome 1](#)

Example [Example region on 1](#)

Assembly GRCh38.p12 (Genome Reference Consortium Human Build 38), INSDC Assembl

Base Pairs 3,609,003,417

Golden Path Length 3,096,649,726

Annotation provider Ensembl

Annotation method Full genebuild

Genebuild started Jan 2014

Genebuild released Jul 2014

Genebuild last updated/patched Jul 2018

Database version 94.38

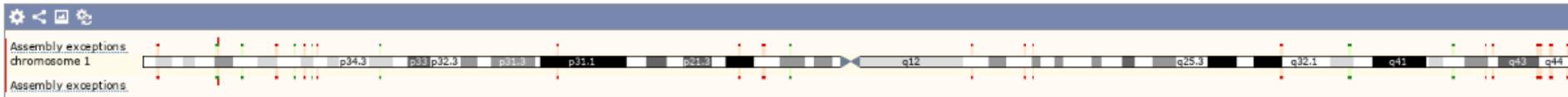
Gencode version GENCODE 29

Gene counts (Primary assembly)

Coding genes	20,418 (incl 650 readthrough)
Non coding genes	22,107
Small non coding genes	4,871
Long non coding genes	15,014 (incl 284 readthrough)
Misc non coding genes	2,222
Pseudogenes	15,195 (incl 8 readthrough)
Gene transcripts	206,762

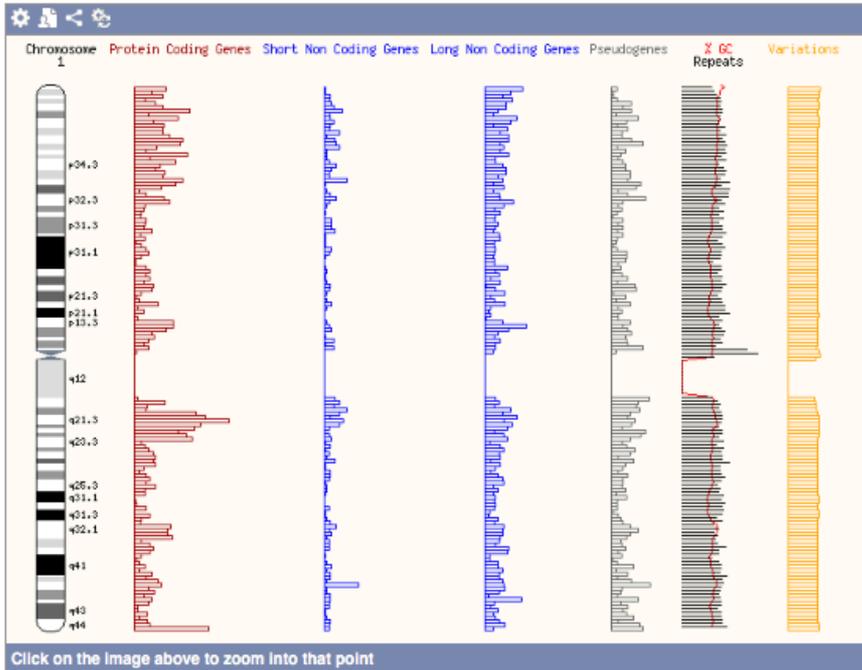
Click for further information

Chromosome 1: 1-1



Chromosome summary

+ Add features



Diğer kromozomları seçin

Change chromosome: Go

Chromosome Statistics	
Length (bps)	248,956,422
Coding genes	2,057 (incl. 49 readthrough)
Non coding genes	2,088
Small non coding genes	482
Long non coding genes	1,414 (incl. 14 readthrough)
Misc non coding genes	192
Pseudogenes	1,293
Short Variants	52,130,200

Protein coding genes
Non coding genes (RNA genes)
Pseudogenes

- Location: 1:1-1
- Location-based displays
- Whole genome
 - Chromosome summary**
 - Region overview
 - Region in detail
 - Comparative Genomics
 - Synteny**
 - Alignments (image)
 - Alignments (text)
 - Region Comparison
 - Genetic Variation
 - Variant table
 - Resequencing
 - Linkage Data
 - Markers
 - Other genome browsers
 - UCSC
 - NCBI
 - Ensembl GRCh37

Chromosome 1: 1-1



Chromosome summary

+ Add features



Synteny regions for comparison between organisms' chromosomes

Synteny regions compared



Synteny

Synteny between Human chromosome 2 and Chimpanzee

Change Species:

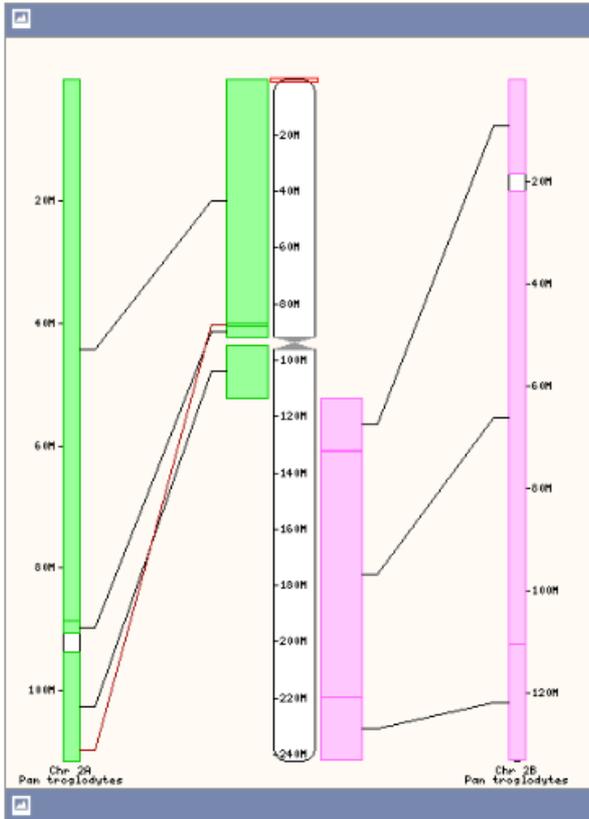
Chimpanzee Go

Select organism

Change chromosome:

2 Go

Select chromosome



Human chromosome 2
Syntenic regions on chimps chromosomes 2A, 2B

The screenshot shows the top section of the HGNC website. On the left is the HGNC logo with the text 'HUGO Gene Nomenclature Committee'. To the right is a search bar with the placeholder text 'Search everything' and 'Search symbols, keywords or IDs'. Below the search bar is a note: 'Use * to search with a root symbol (e.g. *NF*)'. A red arrow points from the text 'Inquiry for gene names and symbols' to the search bar. Below the search bar is a navigation menu with buttons for 'Home', 'Downloads', 'Gene Families', 'Tools', 'Useful links', 'About', 'Newsletters', 'Contact Us', 'Help', 'VGNC', and 'Request Symbol'.

Inquiry for gene names and symbols

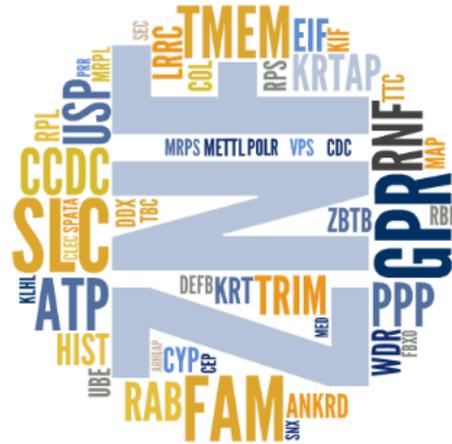
HGNC is responsible for approving unique symbols and names for human loci, including protein coding genes, ncRNA genes and pseudogenes, to allow unambiguous scientific communication.

genenames.org is a curated online repository of HGNC-approved gene nomenclature, gene families and associated resources including links to genomic, proteomic and phenotypic information.

Search our catalogue of more than 40,000 symbol reports using our improved search engine (see [Search help](#)), search lists of symbols using our [Multi-symbol checker](#) and identify possible orthologs using our [HCOP tool](#).

Download our ready-made data files from our [Statistics and Downloads](#) page, create your own datasets using either our [Custom Downloads](#) tool or [BioMart](#) service, or write a script/program utilising our [REST service](#).

Submit your [gene symbol and name proposals](#) to us to be accredited with HGNC approved nomenclature for use in publications, databases and presentations.



FAQ

- [What is the HGNC?](#)
- [What is HGNC-approved nomenclature and why do we need it?](#)
- [Where can I find information about existing human gene symbols?](#)
- [What is a stem symbol?](#)
- [Where can I find the Nomenclature Guidelines?](#)
- [Do I have to use the approved symbols?](#)
- [How should I cite HGNC nomenclature resources?](#)

Latest News

New species data available in HCOP

We are pleased to announce that [HCOP](#) now contains orthology data for two new species, cat (*Felis catus*) and fission yeast (*Schizosaccharomyces pombe*). The addition of *S. pombe* includes gene nomenclature and orthology data from [PomBase](#).

If there is a species or orthology source that you would like to see included in HCOP please contact us via email hgnc@genenames.org.

[genenames.org](#) is now HTTPS compliant



New Beta Site!

Please view [this page](#) on our new HGNC beta site and let us know what you think via the [feedback form](#).

Search

Document type

Gene 200
Family 50
Site 9

Items per page

10 20 50 100 200

Results: 1 to 10 of 259

Page 1 of 26 Next > Last >>

Histones

Document type: Family HGNC Family ID: 864
Matches: Gene name: H1 histone family member 0
Previous gene name: H1 histone family, member 0

Histone deacetylase superfamily

Document type: Family HGNC Family ID: 937 Root symbol: HDAC, SIRT
Matches: Family alias: Histone deacetylases
Family name: Histone deacetylase superfamily

Histone deacetylases, class I

Document type: Family HGNC Family ID: 989 Root symbol: HDAC
Matches: Family name: Histone deacetylases, class I
Gene name: histone deacetylase 1
Previous gene name: histone deacetylase-like 1

Histone deacetylases, class II

Document type: Family HGNC Family ID: 990 Root symbol: HDAC
Matches: Family name: Histone deacetylases, class II

Histone genes listing

www.genenames.org

Histon genleri

[Home](#)
[Downloads](#)
[Gene Families](#)
[Tools](#)
[Useful Links](#)
[About](#)
[Newsletters](#)
[Contact Us](#)
[Help](#)
[VGNC](#)
[Request Symbol](#)

Gene Family: Histones

Histone: In biology, histones are highly alkaline proteins found in eukaryotic cell nuclei that package and order the DNA into structural units called nucleosomes. They are the chief protein components of chromatin, acting as spools around which DNA winds, and play a role in gene regulation. Without histones, the unwound DNA in chromosomes would be very long (a length to width ratio of more than 10 million to 1 in human DNA). For example, each human cell has about 1.8 meters of DNA, (~6 ft) but wound on the histones it has about 90 micrometers (0.09 mm) of chromatin, which, when duplicated and condensed during mitosis, result in about 120 micrometers of chromosomes. [Source: [Wikipedia](#)]

The mapped domains of [P07305](#), encoded by the [H1FO](#) gene, an example gene within the family. [Source: [Pfam](#) & [UniProt](#)]

Genes contained within the family: 116

Approved Symbol	Approved Name	Previous Symbols	Synonyms	Chromosome
HIST1H1A	histone cluster 1 H1 family member a	H1F1	H1.1, H1a	6p22.2
HIST1H1B	histone cluster 1 H1 family member b	H1F5	H1.5, H1b, H1s-3	6p22.1
HIST1H1C	histone cluster 1 H1 family member c	H1F2	H1.2, H1s-1, H1c	6p22.2
HIST1H1D	histone cluster 1 H1 family member d	H1F3	H1.3, H1d, H1s-2	6p22.2
HIST1H1E	histone cluster 1 H1 family member e	H1F4	H1.4, H1e, H1s-4	6p22.2
HIST1H1PS1	histone cluster 1 H1 pseudogene 1		dJ34820.16, FU39701, H1F6P	6p22.2
HIST1H1PS2	histone cluster 1 H1 pseudogene 2	H3FEP	pH3/e	6p22.1
HIST1H1T	histone cluster 1 H1 family member t	H1FT	H1t	6p22.2
HIST1H2AA	histone cluster 1 H2A family member a		bA317E16.2, H2AFR	6p22.2
HIST1H2AB	histone cluster 1 H2A family member b	H2AFM	H2A/m	6p22.2
HIST1H2AC	histone cluster 1 H2A family member c	H2AFL		6p22.2
HIST1H2AD	histone cluster 1 H2A family member d	H2AFG	H2A/g, H2A.3	6p22.2
HIST1H2AE	histone cluster 1 H2A family member e	H2AFA	H2A/a, H2A.1	6p22.2
HIST1H2AG	histone cluster 1 H2A family member g	H2AFP	pH2A/f, H2A/p, H2A.1b	6p22.1
HIST1H2AH	histone cluster 1 H2A family member h		H2AFAl; dJ86C11.1, H2A/5	6p22.1
HIST1H2AI	histone cluster 1 H2A family member i	H2AFC	H2A/c	6p22.1
HIST1H2AJ	histone cluster 1 H2A family member j	H2AFE	H2A/E	6p22.1
HIST1H2AK	histone cluster 1 H2A family member k	H2AFD	H2A/d	6p22.1
HIST1H2AL	histone cluster 1 H2A family member l	H2AFI	H2A/i; dJ193B12.9	6p22.1
HIST1H2AM	histone cluster 1 H2A family member m	H2AFN	H2A/n, H2A.1	6p22.1
HIST1H2APS1	histone cluster 1 H2A pseudogene 1		bA317E16.5, H2AF5P	6p22.2
HIST1H2APS2	histone cluster 1 H2A pseudogene 2	H2AFTP	dJ139G21.2, H2A/T	6p22.2
HIST1H2APS3	histone cluster 1 H2A pseudogene 3		dJ34820.8, H2AFUP	6p22.2
HIST1H2APS4	histone cluster 1 H2A pseudogene 4	H2AFK, H2AFKP	pH2A/k, dJ34820.2	6p22.2

Given below enlarged
Histone cluster 1
H1 gene family member a

Symbol: HIST1H1A
Kromozom lokusu: 6p22.2

Click for details

Genes contained within the family: 116

Approved Symbol	Approved Name	Previous Symbols	Synonyms	Chromosome
HIST1H1A	histone cluster 1 H1 family member a	H1F1	H1.1, H1a	6p22.2

HIST1H1A

HGNC
HUGO Gene Nomenclature Committee

Search everything

Use * to search with a root symbol (eg ZNF*)

Home Downloads Gene Families Tools Useful Links About Newsletters Contact Us Help VGNC Request Symbol

Symbol Report: HIST1H1A

APPROVED SYMBOL	HIST1H1A
APPROVED NAME	histone cluster 1 H1 family member a
HGNC ID	HGNC:4715
PREVIOUS SYMBOLS & NAMES	"H1 histone family, member 1", H1F1, "histone 1, H1a", "histone cluster 1, H1a"
SYNONYMS	H1.1, H1a
LOCUS TYPE	gene with protein product
CHROMOSOMAL LOCATION	6p22.2
GENE FAMILY	Histones
HCOP	Orthology Predictions for HIST1H1A

External links

HOMOLOGS

	Symbol	Database
Mus musculus	Hist1h1a	MG1:1931523 C
Rattus norvegicus	Hist1h1a	RGD:1305706 D

GENE RESOURCES

Entrez Gene: [3024](#) C
Ensembl: [ENSG00000124610](#) C [Region in detail](#) [Sequence](#)
Vega: [OTTHUMG00000016413](#) C [Region in detail](#) [Sequence](#)
UCSC: [uc003nfo.4](#) D [Genome browser](#)

NUCLEOTIDE SEQUENCES

AF531299 C [GenBank](#) [ENA](#) [DDBJ](#)
NM_005325 C [RefSeq](#) [NCBI Sequence Viewer](#)
CCDS4569 C [CCDS](#)

PROTEIN RESOURCES

Q02539 D [UniProt](#) [InterPro](#) [PDBa](#)

CLINICAL RESOURCES

OMIM: [142709](#) D
[GeneTests](#) D
[DECIPHER](#) D
[COSMIC](#) D
[Genetic Testing Registry](#) C

REFERENCES

Human H1 histones: conserved and varied sequence elements in two H1 subtype genes.
Eick S *et al.* *Eur. J. Cell Biol.* 1989 Jun;49(1):110-115
PMID: 2759094 [Europe PMC](#) [PubMed](#)

The human and mouse replication-dependent histone genes.
Marzluff WF *et al.* *Genomics* 2002 Nov;80(5):487-498
PMID: 12408966 [Europe PMC](#) [PubMed](#)

OTHER DATABASE LINKS

[BioGPS](#) D
[GENATLAS](#) D
[GeneCards](#) D
[GOPubmed](#) D
[H-InvDB](#) D
[QuickGO](#) D
[Reactome](#) D
[WikiGenes](#) D

Click to go to Ensembl link

Click sequence



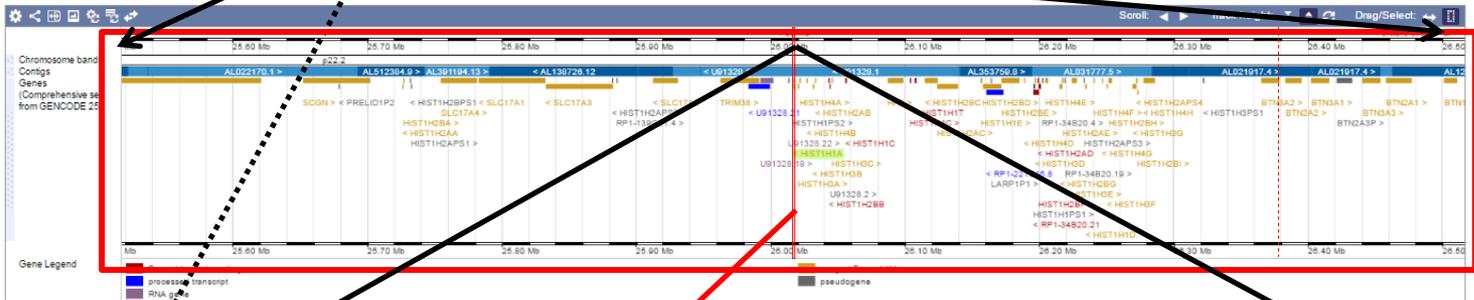
HIST1H1A

Chromosome 6: 26,017,085-26,017,732

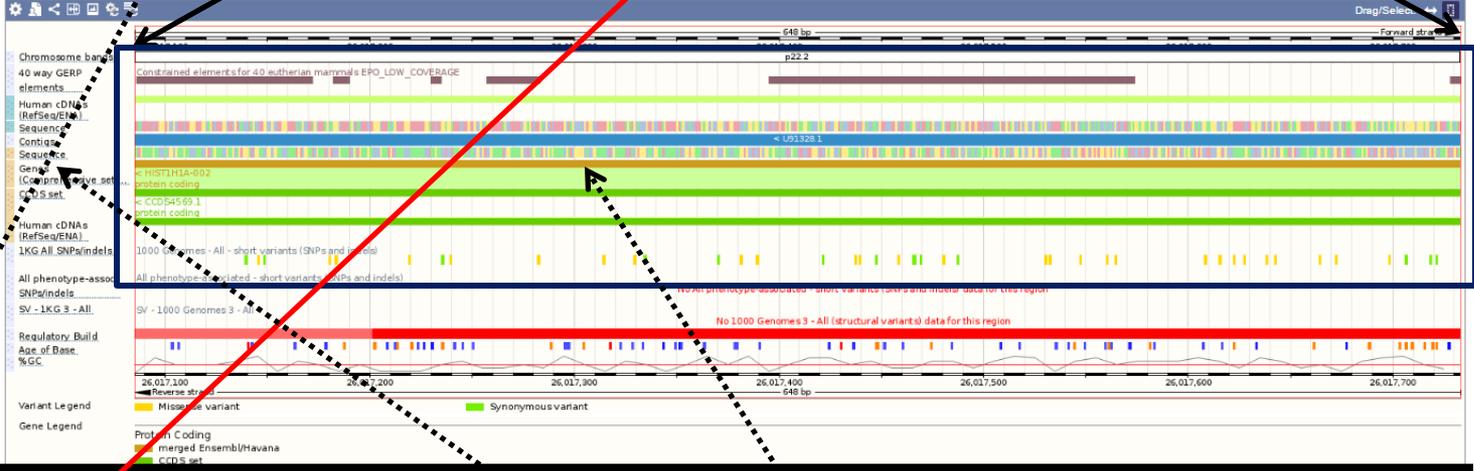
- Region in detail
- Comparative Genomics
 - Synteny
 - Alignments (image)
 - Alignments (text)
 - Region Comparison
- Genetic Variation
 - Resequencing
 - Linkage Data
- Markers
- Other genome browsers
 - UCSC
 - NCBI
 - Vega
 - Ensembl GRCh37
- Configure this page
- Custom tracks
- Export data
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Region in detail



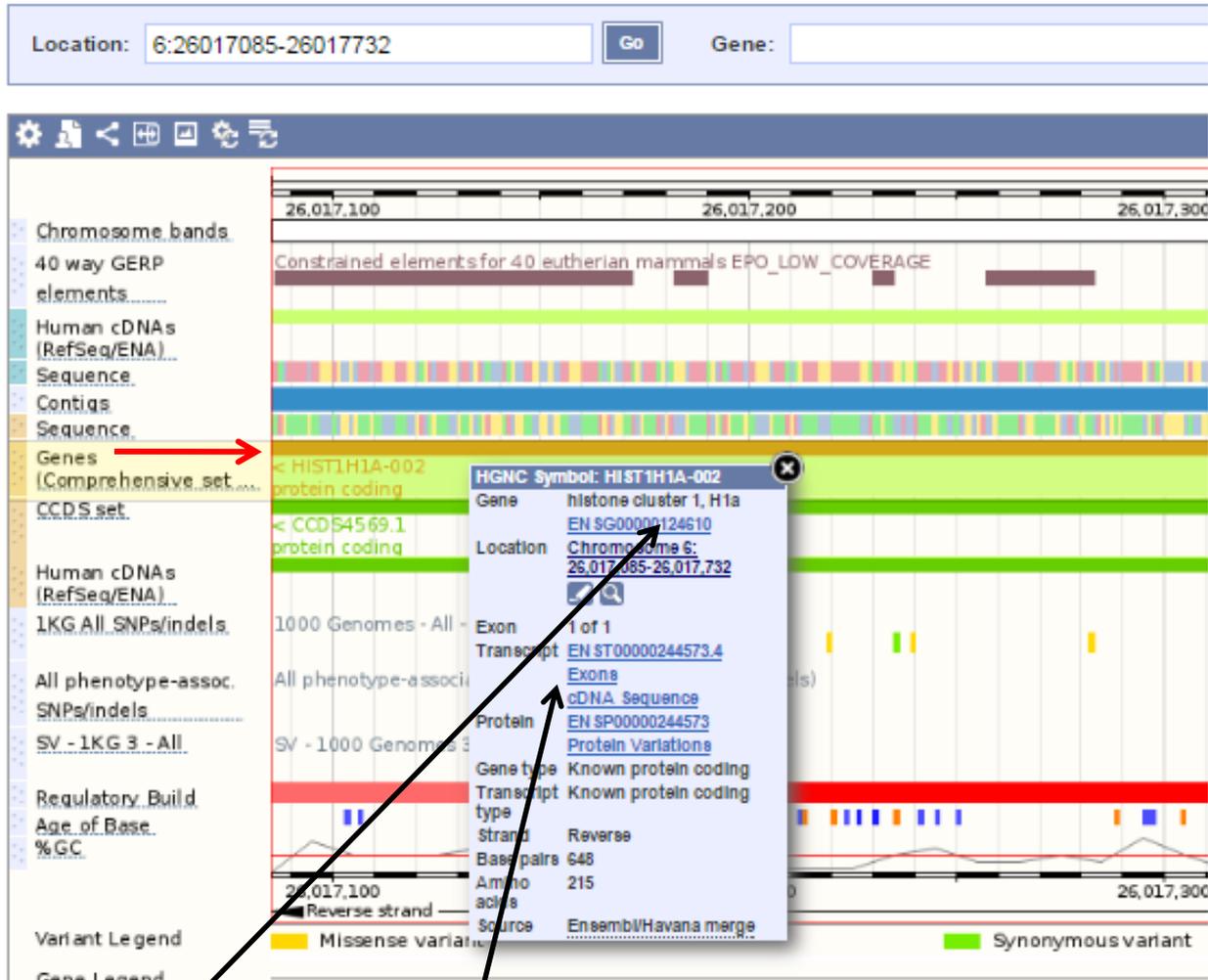
Location: 6:26017085-26017732 Gene:



HIST1H1A gene sequence is reached by clicking the gene row

- Chromosome location in between red bars
- The below window in red rectangle shows the region in detail with neighboring sequences/genes
- Red thin line is enlarged to show the gene of interest.

HIST1H1A geni haritasi



Gene page

Transcripts, exons, cDNA and proteins can be reached

HIST1H1A bilgi sayfası

Location: 6:26,017,085-26,017,732

Gene: HIST1H1A

Transcript: HIST1H1A-201

Gene-based displays

- Summary
 - Splice variants
 - Transcript comparison
 - Gene alleles
- Sequence
 - Secondary Structure
- Comparative Genomics
 - Genomic alignments
 - Gene tree
 - Gene gain/loss tree
 - Orthologues
 - Paralogues
 - Ensembl protein families
- Ontologies
 - GO: Molecular function
 - GO: Cellular component
 - GO: Biological process
- Phenotypes
- Genetic Variation
 - Variant table
 - Variant image
 - Structural variants
- Gene expression
- Pathway
- Regulation
- External references
- Supporting evidence
- ID History
 - Gene history

Gene: HIST1H1A ENSG00000124610

Description histone cluster 1 H1 family member a [Source:HGNC Symbol;Acc:HGNC:4715]

Gene Synonyms H1.1, H1F1, H1a

Location [Chromosome 6: 26,017,085-26,017,732](#) reverse strand.
GRCh38:CM000668.2

About this gene This gene has 1 transcript ([splice variant](#)), [43 orthologues](#) and is a member of [1 Ensembl protein family](#).

Transcripts [Hide transcript table](#)

43 orthologues

Show/hide columns (1 hidden)

Name	Transcript ID	bp	Protein	Biotype	CCDS	UniProt	RefSeq	Flags
HIST1H1A-201	ENST00000244573.4	648	215aa	Protein coding	CCDS4569.1	Q02539	NM_005325 NP_005316	TSL:NA GENCODE basic APPRIS P1

Summary

Name [HIST1H1A](#) (HGNC Symbol)

CCDS This gene is a member of the Human CCDS set: [CCDS4569.1](#)

UniProtKB This gene has proteins that correspond to the following UniProtKB identifiers: [Q02539](#)

RefSeq Overlapping RefSeq annotation not matched

Ensembl version ENSG00000124610.4

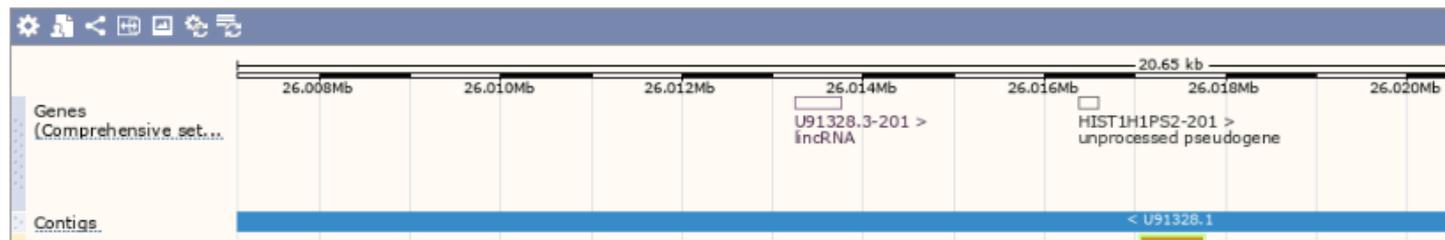
Other assemblies This gene maps to [26,017,313-26,017,960](#) in GRCh37 coordinates.

View this locus in the GRCh37 archive: [ENSG00000124610](#)

Gene type Protein coding

Annotation method Annotation for this gene includes both automatic annotation from Ensembl and [Havana](#) manual curation, see [article](#).

 Go to [Region in Detail](#) for more tracks and navigation options (e.g. zooming)



HIST1H1A orthologues

Ensembl BLAST/BLAT | BioMart | Tools | Downloads | Help & Documentation | Blog | Mirrors Login/Register

Human (GRCh38.p7) Location: 6,26,017,069-26,017,732 Gene: HIST1H1A Transcript: HIST1H1A-002

Gene-based displays

- Summary
 - Splice variants
 - Transcript comparison
 - Gene alleles
- Sequence
 - Secondary Structure
- Comparative Genomics
 - Genomic alignments
 - Gene tree
 - Gene gain/loss tree
 - Orthologues**
 - Paralogues
 - Ensembl protein families
- Ontologies
 - GO: Biological process
 - GO: Molecular function
 - GO: Cellular component
- Phenotypes
 - Genetic Variation
 - Variant table
 - Variant image
 - Structural variants
 - Gene expression
 - Regulation
 - External references
 - Supporting evidence
 - ID History
 - Gene history

Gene: HIST1H1A ENSG00000124610

Description histone cluster 1, H1a [Source:HGNC Symbol;Acc:HGNC:47156]

Synonyms H1F1, H1a, H1A, H1.1, HIST1

Location [Chromosome 6: 26,017,069-26,017,732](#) reverse strand.
GRCh38:CM000668.2

About this gene This gene has 1 transcript ([splice variant](#)), [141 orthologues](#), [5 paralogues](#), is a member of [1 Ensembl protein family](#) and is associated with [1 phenotype](#).

Transcripts [Show transcript table](#)

Orthologues [Download orthologues](#)

Summary of orthologues of this gene

Click on 'Show details' to display the orthologues for one or more groups of species. Alternatively, click on 'Configure this page' to choose a custom list of species.

Species set	Show details	With 1:1 orthologues	With 1:many orthologues	With many:many orthologues	Without orthologues
Primates (11 species) Humans and other primates	<input type="checkbox"/>	7	0	0	4
Rodents (8 species) Rodents, rabbits and related species	<input type="checkbox"/>	7	0	0	1
Laurasiatheria (14 species) Carnivores, ungulates and insectivores	<input type="checkbox"/>	8	0	0	6
Placental Mammals (38 species) All placental mammals	<input type="checkbox"/>	26	0	0	12
Sauropsida (7 species) Birds and Reptiles	<input type="checkbox"/>	0	0	7	0
Fish (11 species) Ray-finned fishes	<input type="checkbox"/>	0	2	9	0
All (68 species) All species, including invertebrates	<input type="checkbox"/>	26	3	20	19

Selected orthologues

Show **All** entries Show/hide columns

Species	Type	Orthologue	dN/dS	Target %id	Query %id	GOC Score	WGA Coverage	High Confidence
Alpaca (Vicugna pacos)	1-to-1	HIST1H1A (ENSVPAG00000008215) View Gene Tree Compare Regions (GeneScaffold_2801:3,291,052-3,291,708-1) View Sequence Alignments	n/a	83.03 %	84.19 %	50	100.00	Y
Amazon molly (Poecilia formosa)	Many-to-many	hist1l (1 of many) (ENSFP0G00000011672) View Gene Tree Compare Regions (K1519952.1:301,208-302,584-1) View Sequence Alignments	n/a	48.25 %	51.16 %	n/a	n/a	Y

İlgilendiğiniz türlerle ilgili seçim yapabilir

Scroll down to see other organisms

HIST1H1A human – primate orthologues

Gene: HIST1H1A | Transcript: HIST1H1A-201

Gene: HIST1H1A ENSG00000124610

Description histone cluster 1 H1 family member a [Source:HGNC Symbol;Acc:HGNC:4715;#]
Gene Synonyms H1.1, H1F1, H1a
Location [Chromosome 6: 26,017,085-26,017,732](#) reverse strand.
GRCh38:CM000668.2
About this gene This gene has 1 transcript ([splice variant](#)), [43 orthologues](#) and is a member of [1 Ensembl protein family](#).
Transcripts [Show transcript table](#)

Orthologues ?

[Download orthologues](#)

Summary of orthologues of this gene [Hide](#)

Click on 'Show details' to display the orthologues for one or more groups of species. Alternatively, click on 'Configure this page' to choose a custom list of species.

Species set	Show details	With 1:1 orthologues	With 1:many orthologues	With many:many orthologues	Without orthologues
Primates (23 species) Humans and other primates	<input checked="" type="checkbox"/>	6	0	0	17
Rodents and related species (24 species) Rodents, lagomorphs and tree shrews	<input type="checkbox"/>	20	0	0	4
Laurasiatheria (17 species) Carnivores, ungulates and insectivores	<input type="checkbox"/>	10	0	0	7
Placental Mammals (69 species) All placental mammals	<input type="checkbox"/>	40	0	0	29
Sauropsida (7 species) Birds and Reptiles	<input type="checkbox"/>	1	0	0	6
Fish (48 species) Ray-finned fishes	<input type="checkbox"/>	2	0	0	46
All (137 species) All species, including invertebrates	<input type="checkbox"/>	43	0	0	94

Selected orthologues [Hide](#)

Show [All](#) entries [Show/hide columns](#)

Species	Type	Orthologue	dN/dS	Target %id	Query %id	GOC Score	WGA Coverage	High Confidence
Bonobo (<i>Pan paniscus</i>)	1-to-1	HIST1H1A (ENSPPAG00000011952) View Gene Tree Compare Regions (6:26,508,481-26,509,128:-1) View Sequence Alignments	0.41600	99.07 %	99.07 %	50	100.00	Yes
Chimpanzee (<i>Pan troglodytes</i>)	1-to-1	HIST1H1A (EN) View Gene Tree Compare Regions (6:26,381,168-26,381,815:-1)	0.46154	98.60 %	98.60 %	50	100.00	Yes

[Orthologue Alignment](#)
[View Protein Alignment](#)
[View cDNA Alignment](#)

See organisms in a phylogenetic tree
(choose the node including the organisms that you want)

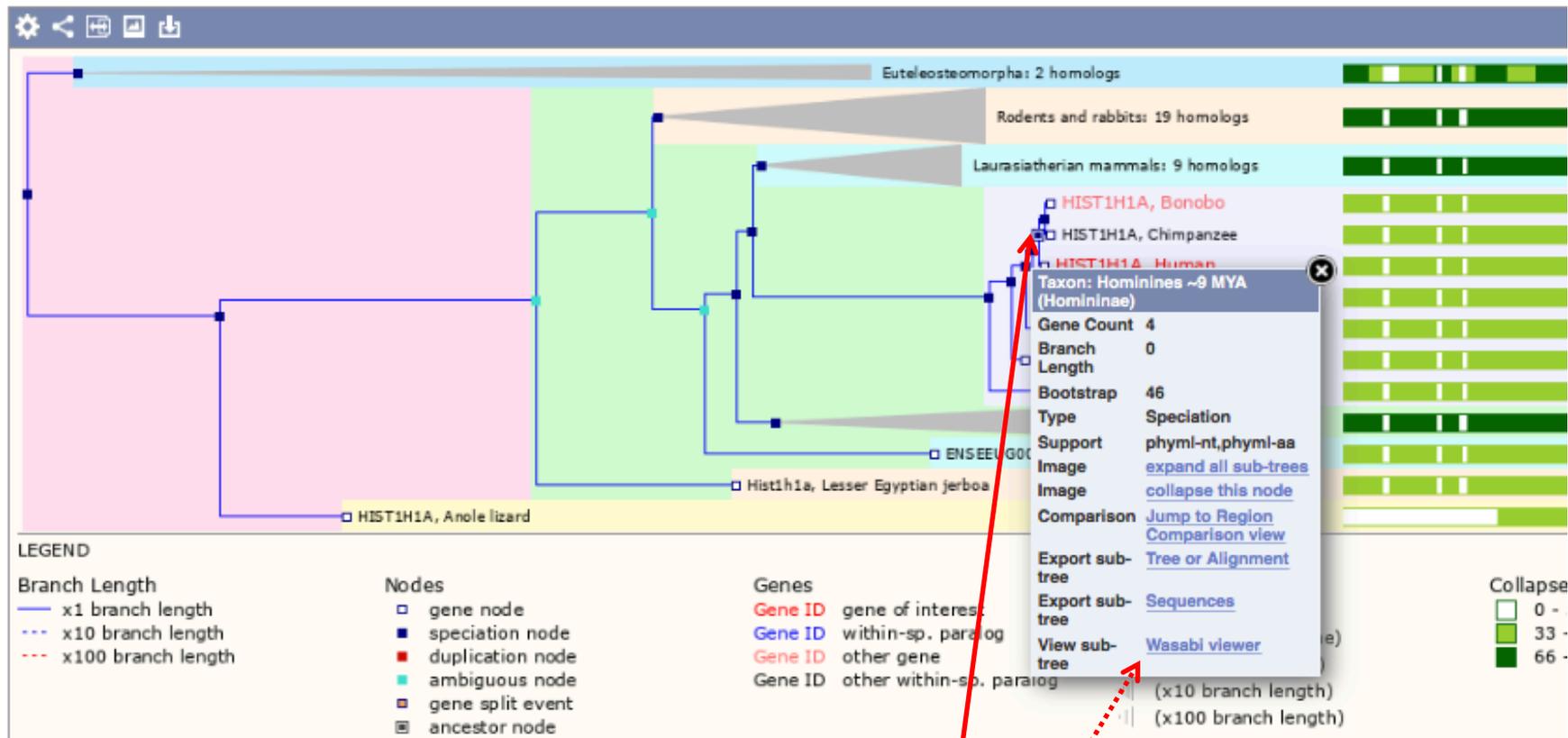
Align DNA/protein sequences

HIST1H1A Gene tree

Number of duplication nodes 0
 Number of ambiguous 3
 Number of gene split events 0

Highlighted genes

The *Human HIST1H1A* gene, its paralogues, its orthologue in *Bonobo*, and paralogues of the *Bonobo* gene, have all been highlighted. [Click here to disable highlighting.](#)



View options:

- [View current genes only](#) (Default)
- [View paralogues of current genes](#)
- [View all duplication nodes](#)
- [View fully expanded tree](#)
- Collapse all the nodes at the taxonomic rank

Click the node human and chimps are separated

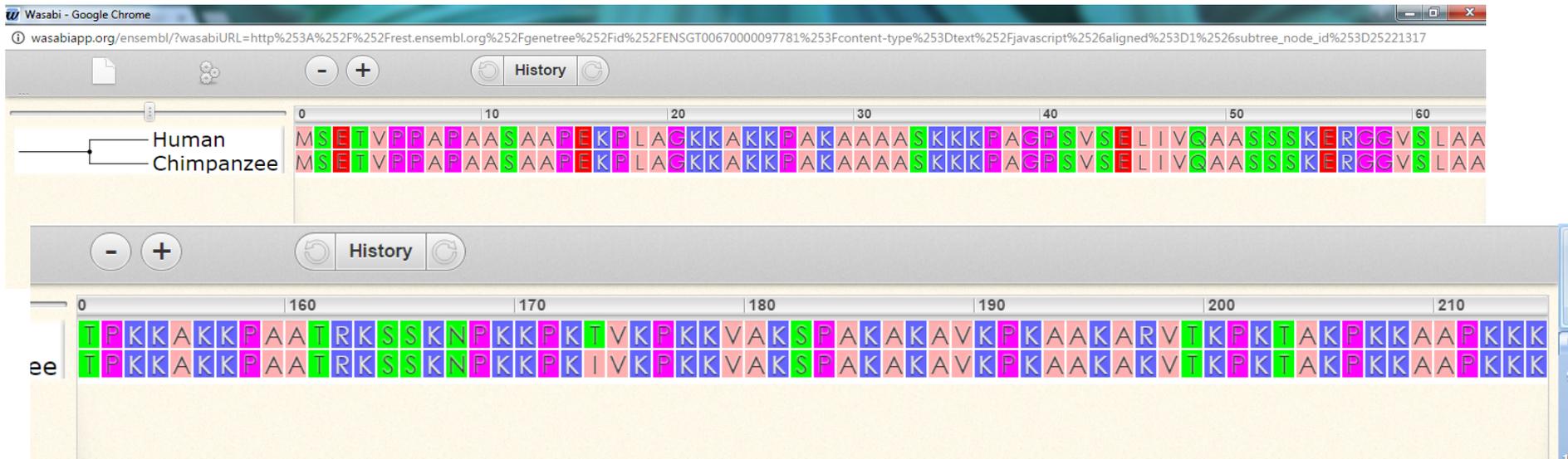
Wasabi shows you peptides

HIST1H1A human – chimp peptide sequence compared



Enlarge

Hold and scroll



HIST1H1A cDNA comparison

Orthologue alignment

Bu bilgilere dikkat ! Peptide length, % identity



Download homology

Type: 1-to-1 orthologues

Species	Gene ID	Peptide ID	Peptide length	% identity (cDNA)	% coverage	Genomic location
Human (<i>Homo sapiens</i>)	ENSG00000124610	ENSP00000244573	215 aa	99 %	100 %	6:26017085-26017732
Bonobo (<i>Pan paniscus</i>)	ENSPPAG00000011952	ENSPPAP00000002729	215 aa	99 %	100 %	6:26508481-26509128

CLUSTAL W (1.81) multiple sequence alignment

```
ENSP00000244573/1-645 ATGTCTGAAACAGTGCCTCCCGCCCCGCGCTTCTGCTGCTCCTGAGAAACCTTTAGCT
ENSPPAP00000002729/1-645 ATGTCTGAAACGGTGCCTCCCGCTCCCGCCGCTTCTGCTGCTCCTGAGAAACCTTTAGCT
*****
ENSP00000244573/1-645 GGCAAGAAGGCCAAGAAAACCTGCTAAGGCTGCAGCAGCCTCCAAGAAAAACCCGCTGGC
ENSPPAP00000002729/1-645 GGCAAGAAGGCCAAGAAAACCTGCTAAGGCTGCAGCAGCCTCCAAGAAAAACCCGCTGGC
*****
ENSP00000244573/1-645 CCTTCCGTGTCAGAGCTGATCGTGCAGGCTGCTTCCTCCTCTAAGGAGCGTGGTGGTGTG
ENSPPAP00000002729/1-645 CCTTCCGTGTCAGAGCTGATCGTGCAGGCTGCTTCCTCCTCTAAGGAGCGTGGTGGTGTG
*****
ENSP00000244573/1-645 TCGTTGGCAGCTCTTAAAAAGGCGCTGGCGGCCGCGAGGCTACGACGTGGAGAAGAACAAC
ENSPPAP00000002729/1-645 TCGTTGGCAGCTCTTAAAAAGGCGCTGGCGGCCGCGAGGCTACGACGTGGAGAAGAACAAC
*****
```

Scroll page down to see the rest



Gene-based displays

- Summary
 - Splice variants
 - Transcript comparison
 - Gene alleles
- Sequence
 - Secondary Structure
- Comparative Genomics
 - Genomic alignments
 - Gene tree
 - Gene gain/loss tree
 - Orthologues
 - Paralogues
 - Ensembl protein families
- Ontologies
 - GO: Molecular function
 - GO: Cellular component
 - GO: Biological process
- Phenotypes
- Genetic Variation
 - Variant table
 - Variant image
 - Structural variants
- Gene expression
- Pathway
- Regulation
- External references
- Supporting evidence
- ID History
 - Gene history

Gene: HBB ENSG00000244734

Description hemoglobin subunit beta [Source:HGNC Symbol;Acc:HGNC:4827 [rⒺ](#)]
Gene Synonyms CD113t-C, beta-globin
Location [Chromosome 11: 5,225,464-5,229,395](#) reverse strand.
GRCh38:CM000673.2

About this gene This gene has 6 transcripts ([splice variants](#)), [23 orthologues](#), [1 paralogue](#), is a member of [1 Ensembl protein family](#) and is associated with [28 phenotypes](#).

Transcripts

Show transcript table

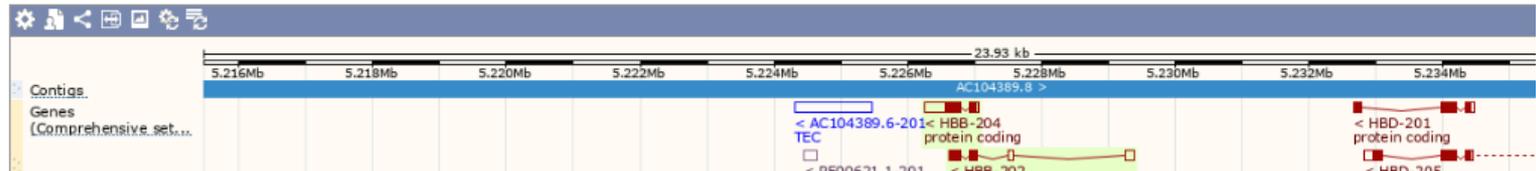


Summary [?](#)

Name [HBB](#) [rⒺ](#) (HGNC Symbol)
CCDS This gene is a member of the Human CCDS set: [CCDS7753.1](#) [rⒺ](#)
UniProtKB This gene has proteins that correspond to the following UniProtKB identifiers: [P68871](#) [rⒺ](#)
RefSeq Overlapping RefSeq annotation not matched
Ensembl version ENSG00000244734.4
Other assemblies This gene maps to [5,246,694-5,250,625](#) in GRCh37 coordinates.
View this locus in the GRCh37 archive: [ENSG00000244734](#) [rⒺ](#)
Gene type Protein coding
Annotation method Annotation for this gene includes both automatic annotation from Ensembl and [Havana](#) [rⒺ](#) manual curation, see [article](#).

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Hemoglobin subunit beta gene

Gene: HBB ENSG00000244734

Description hemoglobin subunit beta [Source:HGNC Symbol;Acc:[HGNC:4827](#)]

Gene Synonyms CD113t-C, beta-globin

Location [Chromosome 11: 5,225,464-5,229,395](#) reverse strand.
GRCh38:CM000673.2

About this gene This gene has 6 transcripts ([splice variants](#)), [23 orthologues](#), [1 paralogue](#), is a member of [1 Ensembl protein family](#) and is associated with

Transcripts [Hide transcript table](#)

Show/hide columns (1 hidden)		Filter						
Name	Transcript ID	bp	Protein	Biotype	CCDS	UniProt	RefSeq	Flags
HBB-206	ENST0000064709.1	754	147aa	Protein coding	CCDS7753.1	D9YZU5 P68871	-	GENCODE basic APPRIS P1
HBB-201	ENST00000335295.4	628	147aa	Protein coding	CCDS7753.1	D9YZU5 P68871	NM_000518 NP_000509	TSL:1 GENCODE basic APPRIS P1
HBB-204	ENST00000485743.1	680	111aa	Protein coding	-	A0A2R8Y7R2	-	TSL:1 GENCODE basic
HBB-202	ENST00000380315.2	502	90aa	Protein coding	-	F8W6P5	-	CDS 3' incomplete TSL:5
HBB-205	ENST00000633227.1	609	55aa	Nonsense mediated decay	-	A0A0J9YWK4	-	TSL:3
HBB-203	ENST00000475226.1	319	No protein	Retained intron	-	-	-	TSL:2

The longest transcript. Note the information: 147aa; 754 bp

Summary

Name [HBB](#) (HGNC Symbol)

CCDS This gene is a member of the Human CCDS set: [CCDS7753.1](#)

UniProtKB This gene has proteins that correspond to the following UniProtKB identifiers: [P68871](#)

RefSeq Overlapping RefSeq annotation not matched

Ensembl version ENSG00000244734.4

Other assemblies This gene maps to [5,246,694-5,250,625](#) in GRCh37 coordinates.
View this locus in the GRCh37 archive: [ENSG00000244734](#)

Gene type Protein coding

Annotation method Annotation for this gene includes both automatic annotation from Ensembl and [Havana](#) manual curation, see [article](#).

Hemoglobin beta altünitesi geni

Location: 11:5,225,464-5,229,395 Gene: HBB Transcript: HBB-206

Transcript-based displays

- Summary
- Sequence
 - Exons
 - cDNA
 - Protein
- Protein Information
 - Protein summary
 - Domains & features
 - Variants
- Genetic Variation
 - Variant table
 - Variant image
 - Haplotypes
 - Population comparison
 - Comparison image
- External References
 - General identifiers
 - Oligo probes
 - Supporting evidence
- ID History
 - Transcript history
 - Protein history

Transcript: HBB-206 ENST00000647020.1

Description hemoglobin subunit beta [Source:HGNC Symbol;Acc:HGNC:4827 [r#](#)]

Gene Synonyms CD113t-C, beta-globin

Location [Chromosome 11: 5,225,464-5,227,197](#) reverse strand.

About this transcript This transcript has [3 exons](#), is annotated with [12 domains and features](#), is associated with [2132 variations](#) and maps to [456 oligo probes](#).

Gene This transcript is a product of gene [ENSG00000244734](#) [Hide transcript table](#)

Name	Transcript ID	bp	Protein	Biotype	CCDS	UniProt	RefSeq	Flags
HBB-206	ENST00000647020.1	754	147aa	Protein coding	CCDS7753 r#	D9YZU5 r# P68871 r#	-	GENCODE basic APPRIS P1
HBB-201	ENST00000335295.4	628	147aa	Protein coding	CCDS7753 r#	D9YZU5 r# P68871 r#	NM_000518 r# NP_000509 r#	TSL:1 GENCODE basic APPRIS P1
HBB-204	ENST00000485743.1	680	111aa	Protein coding	-	A0A2R8Y7R2 r#	-	TSL:1 GENCODE basic
HBB-202	ENST00000380315.2	502	90aa	Protein coding	-	F8W6P5 r#	-	CDS 3' incomplete TSL:5
HBB-205	ENST00000633227.1	609	55aa	Nonsense mediated decay	-	A0A0J9YWK4 r#	-	TSL:3
HBB-203	ENST00000475226.1	319	No protein	Retained intron	-	-	-	TSL:2

Summary

Statistics Exons: 3, Coding exons: 3, Transcript length: 754 bps, Translation length: 147 residues

CCDS This transcript is a member of the Human CCDS set: [CCDS7753](#) [r#](#)

Uniprot This transcript corresponds to the following Uniprot identifiers: [P68871](#) [r#](#)

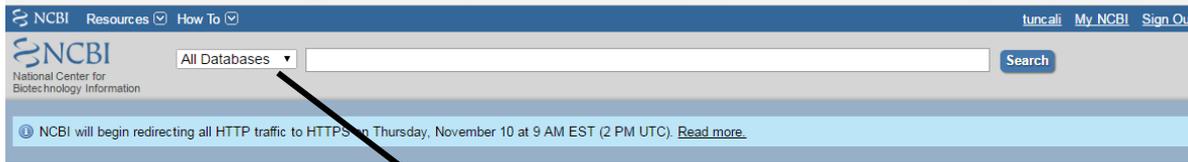
Click to see, cDNA, protein and the sequences

! 3 exons, 754 bp, 147 aa

Exons

Variation database

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



- NCBI Home
- Resource List (A-Z)
- All Resources
- Chemicals & Bioassays
- Data & Software
- DNA & RNA
- Domains & Structures
- Genes & Expression
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 - PubChem

SNP database

This image shows the NCBI homepage with the search dropdown menu open. The menu lists various databases, with "SNP" highlighted in blue. A black arrow points from the text "SNP database" to the "SNP" option in the dropdown. Another black arrow points from the text "Enter SNP reference number (rs xxxxx) or gene symbol" to the search input field. A third black arrow points from the text "Check proper gene name or symbol from HGNC or wikipedia if available." to the search input field. The background shows the same navigation and content as the previous image, but with the search dropdown menu expanded.

NCBI Home

Resource List (A-Z)

All Resources

Chemicals & Bioassays

Data & Software

DNA & RNA

Domains & Structures

Genes & Expression

Genetics & Medicine

Genomes & Maps

Homology

Literature

All Databases

- PMC
- PonSet
- Probe
- Protein
- Protein Clusters
- PubChem BioAssay
- PubChem Compound
- PubChem Substance
- PubMed
- PubMed Health
- SNP**
- Sparcle
- SRA
- Structure
- Taxonomy
- ToolKit
- ToolKitAll
- ToolKitBook
- ToolKitBookgh
- UniGene

Enter SNP reference number (rs xxxxx) or gene symbol

Check proper gene name or symbol from HGNC or wikipedia if available.

HGNC is responsible for approving unique symbols and names for human loci, including protein coding genes, ncRNA genes and pseudogenes, to allow unambiguous scientific communication.

genenames.org is a curated online repository of HGNC-approved gene nomenclature, gene families and associated resources including links to genomic, proteomic and phenotypic information.

Search our catalogue of more than 39,000 symbol reports using our improved search engine (see [Search help](#)), search lists of symbols using our [Multi-symbol checker](#) and identify possible orthologs using our [HCOP tool](#).

Download our ready-made data files from our [Statistics and Downloads](#) page, create your own datasets using either our [Custom Downloads](#) tool or [BioMart](#) service, or write a script/program utilising our [REST service](#).

Submit your [gene symbol and name proposals](#) to us to be accredited with HGNC approved nomenclature for use in publications, databases and presentations.

FAQ

- [What is the HGNC?](#)
- [What is HGNC-approved nomenclature and why do we need it?](#)
- [Where can I find information about existing human gene symbols?](#)
- [What is a stem symbol?](#)
- [Where can I find the Nomenclature Guidelines?](#)
- [Do I have to use the approved symbols?](#)
- [How should I cite HGNC nomenclature resources?](#)
- [Are there nomenclature committees for other species?](#)
- [Does the HGNC collaborate with specialist nomenclature committees and advisors?](#)
- [How should orthologs be identified?](#)
- [How should I refer to the protein encoded by a gene?](#)
- [Do alternative gene transcripts or splice variants have approved symbols?](#)

Latest News

[Announcing the Vertebrate Gene Nomenclature Committee](#)

The Vertebrate Gene Nomenclature Committee (VGNC) is an extension of the HGNC. VGNC is responsible for assigning standardized names to genes in vertebrate species that currently lack a [nomenclature committee](#). The 6 existing vertebrate nomenclature committees and VGNC coordinate to ensure genes are named in line with their human homologs. We also rely on the expertise of [specialist advisors](#) for naming within complex families.

[New features and changes](#)

Our new [BioMart](#) server using the latest BioMart (0.9.0) code has been released. Despite looking very different the new service retains an easy to use interface, include our gene family data in addition to the gene symbol report dataset, and allows our users to access and query a lot more of the HGNC data than was previously possible. See our [BioMart help](#) for more information.

[More...](#)

Get the name or symbol of the gene you are searching for

Check for SNP entry for insulin gene. First get the gene symbol or name

The image shows the top navigation bar of the HGNC website. On the left is the HGNC logo (HUGO Gene Nomenclature Committee). In the center is a search bar with the text 'insulin' and a magnifying glass icon. To the right of the search bar is a note: 'Use * to search with a root symbol (eg ZNF*)'. Below the search bar is a horizontal menu with links: Home, Downloads, Gene Families, Tools, Useful Links, About, Newsletters, Contact Us, Help, VGNC, and a yellow 'Request Symbol' button.

Search

Document type

Gene	63
Family	19
Site	0

Items per page

10 20 50 100 200

Results: 1 to 10 of 90

Page 1 of 9 Next > Last >>

Insulin like growth factor binding proteins

Document type: Family HGNC Family ID: 1065 Root symbol: IGFBP
Matches: Family name: Insulin like growth factor binding proteins
Gene name: insulin like growth factor binding protein 1
Previous gene name: insulin-like growth factor binding protein 1

Endogenous ligands

Document type: Family HGNC Family ID: 542
Matches: Gene name: insulin like growth factor 1
Previous gene name: insulin-like growth factor 1 (somatomedin C)

Relaxin family peptide receptors

Document type: Family HGNC Family ID: 217 Root symbol: RXFP
Matches: Gene name: relaxin/insulin like family peptide receptor 1

CD molecules

Document type: Family HGNC Family ID: 471 Root symbol: CD
Matches: Gene name: insulin like growth factor 1 receptor
Previous gene name: insulin-dependent diabetes mellitus 12

Fibronectin type III domain containing

Document type: Family HGNC Family ID: 555
Matches: Gene name: insulin like growth factor 1 receptor
Previous gene name: insulin-like growth factor 1 receptor

Receptor Tyrosine Kinases

Document type: Family HGNC Family ID: 321
Matches: Gene name: insulin like growth factor 1 receptor
Previous gene name: insulin-like growth factor 1 receptor

RNA binding motif containing

90 entries. 9 pages

Gene search
63 genes.



Symbol Search

Locus groups & types

Protein-coding gene	46
Non-coding RNA	1
- RNA, long non-coding	1
Phenotype	15
Pseudogene	1
- Pseudogene	1

Items per page

10
 20
 50
 100
 200

Results: 1 to 10 of 63

Page of 7 [Next >](#) [Last >>](#)

- IDDM3:** insulin dependent diabetes mellitus 3
 HGNC_ID: HGNC:5374 **Location:** 15q26 **Locus type:** phenotype only
 Matches: Approved name: **insulin** dependent diabetes mellitus 3
- IDDM4:** insulin dependent diabetes mellitus 4
 HGNC_ID: HGNC:5375 **Location:** 11q13 **Locus type:** phenotype only
 Matches: Approved name: **insulin** dependent diabetes mellitus 4
- IDDM6:** insulin dependent diabetes mellitus 6
 HGNC_ID: HGNC:5377 **Location:** 18q21 **Locus type:** phenotype only
 Matches: Approved name: **insulin** dependent diabetes mellitus 6
- IDDM7:** insulin dependent diabetes mellitus 7
 HGNC_ID: HGNC:5378 **Location:** 2q31 **Locus type:** phenotype only
 Matches: Approved name: **insulin** dependent diabetes mellitus 7
- IDDM8:** insulin dependent diabetes mellitus 8

Select protein coding genes.
Decreased to 46.

IGFBP6: insulin like growth factor binding protein 6

HGNC_ID: HGNC:5475 **Location:** 12q13.13 **Locus type:** gene with protein product

Matches: Approved name: insulin like growth factor binding protein 6
Gene family: Insulin like growth factor binding proteins
Previous symbols & names: insulin-like growth factor binding protein 6

IGFBP7: insulin like growth factor binding protein 7

HGNC_ID: HGNC:5476 **Location:** 4q12 **Locus type:** gene with protein product

Matches: Approved name: insulin like growth factor binding protein 7
Previous symbols & names: insulin-like growth factor binding protein 7

IGFBPL1: insulin like growth factor binding protein like 1

HGNC_ID: HGNC:20081 **Location:** 9p13.1 **Locus type:** gene with protein product

Matches: Approved name: insulin like growth factor binding protein like 1
Previous symbols & names: insulin-like growth factor binding protein-like 1

INS: insulin

HGNC_ID: HGNC:6081 **Location:** 11p15.5 **Locus type:** gene with protein product

Matches: Approved name: insulin
Previous symbols & names: insulin-dependent diabetes mellitus 2

INSIG1: insulin induced gene 1

HGNC_ID: HGNC:6083 **Location:** 7q36.3 **Locus type:** gene with protein product

Matches: Approved name: insulin induced gene 1

INSIG2: insulin induced gene 2

HGNC_ID: HGNC:20452 **Location:** 2q14.1-q14.2 **Locus type:** gene with protein product

Matches: Approved name: insulin induced gene 2

INSL3: insulin like 3

HGNC_ID: HGNC:6086 **Location:** 19p13.11 **Locus type:** gene with protein product

Matches: Approved name: insulin like 3
Previous symbols & names: insulin-like 3 (Leydig cell)

INSL4: insulin like 4

HGNC_ID: HGNC:6087 **Location:** 9p24.1 **Locus type:** gene with protein product

Matches: Approved name: insulin like 4
Previous symbols & names: insulin-like 4 (placenta)

INSL5: insulin like 5

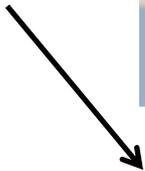
HGNC_ID: HGNC:6088 **Location:** 1p31.3 **Locus type:** gene with protein product

Scroll down to find insulin.

Symbol "INS"

Go back to SNP database.

Select Homo sapiens



- Organism
 - Bos taurus
 - Homo sapiens
 - Customize ...
- Variation Class
 - in del
 - snp
- Clinical Significance
 - benign
 - likely benign
 - pathogenic
 - uncertain significance
 - untested
- Annotation
 - Cited in PubMed
 - OMIM
 - PubMed
 - nucleotide
 - protein
 - structure
- Function Class
 - 3' splice site
 - 3' utr
 - 5' splice site
 - 5' utr
 - coding synonymous
 - frame shift
 - intron
 - missense
 - nonsense
 - stop gained

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

Send to: Filters: Manage

Search results

Items: 1 to 20 of 4162

<< First < Prev Page 1 of 209 Next > Last >>

Total SNPs =4162

- rs5505 [Homo sapiens]
1.
CCCTGCCTGTCTCCCAGATCACTGT[C/T]CTTCTGCCATGGCCCTGTGGATGCG
Chromosome: 11:2160980
Gene: INS-IGF2 (GeneView) INS (GeneView)
Functional Consequence: nc transcript variant,utr variant 5 prime
Allele Origin: T(germline)/C(germline)
Clinical significance: Benign
Validated: by 1000G,by cluster,by frequency
Global MAF: A=0.0058/29
HGVS: NC_000011.10:g.2160980G>A, NC_000011.9:g.2182210G>A, NG_007114.1:g.5215C>T, NM_000207.2:c.-9C>T, NM_001042376.2:c.-9C>T, NM_001185097.1:c.-9C>T, NM_001185098.1:c.-9C>T, NM_001291897.1:c.-9C>T, NR_003512.3:n.51C>T
[Varview](#)
- rs5507 [Homo sapiens]
2.
CCGCTGTTCCGGAACCTGCTCTGCG[C/T]GGCACGTCTGGCAGTGGGGCAGGT
Chromosome: 11:2160013
Gene: INS-IGF2 (GeneView) INS (GeneView)
Functional Consequence: intron variant
Allele Origin: T(germline)/C(germline)
Clinical significance: Benign
Validated: by 1000G,by cluster,by frequency
Global MAF: A=0.0082/41
HGVS: NC_000011.10:g.2160013G>A, NC_000011.9:g.2181243G>A, NG_007114.1:g.6182C>T, NM_000207.2:c.188-16C>T, NM_001042376.2:c.187+772C>T, NM_001185097.1:c.188-

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Search

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INS (4162)

INS AND (44)

INS AND (pathogenic

Homo sapi variant 1, r

insulin AN

NCBI Resources How To

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Organism: clear Display Settings: Summary, 20 per page, Sorted by SNP_ID Send to:

Bos taurus
 Homo sapiens
 Customize ...

Variation Class
 in del
 snp

Clinical Significance
 benign
 likely benign
 pathogenic
 uncertain significance
 untested

Annotation
 Cited in PubMed
 OMIM
 PubMed
 nucleotide
 protein
 structure

Function Class
 3' splice site
 3' utr
 5' utr
 coding synonymous
 intron
 missense

Global MAF
 Custom range...

Validation Status
 by-1000 Genomes
 by-2hit-2allele
 by-cluster
 by-frequency
 no-info

Search results
 Items: 1 to 20 of 428

Filters activated: Homo sapiens. Clear all to show 4162 items.

rs5505 [Homo sapiens]
 1.
 CCCTGCCTGTCTCCCAGATCACTGT [C/T] CTCTGCCATGGCCCTGTGGATGCG
 Chromosome: 11:2160980
 Gene: INS-IGF2 (GeneView) INS (GeneView)
 Functional Consequence: nc transcript variant,utr variant 5 prime
 Allele Origin: T(germline)/C(germline)
 Clinical significance: Benign
 Validated: by 1000G,by cluster,by frequency
 Global MAF: A=0.0058/29
 HGVS: NC_000011.10:g.2160980G>A, NC_000011.9:g.2182210G>A, NG_007114.1:g.5215C>T, NM_000207.2:c.-9C>T, NM_001042376.2:c.-9C>T, NM_001185097.1:c.-9C>T, NM_001185098.1:c.-9C>T, NM_001291897.1:c.-9C>T, NR_003512.3:n.51C>T
[Varview](#)

rs5507 [Homo sapiens]
 2.
 CCGCTGTTCCGGAACCTGCTCTGCG [C/T] GGCACGTCCTGGCAGTGGGGCAGGT
 Chromosome: 11:2160013
 Gene: INS-IGF2 (GeneView) INS (GeneView)
 Functional Consequence: intron variant
 Allele Origin: T(germline)/C(germline)
 Clinical significance: Benign
 Validated: by 1000G,by cluster,by frequency
 Global MAF: A=0.0082/41
 HGVS: NC_000011.10:g.2160013G>A, NC_000011.9:g.2181243G>A, NG_007114.1:g.6182C>T, NM_000207.2:c.188-16C>T, NM_001042376.2:c.187+772C>T, NM_001185097.1:c.188-16C>T, NM_001185098.1:c.188-16C>T, NM_001291897.1:c.188-16C>T, NR_003512.3:n.246+772C>T, XM_005252899.1:c.-2215C>T
[Varview](#)

rs28933985 [Homo sapiens]

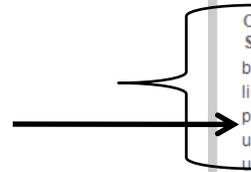
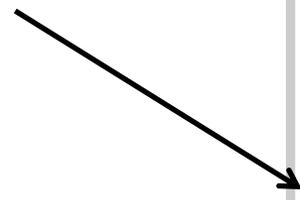
Select variation type SNPs (there are other types as well)

Select pathogenic SNPs

Select mutation type

As missense seçelim

SNP numbers get decreased



NCBI will begin redirecting all HTTP traffic to HTTPS on Thursday, November 10 at 9 AM EST (2 PM UTC). [Read more.](#)

Organism Display Settings: Send to:

Homo sapiens
[Customize ...](#)

Variation Class
snp

Search results

Items: 1 to 20 of 41

<< First < Prev Page 1 of 3 Next > Last >>

Clinical Significance

i Filters activated: Homo sapiens, pathogenic, missense. [Clear all to show 4162 items.](#)

pathogenic

rs28933985 [*Homo sapiens*]

1.

Annotation

Cited in PubMed
 OMIM
 PubMed
 nucleotide
 protein
 structure

GCCCTGGAGGGGTCCTGCAGAAGC[A/C/G/T]TGGCATTGTGGAAACAATGCTGTACC

Chromosome: 11:2159919
Gene: INS-IGF2 (GeneView) INS (GeneView)

Functional Consequence: intron variant,missense
Allele Origin: G(germline)/T(germline)/A(germline)/C(germline)
Clinical significance: Pathogenic
Validated: by cluster

Function Class

3' utr
 coding synonymous
 intron

HGVS: NC_000011.10:g.2159919C>A, NC_000011.10:g.2159919C>G,
NC_000011.10:g.2159919C>T, NC_000011.9:g.2181149C>A,
NC_000011.9:g.2181149C>G, NC_000011.9:g.2181149C>T, NG_007114.1:g.6276G>A,
NG_007114.1:g.6276G>C, NG_007114.1:g.6276G>T, NM_000207.2:c.266G>A,
NM_000207.2:c.266G>C, NM_000207.2:c.266G>T, NM_001042376.2:c.187+866G>A,
NM_001042376.2:c.187+866G>C, NM_001042376.2:c.187+866G>T,
NM_001185097.1:c.266G>A, NM_001185097.1:c.266G>C, NM_001185097.1:c.266G>T,
NM_001185098.1:c.266G>A, NM_001185098.1:c.266G>C, NM_001185098.1:c.266G>T,
NM_001291897.1:c.266G>A, NM_001291897.1:c.266G>C, NM_001291897.1:c.266G>T,
NP_000198.1:p.Arg89His, NP_000198.1:p.Arg89Leu, NP_000198.1:p.Arg89Pro,
NP_001172026.1:p.Arg89His, NP_001172026.1:p.Arg89Leu,
NP_001172026.1:p.Arg89Pro, NP_001172027.1:p.Arg89His,
NP_001172027.1:p.Arg89Leu, NP_001172027.1:p.Arg89Pro,
NP_001278826.1:p.Arg89His, NP_001278826.1:p.Arg89Leu,
NP_001278826.1:p.Arg89Pro, NR_003512.3:n.246+866G>A,
NR_003512.3:n.246+866G>C, NR_003512.3:n.246+866G>T,
XM_005252899.1:c.-2121G>A, XM_005252899.1:c.-2121G>C,
XM_005252899.1:c.-2121G>T

Global MAF

Validation Status
 by-cluster
 by-frequency
 no-info

[Clear all](#)

[Show additional filters](#)

[Varview](#) [Protein3D](#)

rs80356663 [*Homo sapiens*]

2.

CTCTGGGGACCTGACCCAGCCGACG[A/C/T]CTTTGTGAACCAACACCTGTGCGGC

Chromosome: 11:2160001

Human insulin gene missense mutations that are pathogenic.

Click for detailed information On a certain SNP

Known SNP reference number can be searched

HBB gene sickle cell anemia SNP: **rs334**

NCBI Resources How To

dbSNP SNP rs334 Create alert Advanced

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Organism: Homo sapiens
Variation Class: snp
Clinical Significance: pathogenic
Annotation: Cited in PubMed, OMIM, PubMed, nucleotide, protein, structure
Function Class: missense
Global MAF: Custom range...

Display Settings: Summary, Sorted by SNP_ID

Send to: Filters: Manage Filter

Find related data
Database: Select
Find items

Search details
rs334[All Fields:
[Organism] AND
pathogenic[Clini:
missense[Funcio

Recent activity

Search results
Items: 4

Filters activated. Homo sapiens, pathogenic, missense. Clear all to show 4 items.

rs334 [Homo sapiens]
1.

GACACCACTGGTGCATCTGACTCCTG[A/C/G/T]GGAGAAGTCTGCCGTTACTGCCCTG
Chromosome: 11:5227002
Gene: HBB (GeneView)
Functional Consequence: missense
Allele Origin: G(germline)/T(germline)/A(germline)/C(germline)
Clinical significance: Pathogenic
Validated: by 1000G,by cluster,by frequency,by hapmap
Global MAF: A=0.0274/137
HGVS: NC_000011.10:g.5227002T>A, NC_000011.10:g.5227002T>C,
NC_000011.10:g.5227002T>G, NC_000011.9:g.5248232T>A,
NC_000011.9:g.5248232T>C, NC_000011.9:g.5248232T>G, NG_000007.3:g.70614A>C,
NG_000007.3:g.70614A>G, NG_000007.3:g.70614A>T, NM_000518.4:c.20A>C,

Click for details



dbSNP Short Genetic Variations

Example: rs268

Reference SNP (rs) Report ALPHA



[Switch to classic site](#)

rs334

Current Build 151
Released July 17, 2018

FEEDBACK

Organism *Homo sapiens*

Position chr11:5227002 (GRCh38.p7)

Alleles T>A / T>C / T>G

Variation Type SNV Single Nucleotide Variation

Frequency
A=0.00342 (842/245928, GnomAD)
A=0.00438 (532/121340, ExAC)
A=0.0116 (360/30968, GnomAD) [\(+ 1 more\)](#)

Clinical Significance Reported in [ClinVar](#)

Gene : Consequence HBB : Missense Variant

Publications 99 citations

Genomic View [See rs on genome](#)

Variant Details

Clinical Significance

Frequency

Aliases

Submissions

History

Publications

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Study	Population	Group	Sample Size	Ref Allele	Alt Allele
The Genome Aggregation Database	Global	Study-wide	245928	T=0.99658	A=0.00342
The Genome Aggregation Database	European	Sub	133746	T=0.99996	A=0.00004
The Genome Aggregation Database	Asian	Sub	48000	T=0.9996	A=0.0004
The Genome Aggregation Database	American	Sub	33558	T=0.9978	A=0.0022
The Genome Aggregation Database	African	Sub	15302	T=0.9522	A=0.0478
The Genome Aggregation Database	Ashkenazi Jewish	Sub	9840	T=1.000	A=0.000
The Genome Aggregation Database	Other	Sub	5482	T=0.998	A=0.002
The Exome Aggregation Consortium	Global	Study-wide	121340	T=0.99562	A=0.00438

Human genome GRCh38.p13 assembly:

Number of protein coding genes?

Number of non coding genes?

Chromosomes with highest number of protein coding genes?

Chromosomes with lowest number of protein coding genes?

Chromosomes with highest number of non coding genes?

Chromosomes with highest number of pseudogenes?

Human X chromosome synteny regions count on chimp, gorilla and orangutan.

Longest transcript of hemoglobin B gene codes for how many aminoacids?

What is the percent homology of human and chimp beta hemoglobin gene product?

Longest transcript of hemoglobin A1 gene in humans?

Human hemoglobin A1 ve A2 identity?

NCBI SNP database:

Check for the total number of SNPs for hemoglobin beta gene?

Mutation causing sickle cell anemia is represented by SNP code rs334

Determine major and minor allele frequencies