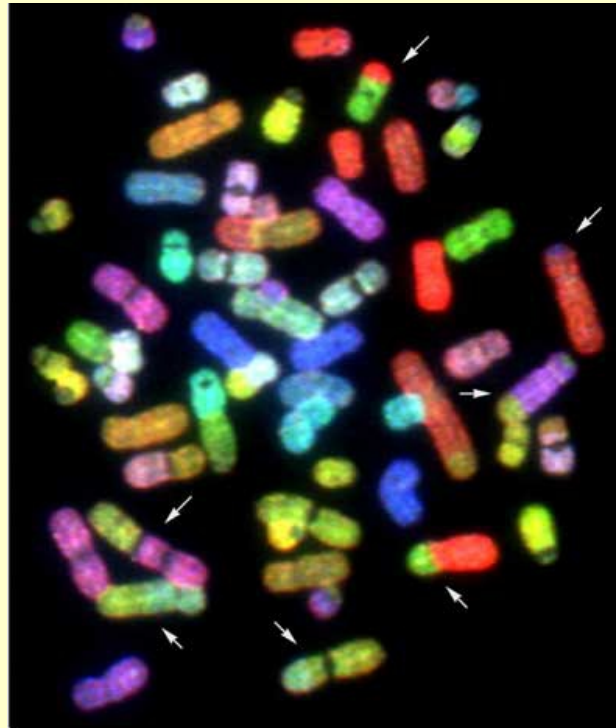


Genome Databases

Genomics, Bioinformatics & Medicine

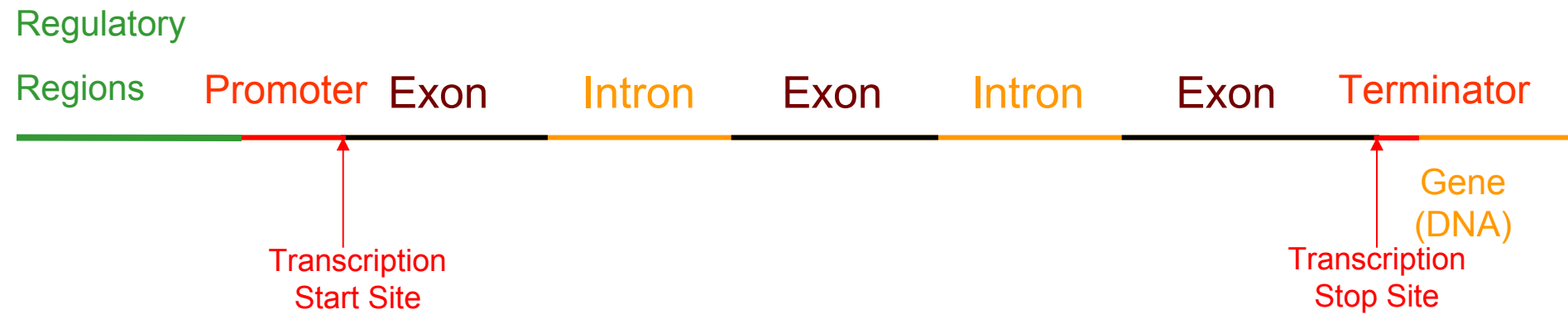
<http://biochem158.stanford.edu/>



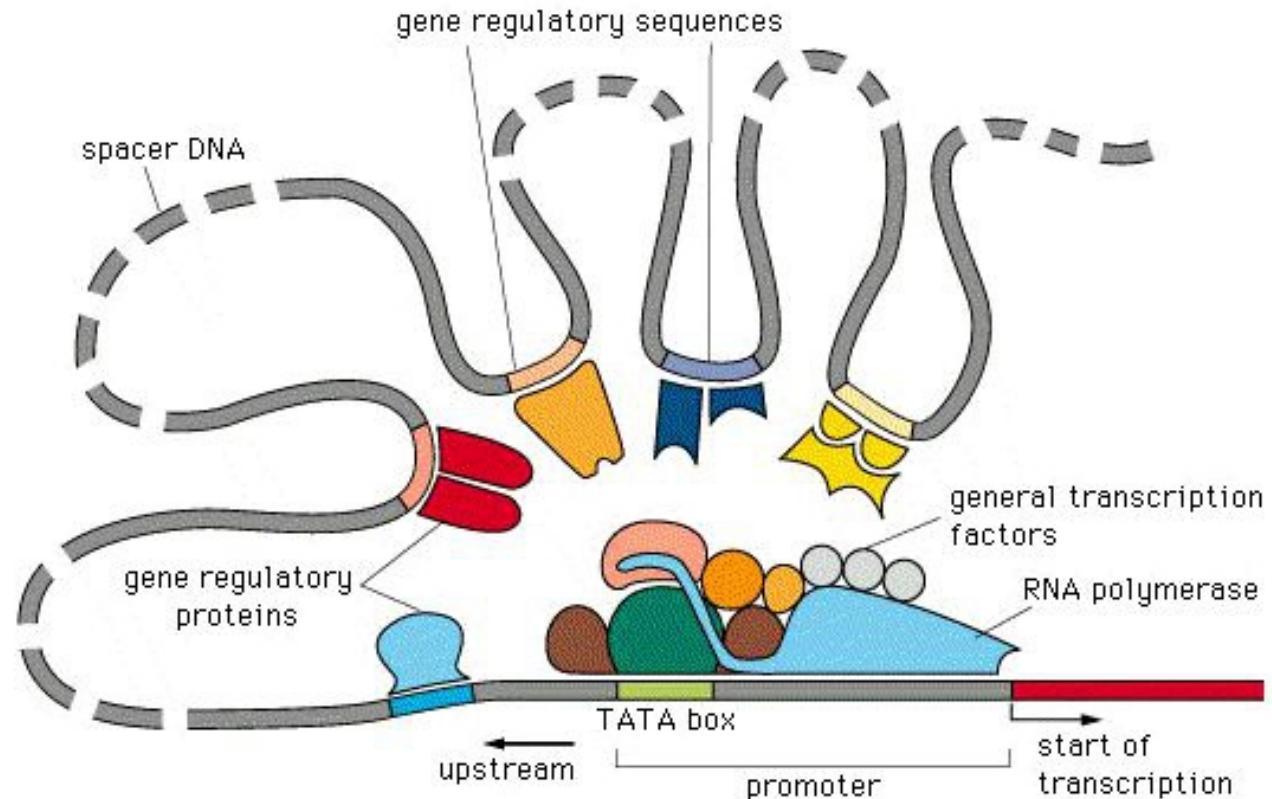
Doug Brutlag

Professor Emeritus of Biochemistry & Medicine
Stanford University School of Medicine

Components of a Typical Human Gene

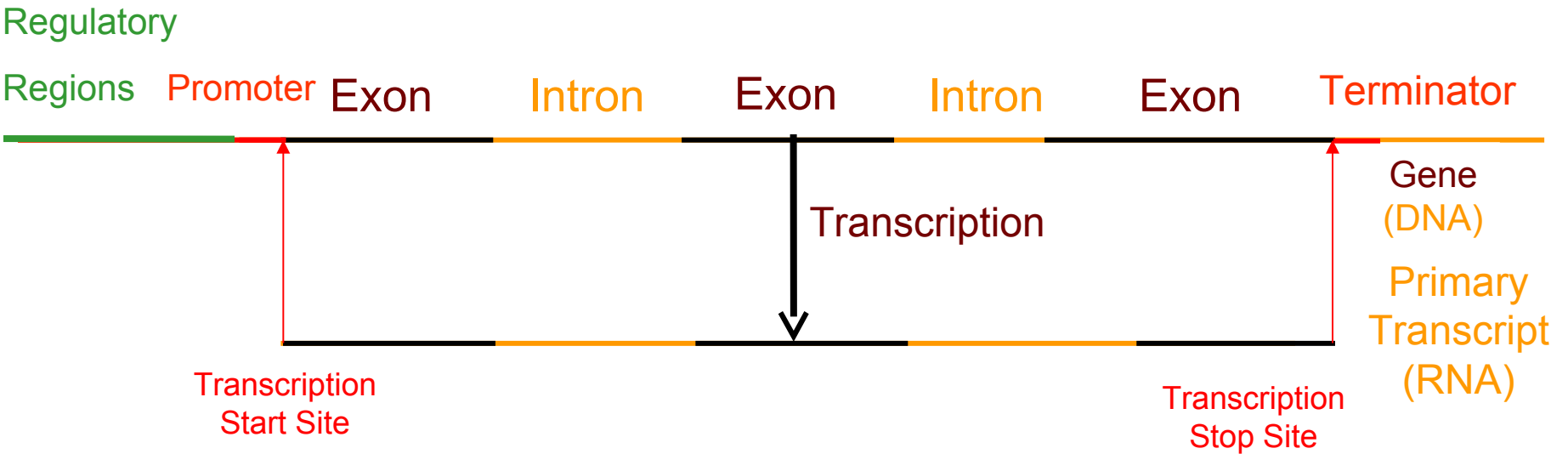


Multiple Enhancer Sequences

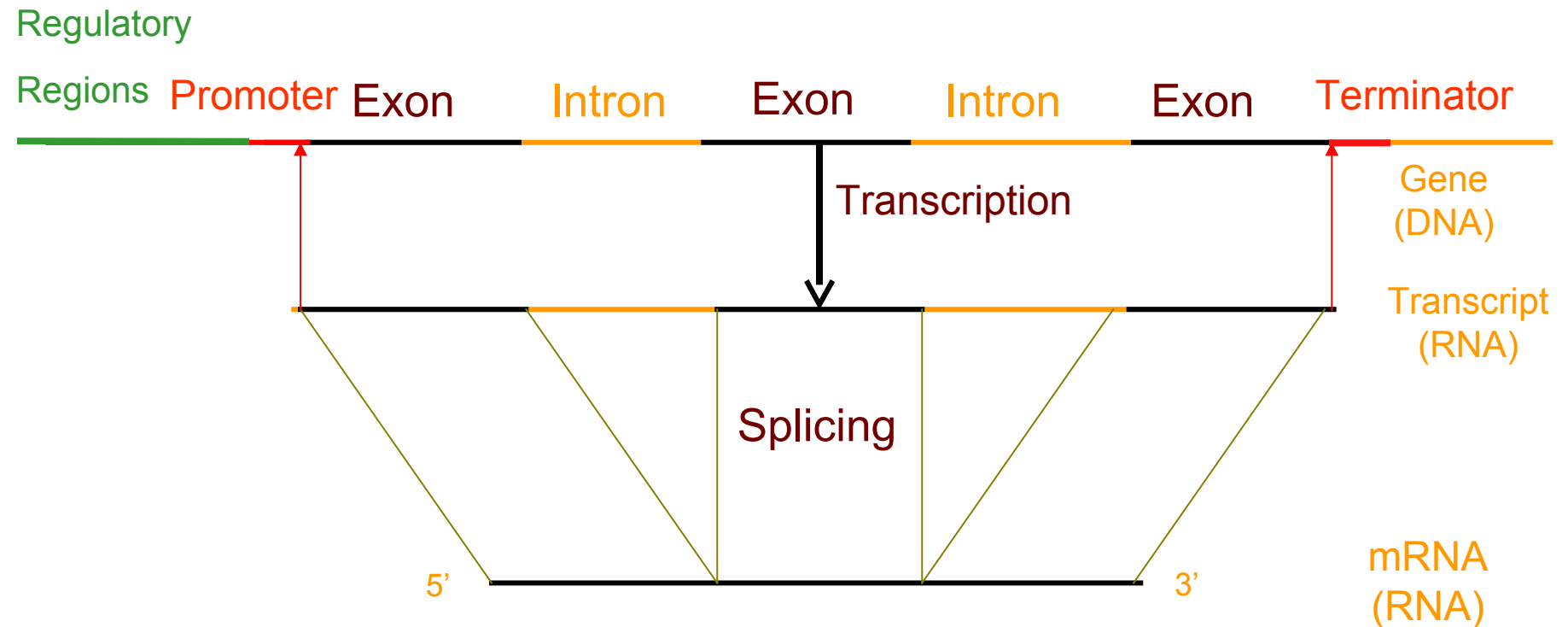


©1996 GARLAND PUBLISHING

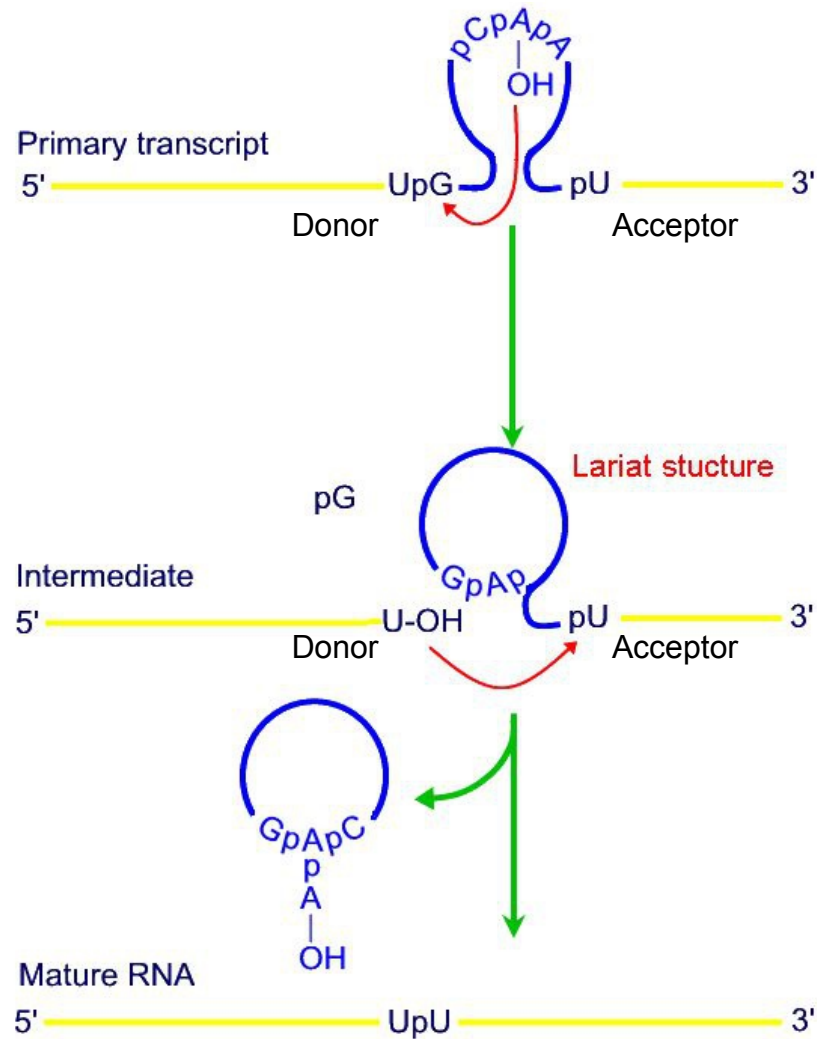
Active Genes are Transcribed into RNA



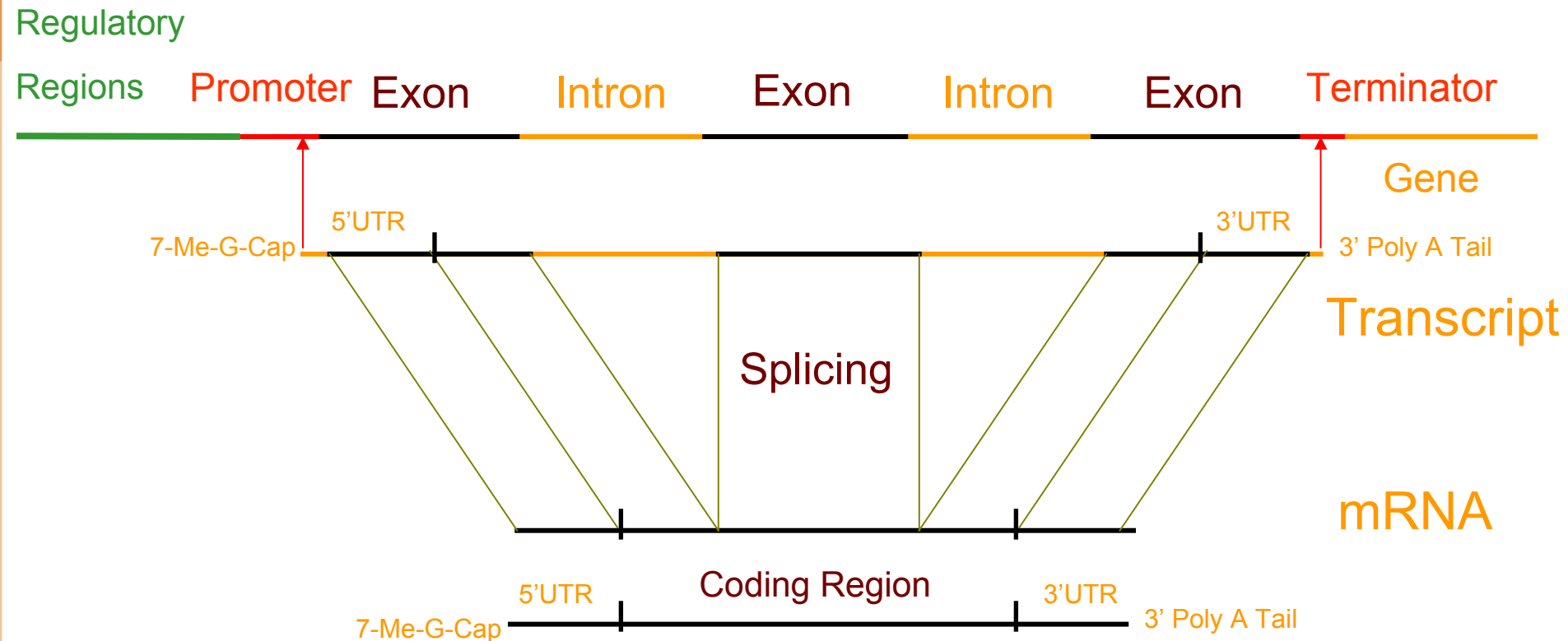
Splicing Transcript Yields Mature mRNA



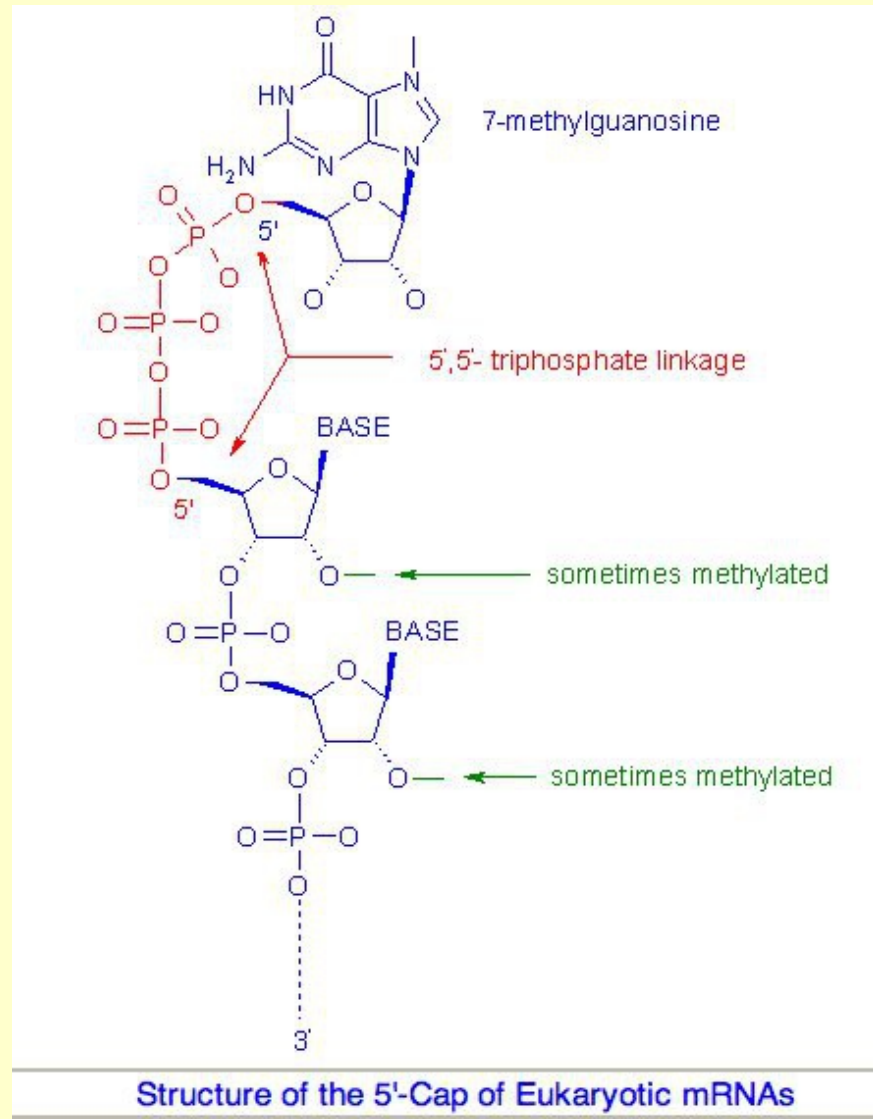
Intron Splicing Mechanism



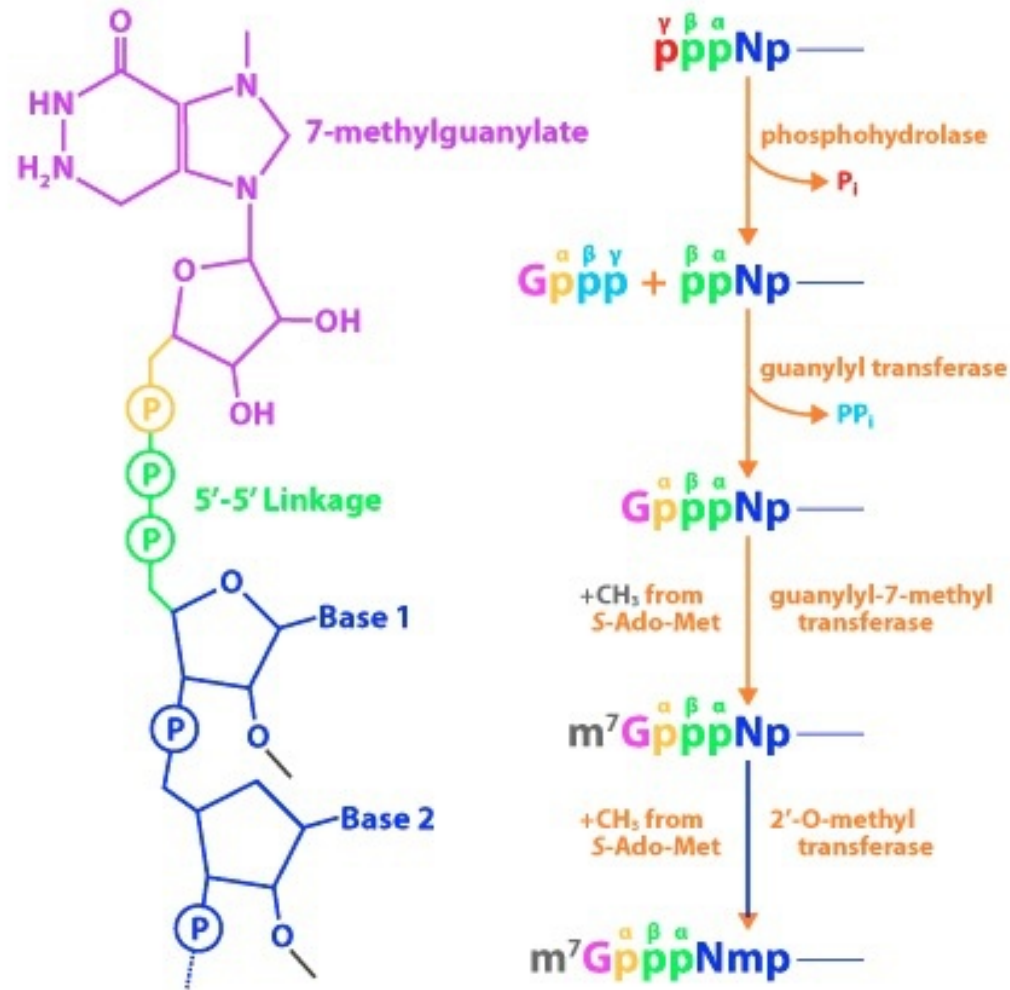
Mature mRNA contains 7-Methyl-Guanylate 5' Cap and 3' Poly A Tail



Structure of 5' CAP

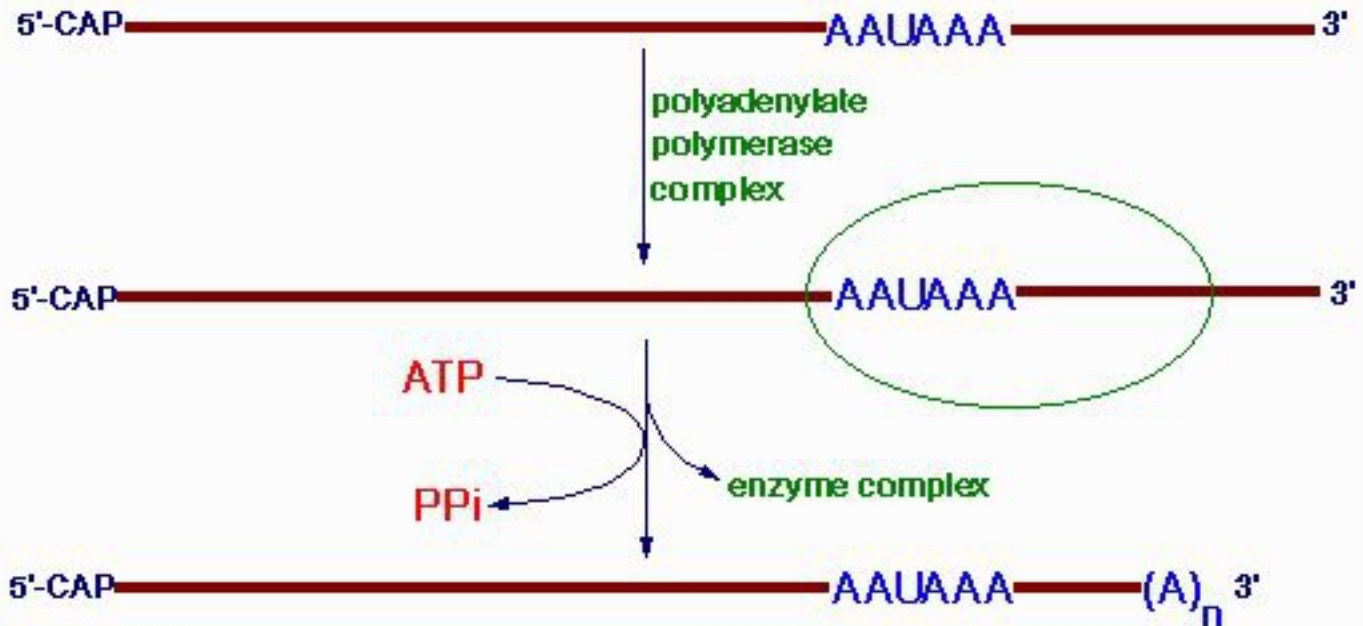


Mature mRNA contains 7-Methyl-Guanylate5' Cap



PolyAdenylation of mRNAs

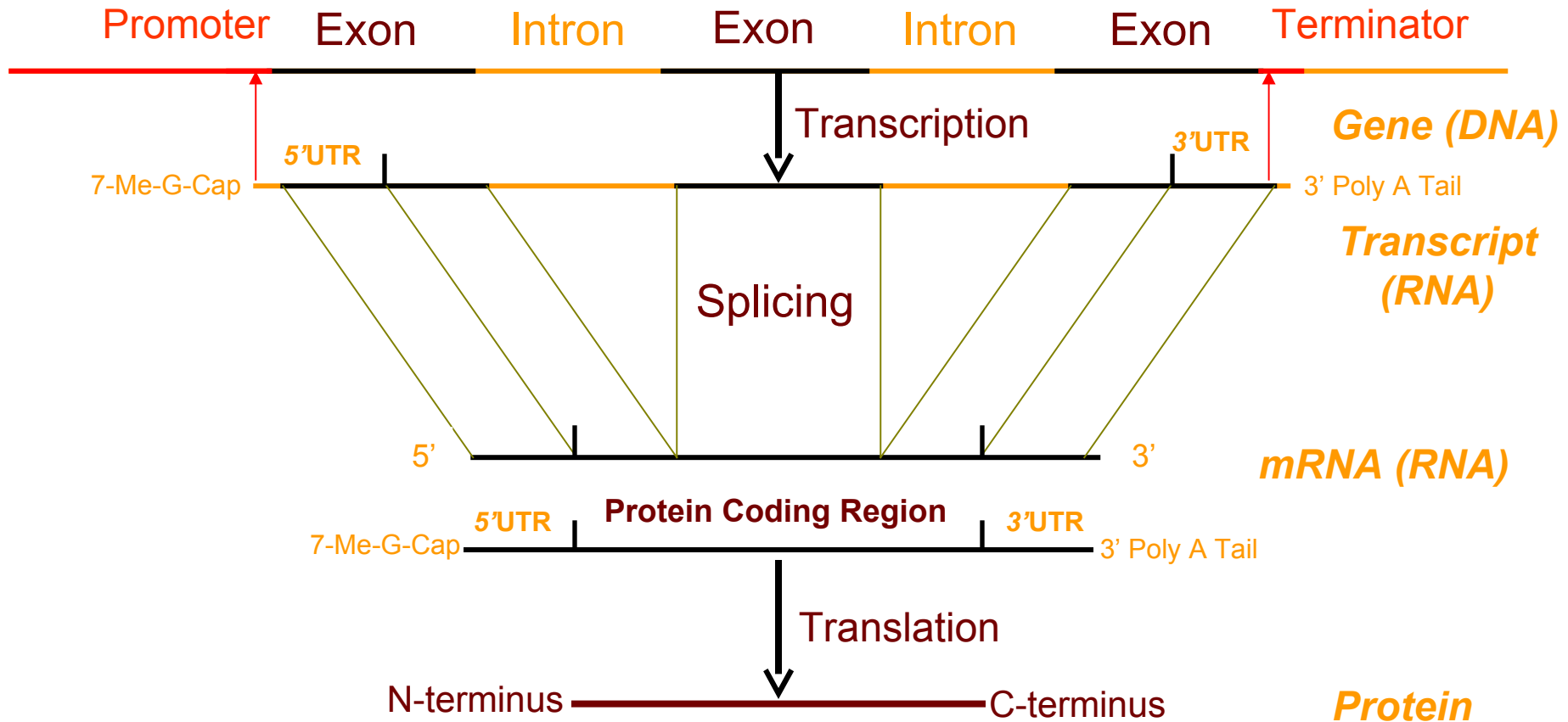
Polyadenylation of mRNAs



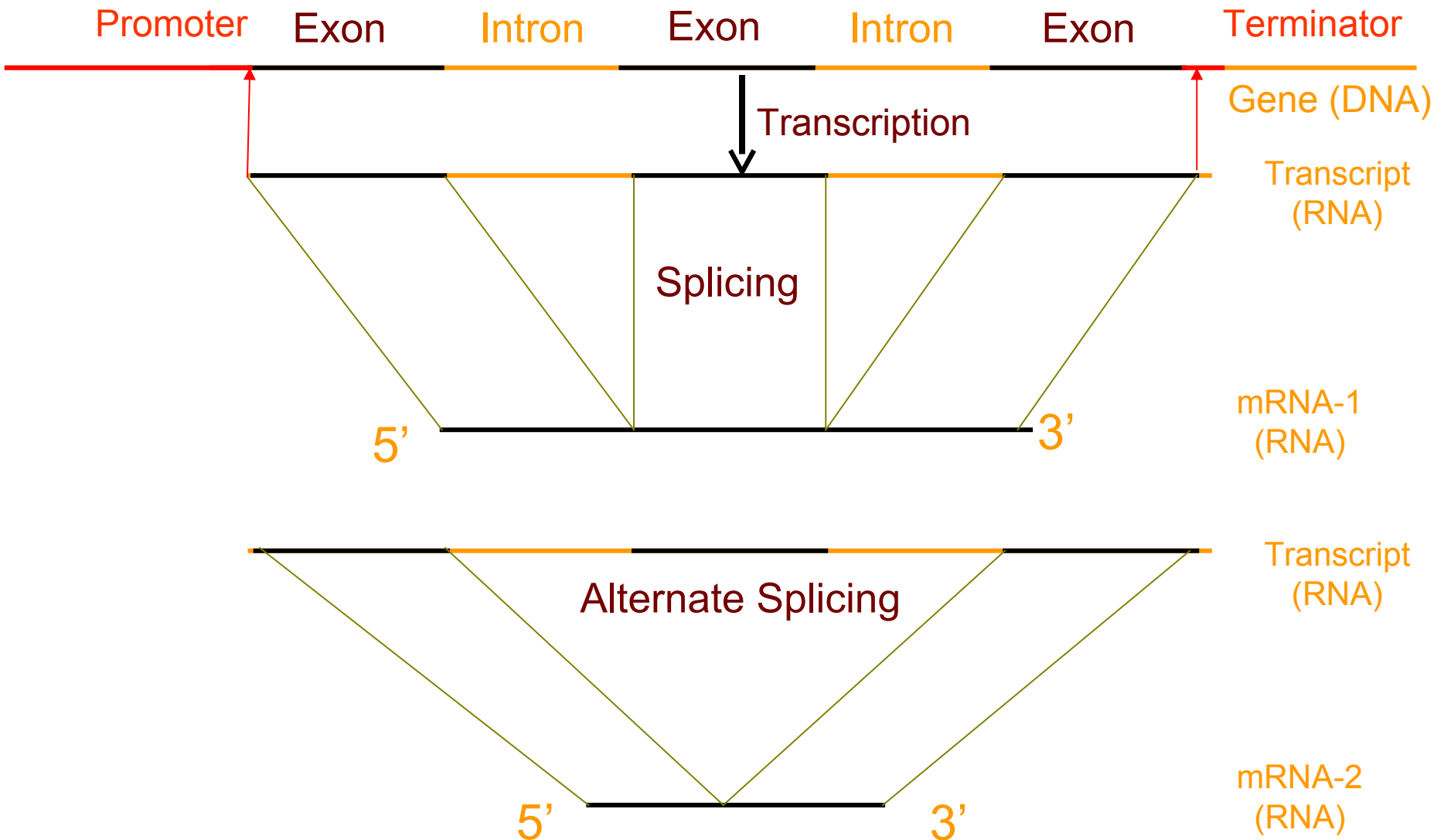
copyright 1996 M.W.King

Processes of Polyadenylation

Mature mRNA contains Coding Region and 5' and 3' Untranslated Regions

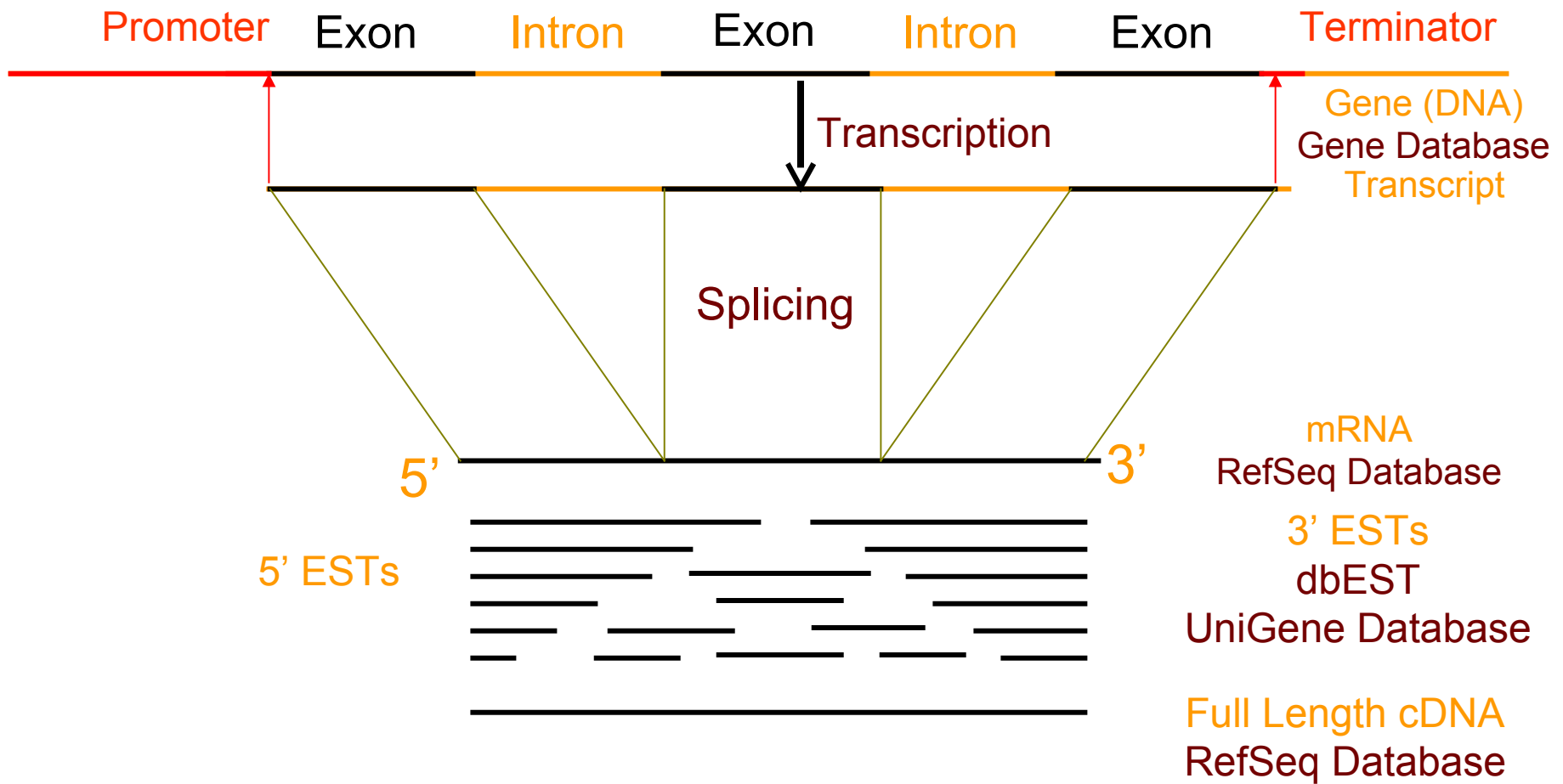


Alternative Splicing Generates Distinct Proteins in Different Tissues

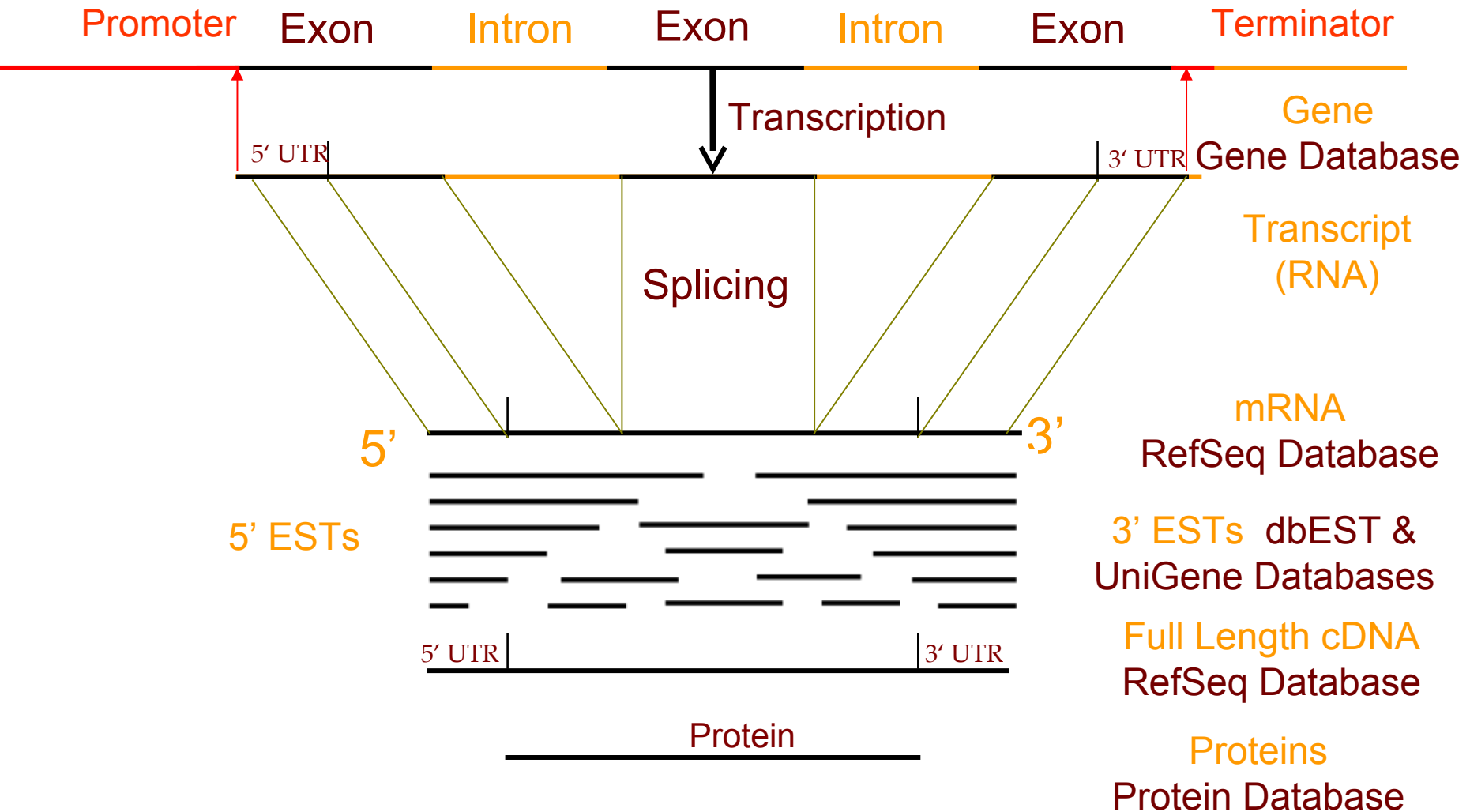


Messenger RNA (mRNA) Databases

ESTs UniGene & Full Length cDNA RefSeq



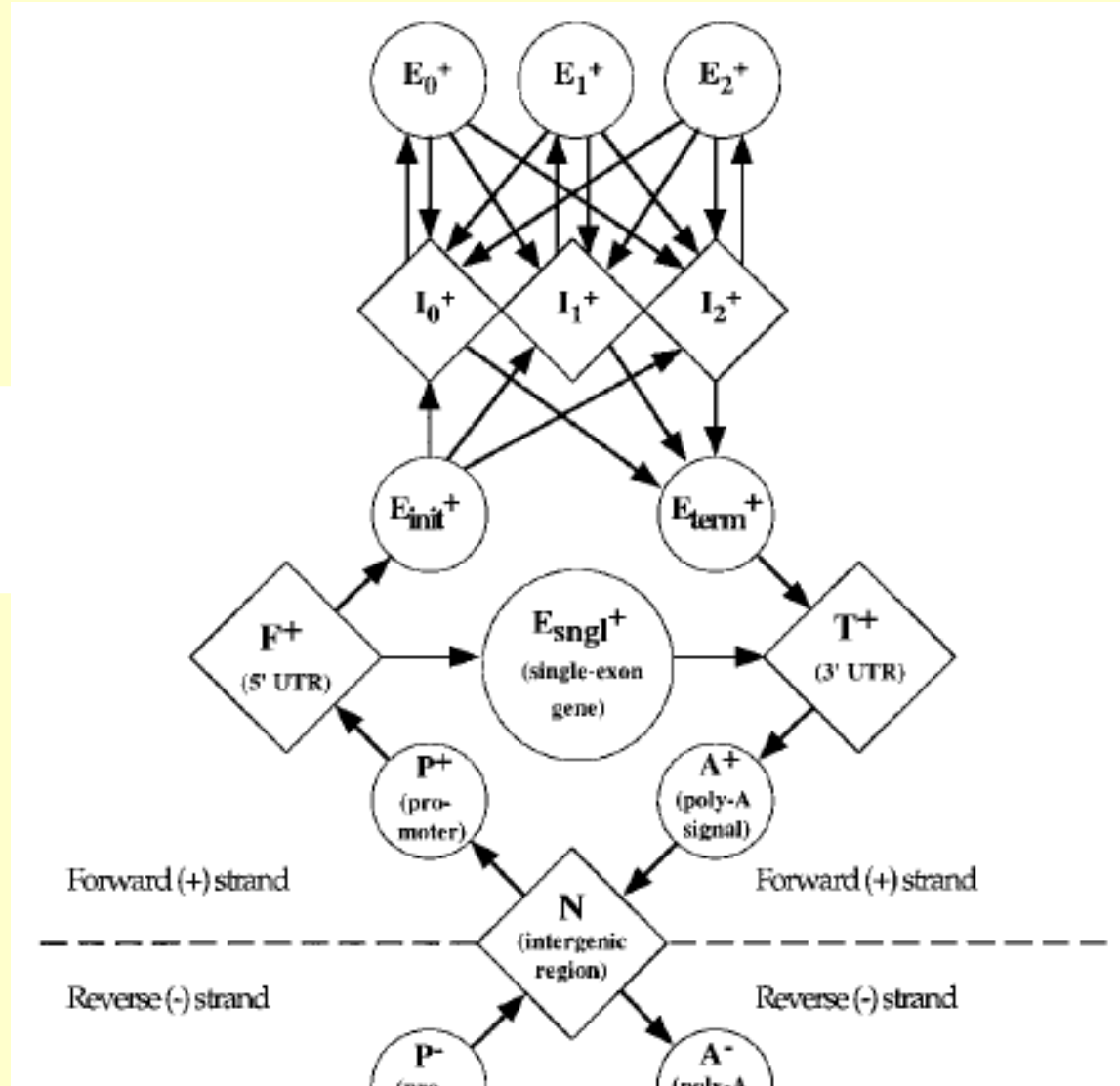
ESTs, Full Length cDNA UniGene & RefSeq Databases



GENSCAN Gene Model

<http://genes.mit.edu/GENSCAN.html>

Hidden Markov models of gene structure



Genome Databases

Assembled contigs

A Mapping

NCBI uniSTS NCBI dbSNP

B Gene Prediction

GrailEXP

GenScan

FGENESH

FGENESH+

GeneMark

C Expression Data

Human
ESTs

NCBI UniGene
Human

NCBI RefSeq
Human

Ensembl
cDNA

Mouse
ESTs

NCBI Gene
Mouse

NCBI RefSeq
Mouse

D Protein Similarity

NCBI nrPRO

EBI pFAM

UniProt

E Additional Data

Promoters

F Summary

NCBI Gene

UCSC Genome Browser

EBI Ensembl

NCBIGene

<http://www.ncbi.nlm.nih.gov/gene>

NR-Pro

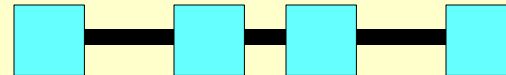


**Inclusive
Exon
Prediction**

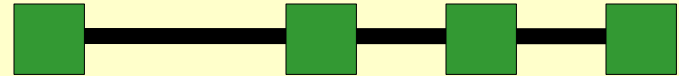
UniGene



RefSeq



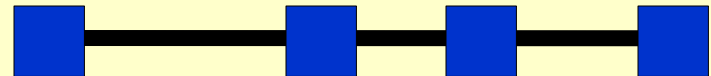
GrailEXP



FGENESH



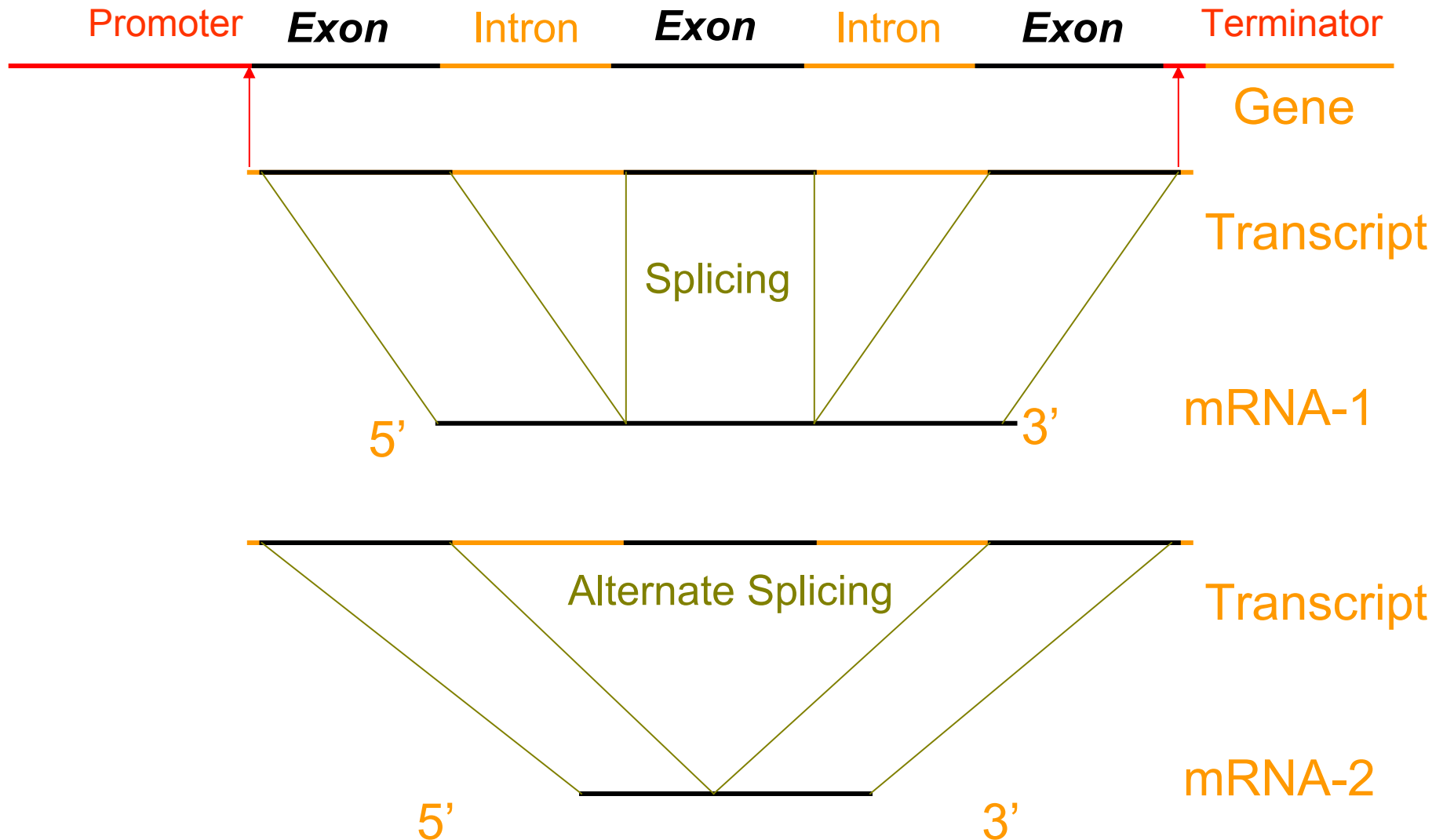
Genscan



NCBI Gene



Alternative Splicing Generates Distinct Proteins in Different Tissues



Genome

Genome ▾

dog



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Genome

This resource organizes information on genomes including sequences, maps, chromosomes, assemblies, and annotations.

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[Genomic groups BLAST](#)

[NCBI remap](#)

[Genome Decoration Page](#)

Genome Annotation and Analysis

[Eukaryotic Genome Annotation](#)

[Prokaryotic Genome Annotation](#)

[PASC \(Pairwise Sequence Comparison\)](#)

[TaxPlot \(3-way Genome Comparison\)](#)

External Resources

[GOLD - Genomes Online Database](#)

[Ensembl Genome Browser](#)

[Bacteria Genomes at Sanger](#)

[Large-Scale Genome Sequencing \(NHGRI\)](#)

Canis lupus familiaris Genome

<http://www.ncbi.nlm.nih.gov/genome/85>

Display Settings: Overview

Send to:

[Organism Overview](#) ; [Genome Project Report](#) ; [Genome Annotation Report](#) ; [Organelle Annotation Report](#)



Canis lupus familiaris (dog)

Model organism that is notable for its extensive genetic diversity and morphological variation

Lineage: [Eukaryota\[2439\]](#); [Metazoa\[952\]](#); [Chordata\[393\]](#); [Craniata\[386\]](#); [Vertebrata\[385\]](#); [Euteleostomi\[377\]](#); [Mammalia\[147\]](#); [Eutheria\[143\]](#); [Laurasiatheria\[56\]](#); [Carnivora\[14\]](#); [Caniformia\[11\]](#); [Canidae\[3\]](#); [Canis\[3\]](#); [Canis lupus\[2\]](#); [Canis lupus familiaris\[1\]](#)

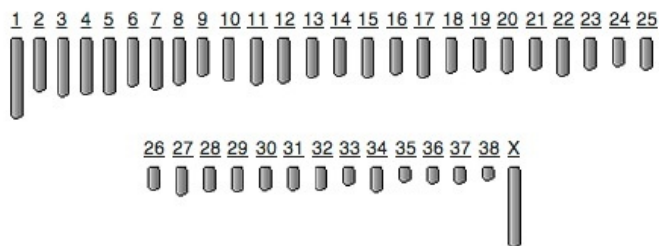
The dog, *Canis lupus familiaris*, is a useful model organism for medical research due to extensive genetic diversity and morphological variation within the species and to aggressive breeding practices that have resulted in inbred populations of dogs. Many breeds of dog are particularly susceptible to inherited diseases that are also common in [More...](#)

Representative

Reference genome:

[Canis lupus familiaris CanFam3.1](#)

Chromosomes



Click on chromosome name to open MapViewer

Genome Sequencing Projects

Chromosomes [1] Scaffolds or contigs [2] SRA or Traces [2]

Organism	BioProject	Assembly	Status	Chrs	Organelles	Size (Mb)	GC%	Gene	Protein
Canis lupus familiaris	PRJNA12384, PRJNA13179	CanFam3.1	<input checked="" type="radio"/>	39	1	2,410.98	35.5	28,864	42,303
Canis lupus familiaris	PRJNA10628	ASM18141v1	<input type="radio"/>	-	-	1,517.48	41.1	-	-
Canis lupus familiaris	PRJNA176193	Beagle	<input type="radio"/>	-	-	2,254.63	40.7	-	-
Canis lupus familiaris	PRJNA167192	-	<input type="radio"/>	-	-	-	-	-	-
Canis lupus familiaris	PRJNA186960	-	<input type="radio"/>	-	-	-	-	-	-

See more...

Human Genome Resources

<http://www.ncbi.nlm.nih.gov/genome/guide/human/>

NCBI Genomic Biology Homo sapiens

Search for

Browse your Genome
Click on the Chromosome to show




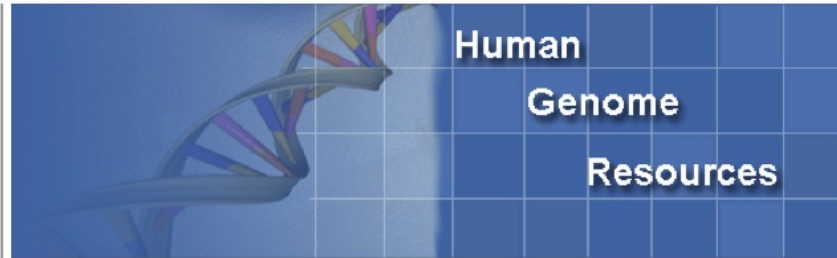
Find A Gene

Search for

from

The NCBI Handbook
An online guide to the use of NCBI resources. Titles of selected chapters that refer to human genome resources are shown below.

 **The Single Nucleotide Polymorphism Database (dbSNP) of Nucleotide Sequence Variation**
Adrienne Kitts and Stephen Sherry



A challenge facing researchers today is that of piecing together and analyzing the plethora of data currently being generated through the Human Genome Project and scores of smaller projects. NCBI's Web site serves an integrated, one-stop, genomic information infrastructure for biomedical researchers from around the world so that they may use these data in their research efforts. [More...](#)

Genes and Human Health

Gene Database

A new database of genes and associated information is now available for searching in Entrez.

OMIM

A guide to human genes and inherited disorders maintained by Johns Hopkins University and collaborators.

dbSNP

A database of single nucleotide polymorphisms (SNPs) and other nucleotide variations.

dbGaP

The database of Genotypes and Phenotypes (dbGaP) was developed to archive and distribute the results of studies that have investigated the interaction of genotype and phenotype.

Epigenomics

NIH Epigenomics Roadmap

Reference epigenomic maps and studies on new epigenetic mechanisms and their relevance to human health.

Roadmap Epigenomics Data

A comprehensive listing of all NIH Roadmap Epigenomics datasets submitted to GEO and SRA.

NCBI Entrez Gene

<http://www.ncbi.nlm.nih.gov/sites/entrez?db=gene>

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Entrez Gene
Genes and mapped phenotypes

Search: Gene

Limits Advanced search Help

human hemoglobin beta Search Clear



Welcome to Entrez Gene

Entrez Gene maintains information about genes from genomes of interest to the RefSeq group.

Using Entrez Gene

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Other Resources

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[RefSeqGene](#)

[UniGene](#)

[Protein Clusters](#)

Human Beta-Hemoglobin Gene Entry

<http://www.ncbi.nlm.nih.gov/gene/3043>

Gene

Genes and mapped phenotypes

Search:

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HBB hemoglobin, beta [*Homo sapiens*]

Gene ID: 3043, updated on 14-Jan-2011

Summary

Official Symbol	HBB provided by HGNC
Official Full Name	hemoglobin, beta provided by HGNC
Primary source	HGNC:4827
See related	Ensembl:ENSG00000223609 ; Ensembl:ENSG00000244734 ; HPRD:00786 ; MIM:141900
Gene type	protein coding
RefSeq status	REVIEWED
Organism	Homo sapiens
Lineage	Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo
Also known as	CD113t-C; beta-globin; HBB
Summary	The alpha (HBA) and beta (HBB) loci determine the structure of the 2 types of polypeptide chains in adult hemoglobin, Hb A. The normal adult hemoglobin tetramer consists of two alpha chains and two beta chains. Mutant beta globin causes sickle cell anemia. Absence of beta chain causes beta-zero-thalassemia. Reduced amounts of detectable beta globin causes beta-plus-thalassemia. The order of the genes in the beta-globin cluster is 5'-epsilon -- gamma-G -- gamma-A -- delta -- beta--3'. [provided by RefSeq]

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Links

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- [BioSystems](#)
- [Books](#)
- [CCDS](#)
- [Conserved Domains](#)
- [Full text in PMC](#)

Human Beta-Hemoglobin Gene Entry

<http://www.ncbi.nlm.nih.gov/gene/3043>

Genomic regions, transcripts, and products ↑ ?

Genomic Sequence NC_000011 chromosome 11 reference GRCh37.p13 Primary Assembly Go to [reference sequence details](#)

Go to nucleotide [Graphics](#) [FASTA](#) [GenBank](#)




NC_000011.9: 5.2M..5.2M (2.1Kbp) C Tools | Configure ?

3,600 | 5,248,400 | 5,248,200 | 5,248 K | 5,247,800 | 5,247,600 | 5,247,400 | 5,247,200 | 5,247 K | 5,246,800 | 5,246,600

Genes

HBB

NM_000518.4  NP_000509.1

SNP

Clinical Channel

Cited Variants

Association Results

Human Hemoglobin Gene Entry

<http://www.ncbi.nlm.nih.gov/gene/3043>

Related articles in PubMed

1. [Distribution of beta-globin haplotypes among the tribes of southern Gujarat, India.](#)
Aggarwal A, *et al.* Gene, 2013 Jun 1. PMID 23500448.
2. [Self-catalytic DNA depurination underlies human \$\beta\$ -globin gene mutations at codon 6 that cause anemias and thalassemias.](#)
Alvarez-Dominguez JR, *et al.* J Biol Chem, 2013 Apr 19. PMID 23457306.
3. [Sistani population: a different spectrum of \$\beta\$ -thalassemia mutations from other ethnic groups of Iran.](#)
Miri-Moghaddam E, *et al.* Hemoglobin, 2013. PMID 23437895.
4. [Association in cis of the mutations +20 \(C>T\) in the 5' untranslated region and IVS-II-745 \(C>G\) on the \$\beta\$ -globin gene.](#)
Ropero P, *et al.* Hemoglobin, 2013. PMID 23425204.
5. [Identification and molecular characterization of a novel 55-kb deletion recurrent in southern Italy: the Italian \(G\) \$\gamma\$ \(\(A\) \$\gamma\delta\beta\$ \)^o-thalassemia.](#)
Lacerra G, *et al.* Eur J Haematol, 2013 Mar. PMID 23281611.

[See all \(600\) citations in PubMed](#)

[See citations in PubMed for homologs of this gene provided by HomoloGene](#)

GeneRIFs: Gene References Into Functions [What's a GeneRIF?](#)

1. [The beta-globin distribution pattern of various haplotypes was consistent with the global pattern.](#)
2. [analysis of the contact region between CD163 and its high affinity ligand Hp-Hb reveals a mechanism of Ca²⁺-dependent coupling and uncoupling of ligand](#)
3. [Data indicate association of the +20 and IVS-II-745 mutations of beta-globin beta-thalassemia \(beta-thal\). Read More: <http://informahealthcare.com/doi/full/10.3109/03630269.2013.766620>](#)
4. [Data indicate twenty-one beta-globin mutations were identified in beta-thalassemia \(beta-thal\), of which the most frequent ones were IVS-I-5 \(G>C\), followed by codon 15 G>A codon -88 \(C>T\), IVS-II-1 \(G>A\), codons 8/9 \(+G\) and IVS-I-1 \(G>T\).](#)
5. [Data show that Hgb-alpha and Hgb-beta are expressed in carcinoma cells of several types of solid tumors.](#)
6. [analysis of a mouse model of the human beta-globin locus](#)

Human Beta-Hemoglobin Phenotypes

<http://www.ncbi.nlm.nih.gov/gene/3043>

Phenotypes

[Find tests for this gene in the NIH Genetic Testing Registry \(GTR\)](#)

[Review eQTL and phenotype association data in this region using PheGenI](#)

Associated conditions

Description	Tests
<p>beta Thalassemia</p> <p>MedGen: C0005283, OMIM: 613985, GeneReviews: Beta-Thalassemia</p>	Compare labs
<p>Beta thalassemia, dominant inclusion body type</p> <p>MedGen: C1858990, OMIM: 603902, GeneReviews: Not available</p>	Compare labs
<p>Fetal hemoglobin quantitative trait locus 1</p> <p>MedGen: C1841621, OMIM: 141749, GeneReviews: Not available</p>	Compare labs
<p>Hb SS disease</p> <p>MedGen: C0002895, OMIM: 603903, GeneReviews: Sickle Cell Disease</p>	Compare labs
<p>Heinz body anemias</p> <p>MedGen: C0700299, OMIM: 140700, GeneReviews: Not available</p>	Compare labs
<p>Susceptibility to malaria</p> <p>MedGen: C1970028, OMIM: 611162, GeneReviews: Not available</p>	not available

Human Beta-Hemoglobin Phenotypes

<http://www.ncbi.nlm.nih.gov/gene/3043>

NHGRI GWAS Catalog

Description

A genome-wide association scan on the levels of markers of inflammation in Sardinians reveals associations that underpin its complex regulation.

NHGRI GWA Catalog
[NHGRI GWA Catalog](#), [PubMed](#)

Genome-wide and fine-resolution association analysis of malaria in West Africa.

NHGRI GWA Catalog
[NHGRI GWA Catalog](#), [PubMed](#)

Genome-wide association study indicates two novel resistance loci for severe malaria.

NHGRI GWA Catalog
[NHGRI GWA Catalog](#), [PubMed](#)

Genome-wide association study shows BCL11A associated with persistent fetal hemoglobin and amelioration of the phenotype of beta-thalassemia.

NHGRI GWA Catalog
[NHGRI GWA Catalog](#), [PubMed](#)

GWAS of blood cell traits identifies novel associated loci and epistatic interactions in Caucasian and African-American children.

NHGRI GWA Catalog
[NHGRI GWA Catalog](#), [PubMed](#)

HbA2 levels in normal adults are influenced by two distinct genetic mechanisms.

NHGRI GWA Catalog
[NHGRI GWA Catalog](#), [PubMed](#)

Hemoglobin Interactions

<http://www.ncbi.nlm.nih.gov/gene/3043>

Interactions



Items 1 - 25 of 31 < Prev Page 1 of 2 Next >

Products	Interactant	Other Gene	Complex	Source	Pubs	Description
NC_000011.8	NP_006181.1	ORC2		BIND	PubMed	Orc2 interacts with beta-globin origin.
NC_000011.8	NP_000928.1	POLR2A		BIND	PubMed	Beta-globin interacts with pol II.
NC_000011.8	NP_066964.1	XRCC5		BIND	PubMed	Ku80 interacts with beta-globin origin.
P68871	Hemoglobin alpha 2	HBA2		HPRD	PubMed	
P68871	P69905	HBA2		HPRD	PubMed	
P68871	P68871	HBB		HPRD	PubMed	
P68871	P69892	HBG2		HPRD	PubMed	
P68871	P02008	HBZ		HPRD	PubMed	
P68871	P00738	HP		HPRD	PubMed	
P68871	Selenoprotein T	SELT		HPRD	PubMed	
BioGRID:109293	BioGRID:106710	AKT1		BioGRID	PubMed	Affinity Capture-MS
BioGRID:109293	BioGRID:107452	CDK2		BioGRID	PubMed	Affinity Capture-MS
BioGRID:109293	BioGRID:108102	DMWD		BioGRID	PubMed	Affinity Capture-MS
BioGRID:109293	BioGRID:115119	EIF4A3		BioGRID	PubMed	Affinity Capture-RNA
BioGRID:109293	BioGRID:119934	GDAP1		BioGRID	PubMed	Co-fractionation
BioGRID:109293	BioGRID:109289	HBA1		BioGRID	PubMed	Co-crystal Structure; Two-hybrid

Display Settings: Full Report

Send to:

HBB hemoglobin, beta [*Homo sapiens*]

Gene ID: 3043, updated on 14-Jan-2011

Summary

Official Symbol HBB provided by [HGNC](#)

Official Full Name hemoglobin, beta provided by [HGNC](#)

Primary source [HGNC:4827](#)

See related [Ensembl:ENSG00000223609](#); [Ensembl:ENSG00000244734](#); [HPRD:00786](#); [MIM:141900](#)

Gene type protein coding

RefSeq status REVIEWED

Organism [Homo sapiens](#)

Lineage Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo

Also known as CD113t-C; beta-globin; HBB

Summary The alpha (HBA) and beta (HBB) loci determine the structure of the 2 types of polypeptide chains in adult hemoglobin, Hb A. The normal adult hemoglobin tetramer consists of two alpha chains and two beta chains. Mutant beta globin causes sickle cell anemia. Absence of beta chain causes beta-zero-thalassemia. Reduced amounts of detectable beta globin causes beta-plus-thalassemia. The order of the genes in the beta-globin cluster is 5'-epsilon -- gamma-G -- gamma-A -- delta -- beta--3'. [provided by RefSeq]

Genomic regions, transcripts, and products

Go to [reference sequence details](#)

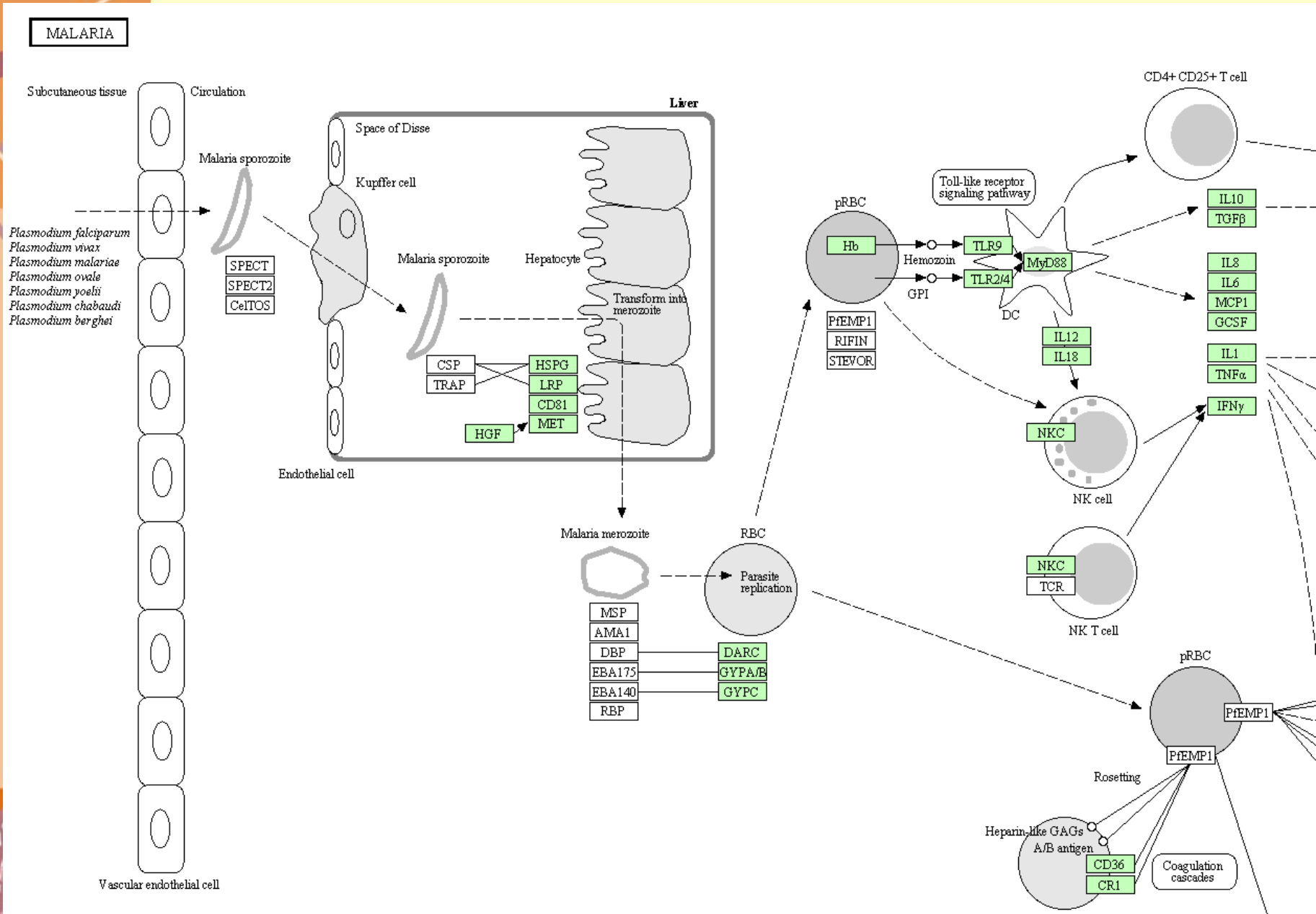
Genomic Sequence

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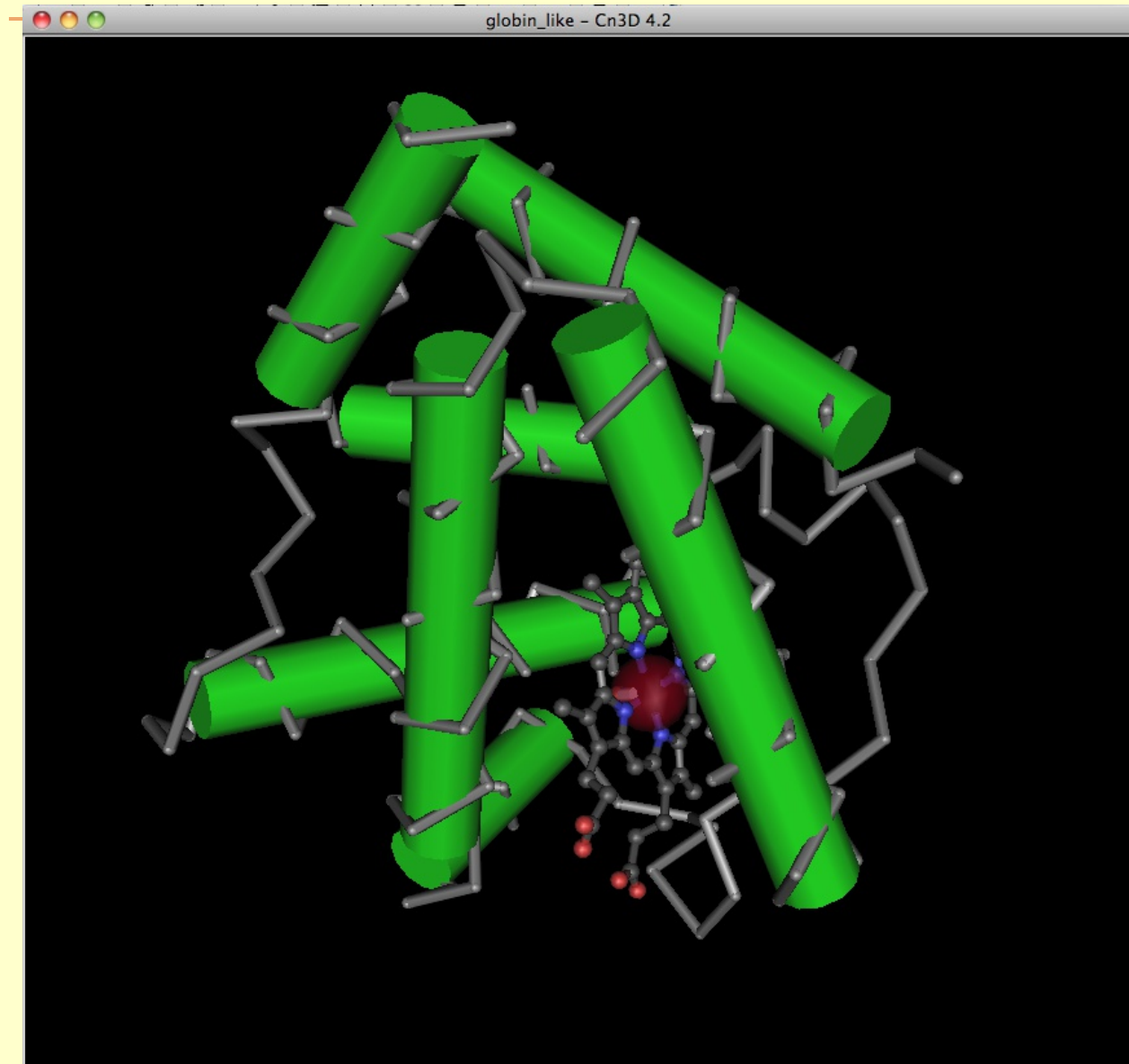
Links

- Order cDNA clone
- BioAssay, by Gene target
- BioSystems
- Books
- CCDS
- Conserved Domains
- Full text in PMC
- GEO Profiles
- Genome
- HomoloGene
- Map Viewer



Human Beta-Hemoglobin Conserved Domain

http://www.ncbi.nlm.nih.gov/cdd?LinkName=gene_cdd&from_uid=3043



Human Beta-Hemoglobin Homologene

http://www.ncbi.nlm.nih.gov/homologene?LinkName=gene_homologene&from_uid=3043h

Display Settings: HomoloGene

HomoloGene:68066. Gene conserved in Eutheria
















Genes

Genes identified as putative homologs of one another during the construction of HomoloGene.

- HBB, *H.sapiens*
hemoglobin, beta
- HBB, *P.troglodytes*
hemoglobin, beta
- HBB, *M.mulatta*
hemoglobin, beta
- LOC609402, *C.lupus*
hemoglobin subunit beta-like
- LOC480784, *C.lupus*
hemoglobin subunit beta-like
- HBD, *C.lupus*
hemoglobin, delta
- LOC100337028, *B.taurus*
hemoglobin fetal subunit beta-like
- HBB, *B.taurus*
hemoglobin, beta
- LOC781674, *B.taurus*
hemoglobin fetal subunit beta-like
- Beta-s, *M.musculus*
hemoglobin subunit beta-1-like
- Hbb-b1, *M.musculus*
hemoglobin, beta adult major chain
- Hbb-b1, *R.norvegicus*
hemoglobin, beta adult major chain
- LOC689064, *R.norvegicus*
beta-globin
- Hbb, *R.norvegicus*
hemoglobin, beta
- LOC100134871, *R.norvegicus*
beta globin minor gene

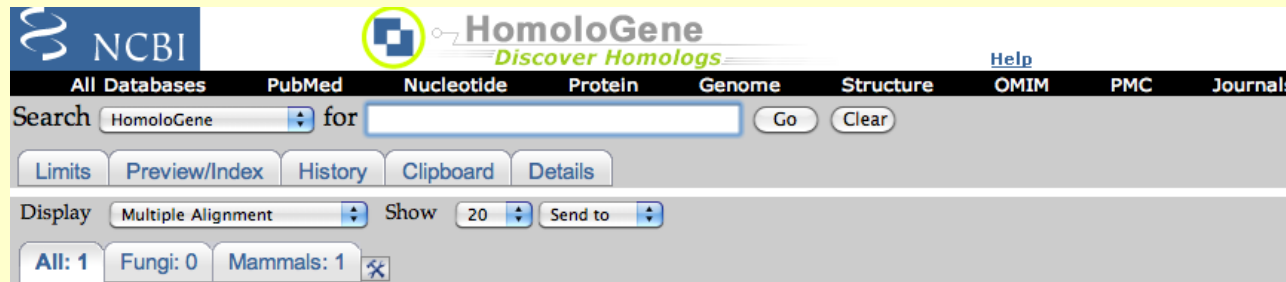
Proteins

Proteins used in sequence comparisons and their conserved domain architectures.

- NP_000509.1 
147 aa
- XP_508242.1 
147 aa
- NP_001157900.1 
147 aa
- XP_862442.1 
162 aa
- XP_003433067.1 
147 aa
- XP_534029.2 
147 aa
- XP_002693271.1 
145 aa
- NP_776342.1 
145 aa
- XP_003587018.1 
145 aa
- NP_001188320.1 
147 aa
- NP_032246.2 
147 aa
- NP_942071.1 
147 aa
- NP_001104739.1 
147 aa
- NP_150237.1 
147 aa
- NP_001106694.1 
147 aa

Human Beta-Hemoglobin Homologene

http://www.ncbi.nlm.nih.gov/homologene?LinkName=gene_homologene&from_uid=3043h



NCBI HomoloGene Discover Homologs

All Databases PubMed Nucleotide Protein Genome Structure OMIM PMC Journals

Search HomoloGene for Go Clear

Limits Preview/Index History Clipboard Details

Display Multiple Alignment Show 20 Send to

All: 1 Fungi: 0 Mammals: 1

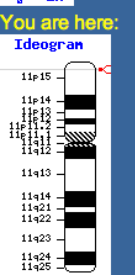
1: HomoloGene:68066. Gene conserved in Eutheria [Download](#) , [Links](#)

Multiple Sequence Alignment

Generated by MUSCLE [\[see reference\]](#) version 3.6 (using option: -maxiters 2).

NP_000509.1	1	MVHLTPEEKSAVTALWGKVVNDEVGGEALGRLLVVYPWTPQRFESFGDLS	50
XP_508242.1	1	MVHLTPEEKSAVTALWGKVVNDEVGGEALGRLLVVYPWTPQRFESFGDLS	50
XP_850823.1	1	MVHLTAEKSLIVSGLWGKVVNDEVGGEALGRLLIVYPWTPQRFESFGDLS	50
XP_534029.2	1	MVHLTAEKSLISSMWGKVVNDEVGGEALGRLLIVYPWTPQRFESFGDLS	50
XP_537902.1	1	MVHLTAEKSLIVSGLWGKVVNDEVGGEALGRLLIVYPWTPQRFESFGDLS	50
XP_001249460.2	1	--MLSAEKAAVTSLFAKVKVDEVGGEALGRLLVVYPWTPQRFESFGDLS	48
NP_001014902.1	1	--MLSAEKAAVTSLFAKVKVDEVGGEALGRLLVVYPWTPQRFESFGDLS	48
XP_001252211.2	1	--MLSAEKAAVTSLFAKVKVDEVGGEALGRLLVVYPWTPQRFESFGDLS	48
XP_001250142.2	1	--MLSAEKAAVTSLFAKVKVDEVGGEALGRLLVVYPWTPQRFESFGDLS	48
NP_001103979.1	1	--MLSAEKAAVTSLFAKVKVDEVGGEALGRLLVVYPWTPQRFESFGDLS	48
NP_776342.1	1	--MLTAEKAAVTAFWGKVKVDEVGGEALGRLLVVYPWTPQRFESFGDLS	48
NP_032246.2	1	MVHLTDAEKAASVGLWGKVNADDEVGGEALGRLLVVYPWTPQRYFDSFGDLS	50
NP_150237.1	1	MVHLTDAEKAASVGLWGKVNADDEVGGEALGRLLVVYPWTPQRYFDSFGDLS	50
XP_001069372.1	1	MVHLTDAEKATVSGLWGKVNADNVGAEALGRLLVVYPWTPQRYFSKFGDLS	50
NP_942071.1	1	MVHLTDAEKATVSGLWGKVNADNVGAEALGRLLVVYPWTPQRYFSKFGDLS	50

NP_000509.1	51	TPDAVMGNPKVKAHGKKVLAFAVSDGLAHLNLDNLKGTFAATLSELHCDKLHVD	100
XP_508242.1	51	TPDAVMGNPKVKAHGKKVLAFAVSDGLAHLNLDNLKGTFAATLSELHCDKLHVD	100
XP_850823.1	51	TPDAVMSNAKVKAHGKKVLSNSFSDGLKLNLDNLKGTFAKLSELHCDKLHVD	100
XP_534029.2	51	TPDAVMSNAKVKAHGKKVLSNSFSDGLKLNLDNLKGTFAKLSELHCDKLHVD	100
XP_537902.1	51	TPDAVMSNAKVKAHGKKVLSNSFSDGLKLNLDNLKGTFAKLSELHCDKLHVD	100
XP_001249460.2	49	SADAILGNPKVKAHGKKVLDVDFCEGLKQLDDDLKGAFAASLSELHCDKLHVD	98
NP_001014902.1	49	SADAILGNPKVKAHGKKVLDVDFCEGLKQLDDDLKGAFAASLSELHCDKLHVD	98
XP_001252211.2	49	SADAILGNPKVKAHGKKVLDVDFCEGLKQLDDDLKGAFAASLSELHCDKLHVD	98
XP_001250142.2	49	SADAILGNPKVKAHGKKVLDVDFCEGLKQLDDDLKGAFAASLSELHCDKLHVD	98
NP_001103979.1	49	SADAILGNPKVKAHGKKVLDVDFCEGLKQLDDDLKGAFAASLSELHCDKLHVD	98
NP_776342.1	49	TADAVMNNPKVKAHGKKVLDVDFSNMGMKHLDDDLKGTFAALSELHCDKLHVD	98
NP_032246.2	51	SASAIMGNAKVKAHGKKVITAFNDGLNHLDSLKGTFAALSELHCDKLHVD	100
NP_150237.1	51	SASAIMGNPKVKAHGKKVINAFNDGLKHLNLDNLKGTFAHLSELHCDKLHVD	100
XP_001069372.1	51	SASAIMGNPQVKAHGKKVINAFNDGLKHLNLDNLKGTFAHLSELHCDKLHVD	100
NP_942071.1	51	SVASAIMGNPQVKAHGKVINAFDDGLKHLNLDNLKGTFAALSELHCDKLHVD	100



NCBI Map Viewer

PubMed Entrez BLAST OMIM Taxonomy Structure

Search

Homo sapiens (human) Build 37.2 (Current) [BLAST The Human Genome](#)

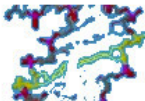
Chromosome: [1](#) [2](#) [3](#) [4](#) [5](#) [6](#) [7](#) [8](#) [9](#) [10](#) | **11** | [12](#) [13](#) [14](#) [15](#) [16](#) [17](#) [18](#) [19](#) [20](#) [21](#) [22](#) [X](#) [Y](#) [MT](#)

Query: 3043[[gene_id](#)] [\[clear\]](#)

Master Map: Genes On Sequence [Summary of Maps](#) [Maps & Options](#)

Region Displayed: 5,180K-5,315K bp [Download/View Sequence/Evidence](#)

Model1	Hs UniG	ensGenes	RefSeq	RNA	Genes_seq	Symbol	Links	E	Cyto	Description
						OR51A1P	HGNC sv prd lev mm	sts		best RefSeq 11p15.4 olfactory receptor, family 51, s
						OR5221P	HGNC sv prd lev mm	sts		best RefSeq 11p15.4 olfactory receptor, family 52, s
						OR52A1				
						OR51V1	HGNC sv prd lev mm hm sts	SNP		best RefSeq 11p15.4 hCG_1647060
						HBB	OMIM HGNC sv prd lev mm hm sts	SNP		best RefSeq 11p15.5 hCG_21979
						HBD	OMIM HGNC sv prd lev mm	sts SNP		best RefSeq 11p15.5 hCG_1641001
						HBBP1	HGNC sv d lev mm	sts		best RefSeq 11p15.5 hemoglobin, beta pseudogene
						HBG1	OMIM HGNC sv prd lev mm hm sts	SNP		best RefSeq 11p15.5 hCG_28329
						HBG2	OMIM HGNC sv prd lev mm hm sts	SNP		best RefSeq 11p15.5 hemoglobin, gamma G
						HBE1	OMIM HGNC sv prd lev mm hm sts	SNP		best RefSeq 11p15.5 hCG_1640999



PubMed Nucleotide Protein Genome Structure PopSet Taxonomy OMIM Books SNP

Search for SNP on NCBI Reference Assembly

Search Entrez for

BUILD 132

Have a question about dbSNP? Try searching the SNP FAQ Archive!

GENERAL

HUMAN VARIATION

Search, Annotate, Submit NEW

Annotate and Submit Batch Data with Clinical Impact NEW

SNP SUBMISSION

DOCUMENTATION

SEARCH

RELATED SITES

SNP linked to Gene HBB(geneID:3043) Via Contig Annotation

rs# on all gene models to Batch Query all rs# to file.

Gene Model (mRNA alignment) information from genome sequence

Total gene model (contig mRNA transcript):				1		
mRNA	transcript	protein	mRNA orientation	Contig	Contig Label	List SNP
NM_000518.4	plus strand	NP_000509.1	forward	NT_009237.18	GRCh37	<- currently shown

Include clinically associated in gene region cSNP has frequency double hit

gene model	Contig Label	Contig	mRNA	protein	mRNA orientation	transcript	snp count
(contig mRNA transcript):	GRCh37	NT_009237.18	NM_000518.4	NP_000509.1	forward	plus strand	15, coding

Region	Chr. position	mRNA pos	dbSNP rs# cluster id	Heterozygosity	Validation	3D	Linkout	Function	dbSNP allele	Protein residue	Codon pos	Amino acid pos	PubMed
	5246854	465	rs41405449	N.D.		Yes		frame shift			1	139	
								frame shift	AGC	Ser [S]	1	139	
								contig reference	GCTA	[AN]	1	139	
	5246870	452	rs113082294	N.D.		Yes		synonymous	C	Val [V]	3	134	
								contig reference	G	Val [V]	3	134	
	5246876	430	rs41511744	N.D.		Yes		frame shift			2	127	
								frame shift (15bp)	[QAAYQ]		2	127	
								contig reference (17bp)	[VQAAYQ]		2	127	
	5246958	365	rs71811954	N.D.				frame shift			3	105	
								frame shift (15bp)			3	105	
								contig reference	G	Arg [R]	3	105	
	5247800	309	rs41539866	N.D.				frame shift			1	87	
								frame shift (17bp)			1	87	
								contig reference	G	Ala [A]	1	87	
	5247855	317	rs11549405	N.D.		Yes		synonymous	C	Leu [L]	3	89	
								contig reference	G	Leu [L]	3	89	
	5247878	294	rs11549406	0.005		Yes		missense	G	Val [V]	1	82	
								contig reference	C	Leu [L]	1	82	
	5247915	257	rs112287010	N.D.		Yes		synonymous	T	Leu [L]	3	69	
								contig reference	C	Leu [L]	3	69	
	5247969	203	rs17850156	N.D.		Yes		synonymous	C	Thr [T]	3	51	
								contig					

HBB Variation Viewer [Download report](#) (86948 bytes)

Gene	HBB; hemoglobin, beta
Description	beta globin chain hemoglobin beta chain hemoglobin subunit beta Also known as: CD113t-C, beta-globin
Species	Homo sapiens
Cyto	11p15.5

Gene Reference Sequences	NG_000007.3 genomic NM_000518.4 transcript NP_000509.1 protein <i>variation locations are based on these acc</i>
Links	HGMD , Panther , Gene , OMIM

Observed Variation | Page 3 of 45 | Displaying results 41 - 60 of 883

Var Class	Genomic	Transcript	Protein	Clinical interpretation	Test status	Freq	Pub...	MIM AI Var	Orig
SNC	g.70610C>G	c.16C>G	p.Pro6Ala			5	5			141900.0451	G
SNC	g.70611C>G	c.17C>G	p.Pro6Arg			3	3			141900.0296	G
DIP	g.70612delT	c.18delT	p.Pro6=fs			2	2				
DIP	g.70612delTins...	c.18delTinsCT	p.Pro6?fs			2	2				
SNC	g.70613G>A	c.19G>A	p.Glu7Lys	probable-pathogenic		18	18		1	141900.0010...	G
SNC	g.70613G>C	c.19G>C	p.Glu7Gln	probable-pathogenic		18	18		1	141900.0010...	G
MIX	g.70614A>C	c.20A>C	p.Glu7Ala			34	34	Q	8	141900.0039...	
MIX	g.70614A>T	c.20A>T	p.Glu7Val			34	34	Q	8	141900.0039...	
DIP	g.70614delA	c.20delA	p.Glu7Glyfs			1	1				
SNC	g.70614A>G	c.20A>G	p.Glu7Gly	pathogenic		7	7	Q		141900.0085...	G
MIX	g.70614delA	c.20delA	p.Glu7Glyfs			34	34	Q	8	141900.0039...	
MIX	g.70614A>C	c.20A>C	p.Glu7Ala			34	34	Q	8	141900.0039...	
MIX	g.70614A>G	c.20A>G	p.Glu7Gly			34	34	Q	8	141900.0039...	
MIX	g.70614A>T	c.20A>T	p.Glu7Val			34	34	Q	8	141900.0039...	
SNC	g.70614A>T	c.20A>T	p.Glu7Val	pathogenic		7	7	Q		141900.0085...	G
DIP	g.70616_7061...	c.22_24delGAG	p.Glu8delGlu			1	1			141900.0156	

Ensembl Home Page

<http://www.ensembl.org/>

Do you want Google Chrome to save your password?

Never for this site Save password

Search: for

e.g. **BRCA2** or **rat X:100000..200000** or **coronary heart disease**

Browse a Genome

The Ensembl project produces genome databases for vertebrates and other eukaryotic species, and makes this information freely available online.

Favourite genomes



Human
GRCh37



Mouse
GRCm38



Zebrafish
Zv9

★ [Change favourites](#)

All genomes

-- Select a species --

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Other species are available in [Ensembl Pre!](#) and [EnsemblGenomes](#)

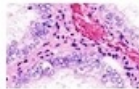
ENCODE data in Ensembl



Variant Effect Predictor



Gene expression in different tissues



Find SNPs and other variants for my gene

```
GTATACATTC
CRTRAAAGTCTT
CTTCTAAATTCT
GRBACATTTCC
```

Retrieve gene sequence

```
GCTGACTTCCGGTGG:
GGGCTTGTGGCGGAGC:
GCGCCTCTGCTGGGCT:
AGGGGACAGATTTGTG:
CACCTCTGGAGCGGTT:
CCCACTCCAGCGTGGCG:
```

Compare genes across species



Use my own data in Ensembl



Learn about a disease or phenotype



What's New in Release 74 (December 2013)

- [ncRNA secondary structure now displayed on the Gene Summary page](#)
- [New matrix configuration for RNASeq models](#)
- [New species: sheep \(*Ovis aries*\), cave fish \(*Astyanax mexicanus*\) and spotted gar \(*Lepisosteus oculatus*\)](#)
- [Updated patches for the human assembly \(GRCh37.p13\) and mouse assembly \(GRCm38.p2\)](#)

[Full details of this release](#)

[More release news on our blog →](#)

Latest blog posts

- 21 Jan 2014: [A GeneBuilder's Perspective on the New Human Genome Assembly](#)
- 14 Jan 2014: [Ensembl workshop at the Avian Model Systems meeting](#)
- 13 Jan 2014: [Retirement of archive 61](#)

[Go to Ensembl blog →](#)

Did you know...?



Come to our [YouTube](#) channel for tutorial videos.

Human (GRCh37) ▾



Human

Homo sapiens

Search all categories ▾ Search Human...

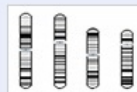
e.g. [BRCA2](#) or [6:133017695-133161157](#) or [osteoarthritis](#)

Genome assembly: GRCh37 (GCA_000001405.13)

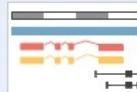
- More information and statistics
- Download DNA sequence (FASTA)
- Convert your data to GRCh37 coordinates
- Display your data in Ensembl

Other assemblies

- [NCBI36](#) (Ensembl release 54)



View karyotype



Example region

Comparative genomics

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

- More about comparative analysis
- Download alignments (EMF)



Example gene tree

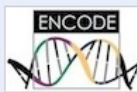
Regulation

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
- Download all regulatory features (GFF)



Example regulatory feature



ENCODE data in Ensembl

What's New in Human release 73

- Update to Ensembl-Havana GENCODE gene set (release 18)
- Human: assembly updated to GRCh37.p12
- Human: updated RefSeq gene import

[More news...](#)

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 genes, cDNAs, ncRNA, proteins (FASTA)
- Update your old Ensembl IDs

Additional manual annotation can be found in Vega



Example gene



Example transcript

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



Example variant



Example phenotype



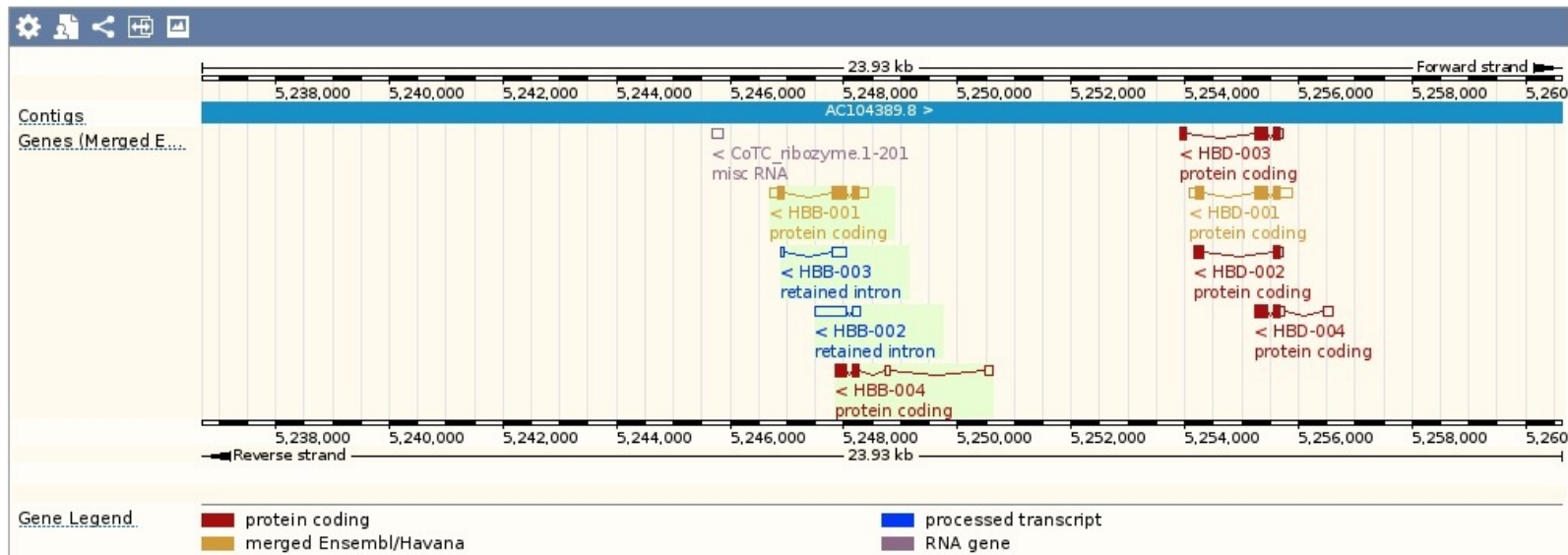
Example structural variant

Description hemoglobin, beta [Source:HGNC Symbol;Acc:4827]
Location [Chromosome 11: 5,246,694-5,250,625](#) reverse strand.
INSDC coordinates chromosome:GRCh37:CM000673.1:5246694:5250625:1
Transcripts This gene has 4 transcripts (splice variants) [Show transcript table](#)

Gene summary

Name [HBB](#) (HGNC Symbol)
Synonyms beta-globin, CD113t-C, HBD [To view all Ensembl genes linked to the name [click here.](#)]
CCDS This gene is a member of the Human CCDS set: [CCDS7753](#)
Ensembl version ENSG00000244734.1
Gene type Known protein coding
Prediction Method Annotation for this gene includes both automatic annotation from Ensembl and [Havana](#) manual curation, see [article](#).
Alternative genes **This gene corresponds to the following database identifiers:**
Havana gene: [OTTHUMG0000066678](#) (version 2)

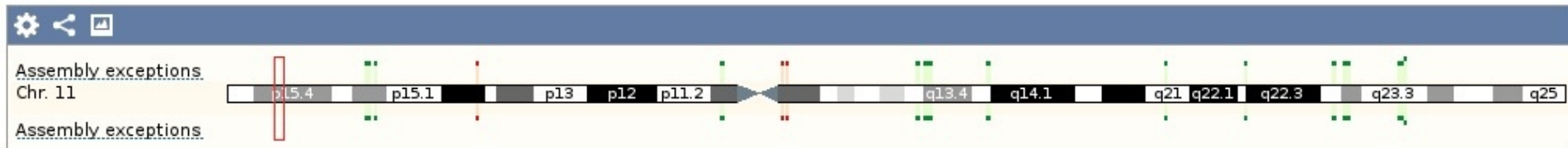
[Go to Region in Detail](#) for more tracks and navigation options (e.g. zooming)



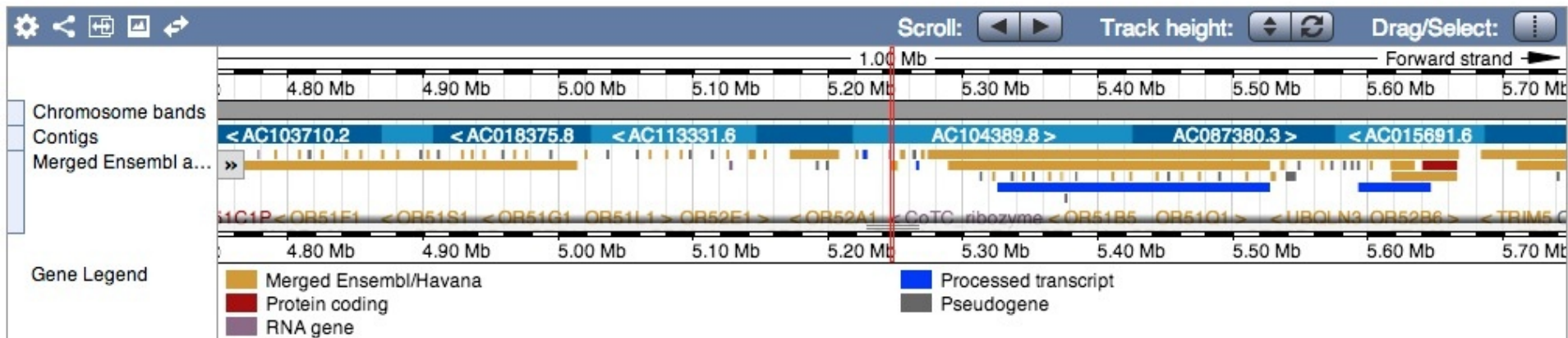
Ensembl Human HBB Gene

http://uswest.ensembl.org/Homo_sapiens/Gene/Summary?db=core;g=ENSG00000244734;r=11:5246694-5

Chromosome 11: 5,246,116-5,250,047



Region in detail



Location:

Gene:

Navigation controls: [Left] [Left] [Zoom In] [Slider] [Zoom Out] [Right] [Right]



Ensembl Human HBB Gene Transcripts

http://uswest.ensembl.org/Homo_sapiens/Gene/Summary?db=core;g=ENSG00000244734;r=11:5246694-5250625

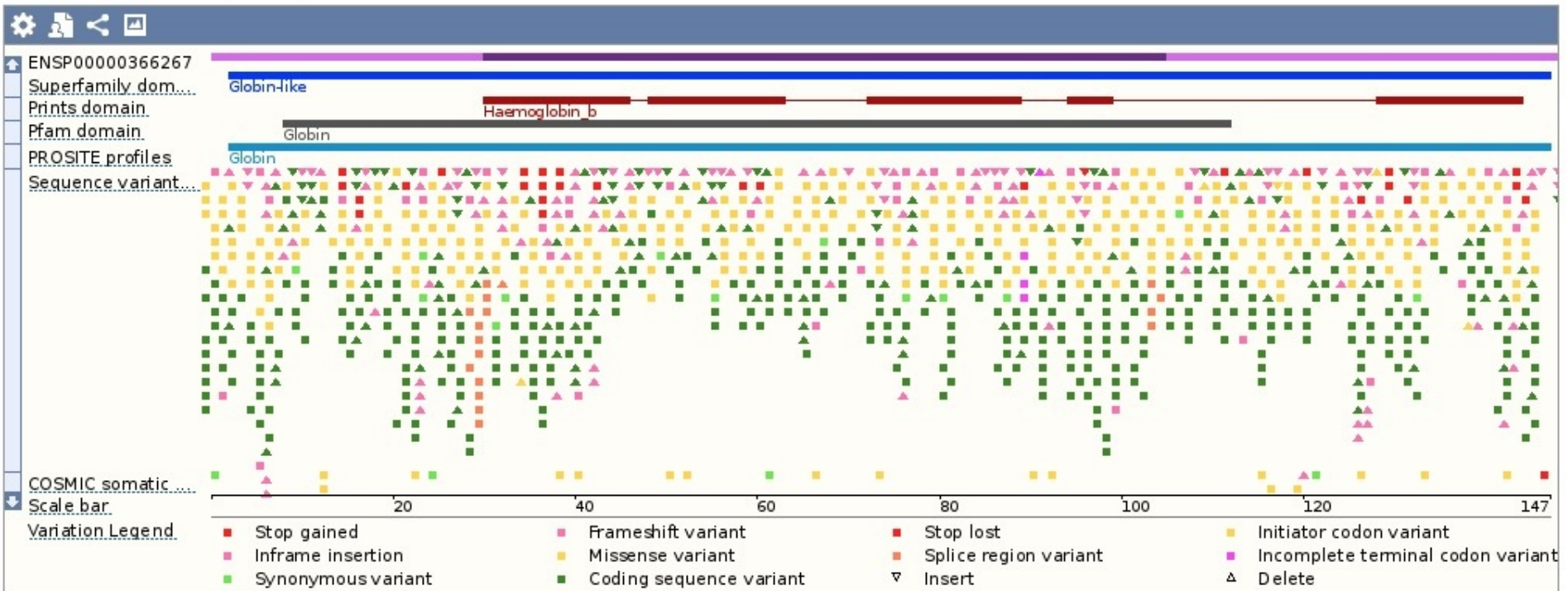


Ensembl Human HBB Protein Summary

http://uswest.ensembl.org/Homo_sapiens/Transcript/ProteinSummary?db=core;g=ENSG00000244734;r=11:52

Protein summary

Protein domains for ENSP00000333994.3



Statistics

Ave. residue weight: 108.833 g/mol
 Charge: 3.5
 Isoelectric point: 7.3230
 Molecular weight: 15,998.41 g/mol
 Number of residues: 147 aa



Ensembl Tutorials

<http://uswest.ensembl.org/info/website/tutorials/index.html>



Ensembl Tutorials and Worked Examples

Online tutorials

- [Introduction to Browsing Chordate Genomes](#) - a 3-hour comprehensive interactive tutorial aimed at beginners

Video tutorials

The tutorials listed below are Flash animations of some of our training presentations. We are gradually adding to the list, so please check back regularly.



Note that we are now hosting all our tutorials on [YouTube](#) (and [优酷网](#) for users in China) for ease of maintenance. A selection of tutorials is also available on the [EBI E-Video website](#).

Title	Running time (minutes)
The Ensembl Genome Browser	10:00
EnsemblGenomes - Extending Ensembl across the taxonomic space	4:54
Comparative Genomics	9:58
SNPs and other Variations - 1 of 2	7:06
SNPs and other Variations - 2 of 2	6:30
Introduction to BioMart	4.27
BioMart: Variation IDs to HGNC Symbols	2:58
Clip: Exons and Introns	1:12
Clip: Export Sequence	1:08
Clip: Genome Variation	0:37
Clip: Transcriptomics (ArrayExpress)	1:12
Clip: View Conserved Sequence	1:26
Clip: View External Data (DAS)	1:53

UCSC Genome Bioinformatics

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About the UCSC Genome Bioinformatics Site

Welcome to the UCSC Genome Browser website. This site contains the reference sequence and working draft assemblies for a large collection of genomes. It also provides portals to the [ENCODE](#) and [Neandertal](#) projects.

We encourage you to explore these sequences with our tools. The [Genome Browser](#) zooms and scrolls over chromosomes, showing the work of annotators worldwide. The [Gene Sorter](#) shows expression, homology and other information on groups of genes that can be related in many ways. [Blat](#) quickly maps your sequence to the genome. The [Table Browser](#) provides convenient access to the underlying database. [VisiGene](#) lets you browse through a large collection of *in situ* mouse and frog images to examine expression patterns. [Genome Graphs](#) allows you to upload and display genome-wide data sets.

The UCSC Genome Browser is developed and maintained by the Genome Bioinformatics Group, a cross-departmental team within the Center for Biomolecular Science and Engineering ([CBSE](#)) at the University of California Santa Cruz ([UCSC](#)). If you have feedback or questions concerning the tools or data on this website, feel free to contact us on our [public mailing list](#).



Genomes

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Human (*Homo sapiens*) Genome Browser Gateway

The UCSC Genome Browser was created by the [Genome Bioinformatics Group of UC Santa Cruz](#).
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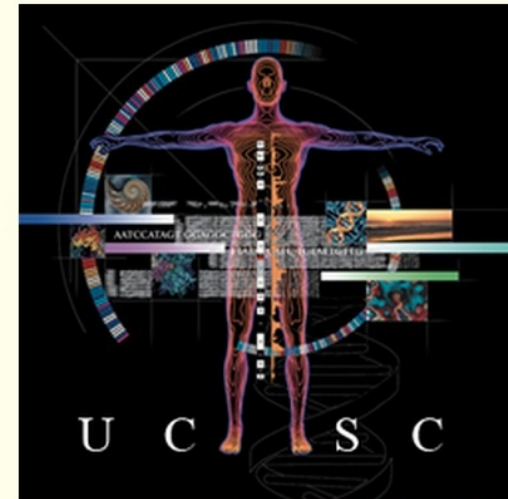
group	genome	assembly	position	search term
Mammal	Human	Feb. 2009 (GRCh37/hg19)	chr11:4,444,499-6,050,498	enter position, gene symbol or search term: <input type="button" value="submit"/>
Click here to reset the browser user interface settings to their defaults.				
<input type="button" value="track search"/> <input type="button" value="add custom tracks"/> <input type="button" value="track hubs"/> <input type="button" value="configure tracks and display"/>				

Human Genome Browser – hg19 assembly ([sequences](#))

The February 2009 human reference sequence (GRCh37) was produced by the [Genome Reference Consortium](#). For more information about this assembly, see [GRCh37](#) in the NCBI Assembly database.

Sample position queries

A genome position can be specified by the accession number of a sequenced genomic clone, an mRNA or EST or STS marker, a chromosomal coordinate range, or keywords from the GenBank description of an mRNA. The following list shows examples of valid position queries for the human genome. See the [User's Guide](#) for more information.



Homo sapiens
(Graphic courtesy of [CBSE](#))

UCSC Genes

[HBB \(uc001mae.1\) at chr11:5246696-5248301](#) - Homo sapiens hemoglobin, beta (HBB), mRNA.
[HBD \(uc001maf.1\) at chr11:5254059-5255858](#) - Homo sapiens hemoglobin, delta (HBD), mRNA.
[RBM17 \(uc010gav.2\) at chr10:6131309-6159422](#) - Homo sapiens RNA binding motif protein 17
[RBM17 \(uc001ijb.3\) at chr10:6130949-6159422](#) - Homo sapiens RNA binding motif protein 17
[HBA1 \(uc002cfx.1\) at chr16:226679-227520](#) - Homo sapiens hemoglobin, alpha 1 (HBA1), mRNA
[HBA2 \(uc002cfv.4\) at chr16:222846-223709](#) - Homo sapiens hemoglobin, alpha 2 (HBA2), mRNA
[HBBP1 \(uc001mag.3\) at chr11:5263185-5264822](#) - Homo sapiens hemoglobin, beta pseudogene 1
[TMEM158 \(uc011baf.2\) at chr3:45265956-45267814](#) - Homo sapiens transmembrane protein 158

RefSeq Genes

[HBB at chr11:5246696-5248301](#) - (NM_000518) hemoglobin subunit beta
[HBBP1 at chr11:5263185-5264822](#) - (NR_001589)

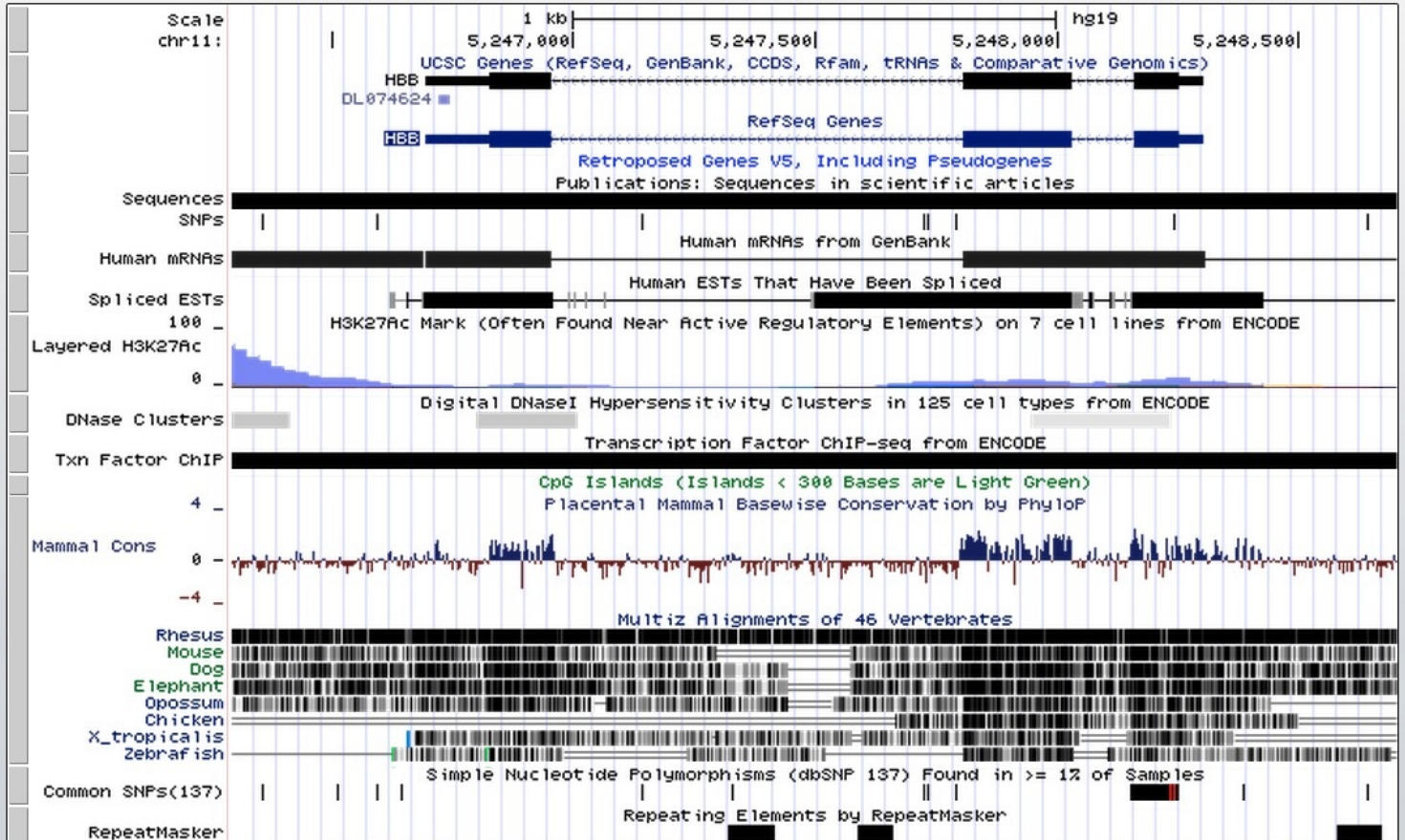
Non-Human RefSeq Genes

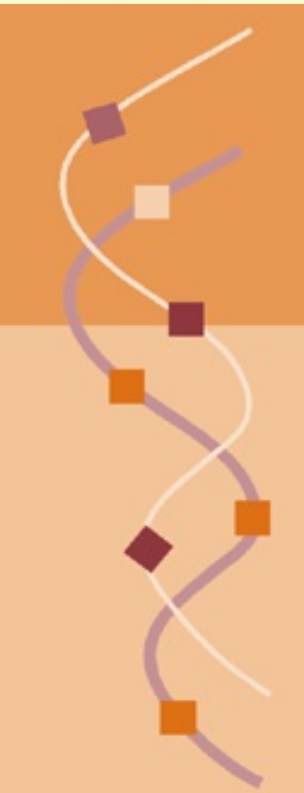
[hbb at chr11:5247810-5275746](#) - (NM_001201019) hemoglobin subunit beta
[HBB at chr11:5246810-5248260](#) - (NM_001164428) hemoglobin subunit beta
[HBB at chr11:5246828-5248251](#) - (NM_001168847) hemoglobin subunit beta
[HBB at chr11:5254194-5255663](#) - (NM_001164018) hemoglobin subunit beta
[HBB at chr11:5246828-5248251](#) - (NM_001164018) hemoglobin subunit beta
[HBB at chr11:5246828-5248301](#) - (NM_001144841) hemoglobin subunit beta

UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly

move <<< << < > >> >>> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x

chr11:5,246,295-5,248,703 2,409 bp.





Mapping and Sequencing Tracks

[refresh](#)

Base Position	Chromosome Band	STS Markers	18 FISH Clones	Recomb Rate	18 deCODE Recomb
<input type="button" value="dense"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>
<input checked="" type="checkbox"/> ENCODE Pilot	Map Contigs	Assembly	GRC Map Contigs	INSDC	Gap
<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>
BAC End Pairs	18 Fosmid End Pairs	GC Percent	GRC Patch Release	Hg18 Diff	GRC Incident
<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>
Hi Seq Depth	Wiki Track	<input checked="" type="checkbox"/> BU ORChID	<input checked="" type="checkbox"/> Mapability	Short Match	Restr Enzymes
<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>

Phenotype and Disease Associations

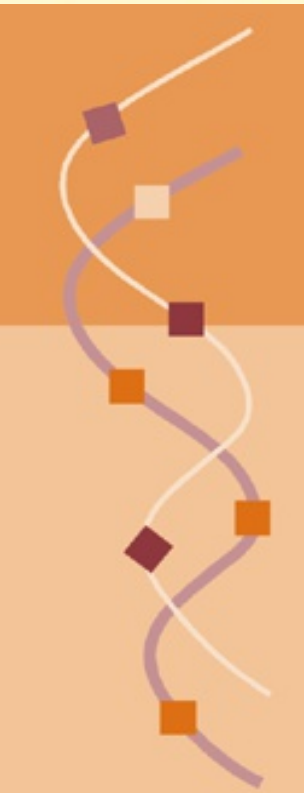
[refresh](#)

18 GAD View	DECIPHER	OMIM AV SNPs	OMIM Genes	OMIM Pheno Loci	COSMIC
<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>
LOVD Variants	HGMD Variants	UniProt Variants	GWAS Catalog	ISCA	Coriell CNVs
<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>
18 RGD Human QTL	18 RGD Rat QTL	18 MGI Mouse QTL	GeneReviews		
<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>		

Genes and Gene Prediction Tracks

[refresh](#)

UCSC Genes	GENCODE...	Old UCSC Genes	UCSC Alt Events	CCDS	RefSeq Genes
<input type="button" value="pack"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="pack"/>
Other RefSeq	MGC Genes	ORFeome Clones	TransMap...	Vega Genes	Pfam in UCSC Gene
<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>
Retroposed Genes	Ensembl Genes	AceView Genes	SIB Genes	N-SCAN	SGP Genes
<input type="button" value="pack"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>
Geneid Genes	Genscan Genes	Exoniphy	Yale Pseudo60	tRNA Genes	H-Inv 7.0
<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>	<input type="button" value="hide"/>
17 EvoFold	sno/miRNA	IKMC Genes Mapped	lincRNAs...		



mRNA and EST Tracks refresh

Human mRNAs <input type="button" value="dense"/>	Spliced ESTs <input type="button" value="dense"/>	Human ESTs <input type="button" value="hide"/>	Other mRNAs <input type="button" value="hide"/>	Other ESTs <input type="button" value="hide"/>	<input checked="" type="checkbox"/> H-Inv <input type="button" value="hide"/>
UniGene <input type="button" value="hide"/>	Gene Bounds <input type="button" value="hide"/>	SIB Alt-Splicing <input type="button" value="hide"/>	<input checked="" type="checkbox"/> Poly(A) <input type="button" value="hide"/>	PolyA-Seq <input type="button" value="hide"/>	<input checked="" type="checkbox"/> CGAP SAGE <input type="button" value="hide"/>
Human RNA Editing <input type="button" value="hide"/>					

Expression refresh

Affy Exon Array <input type="button" value="hide"/>	Affy GNF1H <input type="button" value="hide"/>	<input checked="" type="checkbox"/> Affy RNA Loc <input type="button" value="hide"/>	Affy U133 <input type="button" value="hide"/>	Affy U133Plus2 <input type="button" value="hide"/>	Affy U95 <input type="button" value="hide"/>
Allen Brain <input type="button" value="hide"/>	Burge RNA-seq <input type="button" value="hide"/>	<input checked="" type="checkbox"/> CSHL Small RNA-seq <input type="button" value="hide"/>	<input checked="" type="checkbox"/> ENC Exon Array... <input type="button" value="hide"/>	<input checked="" type="checkbox"/> ENC ProtGeno... <input type="button" value="hide"/>	<input checked="" type="checkbox"/> ENC RNA-seq... <input type="button" value="hide"/>
<input checked="" type="checkbox"/> GIS RNA PET <input type="button" value="hide"/>	GNF Atlas 2 <input type="button" value="hide"/>	<input checked="" type="checkbox"/> Illumina WG-6 <input type="button" value="hide"/>	qPCR Primers <input type="button" value="hide"/>	<input checked="" type="checkbox"/> RIKEN CAGE Loc <input type="button" value="hide"/>	<input checked="" type="checkbox"/> Sestan Brain <input type="button" value="hide"/>

Regulation refresh

<input checked="" type="checkbox"/> ENCODE Regulation... <input type="button" value="show"/>	<input checked="" type="checkbox"/> CD34 Dnase1 <input type="button" value="hide"/>	CpG Islands <input type="button" value="pack"/>	<input checked="" type="checkbox"/> ENC Chromatin... <input type="button" value="hide"/>	<input checked="" type="checkbox"/> ENC DNA Methyl... <input type="button" value="hide"/>	<input checked="" type="checkbox"/> ENC DNase/FAIRE... <input type="button" value="hide"/>
<input checked="" type="checkbox"/> ENC Histone... <input type="button" value="hide"/>	<input checked="" type="checkbox"/> ENC RNA Binding... <input type="button" value="hide"/>	<input checked="" type="checkbox"/> ENC TF Binding... <input type="button" value="hide"/>	<input checked="" type="checkbox"/> FSU Repli-chip <input type="button" value="hide"/>	<input checked="" type="checkbox"/> ORegAnno <input type="button" value="hide"/>	<input checked="" type="checkbox"/> Stanf Nucleosome <input type="button" value="hide"/>
<input checked="" type="checkbox"/> SUNY SwitchGear <input type="button" value="hide"/>	<input checked="" type="checkbox"/> SwitchGear TSS <input type="button" value="hide"/>	TFBS Conserved <input type="button" value="hide"/>	TS miRNA sites <input type="button" value="hide"/>	UMMS Brain Hist <input type="button" value="hide"/>	<input checked="" type="checkbox"/> UW Repli-seq <input type="button" value="hide"/>
Vista Enhancers <input type="button" value="hide"/>	<input checked="" type="checkbox"/> NKI Nuc Lamina... <input type="button" value="hide"/>	<input checked="" type="checkbox"/> UCSF Brain Methyl <input type="button" value="hide"/>			

Comparative Genomics

Conservation full ▾	18 Cons Indels MmCf hide ▾	GERP hide ▾	18 Evo Cpg hide ▾	Primate Chain/Net hide ▾	Placental Chain/Net hide ▾
Vertebrate Chain/Net hide ▾					

Neandertal Assembly and Analysis

18 H-C Coding Diffs hide ▾	18 Sel Swp Scan (S) hide ▾	18 5% Lowest S hide ▾	18 S SNPs hide ▾	18 Cand. Gene Flow [No data-chr11]	Neandertal Seq hide ▾
18 Neandertal Mito [No data-chr11]					

Denisova Assembly and Analysis

Variation and Repeats

Common SNPs(137) dense ▾	Flagged SNPs(137) hide ▾	Mult. SNPs(137) hide ▾	All SNPs(137) hide ▾	Common SNPs(135) hide ▾	Flagged SNPs(135) hide ▾
Mult. SNPs(135) hide ▾	All SNPs(135) hide ▾	Common SNPs(132) hide ▾	Flagged SNPs(132) hide ▾	Mult. SNPs(132) hide ▾	All SNPs(132) hide ▾
SNPs (131) hide ▾	1000G Ph1 Vars hide ▾	1000G Ph1 Accsbl hide ▾	<input checked="" type="checkbox"/> GIS DNA PET hide ▾	<input checked="" type="checkbox"/> HAIB Genotype hide ▾	SNP/CNV Arrays hide ▾
HGDP Allele Freq hide ▾	18 HapMap SNPs hide ▾	DGV Struct Var hide ▾	Segmental Dups hide ▾	RepeatMasker dense ▾	Interrupted Rpts hide ▾
Simple Repeats hide ▾	Microsatellite hide ▾	Self Chain hide ▾	18 Genome Variants hide ▾	NumtS Sequence hide ▾	

UCSC Genome Bioinformatics

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[Session](#) - [FAQ](#)

Genome Browser User Guide

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Search the Genome Browser help pages:

Search the Genome mailing list archives:

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[Questions and feedback are welcome.](#)

What does the Genome Browser do?

Ensembl Human Genome

http://uswest.ensembl.org/Homo_sapiens/Info/Index/

Human (GRCh37) ▾



Human

Homo sapiens

Search all categories ▾ opsin

e.g. **BRCA2** or **6:133017695-133161157** or **osteoarthritis**

Genome assembly: GRCh37 (GCA_000001405.14)

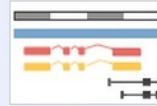
- More information and statistics
- Download DNA sequence (FASTA)
- Convert your data to GRCh37 coordinates
- Display your data in Ensembl

Other assemblies

- NCBI36 (Ensembl release 54)



View karyotype



Example region

What's New in Human release 74

- New dbSNP imports
- Update to Ensembl-Havana GENCODE gene set (release 19)
- Human: assembly updated to GRCh37.p13

[More news...](#)

Gene annotation

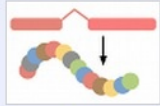
What can I find? Protein-coding and non-coding genes, splice variants, cDNA and protein sequences, non-coding RNAs.

- More about this genebuild
- Download genes, cDNAs, ncRNA, proteins (FASTA)
- Update your old Ensembl IDs

Vega Additional manual annotation can be found in Vega



Example gene

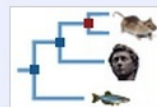


Example transcript

Comparative genomics

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

- More about comparative analysis
- Download alignments (EMF)



Example gene tree

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



Example variant



Example phenotype

Regulation



[Genome Browser](#)[ENCODE](#)[Neandertal](#)[Blat](#)[Table Browser](#)[Gene Sorter](#)[In Silico PCR](#)[Genome Graphs](#)[Galaxy](#)[VisiGene](#)[Proteome Browser](#)[Utilities](#)[Downloads](#)[Release Log](#)[Custom Tracks](#)

About the UCSC Genome Bioinformatics Site

Welcome to the UCSC Genome Browser website. This site contains the reference sequence and working draft assemblies for a large collection of genomes. It also provides portals to the [ENCODE](#) and [Neandertal](#) projects.

We encourage you to explore these sequences with our tools. The [Genome Browser](#) zooms and scrolls over chromosomes, showing the work of annotators worldwide. The [Gene Sorter](#) shows expression, homology and other information on groups of genes that can be related in many ways. [Blat](#) quickly maps your sequence to the genome. The [Table Browser](#) provides convenient access to the underlying database. [VisiGene](#) lets you browse through a large collection of *in situ* mouse and frog images to examine expression patterns. [Genome Graphs](#) allows you to upload and display genome-wide data sets.

The UCSC Genome Browser is developed and maintained by the Genome Bioinformatics Group, a cross-departmental team within the Center for Biomolecular Science and Engineering ([CBSE](#)) at the University of California Santa Cruz ([UCSC](#)). If you have feedback or questions concerning the tools or data on this website, feel free to contact us on our [public mailing list](#).

News

[News Archives](#) ►

To receive announcements of new genome assembly releases, new software features, updates and training seminars by email, subscribe to the [genome-announce](#) mailing list.

10 October 2011 - Updated Yeast Browser Released

We are happy to announce an updated Yeast Genome Browser for the initial release of *Saccharomyces cerevisiae*, sacCer3. The April 2011 *Saccharomyces cerevisiae* genome assembly (Saccharomyces cerevisiae S288c assembly from Saccharomyces Genome Database (GCA_000146055.2)) was produced by the [Saccharomyces Genome Database \(SGD™\)](#) project.

Chromosomes available in this assembly: chrI, chrII, chrIII, chrIV ... etc ... chrXVI, and chrM. See also: [SGD™ genome snapshot/overview](#).

Downloads of the yeast data and annotations may be obtained from the UCSC Genome Browser [FTP server](#) or [Downloads](#) page. The *S. cerevisiae* annotation tracks were generated by UCSC and collaborators worldwide.

We'd like to thank the Saccharomyces Genome Database (SGD™). The *S. cerevisiae* Genome Browser and annotation tracks were produced by Hiram Clawson, Greg Roe, and Steve Heitner. See the [Credits](#) page for a detailed list of the organizations and individuals who contributed to this release.

Human (*Homo sapiens*) Genome Browser Gateway

The UCSC Genome Browser was created by the [Genome Bioinformatics Group of UC Santa Cruz](#).
Software Copyright (c) The Regents of the University of California. All rights reserved.

clade	genome	assembly	position or search term	gene	
Mammal ▾	Human ▾	Feb. 2009 (GRCh37/hg19) ▾	opsin		submit
Click here to reset the browser user interface settings to their defaults.					
track search		add custom tracks		track hubs	
configure tracks and display			clear position		

About the Human Feb. 2009 (GRCh37/hg19) assembly ([sequences](#))

The February 2009 human reference sequence (GRCh37) was produced by the [Genome Reference Consortium](#).

Sample position queries

A genome position can be specified by the accession number of a sequenced genomic clone, an mRNA or EST or STS marker, a chromosomal coordinate range, or keywords from the GenBank description of an mRNA. The following list shows examples of valid position queries for the human genome. See the [User's Guide](#) for more information.

Request:**Genome Browser Response:**

chr7	Displays all of chromosome 7
chrUn_gl000212	Displays all of the unplaced contig gl000212
chr3:1-1000000	Displays first million bases of chr 3, counting from p-arm telomere
chr3:1000000+2000	Displays a region of chr3 that spans 2000 bases, starting with position 1000000
RH18061;RH80175	Displays region between genome landmarks, such as the STS markers RH18061 and RH80175. This syntax may also be used for other range queries, such as between uniquely determined ESTs, mRNAs, refSeqs, etc.



Homo sapiens
(Graphic courtesy of [CBSE](#))

[OPN4 \(uc010gmk.1\) at chr10:88414314-88426605](#) - opsin 4 isoform 2
[OPN4 \(uc009xss.1\) at chr10:88420233-88423870](#) - opsin 4 isoform 1
[OPN1MW \(uc004fkd.2\) at chrX:153485203-153499469](#) - opsin 1 (cone pigments), medium-wave-sensitive
[OPN1MW \(uc004fkb.2\) at chrX:153448085-153462351](#) - opsin 1 (cone pigments), medium-wave-sensitive
[OPN1LW \(uc004fjz.3\) at chrX:153409725-153424505](#) - opsin 1 (cone pigments), long-wave-sensitive
[OPN1SW \(uc003vnt.3\) at chr7:128412545-128415844](#) - opsin 1 (cone pigments), short-wave-sensitive
[OPN5 \(uc003ozd.2\) at chr6:47754862-47794114](#) - opsin 5 isoform 2
[OPN5 \(uc003ozc.2\) at chr6:47749798-47794114](#) - opsin 5 isoform 1
[OPN4 \(uc001kdq.2\) at chr10:88414314-88426214](#) - opsin 4 isoform 1
[OPN4 \(uc001kdp.2\) at chr10:88414314-88426214](#) - opsin 4 isoform 2
[OPN3 \(uc001hza.2\) at chr1:241756453-241803701](#) - opsin 3
[RGR \(uc001kde.1\) at chr10:86004809-86018944](#) - retinal G-protein coupled receptor isoform 3
[RGR \(uc001kdd.1\) at chr10:86004809-86018944](#) - retinal G-protein coupled receptor isoform 1
[RGR \(uc001kdc.1\) at chr10:86004809-86018944](#) - retinal G-protein coupled receptor isoform 2
[VSX1 \(uc010gde.1\) at chr20:25052132-25062767](#) - visual system homeobox 1 isoform b
[VSX1 \(uc002wue.2\) at chr20:25052132-25062767](#) - visual system homeobox 1 isoform b
[VSX1 \(uc002wug.1\) at chr20:25059195-25062767](#) - visual system homeobox 1 isoform b
[VSX1 \(uc010gdf.1\) at chr20:25052132-25062767](#) - visual system homeobox 1 isoform b
[VSX1 \(uc010gdd.1\) at chr20:25052132-25062767](#) - visual system homeobox 1 isoform b
[VSX1 \(uc002wuf.2\) at chr20:25056100-25062767](#) - visual system homeobox 1 isoform a
[CRX \(uc002phq.3\) at chr19:48325099-48346584](#) - cone-rod homeobox protein
[RHO \(uc003emt.2\) at chr3:129247482-129254186](#) - rhodopsin
[RRH \(uc003hzv.2\) at chr4:110749150-110765859](#) - peropsin
[ARR3 \(uc004dyb.2\) at chrX:69488185-69501690](#) - arrestin 3, retinal (X-arrestin)
[GRK7 \(uc011bnd.1\) at chr3:141497043-141535890](#) - G-protein-coupled receptor kinase 7 precursor

RefSeq Genes

[OPN1MW at chrX:153485203-153499470](#) - (NM_000513) medium-wave-sensitive opsin 1
[OPN1MW at chrX:153448085-153462352](#) - (NM_000513) medium-wave-sensitive opsin 1
[OPN5 at chr6:47749775-47794116](#) - (NM_181744) opsin-5
[OPN1SW at chr7:128412543-128415844](#) - (NM_001708) short-wave-sensitive opsin 1
[OPN1LW at chrX:153409725-153424507](#) - (NM_020061) long-wave-sensitive opsin 1
[OPN1MW2 at chrX:153485203-153498755](#) - (NM_001048181) medium-wave-sensitive opsin 1
[OPN1MW2 at chrX:153448085-153461637](#) - (NM_001048181) medium-wave-sensitive opsin 1
[OPN3 at chr1:241756452-241803701](#) - (NM_014322) opsin-3

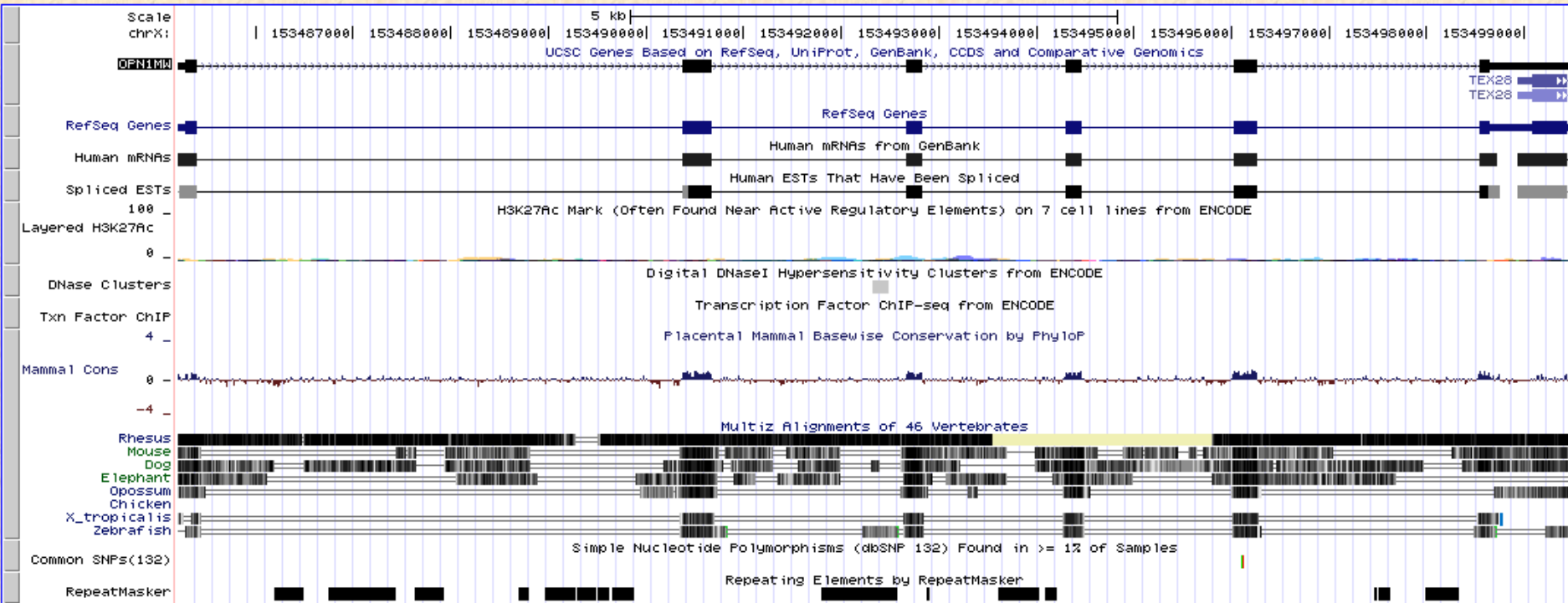
Non-Human RefSeq Genes

[OPN5 at chr6:47749803-47779473](#) - (NM_001206080) opsin-5
[OPN1SW at chr7:128413703-128415835](#) - (NM_001076704) blue-sensitive opsin
[LOC751972 at chr3:129247577-129252528](#) - (NM_001076696) RH2 opsin

UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly

move <<< << < > >> >>> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x

position/search chrX:153,485,203-153,499,469: jump clear size 14,267 bp.

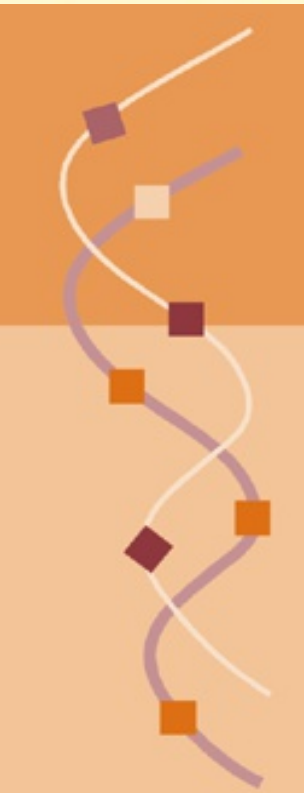


move start < 2.0 >

Click on a feature for details. Click or drag in the base position track to zoom in. Click side bars for track options. Drag side bars or labels up or down to reorder tracks. Drag tracks left or right to new position.

move end < 2.0 >





http

collapse all expand all
 Use drop-down controls below and press refresh to alter tracks displayed.
 Tracks with lots of items will automatically be displayed in more compact modes.

Mapping and Sequencing Tracks refresh

Base Position dense ▾	Chromosome Band hide ▾	STS Markers hide ▾	18 FISH Clones hide ▾	Recomb Rate hide ▾	Map Contigs hide ▾
Assembly hide ▾	GRC Map Contigs hide ▾	Gap hide ▾	BAC End Pairs hide ▾	18 Fosmid End Pairs hide ▾	GC Percent hide ▾
GRC Patch Release [No data-chrX]	Hg18 Diff hide ▾	NCBI Incident hide ▾	Short Match hide ▾	Restr Enzymes hide ▾	Wiki Track hide ▾
<input checked="" type="checkbox"/> BU ORChID hide ▾	<input checked="" type="checkbox"/> Mapability hide ▾				

983641&known

Phenotype and Disease Associations refresh

18 GAD View hide ▾	DECIPHER hide ▾	OMIM AV SNPs hide ▾	OMIM Genes hide ▾	OMIM Pheno Loci hide ▾	GWAS Catalog hide ▾
18 RGD Human QTL hide ▾	18 RGD Rat QTL hide ▾	18 MGI Mouse QTL hide ▾			

Genes and Gene Prediction Tracks refresh

UCSC Genes pack ▾	Alt Events hide ▾	<input checked="" type="checkbox"/> GENCODE Genes V4 hide ▾	<input checked="" type="checkbox"/> GENCODE Genes V7 hide ▾	CCDS hide ▾	RefSeq Genes dense ▾
Other RefSeq hide ▾	MGC Genes hide ▾	ORFeome Clones hide ▾	TransMap... hide ▾	Vega Genes hide ▾	Ensembl Genes hide ▾
AceView Genes hide ▾	N-SCAN hide ▾	SGP Genes hide ▾	Geneid Genes hide ▾	Genscan Genes hide ▾	Exoniphy hide ▾
tRNA Genes hide ▾	H-Inv 7.0 hide ▾	17 EvoFold hide ▾	sno/miRNA hide ▾	IKMC Genes Mapped hide ▾	

mRNA and EST Tracks refresh

Human mRNAs dense ▾	Spliced ESTs dense ▾	Human ESTs hide ▾	Other mRNAs hide ▾	Other ESTs hide ▾	18 H-Inv hide ▾
Gene Bounds hide ▾	18 SIB Alt-Splicing hide ▾	18 Poly(A) hide ▾	18 CGAP SAGE hide ▾	Human RNA Editing hide ▾	

Expression refresh

Affy Exon Array hide ▾	Affy GNF1H hide ▾	<input checked="" type="checkbox"/> Affy RNA Loc hide ▾	Affy U133 hide ▾	Affy U133Plus2 hide ▾	Affy U95 hide ▾
Allen Brain hide ▾	Burge RNA-seq hide ▾	<input checked="" type="checkbox"/> ENC Exon Array... hide ▾	<input checked="" type="checkbox"/> ENC ProtGeno... hide ▾	<input checked="" type="checkbox"/> ENC RNA-seq... hide ▾	GNF Atlas 2 hide ▾
18 Illumina WG-6	18 Sestan Brain				

UCSC Genome Bioinformatics

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What does the Genome Browser do?

Gene

Gene

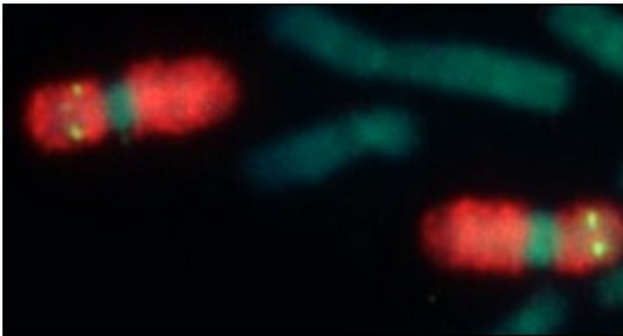
human opsin



Search

[Advanced](#)

[Help](#)



Gene

Gene integrates information from a wide range of species. A record may include nomenclature, Reference Sequences (RefSeqs), maps, pathways, variations, phenotypes, and links to genome-, phenotype-, and locus-specific resources worldwide.

Using Gene

[Gene Quick Start](#)

[FAQ](#)

[Download/FTP](#)

[RefSeq Mailing List](#)

[Gene News](#) 

[Factsheet](#)

Gene Tools

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Other Resources

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[OMIM](#)

[RefSeq](#)

[RefSeqGene](#)

[UniGene](#)

[Protein Clusters](#)



NCBI Gene: Human Opsin

<http://www.ncbi.nlm.nih.gov/gene/?term=human+opsin>

[NCBI](#)
[Resources](#)
[How To](#)

[brutlag](#)
[My NCBI](#)
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Gene

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[Show additional filters](#)

Display Settings: Tabular, 20 per page, Sorted by Relevance
 Send to:

Filters: [Manage Filters](#)

[<< First](#)
[< Prev](#)
 Page of 15
 [Next >](#)
[Last >>](#)

Results: 1 to 20 of 300

- Gene sources**
- Genomic
- Categories**
- Alternatively spliced
- NEWENTRY
- Protein-coding
- Pseudogene
- Sequence content**
- CCDS
- Ensembl
- RefSeq
- RefSeqGene

Status

Current only

Chromos... locations

Select ...

[Clear all](#)

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Name/Gene ID	Description	Location	Aliases
<input type="checkbox"/> RHO ID: 6010	rhodopsin [<i>Homo sapiens</i> (human)]	Chromosome 3, NC_000003.11 (129247482..129254187)	CSNBAD OPN2, R
<input type="checkbox"/> OPN1LW ID: 5956	opsin 1 (cone pigments), long-wave-sensitive [<i>Homo sapiens</i> (human)]	Chromosome X, NC_000023.10 (153409725..153424507)	hCG_413 CBBM, CBP, CO RCP, RO
<input type="checkbox"/> OPN1SW ID: 611	opsin 1 (cone pigments), short-wave-sensitive [<i>Homo sapiens</i> (human)]	Chromosome 7, NC_000007.13 (128412543..128415844, complement)	BCP, BO CBT
<input type="checkbox"/> OPN4 ID: 94233	opsin 4 [<i>Homo sapiens</i> (human)]	Chromosome 10, NC_000010.10 (88414314..88426217)	MOP
<input type="checkbox"/> OPN1MW ID: 2652	opsin 1 (cone pigments), medium-wave-sensitive [<i>Homo sapiens</i> (human)]	Chromosome X, NC_000023.10 (153448085..153462352)	CBBM, CBD, COD5, GCP, GO OPN1MV

Top Organisms [Tree]

- Mus musculus (29)
- Homo sapiens (27)
- Rattus norvegicus (8)
- Bos taurus (8)
- Pan troglodytes (6)
- All other taxa (222)

[More...](#)

Find related data

Database:

Search details

("Homo sapiens"
 [Organism] OR human[All
 Fields]) AND opsin[All
 Fields]

[See more...](#)

Recent activity

OPN1MW opsin 1 (cone pigments), medium-wave-sensitive [*Homo sapiens* (human)]

Gene ID: 2652, updated on 15-Jan-2014

Summary

Official Symbol	OPN1MW <small>provided by HGNC</small>
Official Full Name	opsin 1 (cone pigments), medium-wave-sensitive <small>provided by HGNC</small>
Primary source	HGNC:4206
See related	HPRD:02365 ; MIM:300821
Gene type	protein coding
RefSeq status	REVIEWED
Organism	Homo sapiens
Lineage	Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo
Also known as	CBD; GCP; GOP; CBBM; COD5; OPN1MW1

Summary

This gene encodes for a light absorbing visual pigment of the opsin gene family. The encoded protein is called green cone photopigment or medium-wavelength sensitive opsin. Opsins are G-protein coupled receptors with seven transmembrane domains, an N-terminal extracellular domain, and a C-terminal cytoplasmic domain. The long-wavelength opsin gene and multiple copies of the medium-wavelength opsin gene are tandemly arrayed on the X chromosome and frequent unequal recombination and gene conversion may occur between these sequences. X chromosomes may have fusions of the medium- and long-wavelength opsin genes or may have more than one copy of these genes. Defects in this gene are the cause of deutanopic colorblindness. [provided by RefSeq, Mar 2009]

Table of contents

- Summary
- Genomic context
- Genomic regions, transcripts, and products
- Bibliography
- Phenotypes
- Variation
- Pathways
- General gene information
 - Markers, Clone Names
- General protein information
- Reference sequences
- Related sequences
- Additional links
 - Locus-specific Databases

Related information

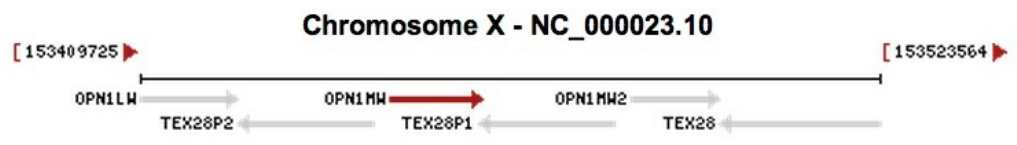
- Order cDNA clone
- BioAssay
- BioAssay by Target (List)
- BioAssay by Target (Summary)
- BioAssay, RNAi Target, Active
- BioProjects
- BioSystems
- Books
- CCDS

NCBI Gene: Human Opsin OPN1MW

<http://www.ncbi.nlm.nih.gov/gene/2652>

Genomic context

Location: Xq28 See OPN1MW in [Epigenomics](#), [MapViewer](#)
Sequence: Chromosome: X; NC_000023.10
 (153448085..153462352)

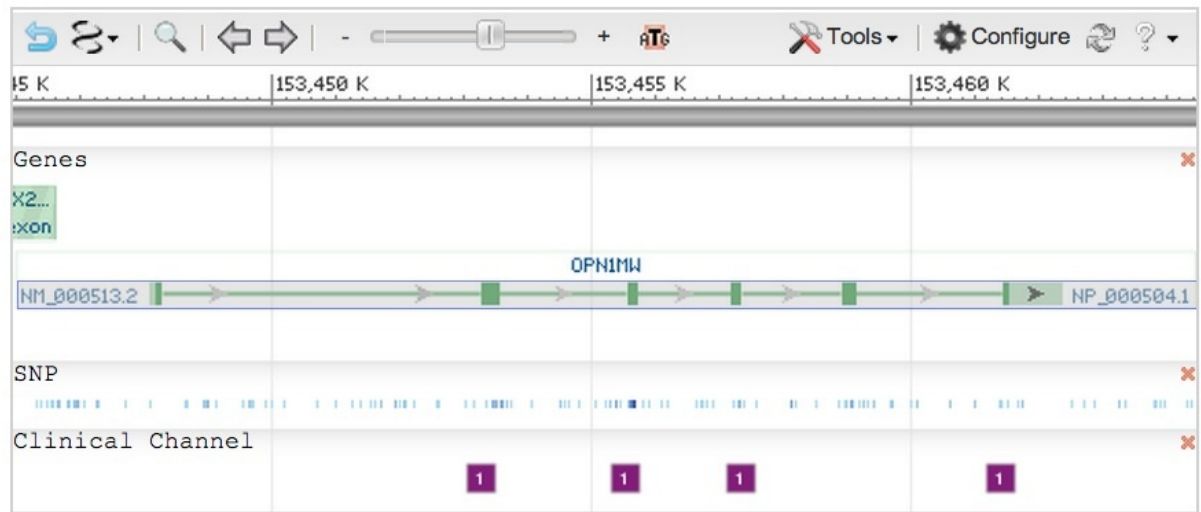


Genomic regions, transcripts, and products

Genomic Sequence

[Go to reference sequence details](#)

[Go to nucleotide](#) [Graphics](#) [FASTA](#) [GenBank](#)



15 K | 153,450 K | 153,455 K | 153,460 K

Genes

X2...
:exon

OPN1MW

NM_000513.2 | NP_000504.1

SNP

Clinical Channel

1 1 1 1

- [ClinVar](#)
- [Conserved Domains](#)
- [dbVar](#)
- [Full text in PMC](#)
- [Full text in PMC_nucleotide](#)
- [Gene neighbors](#)
- [Genome](#)
- [GEO Profiles](#)
- [GTR](#)
- [HomoloGene](#)
- [Map Viewer](#)
- [MedGen](#)
- [Nucleotide](#)
- [OMIM](#)
- [Probe](#)
- [Protein](#)
- [PubChem Substance](#)
- [PubMed](#)
- [PubMed \(GeneRIF\)](#)
- [PubMed \(OMIM\)](#)
- [PubMed\(nucleotide/PMC\)](#)
- [RefSeq Proteins](#)
- [RefSeq RNAs](#)
- [RefSeqGene](#)
- [SNP](#)

MapView: Human Opsin OPN1MW

[http://www.ncbi.nlm.nih.gov/mapview/maps.cgi?TAXID=9606&QSTR=2652\[gene_id\]&QUERY=uid\(-2121809236,-2146](http://www.ncbi.nlm.nih.gov/mapview/maps.cgi?TAXID=9606&QSTR=2652[gene_id]&QUERY=uid(-2121809236,-2146)

Human genome overview page (Annotation Release 105)
Human genome overview page (Build 36.3)
[Map Viewer Home](#)
Map Viewer Help
Human Maps Help
FTP
Data As Table View
[Maps & Options](#)
Region Shown:
153,378K
153,533K

You are here:

 default
 master

NCBI Map Viewer

PubMed Entrez BLAST OMIM Taxonomy Structure

Search

[Homo sapiens \(human\) Annotation Release 105 \(Current\)](#) [BLAST human sequences](#)

Chromosome: 1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 [X] Y MT

Query: 2652[[gene_id](#)]

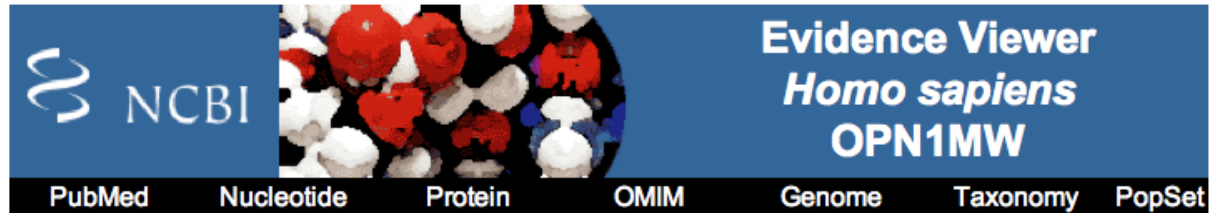
Master Map: [Genes On Sequence](#) [Summary of Maps](#) [Maps & Options](#)

Region Displayed: 153,378K-153,533K bp [Download/View Sequence/Evidence](#)

Hs UniG	ensGenes	RefSeq	RNA	Genes_seq	Symbol	Links	E	Cyto	Description
Hs.658154									
Hs.200716					MECP2				
Hs.247787					OPN1LW	+ OMIM HGNC sv pr dl ev hm sts CCDS SNP			best RefSeq Xq28 opsin 1 (cone pigments), long-wave-sensitive
Hs.592247									
Hs.681381									
Hs.718839									
Hs.666399									
Hs.710323					TEX28P2	+ HGNC sv dl ev sts			best RefSeq Xq28 testis expressed 28 pseudogene 2
Hs.672696									
Hs.247787					OPN1MW	+ OMIM HGNC sv pr dl ev hm sts CCDS SNP			best RefSeq Xq28 opsin 1 (cone pigments), medium-wave-sensitive
Hs.592247									
Hs.681381									
Hs.718839									
Hs.666399									
Hs.710323					TEX28P1	+ HGNC sv dl ev sts			best RefSeq Xq28 testis expressed 28 pseudogene 1
Hs.821									
Hs.672696									
Hs.247787									
Hs.571751					OPN1MW2	+ HGNC sv pr dl ev hm sts CCDS SNP			best RefSeq Xq28 OTTHUMP00000026023
Hs.592247									
Hs.681381									
Hs.718839									
Hs.666399									
Hs.710323					TEX28	+ OMIM HGNC sv pr dl ev hm sts CCDS SNP			best RefSeq Xq28 testis expressed 28
Hs.672696									

Evidence Viewer for OPN1MW

http://www.ncbi.nlm.nih.gov/sutils/evv.cgi?taxid=9606&contig=NT_167198.1&gene=OPN1MW



NCBI Evidence Viewer
Homo sapiens
OPN1MW

PubMed Nucleotide Protein OMIM Genome Taxonomy PopSet

Key for display of mRNAs aligning in this region:

[MapView](#)

[Evidence Viewer Help](#)

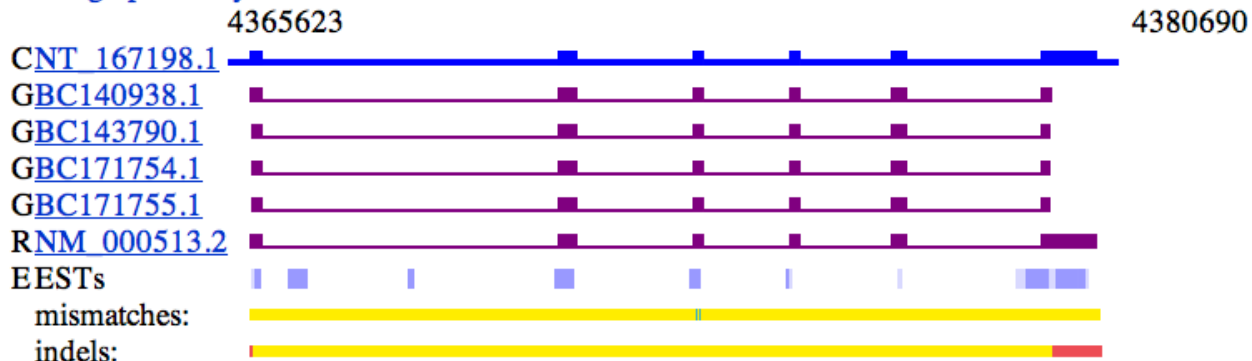
- Genomic sequence (C)
 - model exons, single (M)
 - mRNA exons, single (G, R)
 - model exons, [overlapping](#) (M)
 - mRNA exons, [overlapping](#) (G, R)
- C = contig; M = model mRNA; R = RefSeq mRNA; G = GenBank mRNA
R = new since last genome build; **R** = updated since last genome build

EST density key (E):

- 1 EST
- 2-5 ESTs
- 6-20 ESTs
- 21-99 ESTs
- >100 ESTs

6 exons and 1 gene found in this genomic region spanning 15068 bp.

[View graphic only](#)



Mouse over mismatches, indels and unaligned regions to see their exon number.

NCBI OMIM Home Page

<http://www.ncbi.nlm.nih.gov/omim>

OMIM

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OMIM

OMIM is a comprehensive, authoritative compendium of human genes and genetic phenotypes that is freely available and updated daily. OMIM is authored and edited at the McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins University School of Medicine, under the direction of Dr. Ada Hamosh. Its official home is omim.org.

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Mirror sites: us-east.omim.org, europe.omim.org

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OMIM Coverage

<http://www.ncbi.nlm.nih.gov/Omim/mimstats.html>

January 27, 2014

OMIM Entry Statistics:

Number of Entries in OMIM (Updated 27 January 2014) :

Prefix	Autosomal	X Linked	Y Linked	Mitochondrial	Totals
* Gene description	13,718	670	48	35	14,471
+ Gene and phenotype, combined	103	2	0	2	107
# Phenotype description, molecular basis known	3,709	280	4	28	4,021
% Phenotype description or locus, molecular basis unknown	1,580	135	5	0	1,720
Other, mainly phenotypes with suspected mendelian basis	1,745	116	2	0	1,863
Totals	20,855	1,203	59	65	22,182

➤ 67% Genes

➤ 33% Phenotypes

Mirror sites: us-east.omim.org, europe.omim.org

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Colorblindness in OMIM

http://omim.org/search?index=entry&sort=score+desc%2C+prefix_so

Sort by: Relevance Date updated

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Search History: [View](#), [Clear](#) |

Toggle: [search terms highlighted](#)

Retrieve corresponding:

Search: 'colorblindness'

Results: 1 - 10 of 56 | [Show all](#) | [1](#) [2](#) [3](#) [4](#) [5](#) [6](#) [Next](#) [Last](#)

- | | | |
|-----|---|-----------------------------|
| 1 : | <p># 303800. COLORBLINDNESS, PARTIAL, DEUTAN SERIES; CBD</p> <p>DEUTERANOMALY, INCLUDED
 Cytogenetic location: Xq28
 Matching terms: colorblindness, colourblindness</p> | ICD+, Links |
| 2 : | <p># 190900. TRITANOPIA</p> <p>Cytogenetic location: 7q32.1
 Matching terms: colorblindness</p> | Links |
| 3 : | <p># 303900. COLORBLINDNESS, PARTIAL, PROTAN SERIES; CBP</p> <p>PROTANOMALY, INCLUDED
 Cytogenetic location: Xq28
 Matching terms: colorblindness</p> | ICD+, Links |
| 4 : | <p># 303700. BLUE CONE MONOCHROMACY; BCM</p> <p>CONE DYSTROPHY 5, X-LINKED, INCLUDED
 Cytogenetic locations: Xq28 , Xq28
 Matching terms: colorblindness</p> | ICD+, Links |

Colorblindness in OMIM

http://omim.org/search?index=entry&sort=score+desc%2C+prefix_so

Sort by: Relevance Date updated
 Advanced Search: [OMIM](#), [Clinical Synopses](#), [OMIM Gene Map](#) |
 Search History: [View](#), [Clear](#) |
 Toggle: [search terms highlighted](#) Retrieve corresponding: [gene map](#) [clinical synopses](#)

Search: 'colorblindness'

Results: 1 - 10 of 56 | [Show all](#) | [1](#) [2](#) [3](#) [4](#) [5](#) [6](#) [Next](#) [Last](#)

1 : [# 303800. COLORBLINDNESS, PARTIAL, DEUTAN SERIES; CBD](#)

DEUTERANOMALY, INCLUDED
 Cytogenetic location: Xq28
 Matching terms: colorblindness, colourblindness

2 : [# 190900. TRITANOPIA](#)

Cytogenetic location: 7q32.1
 Matching terms: colorblindness

3 : [# 303900. COLORBLINDNESS, PARTIAL, PROTAN SERIES; CBP](#)

PROTANOMALY, INCLUDED
 Cytogenetic location: Xq28
 Matching terms: colorblindness

4 : [# 303700. BLUE CONE MONOCHROMACY; BCM](#)

CONE DYSTROPHY 5, X-LINKED, INCLUDED
 Cytogenetic locations: Xq28 , Xq28
 Matching terms: colorblindness

External Links for #303800 [x]

- [Clinical Resources](#)
- [Clinical Trials](#)
- [Gene Reviews](#)
- [OrphaNet](#)
- [Genetic Alliance](#)
- [Genetics Home Reference](#)
- [Variation](#)
- [Locus Specific DBs](#)
- [Animal Models](#)
- [NCBI HomoloGene](#)
- [Cellular Pathways](#)
- [Reactome](#)

[ICD+](#), [Links](#)

Colorblindness in OMIM

<http://omim.org/entry/303800>

#303800

ICD+

COLORBLINDNESS, PARTIAL, DEUTAN SERIES; CBD

Alternative titles; symbols

DEUTAN COLORBLINDNESS; DCB

DEUTERANOPIA

GREEN COLORBLINDNESS

Other entities represented in this entry:

DEUTERANOMALY, INCLUDED

Phenotype-Gene Relationships

Location	Phenotype	Phenotype MIM number	Gene/Locus	Gene/Locus MIM number
Xq28	Colorblindness, deutan	303800	OPN1MW	300821

[Clinical Synopsis](#)

TEXT

A number sign (#) is used with this entry because deutan colorblindness is caused by mutation in the OPN1MW gene (300821), which encodes green cone pigment.

Description

Normal color vision in humans is trichromatic, being based on 3 classes of cone that are maximally sensitive to light at approximately 420 nm (blue cones; 613522), 530 nm (green cones; 300821), and 560 nm (red cones; 300822). Comparison by neural circuits of light absorption by the 3 classes of cone photoreceptors allows perception of red, yellow, green,

Table of Contents for #303800

- Title
- Phenotype-Gene Relationships
- Text
 - Description
 - Clinical Features
 - Mapping
 - Population Genetics
 - Inheritance
 - Evolution
 - Molecular Genetics
 - History
- Clinical Synopsis
- See Also
- References
- Contributors
- Creation Date
- Edit History

External Links for Entry:

- ▶ [Clinical Resources](#)
- ▶ [Variation](#)
- ▶ [Animal Models](#)
- ▶ [Cellular Pathways](#)

[Centers for Mendelian Genomics](#)

Genome

Ensembl	Genome databases for vertebrates and other eukaryotic species.
MITOMAP	A curated repository of published and unpublished data on human mitochondrial DNA variation.
NCBI Map Viewer	Detailed views of the complete genomes of selected organisms from vertebrates to protozoa.
UCSC	UCSC Genome Browser; reference sequences and working draft assemblies for a large collection of genomes.

DNA

Ensembl	Transcript-based views for coding and noncoding DNA.
NCBI RefSeq	A collection of genome, gene, and transcript sequence data from several sources, including GenBank, RefSeq.
UCSC	UCSC Genome Browser; reference sequences and working draft assemblies for a large collection of genomes.

Protein

UniProt	Comprehensive protein sequence and functional information, including supporting data.
HPRD	The Human Protein Reference Database; manually extracted and visually depicted information on human proteins.

Gene Info

BioGPS	The Gene Portal Hub; customizable portal of gene and protein function information.
Ensembl	Orthologs, paralogs, regulatory regions, and splice variants.
NCBI Gene	Gene-specific map, sequence, expression, structure, function, citation, and homology data.
GeneCards	The Human Genome Compendium; web-based cards integrating automatically mined information on human genes.
Gene Ontology	Terms, defined using controlled vocabulary, representing gene product properties (biologic process, cellular component, molecular function) across species.
KEGG	Kyoto Encyclopedia of Genes and Genomes; diagrams of signaling pathways.
PharmGKB	Pharmacogenomics Knowledge Base; curated and annotated information regarding the effects of human genetic variations on drug response.
UCSC	UCSC Genome Bioinformatics; gene-specific structure and function information with links to other databases.
HGNC	HUGO Gene Nomenclature Committee.

Clinical

ClinicalTrials.gov	A registry of federally and privately supported clinical trials conducted in the United States and around the world.
DECIPHER	Database of chromosomal aberration including clinical and genomic information.
Gene Tests	Information on genetic testing and its use in diagnosis, management, and genetic counseling.
Gene Reviews	Expert-authored, peer-reviewed descriptions of inherited disorders including the uses of genetic testing in diagnosis, management, and genetic counseling.
OrphaNet	European reference portal for information on rare diseases and orphan drugs.
EuroGentest	A list of European laboratories that offer genetic testing.
Genetic Alliance	Network of disease-specific advocacy organizations, universities, private companies, government agencies, and public policy organizations.
Newborn Screening	Information and resources for newborn screening and genetics.
POSSUM	A dysmorphology database of multiple malformations; metabolic, teratogenic, chromosomal, and skeletal syndromes; and their images.
DermAtlas	Images and descriptions of lesions seen in dermatologic disorders.
GARD	Genetic and Rare Diseases Information Center; information on rare and/or genetic disorders including portal to clinician and patient resources.
GTR	Genetic Testing Registry.
NextGxDx	NextGxDx.

Variation

Genetics Association DB	An archive of human genetic association studies of complex diseases and disorders.
HGMD	Human Gene Mutation Database; published mutations causing or associated with human inherited disease; disease-associated/functional polymorphisms.
GWAS Central	GWAS Central; summary level genotype-to-phenotype information from genetic association studies.
HGVS	Human Genome Variation Society; maintains lists of and links to locus-specific mutation databases; guidelines for description of sequence variants.
Locus Specific DBs	A gene-specific database of variation.
LOVD	Colon cancer gene variant databases.
ClinVar	ClinVar aggregates information about sequence variation and its relationship to human health.
inSIGHT	International Society for Gastrointestinal Hereditary Tumors.

Animal Models

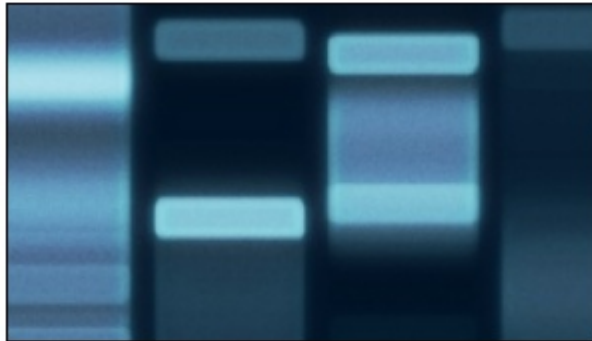
FlyBase	A Database of Drosophila Genes and Genomes.
NCBI HomoloGene	A system for automated detection of homologs among the annotated genes of several completely sequenced eukaryotic genomes.
MGI Mouse Phenotype	Phenotypes, alleles, and disease models from Mouse Genome Informatics.



RefSeqGene

Gene

Search



RefSeqGene

RefSeqGene defines genomic sequences to be used as reference standards for well-characterized genes and is part of the Locus Reference Genomic (LRG) Project.

Using RefSeqGene

- [About](#)
- [Browse Genes with RefSeqGene Sequences](#)
- [See RefSeqGene Sequences in the Nucleotide Database](#)
- [Download Data](#)
- [New RefSeqGene Sequences \(last 30 days\)](#)

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- [Clinical Remap](#)
- [Genome Workbench](#)
- [Map Viewer](#)
- [RefSeqGene BLAST](#)

Related Sites

- [Entrez Gene](#)
- [RefSeq](#)
- [Locus Reference Genomic \(LRG\)](#)
- [Variation Databases \(dbSNP, dbVar\)](#)
- [Online Mendelian Inheritance In Man](#)
- [Contact Us](#)

UniGene

UniGene ▾

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UniGene

UniGene computationally identifies transcripts from the same locus; analyzes expression by tissue, age, and health status; and reports related proteins (protEST) and clone resources.

Using UniGene

[Getting Started](#)[FAQ](#)[Query Tips](#)[ProtEST](#)[DDD](#)[Clustering by transcripts](#)[Clustering by genomes](#)

UniGene Tools

[UniGene Statistics](#)[Library browser](#)[Digital Differential Display \(DDD\)](#)[Downloads/FTP](#)

Also of interest

[BioSample](#)[EST sequences](#)[Gene](#)[HomoloGene](#)[Map Viewer](#)[Short Read Archive \(SRA\)](#)



HomoloGene

An automated system for constructing putative homology groups from the complete gene sets of a wide range of eukaryotic species.

Getting Started

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[FAQ](#)

In Depth

[Build Procedure](#)

[Release Statistics](#)

[Download from FTP site](#)

Other Databases

[Gene](#)

[Genome](#)

[Taxonomy](#)

Release 67 Statistics

Release date:2012-12-12

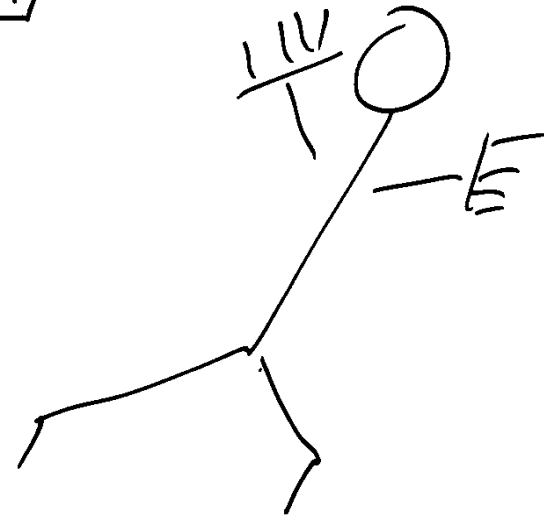
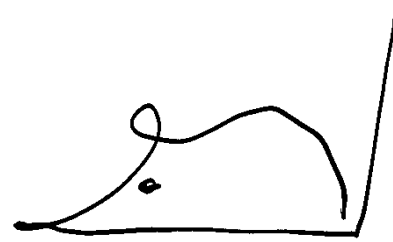
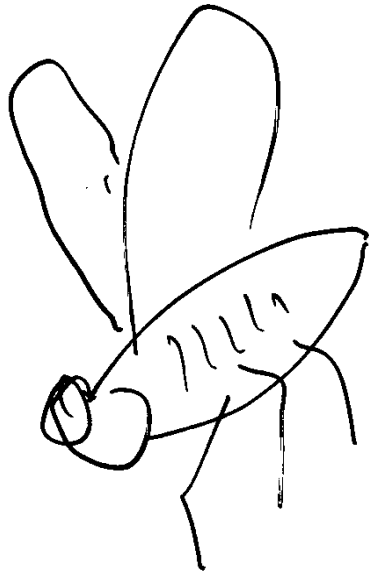
Total species:20

Total HomoloGene groups:43074

[Download](#)

Scientific name	Common name	Genome release	Annotation release	Input genes	Grouped genes	Groups
Homo sapiens	human	GRCh37.p5	Build 37.3	19217	18867	18473
Pan troglodytes	chimpanzee	Pan_troglodytes-2.1.4	Build 3.1	19655	17367	17078
Macaca mulatta	Rhesus monkey	Mmul_051212	Build 1.2	22566	17151	16706
Canis lupus familiaris	dog	CanFam3.1	Build 3.1	19176	17800	17186
Bos taurus	cow	Bos_taurus_UMD_3.1	Build 6.1	21121	18771	17458
Mus musculus	mouse	GRCm38	Build 38.1	21432	20744	18858
Rattus norvegicus	rat	RGSC_v3.4	Build 4.2	21943	19868	17882
Gallus gallus	chicken	Gallus_gallus-4.0	Build 3.1	16731	14171	13149
Danio rerio	zebrafish	Zv9	Build 5.1	26326	20504	14183
Drosophila melanogaster	fruit fly	Release 5	Build 9.4	13555	9252	7713
Anopheles gambiae	malaria mosquito	AgamP3	Build 3.1	12460	8827	7500
Caenorhabditis elegans	nematode	WS195	Build 9.1	20185	8698	4811
Saccharomyces cerevisiae	budding yeast	R64-1-1	Build 3.1	5882	4854	4373
Kluyveromyces lactis	None	ASM251v1	Build 1.1	4978	4404	4338
Eremothecium gossypii	None	ASM9102v1	Build 3.1	4722	3932	3888
Schizosaccharomyces pombe	fission yeast	ASM294v2	Build 1.1	5018	3238	2946

Comparative Genomics



L'homme et ses Cousins germains
F. Jacob

Ensembl Human Opsin Search

http://uswest.ensembl.org/Homo_sapiens/Search/Results?species=Homo_sapiens;idx=;q=

Human (GRCh37) ▾

Current selection:

< all Species

Only searching Human

Restrict category to:

Gene 13

Protein Family 2

Per page:

10 25 50 100

Show all results in one page

Layout:

Standard Table

Tip:

You can choose which results appear near the top of your search by updating your favourite species.

Only searching Human ▾ opsin gene



42609736 results match opsin gene when restricted to species: Human ✕

Did you mean... ▾

OPN5 (Human Gene)

ENSG00000124818 6:47749718-47800516:1

opsin 5 [Source:HGNC Symbol;Acc:19992]

[Variation table](#) • [Location](#) • [Regulation](#) • [Orthologues](#) • [Gene tree](#)

OPN3 (Human Gene)

ENSG00000054277 1:241753404-241840678:-1

opsin 3 [Source:HGNC Symbol;Acc:14007]

[Variation table](#) • [Location](#) • [Regulation](#) • [Orthologues](#) • [Gene tree](#)

OPN1LW (Human Gene)

ENSG00000102076 X:153409698-153424507:1

opsin 1 (cone pigments), long-wave-sensitive [Source:HGNC Symbol;Acc:9936]

[Variation table](#) • [Location](#) • [Regulation](#) • [Orthologues](#) • [Gene tree](#)

OPN1SW (Human Gene)

ENSG00000128617 7:128412545-128415844:-1

opsin 1 (cone pigments), short-wave-sensitive [Source:HGNC Symbol;Acc:1012]

[Variation table](#) • [Location](#) • [Regulation](#) • [Orthologues](#) • [Gene tree](#)

OPN1MW2 (Human Gene)

ENSG00000166160 X:153485225-153498755:1

opsin 1 (cone pigments), medium-wave-sensitive 2 [Source:HGNC Symbol;Acc:26952]

[Variation table](#) • [Location](#) • [Regulation](#) • [Orthologues](#) • [Gene tree](#)

OPN1MW (Human Gene)

ENSG00000147380 X:153448107-153461633:1

opsin 1 (cone pigments), medium-wave-sensitive [Source:HGNC Symbol;Acc:4206]

[Variation table](#) • [Location](#) • [Regulation](#) • [Orthologues](#) • [Gene tree](#)

OPN4 (Human Gene)

ENSG00000122375 10:88414314-88426605:1

opsin 4 [Source:HGNC Symbol;Acc:14449]

[Variation table](#) • [Location](#) • [Regulation](#) • [Orthologues](#) • [Gene tree](#)

Ensembl Human OPN1MW Gene

http://uswest.ensembl.org/Homo_sapiens/Gene/Summary?g=ENSG00000147380

Human (GRCh37) ▾

Location: X:153,448,107-153,461,633 ▾

Gene: OPN1MW ▾

Gene-based displays

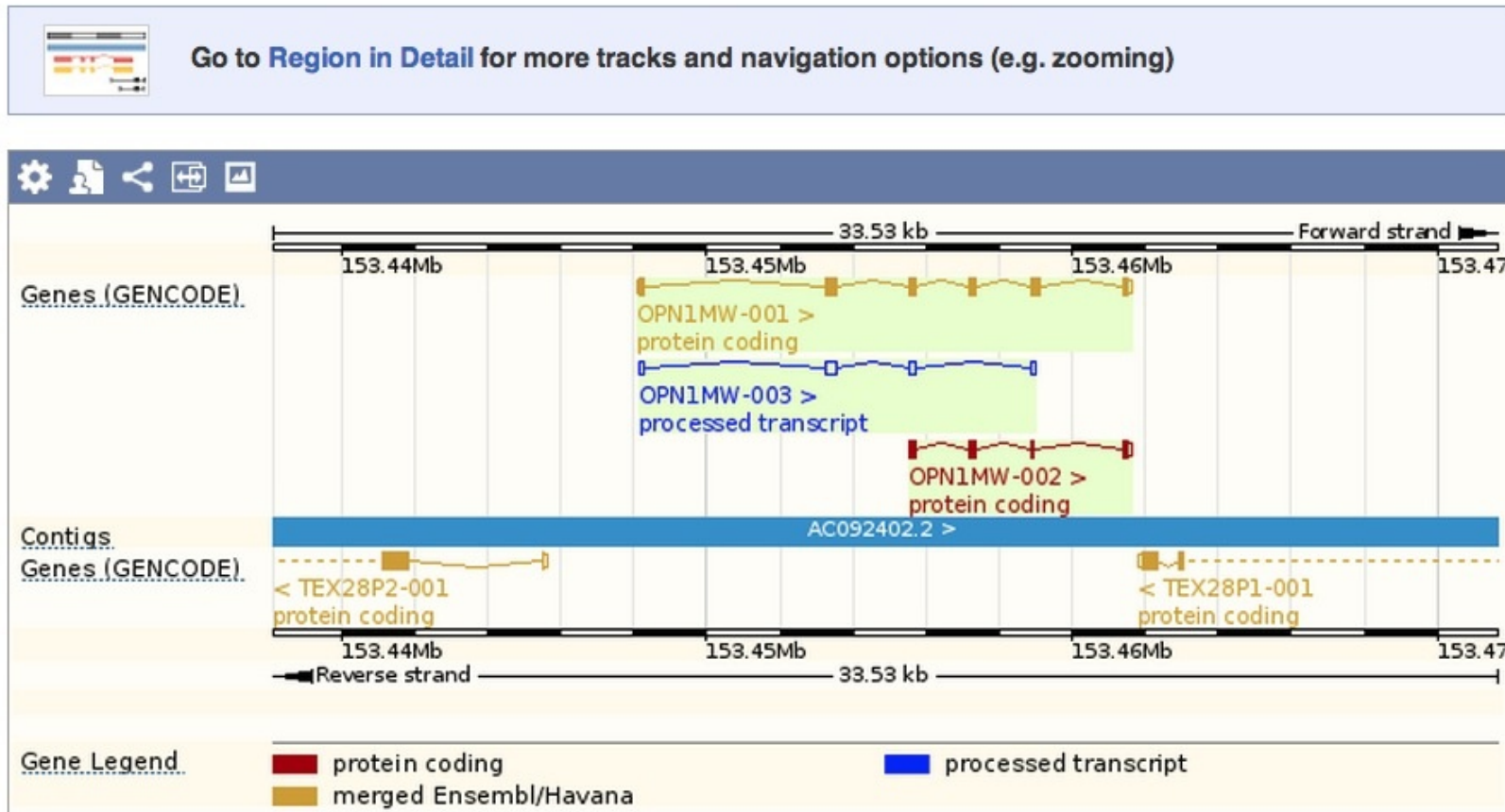
- Summary
- Splice variants (3)
- Transcript comparison
- Supporting evidence
- Sequence
 - Secondary Structure
- External references
- Regulation
- Expression
- Comparative Genomics
 - Genomic alignments
 - Gene tree (image)
 - Gene tree (text)
 - Gene tree (alignment)
 - Gene gain/loss tree
 - Orthologues (60)
 - Paralogues (9)
 - Protein families (1)
- Phenotype
- Genetic Variation
 - Variation table
 - Variation image
 - Structural variation
- External data
 - Personal annotation
- ID History
 - Gene history

Gene: OPN1MW ENSG00000147380

Description	opsin 1 (cone pigments), medium-wave-sensitive [Source:HGNC Symbol;Acc:4206]
Location	Chromosome X: 153,448,107-153,461,633 forward strand.
INSDC coordinates	chromosome:GRCh37:CM000685.1:153448107:153461633:1
Transcripts	This gene has 3 transcripts (splice variants) Show transcript table
Summary ⓘ	
Name	OPN1MW (HGNC Symbol)
Synonyms	CBBM, CBD, COD5, GCP, OPN1MW1 [To view all Ensembl genes linked to the name click here.]
CCDS	This gene is a member of the Human CCDS set: CCDS14743
Ensembl version	ENSG00000147380.6
Gene type	Known protein coding
Prediction Method	Annotation for this gene includes both automatic annotation from Ensembl and Havana manual curation, see article .
Alternative genes	This gene corresponds to the following database identifiers: Havana gene: OTTHUMG00000022652 (version 6)
Secondary structure	Sorry, no secondary structure is available for this gene

Ensembl Opsin OPN1MW Gene Location

http://uswest.ensembl.org/Homo_sapiens/Location/View?h=Havana%20gene;r=X

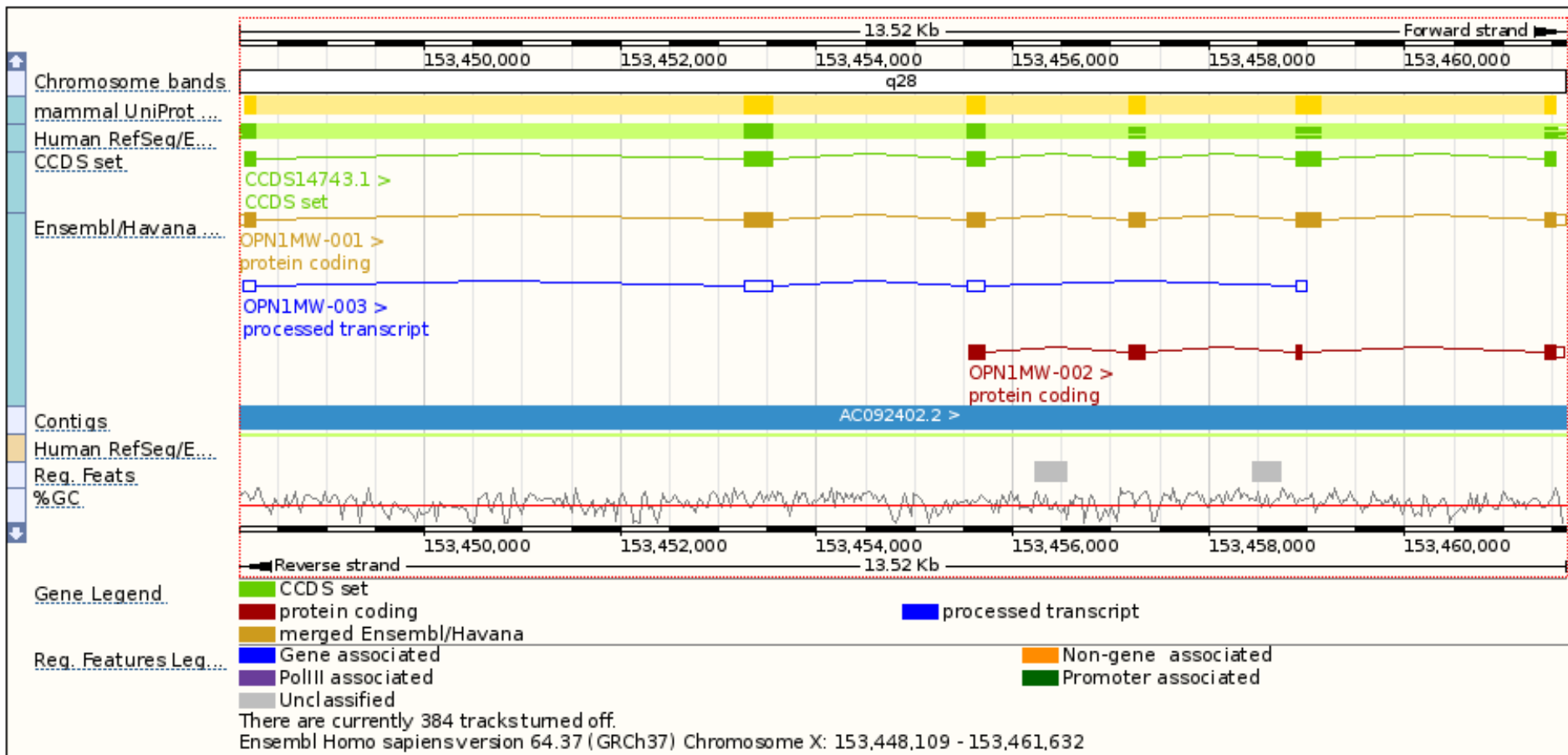


Configuring the display

Tip: use the "Configure this page" link on the left to show additional data in this region.

Ensembl OPN1MW Transcripts

http://uswest.ensembl.org/Homo_sapiens/Location/View?h=Havana%20gene;r=X



[Export Image](#)



Ensembl OPN1MW Opsin Protein

http://uswest.ensembl.org/Homo_sapiens/Transcript/ProteinSummary?db=core;g=ENSG00000369935

Transcript: OPN1MW-001 (ENST00000369935)

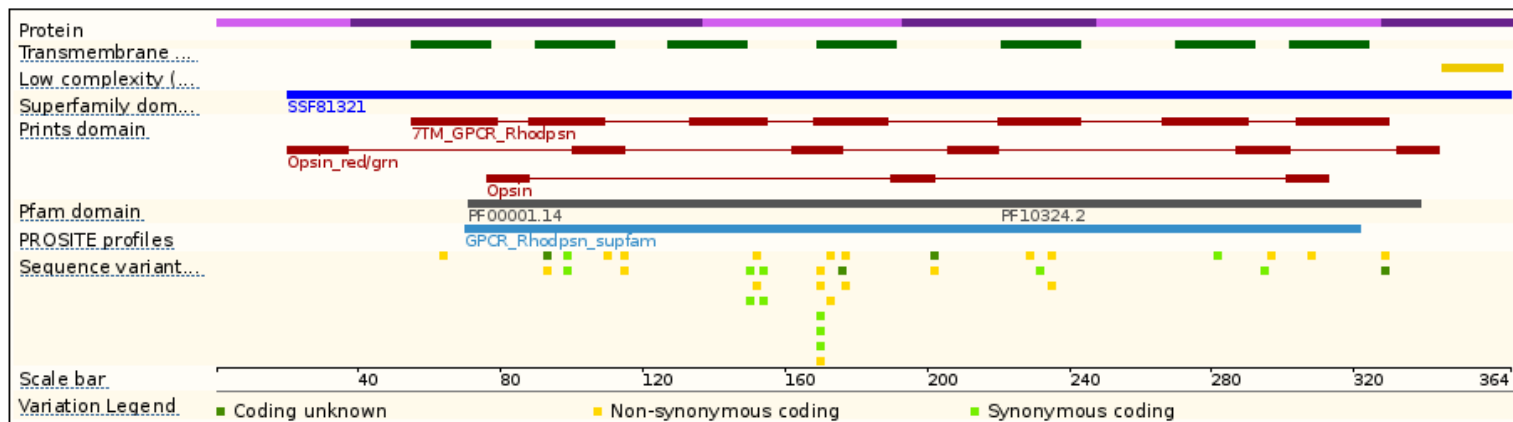
Description opsin 1 (cone pigments), medium-wave-sensitive [Source:HGNC Symbol;Acc:4206]
Location [Chromosome X: 153,448,107-153,461,633](#) forward strand.
Gene This transcript is a product of gene [ENSG00000147380](#) - There are 3 transcripts in this gene

Show/hide columns		Filter				
Name	Transcript ID	Length (bp)	Protein ID	Length (aa)	Biotype	CCDS
OPN1MW-001	ENST00000369935	1257	ENSP00000358951	364	Protein coding	CCDS14743
OPN1MW-002	ENST00000430054	590	ENSP00000394838	164	Protein coding	-
OPN1MW-003	ENST00000468495	692	No protein product	-	Processed transcript	-

Transcript and Gene level displays

Views in Ensembl are separated into gene based views and transcript based views according to which level the information is more appropriately associated with. This view is a transcript level view. To flip between the two sets of views you can click on the Gene and Transcript tabs in the menu bar at the top of the page.

Protein summary [help](#)



[Export Image](#)

Statistics

Ave. residue weight: 111.495 g/mol
 Charge: 11.5
 Isoelectric point: 8.7067
 Molecular weight: 40,584.28 g/mol
 Number of residues: 364 aa

Ensembl Tutorials

<http://uswest.ensembl.org/info/website/tutorials/index.html>



Ensembl Tutorials and Worked Examples

Online tutorials

- [Introduction to Browsing Chordate Genomes](#) - a 3-hour comprehensive interactive tutorial aimed at beginners

Video tutorials

The tutorials listed below are Flash animations of some of our training presentations. We are gradually adding to the list, so please check back regularly.



Note that we are now hosting all our tutorials on [YouTube](#) (and [优酷网](#) for users in China) for ease of maintenance. A selection of tutorials is also available on the [EBI E-Video website](#).

Title	Running time (minutes)
The Ensembl Genome Browser	10:00
EnsemblGenomes - Extending Ensembl across the taxonomic space	4:54
Comparative Genomics	9:58
SNPs and other Variations - 1 of 2	7:06
SNPs and other Variations - 2 of 2	6:30
Introduction to BioMart	4.27
BioMart: Variation IDs to HGNC Symbols	2:58
Clip: Exons and Introns	1:12
Clip: Export Sequence	1:08
Clip: Genome Variation	0:37
Clip: Transcriptomics (ArrayExpress)	1:12
Clip: View Conserved Sequence	1:26
Clip: View External Data (DAS)	1:53