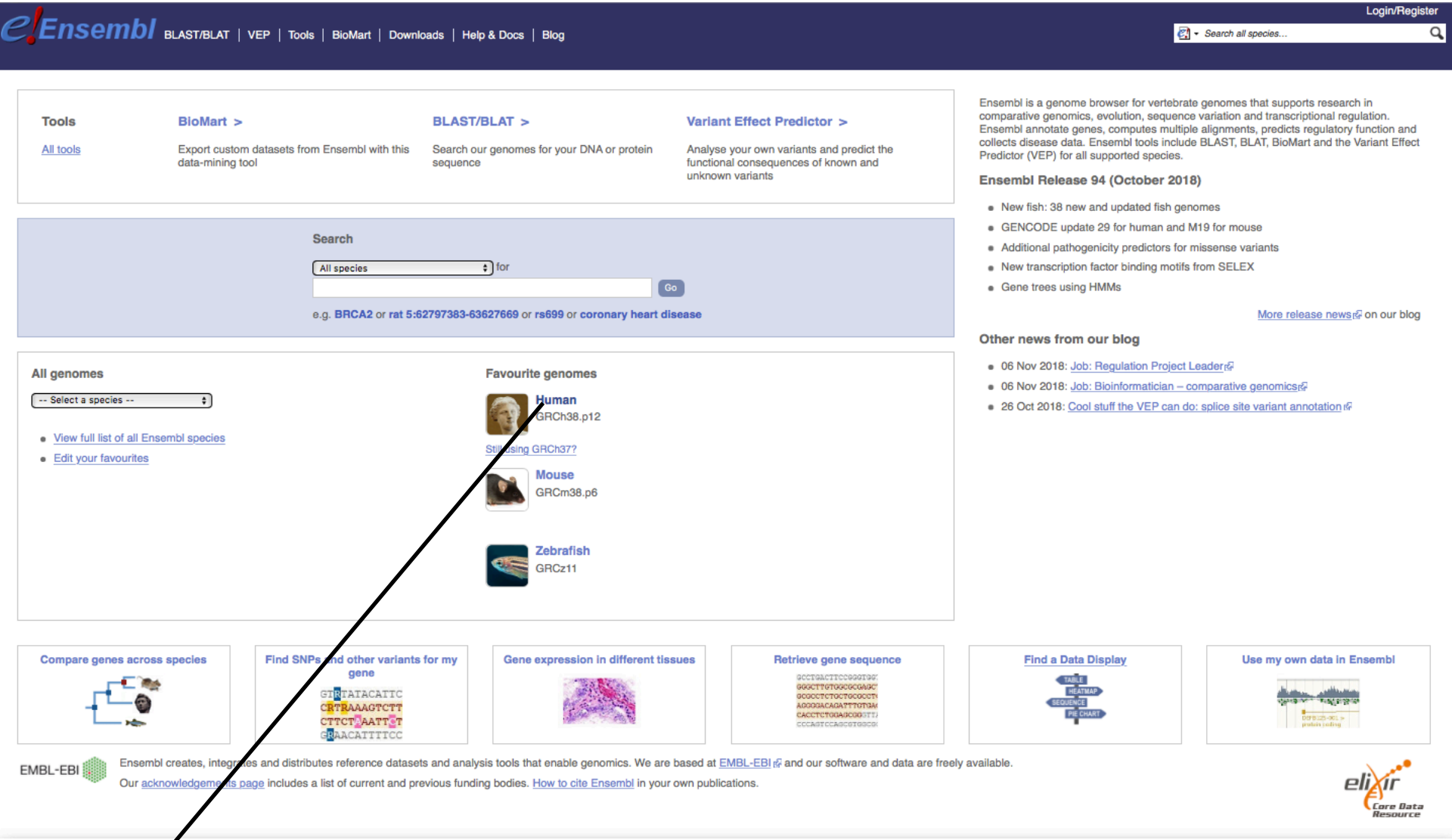
ONLINE GENOME DATABASES

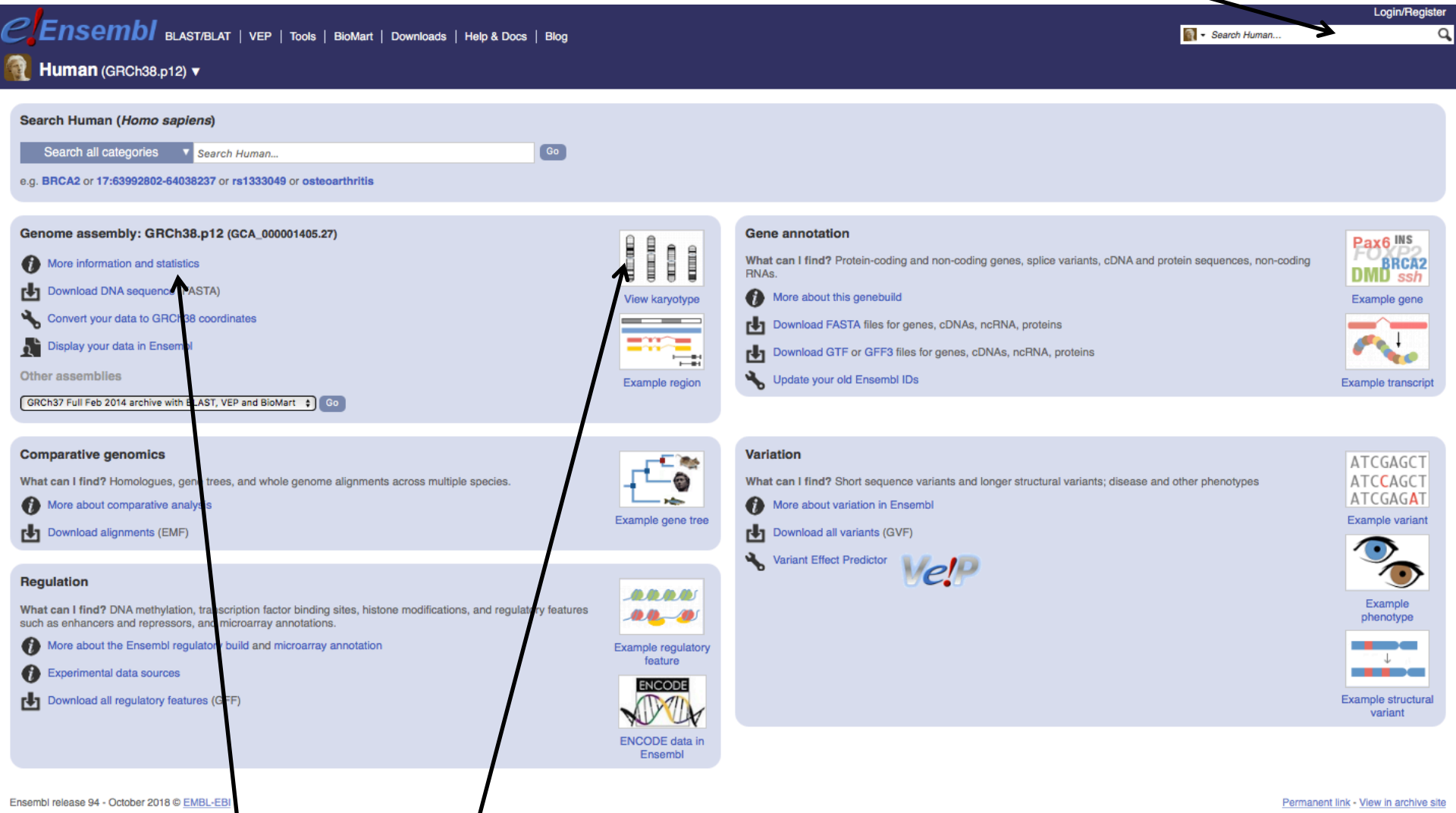
AÜTF D1M1, 2019

www.ensemble.org



Ensemble's latest human genome "assembly"

İnsan genomunda arama penceresi



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

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

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)

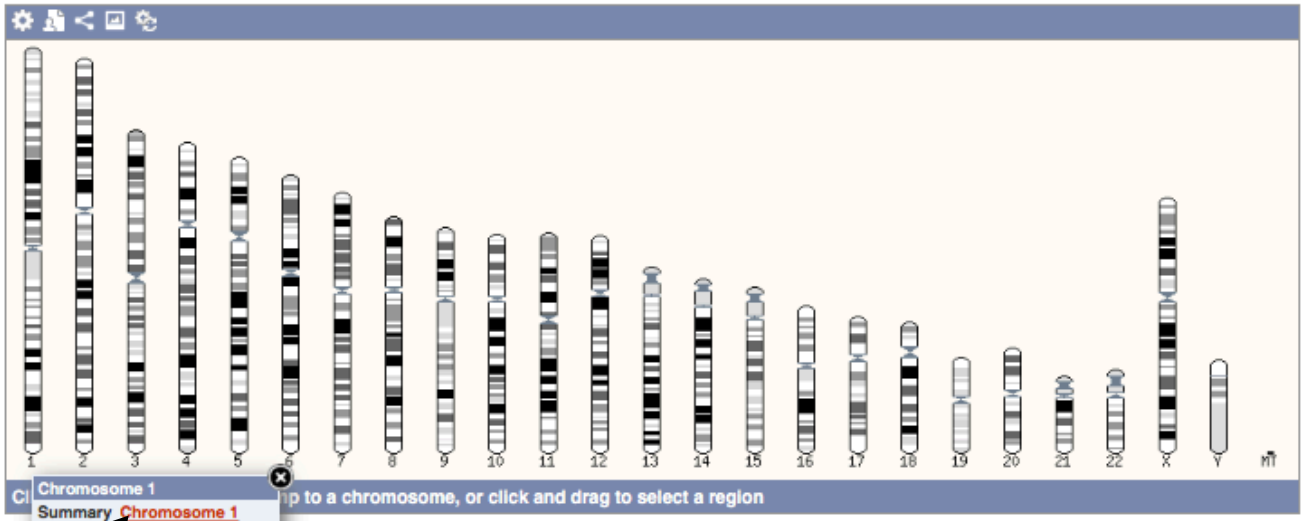
Coding genes	3,054 (incl 44 readthrough)
Non coding genes	1,555
Small non coding genes	297
Long non coding genes	1,071 (incl 45 readthrough)
Misc non coding genes	187
Pseudogenes	1,798
Gene transcripts	21,636

Other

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

İnsan karyotip görüntüsü

Add features



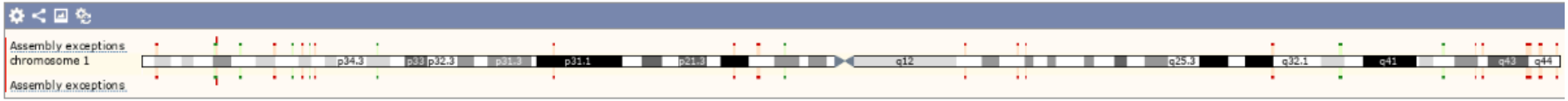
Assembly	GRCh38.p12 (Genome Reference Consortium Human Build 38), INSDC Assembl
Base Pairs	3,609,003,417
Golden Path Length	3,096,649,726
Annotation provider	Ensembl
Annotation method	Full genebuild
Genebuild started	Jan 2014
Genebuild released	Jul 2014
Genebuild last updated/patched	Jul 2018
Database 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

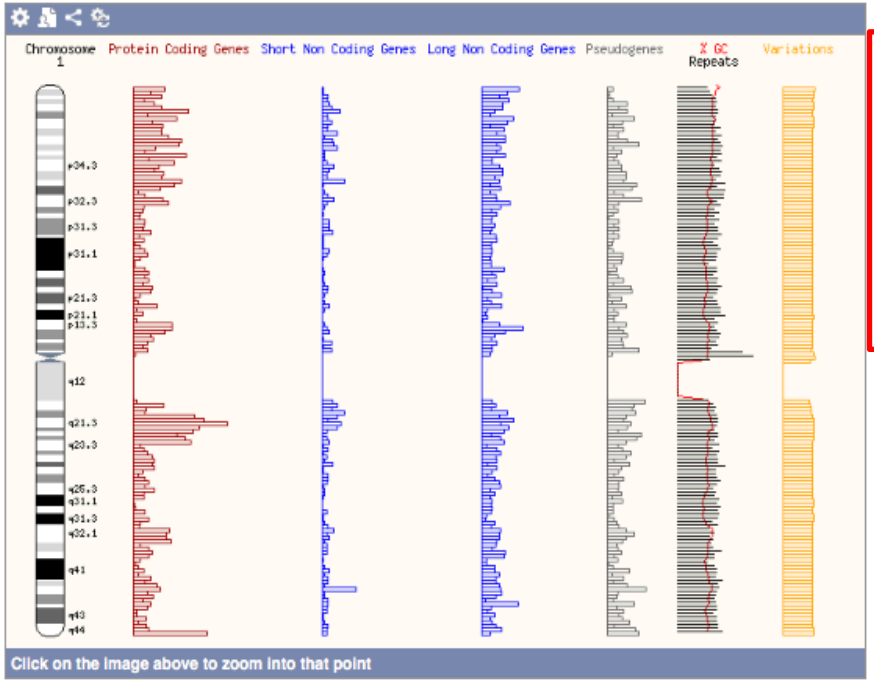
Click for further information

Chromosome 1: 1-1



Chromosome summary

Add features



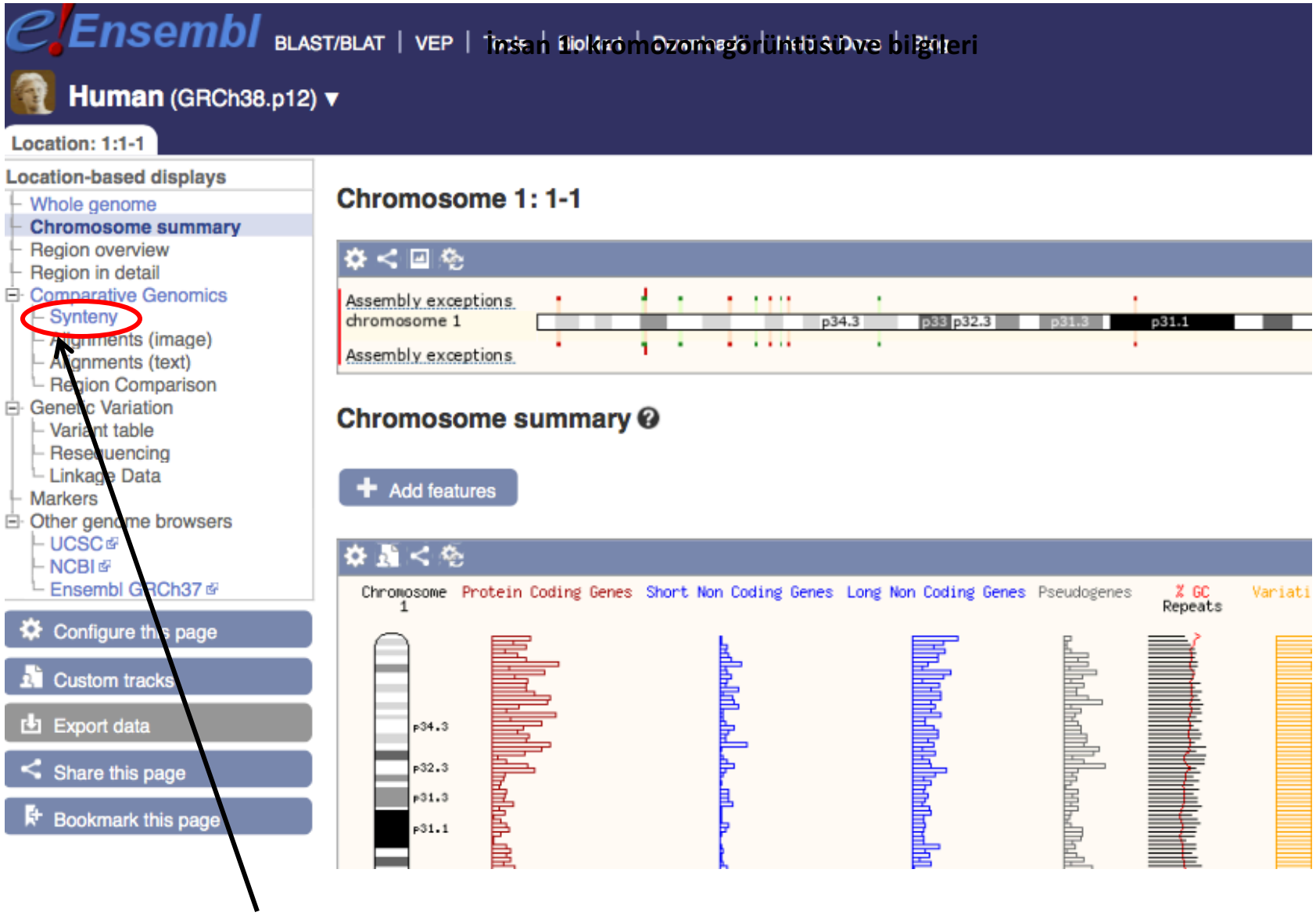
Diğer kromozomları seçin

Change chromosome: 1 Go

Chromosome Statistics

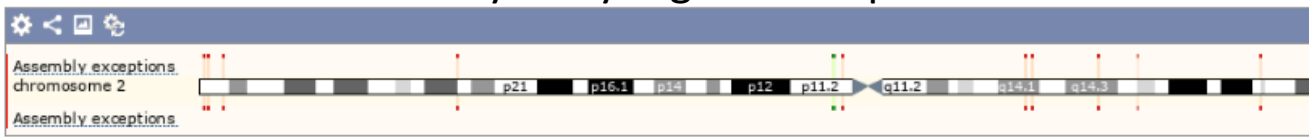
Length (bps)	248,956,422
Coding genes	2,057 (incl. 49 readthrough)
Non coding genes	2,088
Small non coding genes	482
Long non coding genes	1,414 (incl. 14 readthrough)
Misc non coding genes	192
Pseudogenes	1,293
Short Variants	52,130,200

Protein coding genes
Non coding genes (RNA genes)
Pseudogenes



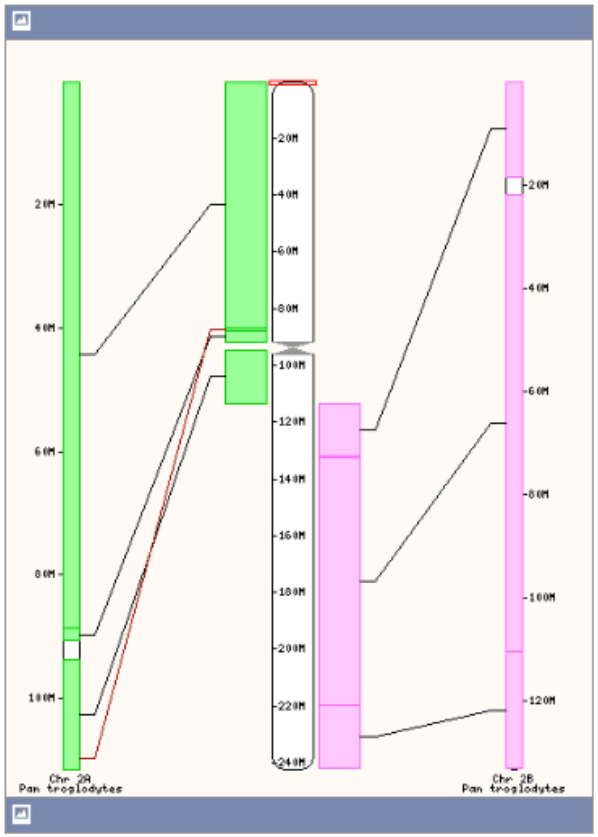
Synteny regions for comparison between organisms' chromosomes

Synteny regions compared



Synteny

Synteny between Human chromosome 2 and Chimpanzee



Change Species: Chimpanzee Go

Change chromosome: 2 Go

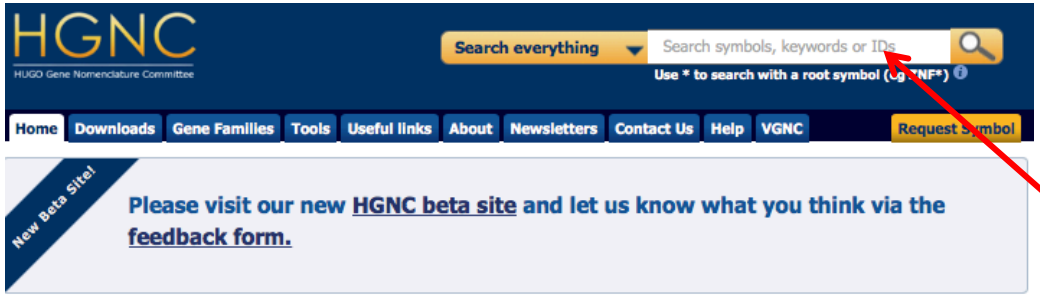
Select organism

Select chromosome

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

www.genenames.org

HGNC (Human Genome Organization Gene Nomenclature Committee)



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

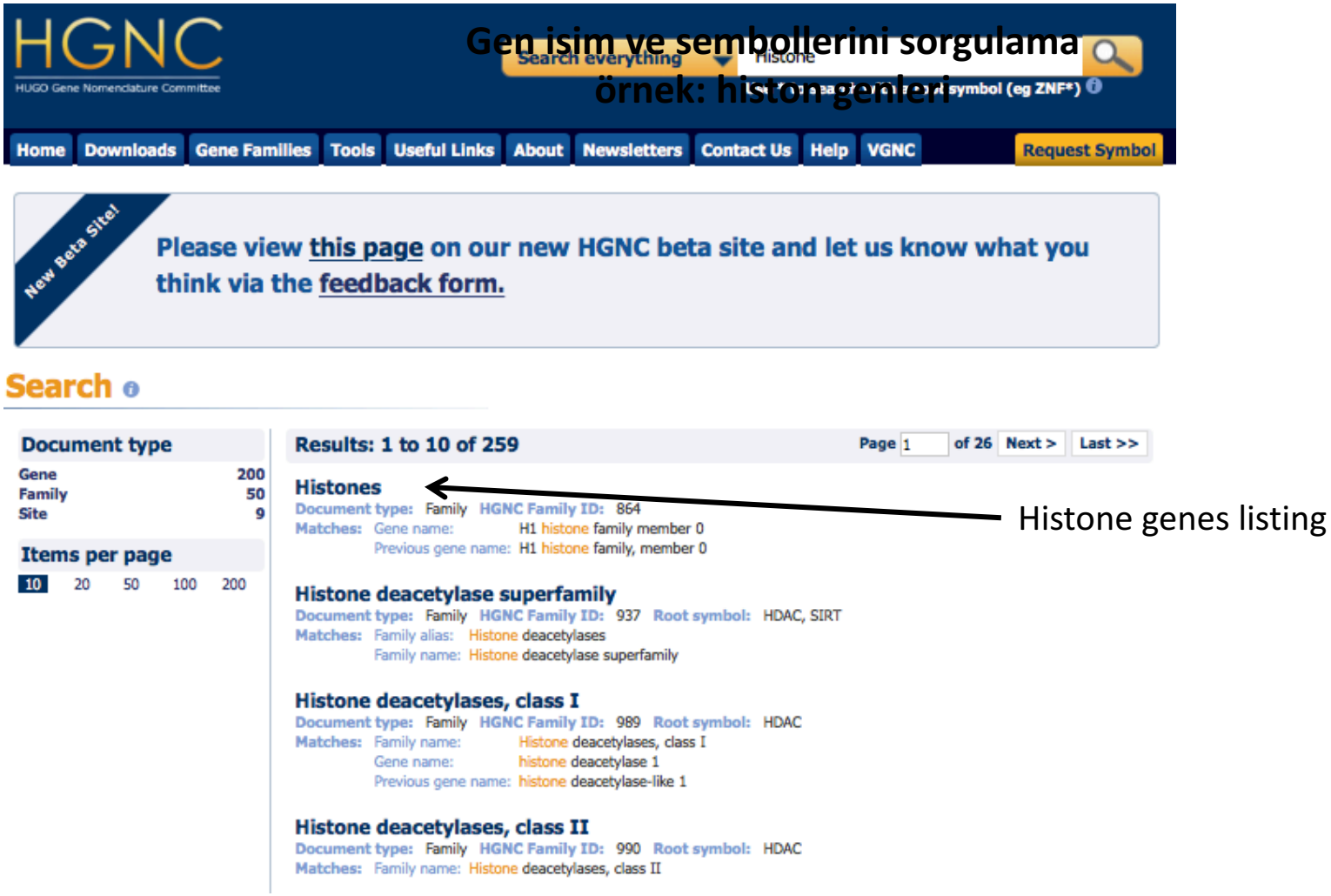
New species data available in HCOP

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

Gen isim ve sembollerini sorgulama
örnek: histon genleri



Histone genes listing

www.genenames.org

Histon genleri

HGNC
HUGO Gene Nomenclature Committee

Search everything | Search symbols, keywords or IDs
Use * to search with a root symbol (eg ZNF*)
Press F11 to exit full screen

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 H1F0 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		dJ34B20.16, FLJ39701, 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		H2AFALii, dJ86C11.1, H2A/S	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, H2AFSP	6p22.2
HIST1H2APS2	histone cluster 1 H2A pseudogene 2	H2AFTP	dJ139G21.2, H2A/T	6p22.2
HIST1H2APS3	histone cluster 1 H2A pseudogene 3		dJ34B20.8, H2AFUP	6p22.2
HIST1H2APS4	histone cluster 1 H2A pseudogene 4	H2AFK, H2AFKP	pH2A/k, dJ34B20.2	6p22.2

Given below enlarged
Histone cluster 1
H1 gene family member a

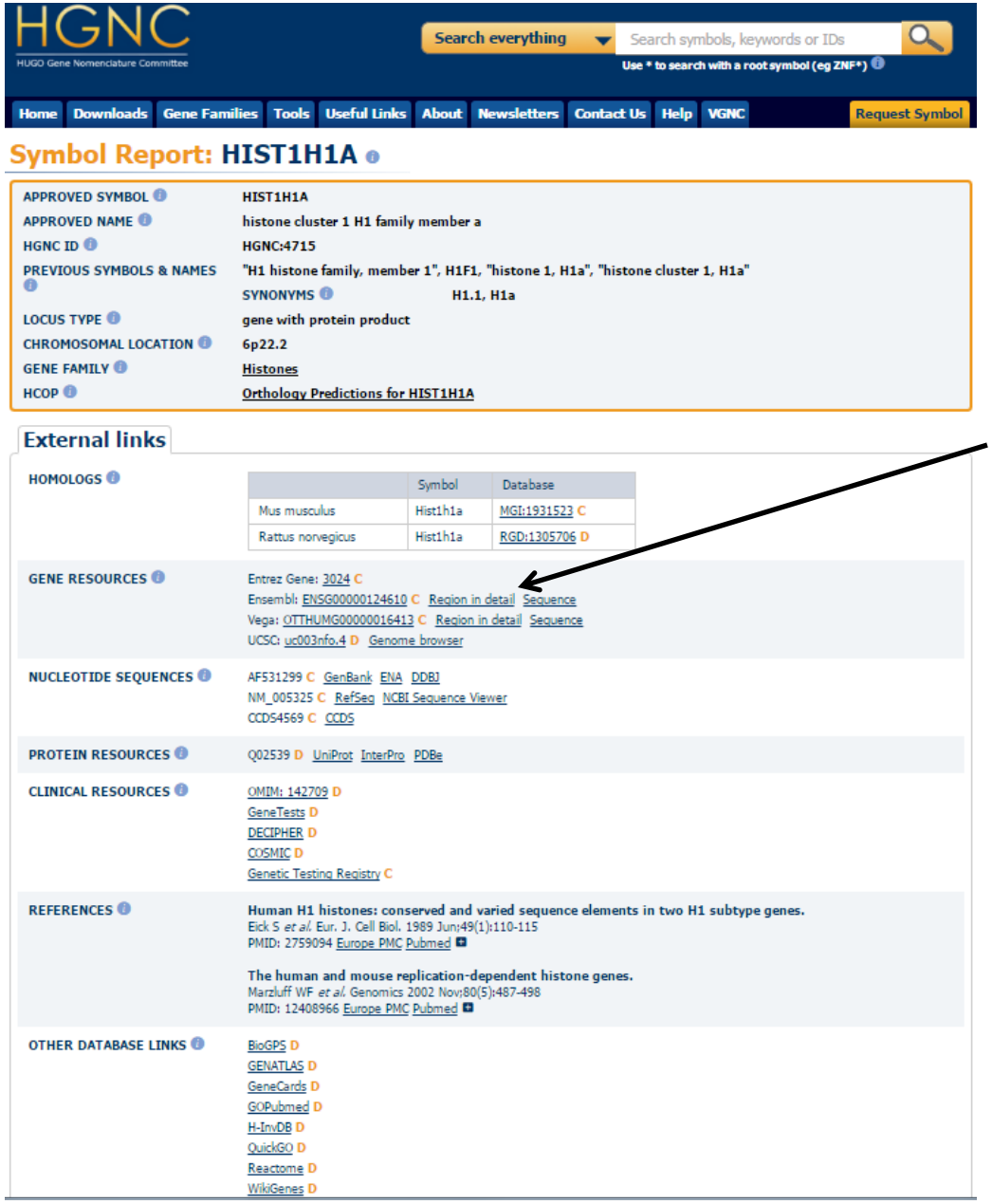
Symbol: HIST1H1A
Kromozom lokusu: 6p22.2

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

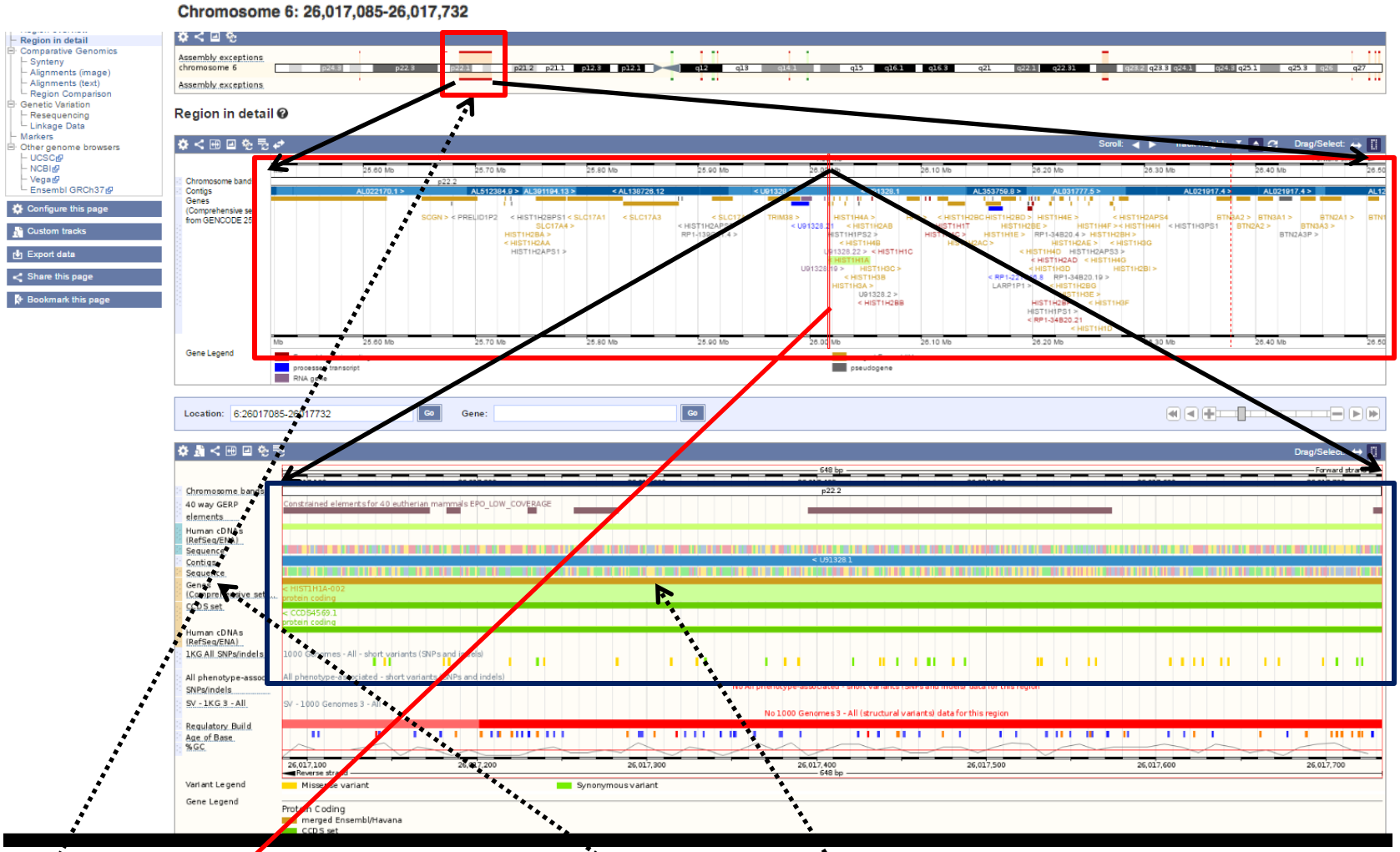
HIST1H1A



Click to go to Ensemble link

Click sequence

HIST1H1A



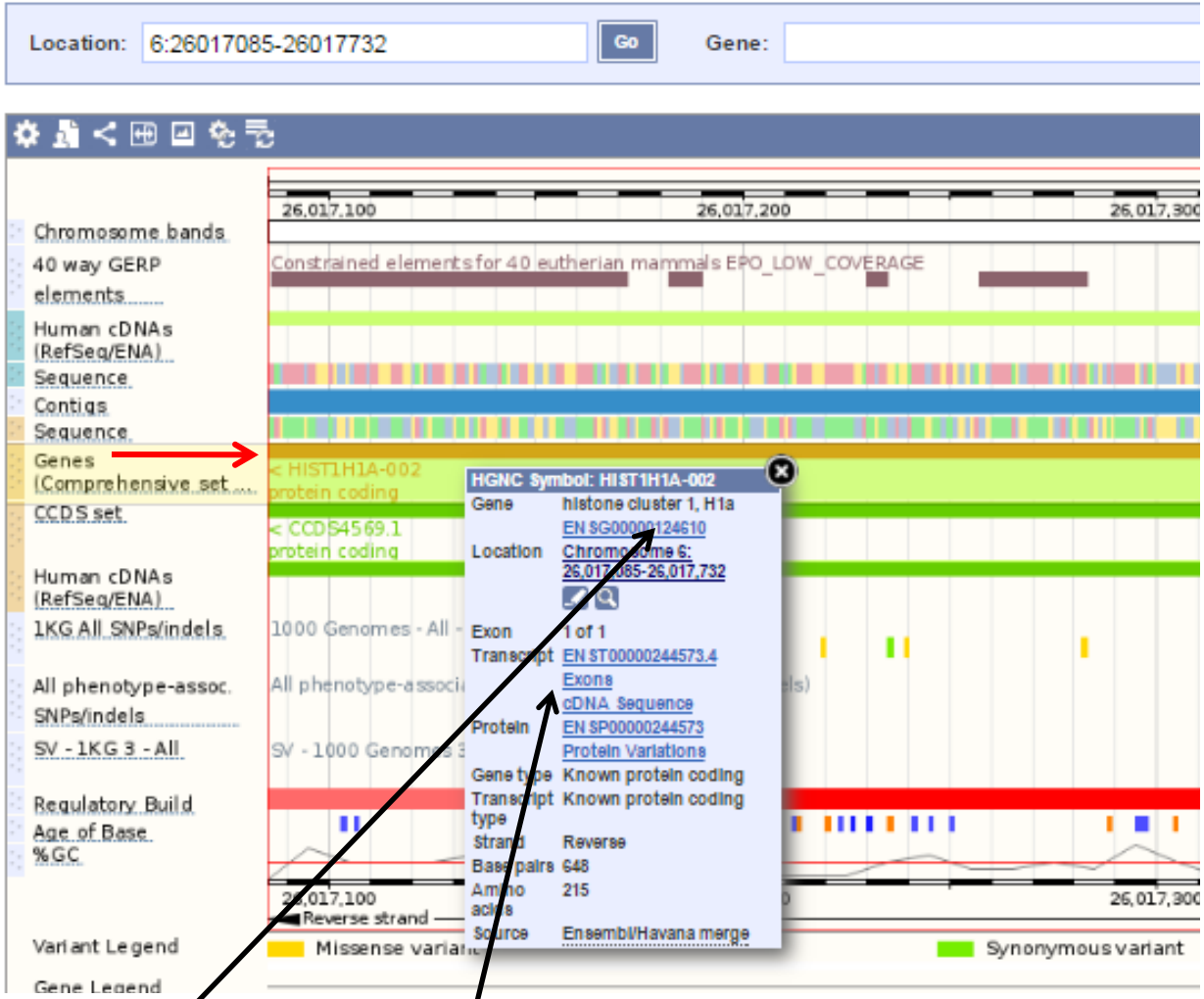
HIST1H1A gene sequence is reached by clicking the gene row

Chromosome location in between red bars

The below window in red rectangle shows the region in detail with neighboring sequneces/genes

Red thin line is enlarged to show the gene of interest in detail.

HIST1H1A geni haritası



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

Configure this page

Custom tracks

Export data

Share this page

Bookmark this page

Gene: HIST1H1A ENSG00000124610

43 orthologues

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

Show/hide columns (1 hidden) Filter

Name	Transcript ID	bp	Protein	Biotype	CCDS	UniProt	RefSeq	Flags
HIST1H1A-201	ENST00000244573.4	648	215aa	Protein coding	CCDS4569	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

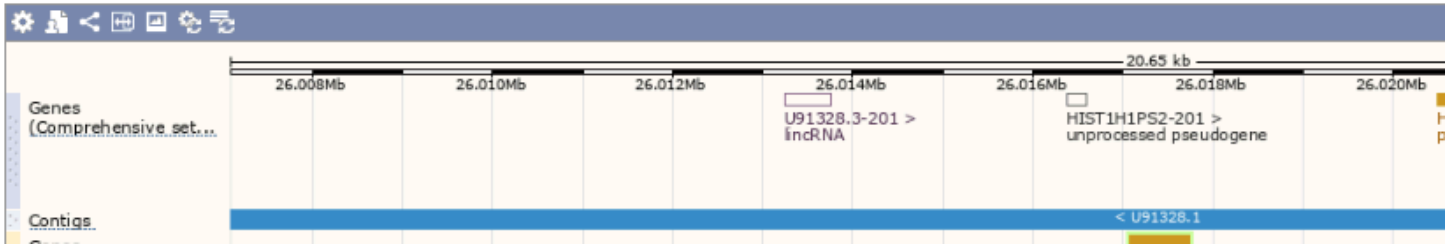
Ensembl version ENSG00000124610.4

Other assemblies This gene maps to 26,017,313-26,017,960 in GRCh37 coordinates.
View this locus in the GRCh37 archive: ENSG00000124610

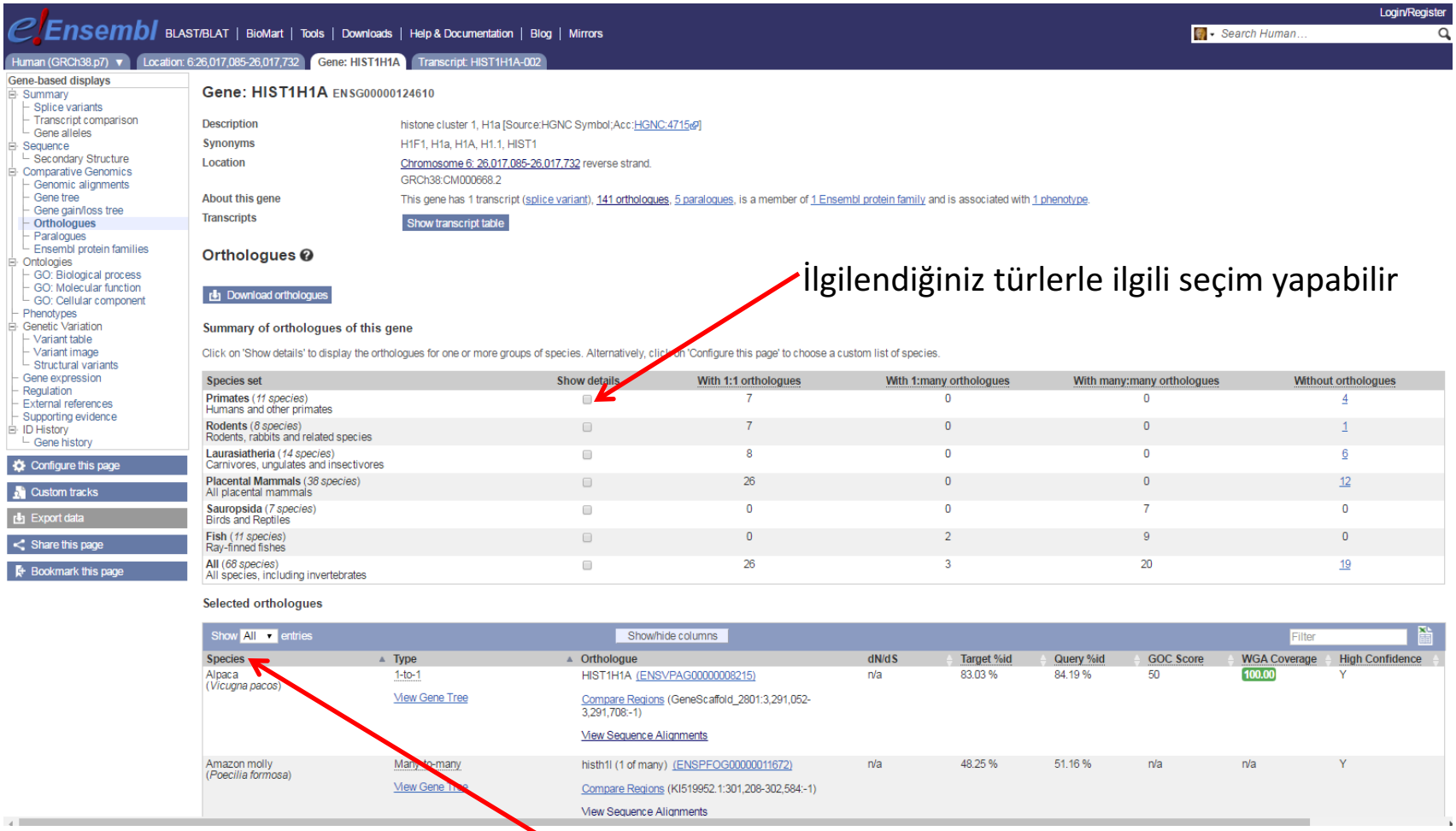
Gene type Protein coding

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



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	☑	6	0	0	17
Rodents and related species (*24 species*) Rodents, lagomorphs and tree shrews	☐	20	0	0	4
Laurasiatheria (*17 species*) Carnivores, ungulates and insectivores	☐	10	0	0	7
Placental Mammals (*69 species*) All placental mammals	☐	40	0	0	29
Sauropsida (*7 species*) Birds and Reptiles	☐	1	0	0	6
Fish (*48 species*) Ray-finned fishes	☐	2	0	0	46
All (*137 species*) All species, including invertebrates	☐	43	0	0	94

Selected orthologues Hide

Show All entries | Show/hide columns | Filter

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

Orthologue Aligment
View Protein Alignment
View cDNA Alignment

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

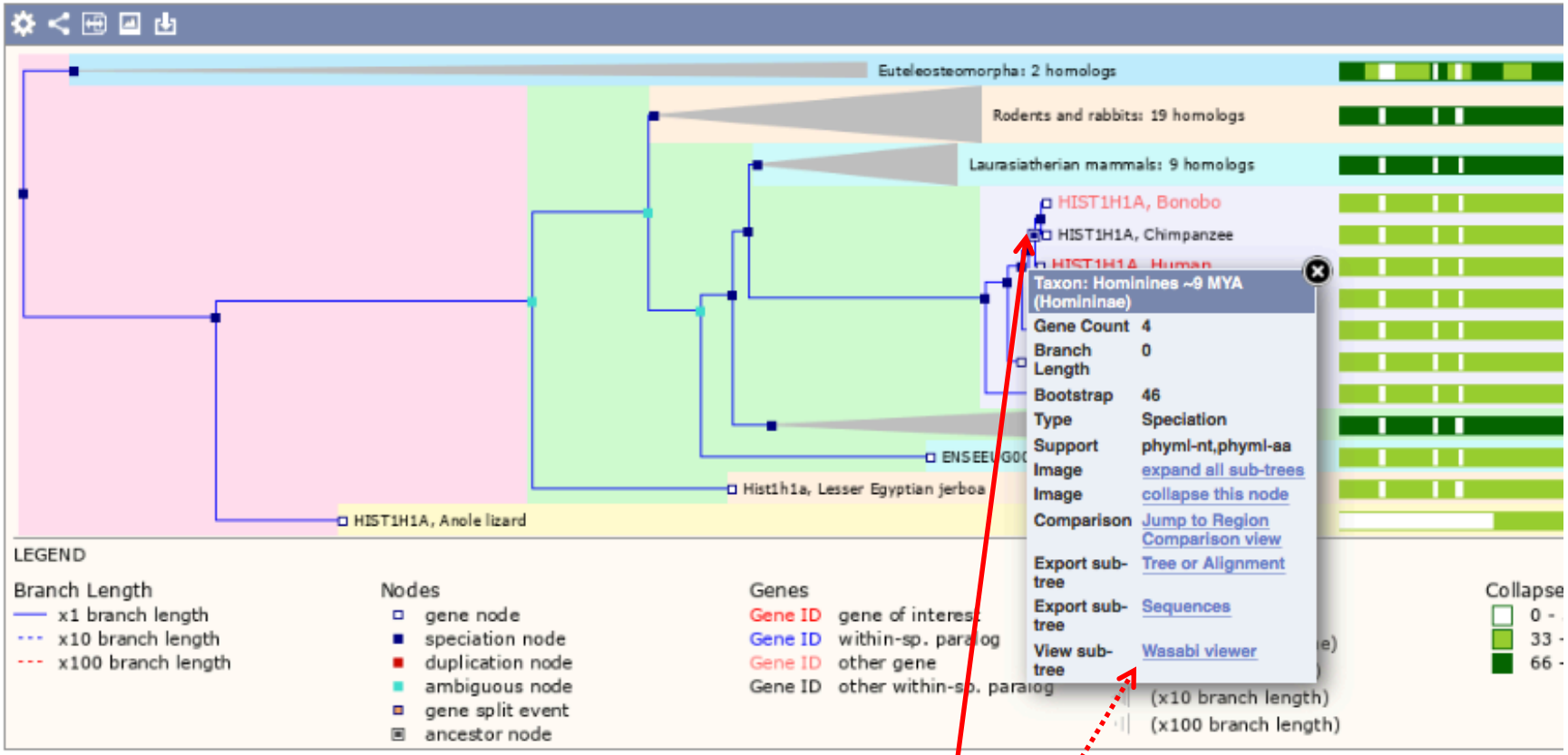
Align DNA/protein sequences

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

HIST1H1A Gene tree

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.



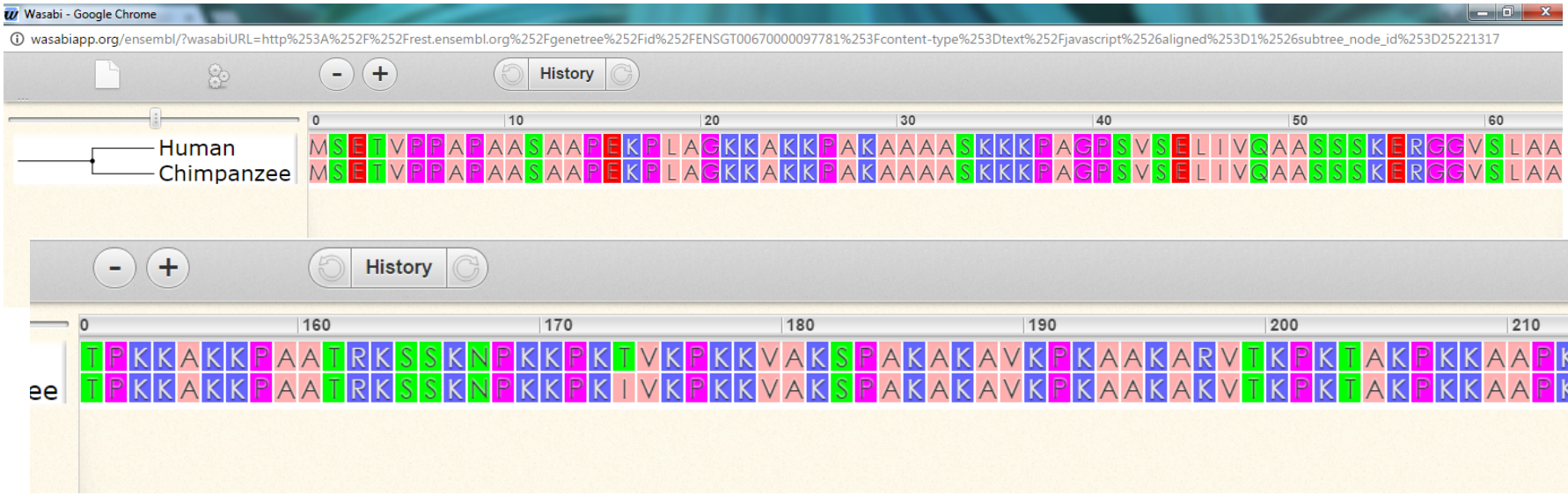
View options:

- View current genes only (Default)
- View paralogs of current genes
- View all duplication nodes
- View fully expanded tree
- Collapse all the nodes at the taxonomic rank -- Select a rank--

Click the node human and chimps are seperated

Wasabi shows you peptides

HIST1H1A human – chimp peptide sequence compared



***HIST1H1A* insan – şempanze** DNA/protein dizilerini hizalamak (diğer bir seçenek)

Sauropsida (*7 species*) Birds and Reptiles	☐	0	0	7	0
Fish (*11 species*) Ray-finned fishes	☐	0	2	9	0
All (*68 species*) All species, including invertebrates	☐	26	3	20	19

Selected orthologues

Show All entries | Show/hide columns | Filter

Species	Type	Orthologue	dN/dS	Target %id	Query %id	GOC Score	WGA Coverage	High Confidence
Chimpanzee (*Pan troglodytes*)	1-to-1 View Gene Tree	HIST1H1A (ENSPTRG00000017796) Compare Regions (6:26,281,104-26,281,751:-1) View Sequence Alignments	0.46154	98.60 %	98.60 %	50	100.00	Y
Gibbon (*Nomascus leucogenys*)	1-to-1 View Gene Tree	HIST1H1A (ENSNLEG[illegible] Compare Regions (GL397291.1:17,127,417-17,128,055:-1) View Sequence Alignments		[illegible] %	95.81 %	50	100.00	Y
Mouse Lemur	1-to-1	HIST1H1A (ENSMICG00000035620)	0.14752	82.33 %	82.33 %	n/a	100.00	Y

Orthologue Aligment
View Protein Alignment
View cDNA Alignment

Click first

Select cDNA or protein in the popup window

HIST1H1A cDNA comparison

Orthologue alignment

Download homology

Bu bilgilere dikkat ! Peptide length, % identity

Type: 1-to-1 orthologues

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

```
CLUSTAL W (1.81) multiple sequence alignment

ENSP00000244573/1-645     ATGTCTGAAACAGTGCCTCCCGCCCCCGCCGCTTCTGCTGCTCCTGAGAAACCTTTAGCT
ENSPPAP00000002729/1-645  ATGTCTGAAACGGTGCCTCCCGCTCCCGCCGCTTCTGCTGCTCCTGAGAAACCTTTAGCT
                          ********** *********** ***********************************

ENSP00000244573/1-645     GGCAAGAAGGCAAAGAAACCTGCTAAGGCTGCAGCAGCCTCCAAGAAAAAACCCGCTGGC
ENSPPAP00000002729/1-645  GGCAAGAAGGCAAAGAAACCTGCTAAGGCTGCAGCAGCCTCCAAGAAAAAACCCGCTGGC
                          ************************************************************

ENSP00000244573/1-645     CCTTCCGTGTCAGAGCTGATCGTGCAGGCTGCTTCCTCCTCTAAGGAGCGTGGTGGTGTG
ENSPPAP00000002729/1-645  CCTTCCGTGTCAGAGCTGATCGTGCAGGCTGCTTCCTCCTCTAAGGAGCGTGGTGGTGTG
                          ************************************************************

ENSP00000244573/1-645     TCGTTGGCAGCTCTTAAAAAGGCGCTGGCGGCCGCAGGCTACGACGTGGAGAAGAACAAC
ENSPPAP00000002729/1-645  TCGTTGGCAGCTCTTAAAAAGGCGCTGGCGGCCGCAGGCTACGACGTGGAGAAGAACAAC
                          ************************************************************
```

Scroll page down to see the rest

Searching genes in Ensemble

Homo sapiens - Ensembl

www.ensembl.org/Homo_sapiens/Info/Index

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

Login/Register

Search Human…

Human (GRCh38.p7)

Human

Homo sapiens

Search all categories | Search Human… | Go

e.g. BRCA2 or 17:63973115-64437414 or rs1333049 or osteoarthritis

What's New in Human release 86

- Human: updated cDNA alignments
- Human: updated RefSeq gene import
- External database references update

More news…

Hemoglobin beta gene

Check HGNC for proper name or symbol

Hemoglobin subunit beta; *HBB. Enter* HBB in search area

Click

Current selection:

< all Species

Only searching Human

Restrict category to:

Gene	13
Transcript	38
Phenotype	1
Somatic Mutation	80
GeneTree	1
GenomicAlignment	8
ProbeFeature	49
Protein Family	1
Variant	2306

Per page: 10 25 50 100

Layout: Standard Table

Tip:

Help and Documentation can be searched from the homepage! Just type in a term you want to know more about, like non-synonymous SNP.

Only searching Human | HBB

2497 results match **HBB** when restricted to species: Human

HBB (Human Gene)
ENSG00000244734 11:5225464-5229395:-1
Hemoglobin subunit beta [Source:HGNC Symbol;Acc:HGNC:4827]

HEMOGLOBIN--BETA LOCUS; *HBB* [*141900] (MIM gene record; description: HEMOGLOBIN--BETA LOCUS; *HBB*,) is an external reference matched to Gene ENSG00000244734

Variant table • Phenotypes • Location • External Refs. • Regulation • Orthologues • Gene tree

HBB-201 (Human Transcript)
ENST00000335295 11:5225464-5227071:-1
Hemoglobin subunit beta [Source:HGNC Symbol;Acc:HGNC:4827].
Location • External Refs. • cDNA seq. • Exons • Variant table • Protein seq. • Population • Protein summary

HBB-203 (Human Transcript)
ENST00000475226 11:5225655-5226823:-1
Hemoglobin subunit beta [Source:HGNC Symbol;Acc:HGNC:4827].
Location • External Refs. • cDNA seq. • Exons • Variant table • Population

HBB-204 (Human Transcript)
ENST00000485743 11:5226263-5227072:-1
Hemoglobin subunit beta [Source:HGNC Symbol;Acc:HGNC:4827].
Location • External Refs. • cDNA seq. • Exons • Variant table • Protein seq. • Population • Protein summary

HBB-205 (Human Transcript)
ENST00000633227 11:5225467-5227071:-1
Hemoglobin subunit beta [Source:HGNC Symbol;Acc:HGNC:4827].
Location • External Refs. • cDNA seq. • Exons • Variant table • Protein seq. • Population • Protein summary

HBB-202 (Human Transcript)
ENST00000380315 11:5226620-5229395:-1
Hemoglobin subunit beta [Source:HGNC Symbol;Acc:HGNC:4827].
Location • External Refs. • cDNA seq. • Exons • Variant table • Protein seq. • Population • Protein summary

HBB-206 (Human Transcript)
ENST00000647020 11:5225464-5227197:-1
Hemoglobin subunit beta [Source:HGNC Symbol;Acc:HGNC:4827].
Location • External Refs. • cDNA seq. • Exons • Variant table • Protein seq. • Population • Protein summary

HBB-Related Disorders (Human Phenotype)

Best gene match

Human Gene

HBB

HGNC Symbol; Acc:HGNC:4827

Human

< HBB 10kb 11:5225464-5229395:-1

Protein coding gene

hemoglobin subunit beta

Suggestions

hba hbe hbi heb hub hbub1 inhbb haab habc habh habp hbda hbii hboa ha hac had hae haf hag hai hak hal ham hao hap has hat hav haw hay hca hce hci he hec hed heg hei hek hel hem hen hep her hes het hev hex hey

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

Human (GRCh38.p12) ▾

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

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

Configure this page

Custom tracks

Export data

Share this page

Bookmark this page

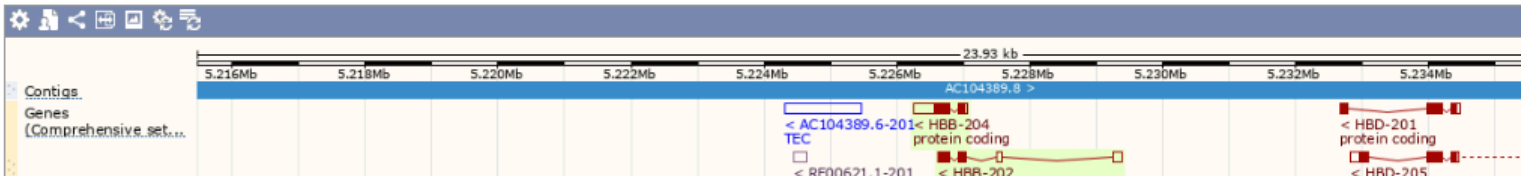
Gene: HBB ENSG00000244734

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 28 phenotypes.
Transcripts	Show transcript table ← Click

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.

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



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

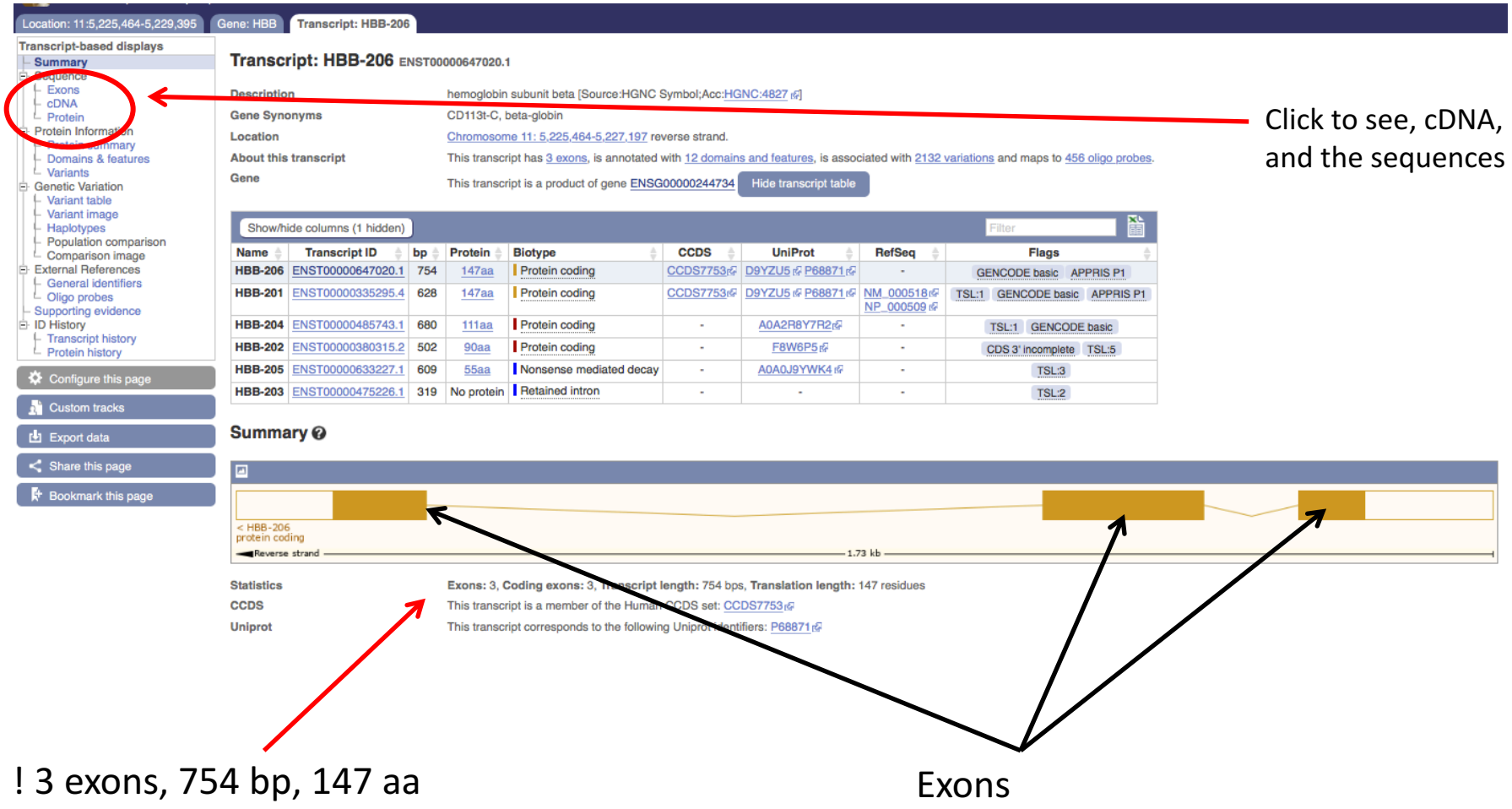
Name	Transcript ID	bp	Protein	Biotype	CCDS	UniProt	RefSeq	Flags
HBB-206	ENST00000647[illegible]0.1	754	147aa	Protein coding	CCDS7753	D9YZU5 P68871	-	GENCODE basic APPRIS P1
HBB-201	ENST00000335295.4	628	147aa	Protein coding	CCDS7753	D9YZU5 P68871	NM_000518 NP_000509	TSL:1 GENCODE basic APPRIS P1
HBB-204	ENST00000485743.1	680	111aa	Protein coding	-	A0A2R8Y7R2	-	TSL:1 GENCODE basic
HBB-202	ENST00000380315.2	582	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



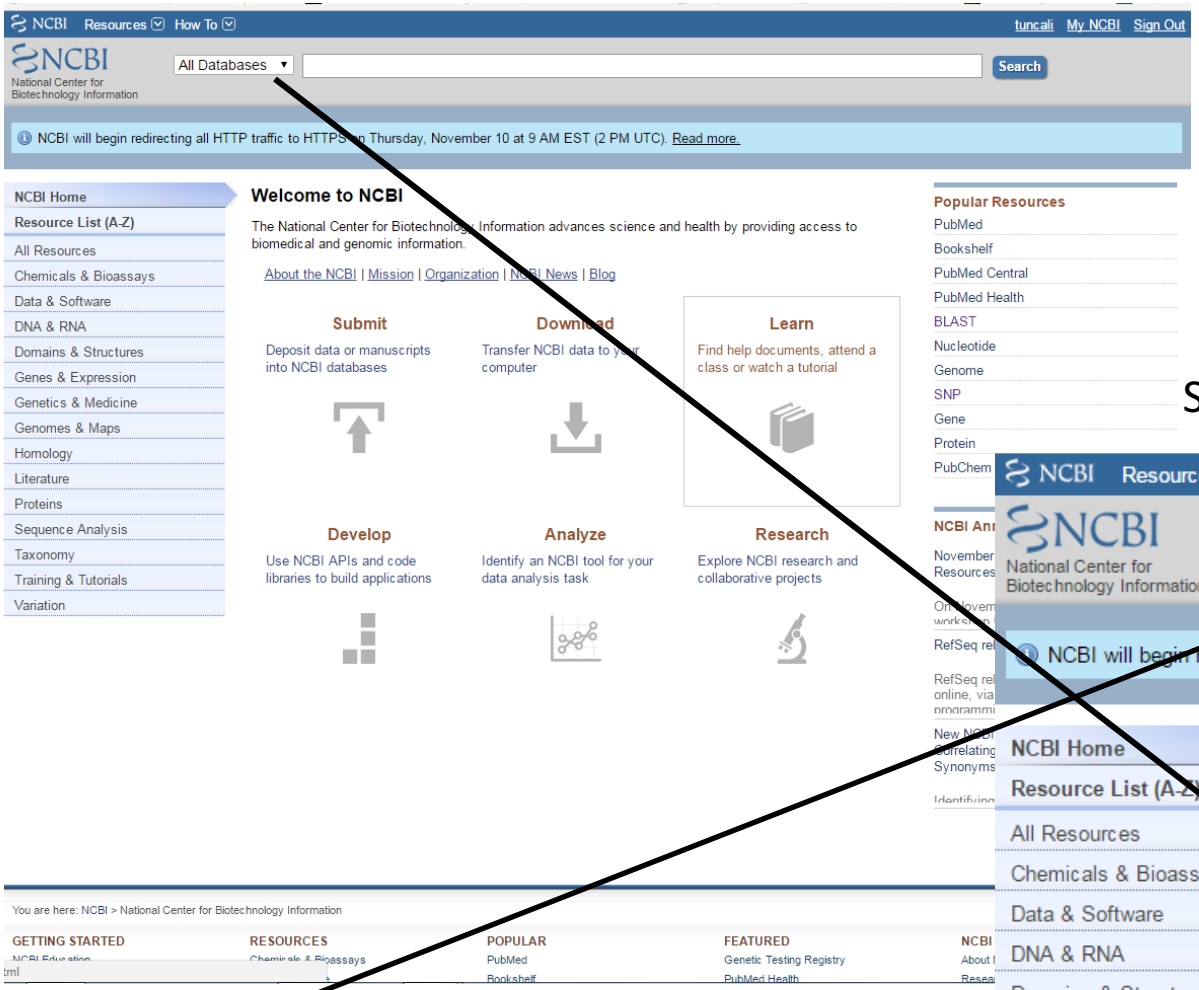
Click to see, cDNA, protein and the sequences

! 3 exons, 754 bp, 147 aa

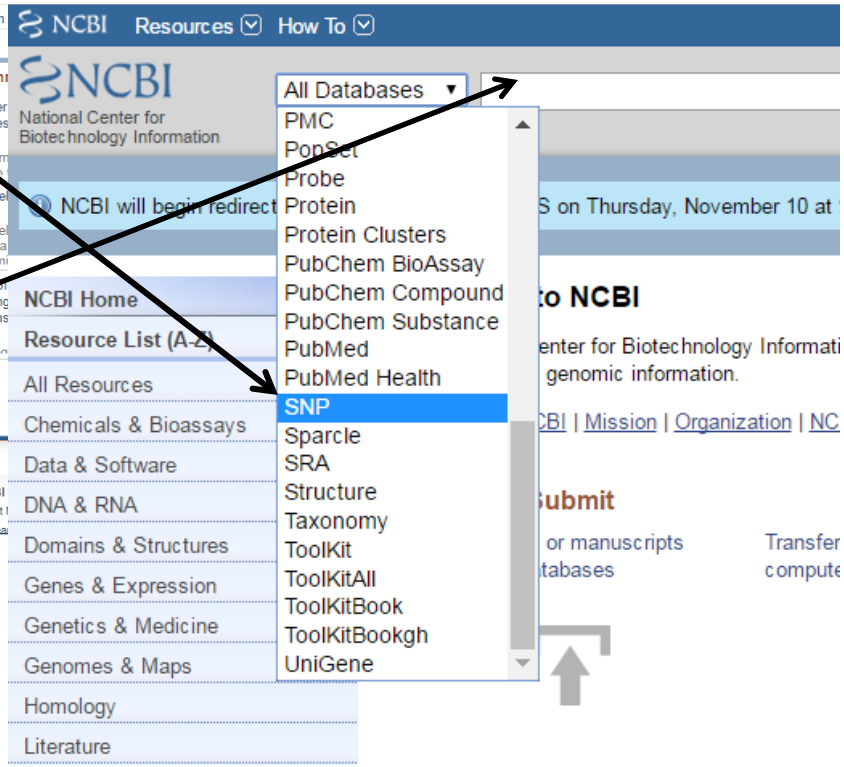
Exons

Variation database

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



SNP database

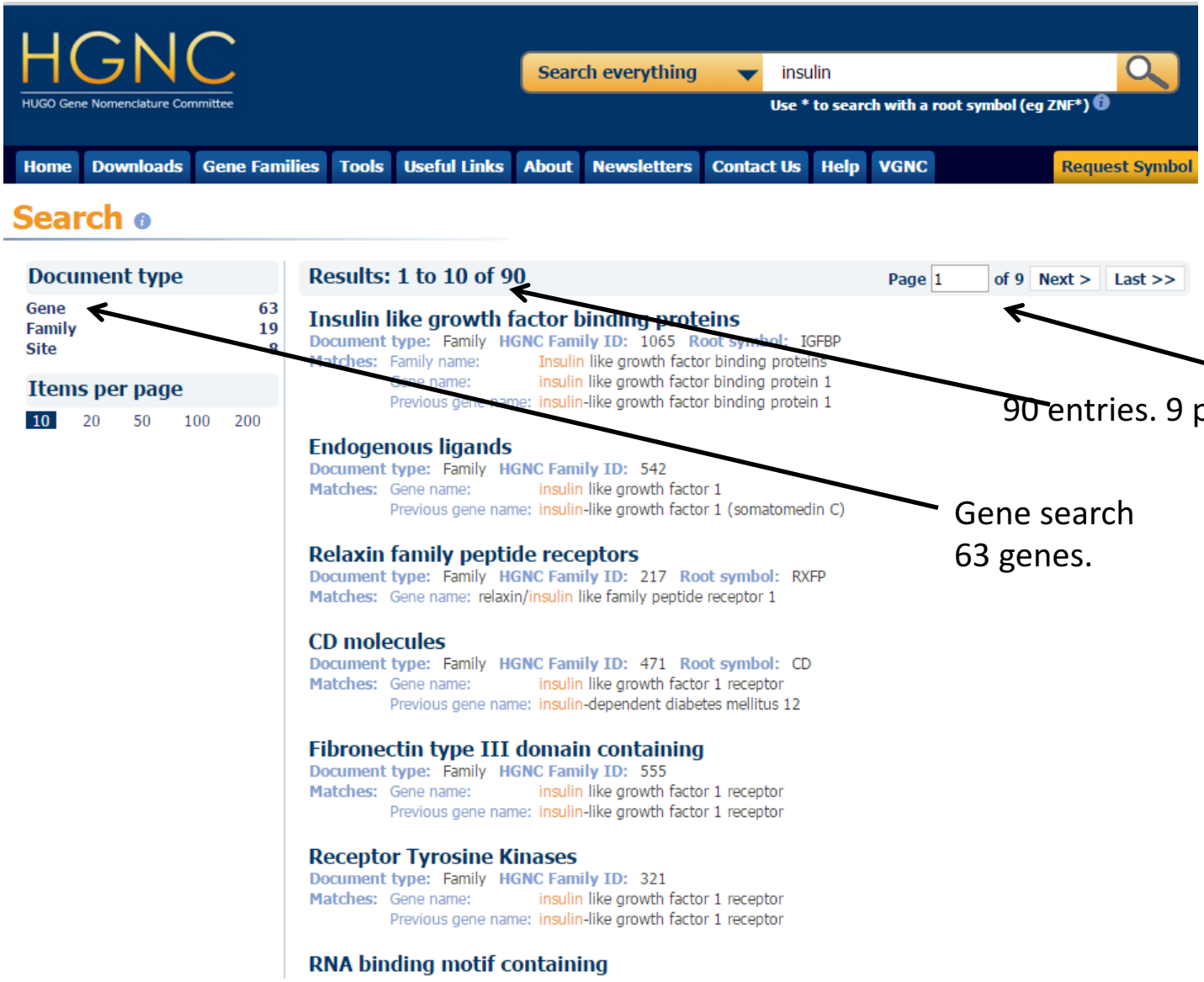


Enter SNP reference number
(rs xxxxx) or gene symbol
Check proper gene name or symbol from
HGNC or wikipedia if available.

www.genenames.org

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



90 entries. 9 pages

Gene search
63 genes.

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)

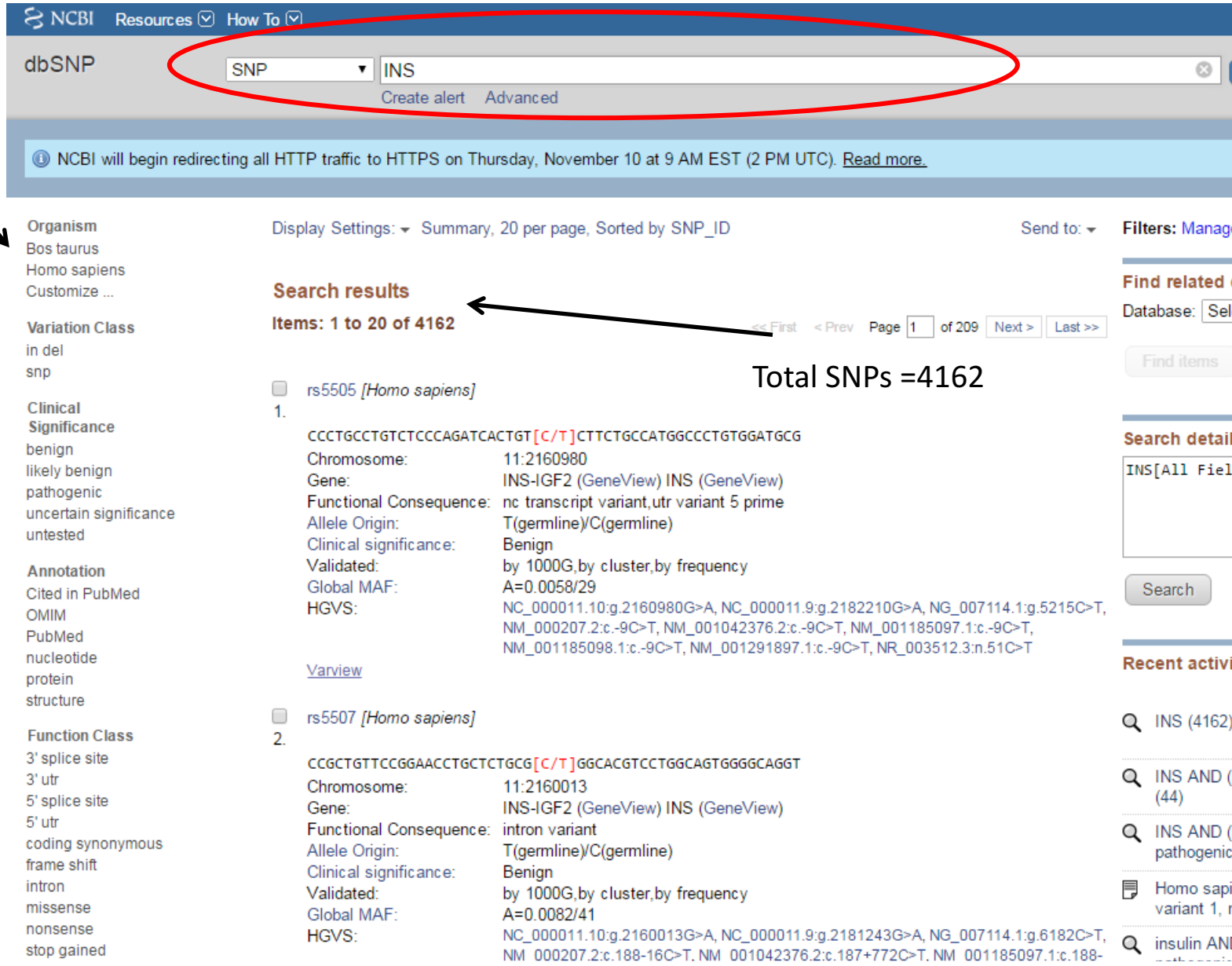
INSL5: insulin like 5
HGNC_ID: HGNC:6088 Location: 1p31.3 Locus type: gene with protein product

Scroll down to find insulin.

Symbol “INS”

Go back to SNP database.

Select Homo sapiens

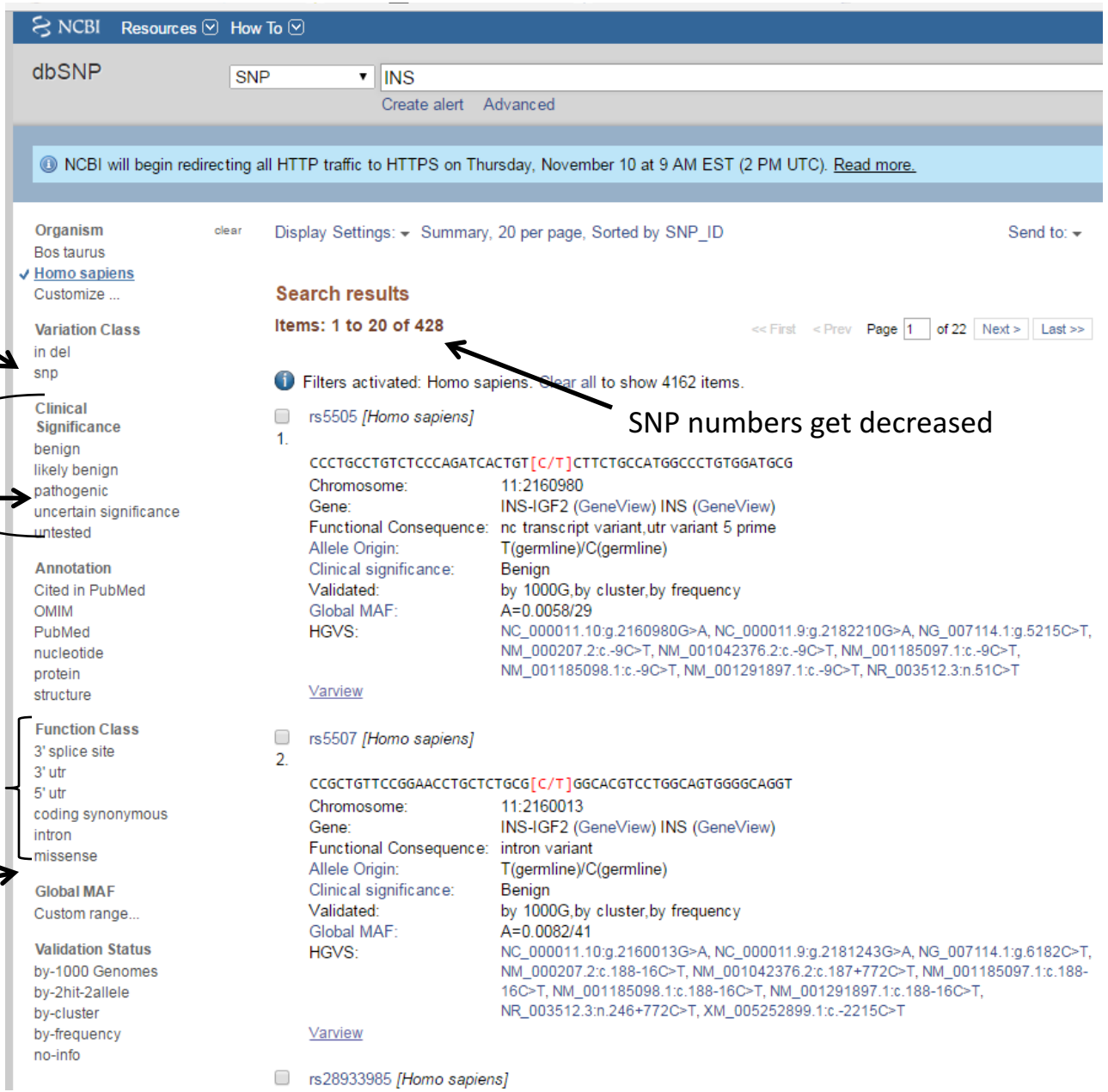


Select variation type
SNPs
(there are other types as well)

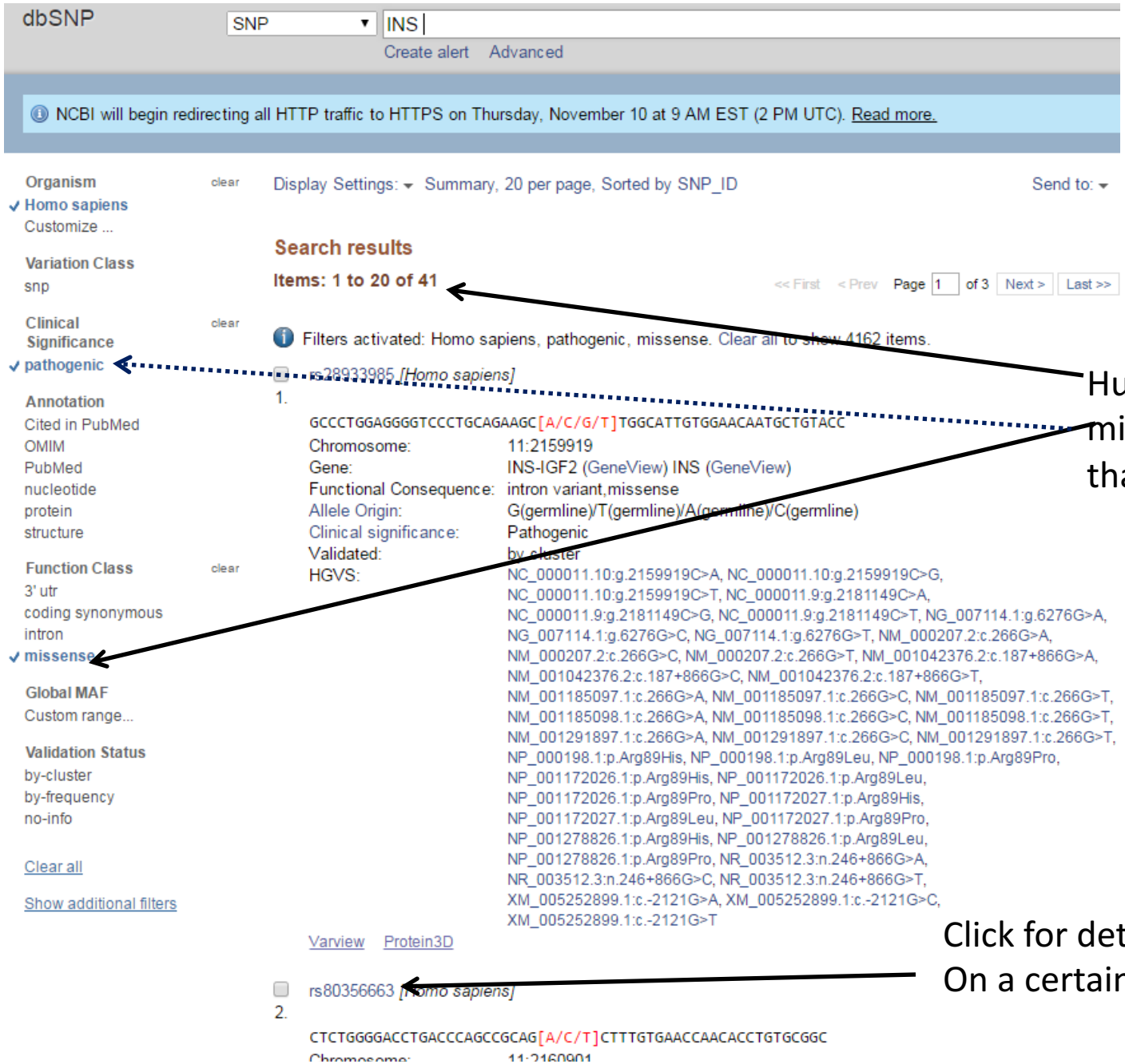
Select pathogenic SNPs

Select mutation type

As missense seçelim



SNP numbers get decreased

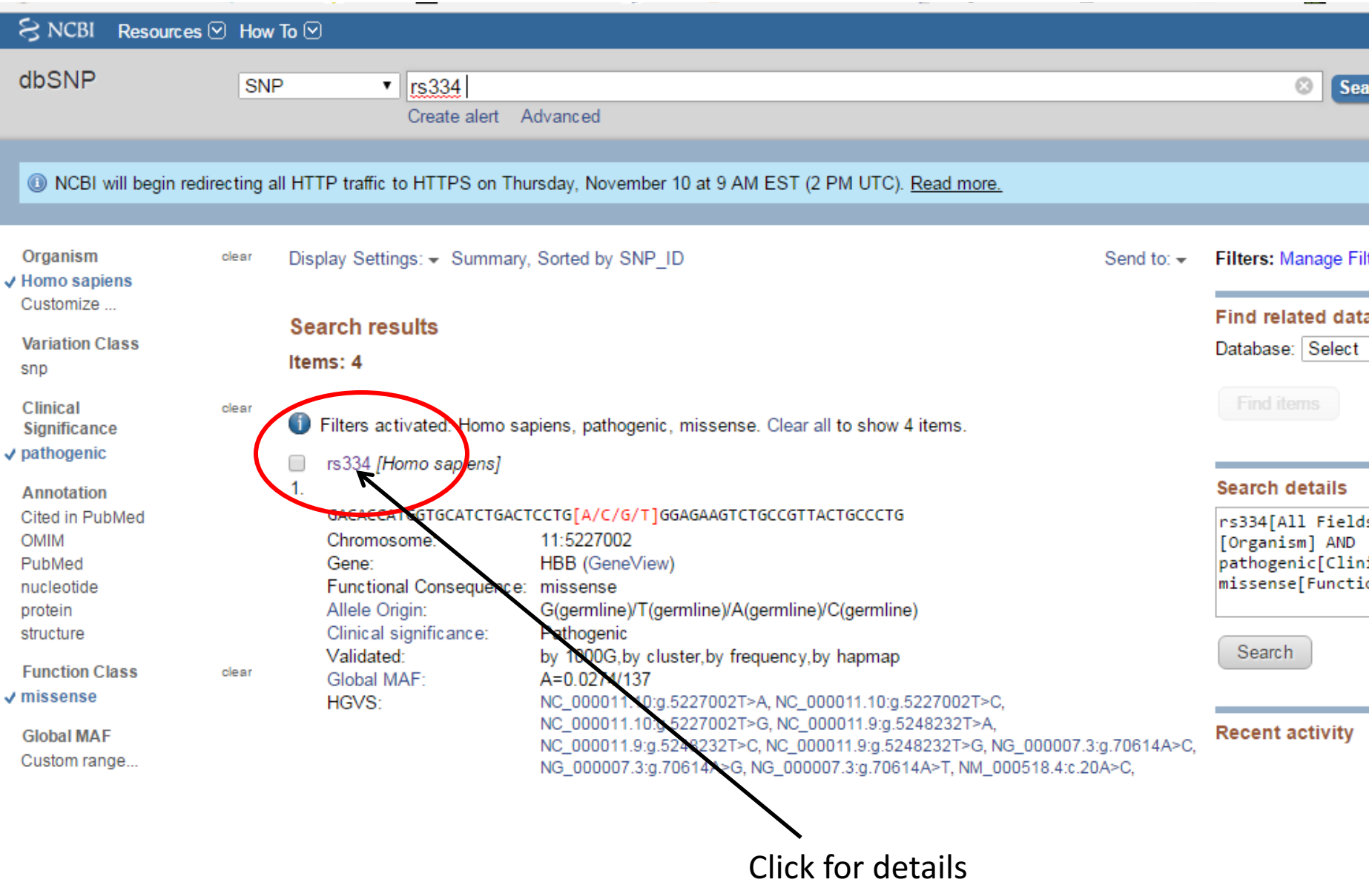


Human insulin gene missense mutations that are pathogenic.

Click for detailed information On a certain SNP

Known SNP reference number can be searched

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



Click for details

Reference SNP (rs) Report ALPHA

API

Switch to classic site

rs334

Current Build 151
Released July 17, 2018

Organism	*Homo sapiens*	**Clinical Significance**	Reported in ClinVar
Position	chr11:5227002 (GRCh38.p7)	**Gene : Consequence**	HBB : Missense Variant
Alleles	T>A / T>C / T>G	**Publications**	99 citations
Variation Type	SNV Single Nucleotide Variation	**Genomic View**	See rs on genome
Frequency	A=0.00342 (842/245928, GnomAD) A=0.00438 (532/121340, ExAC) A=0.0116 (360/30968, GnomAD) (+ 1 more)		

FEEDBACK

Variant Details
Clinical Significance
Frequency
Aliases
Submissions
History
Publications

Filter:

Download

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 frequncies