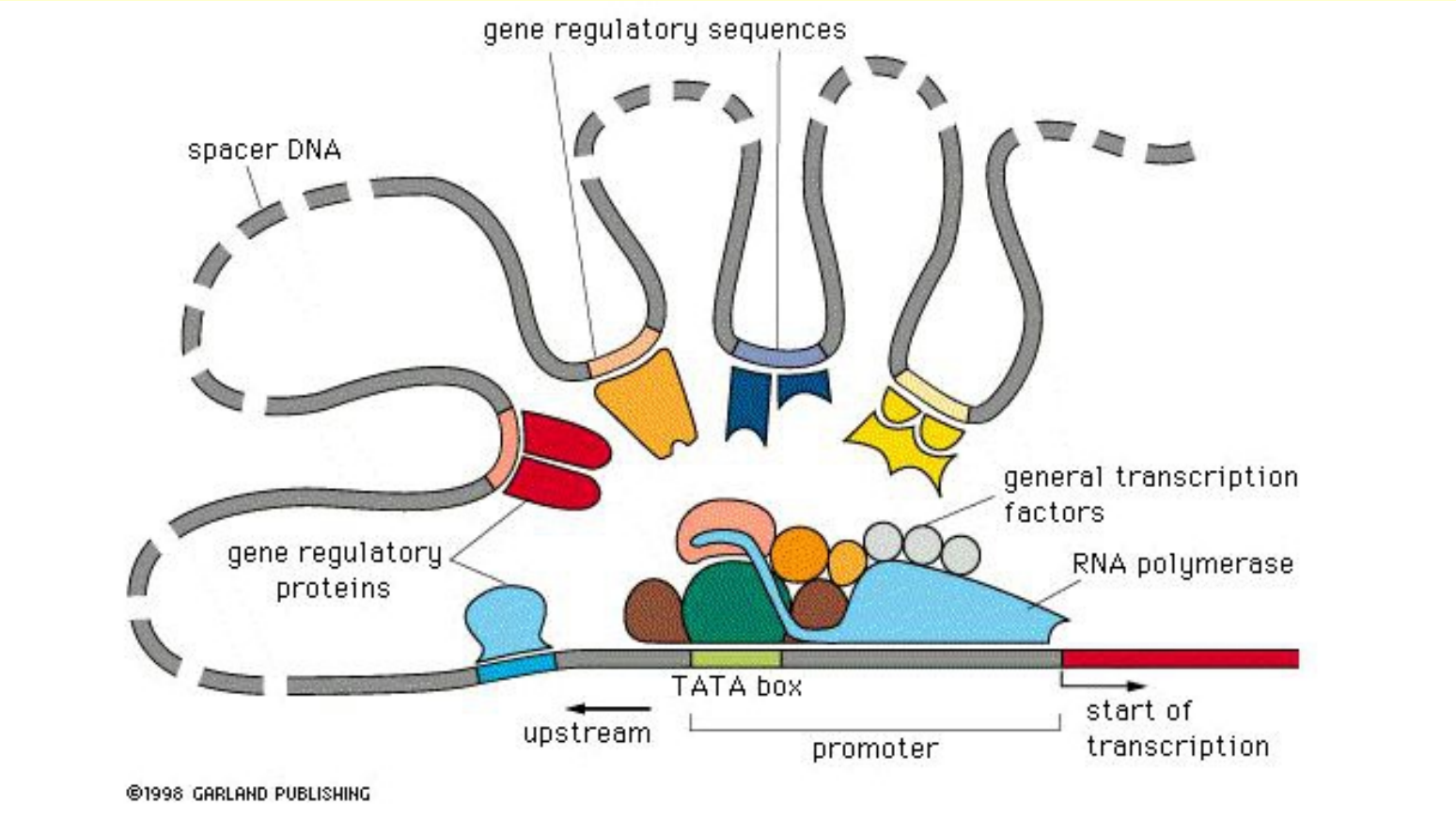
Genome Databases

Genomics, Bioinformatics & Medicine
http://biochem158.stanford.edu/

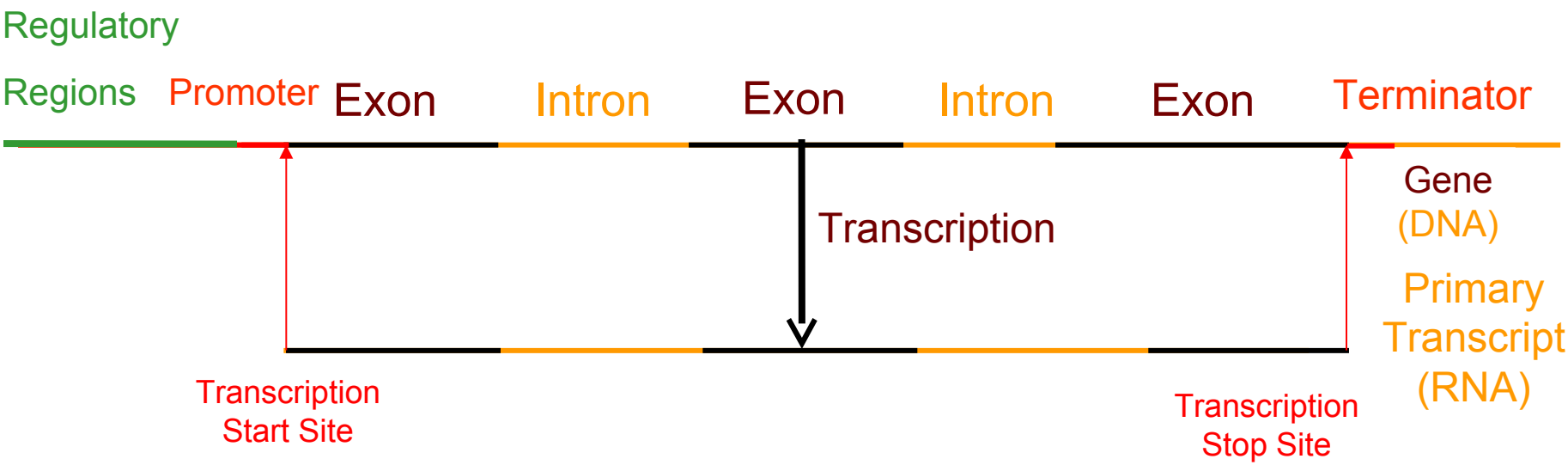
Doug Brutlag
Professor Emeritus of Biochemistry & Medicine
Stanford University School of Medicine

Components of a Typical Human Gene

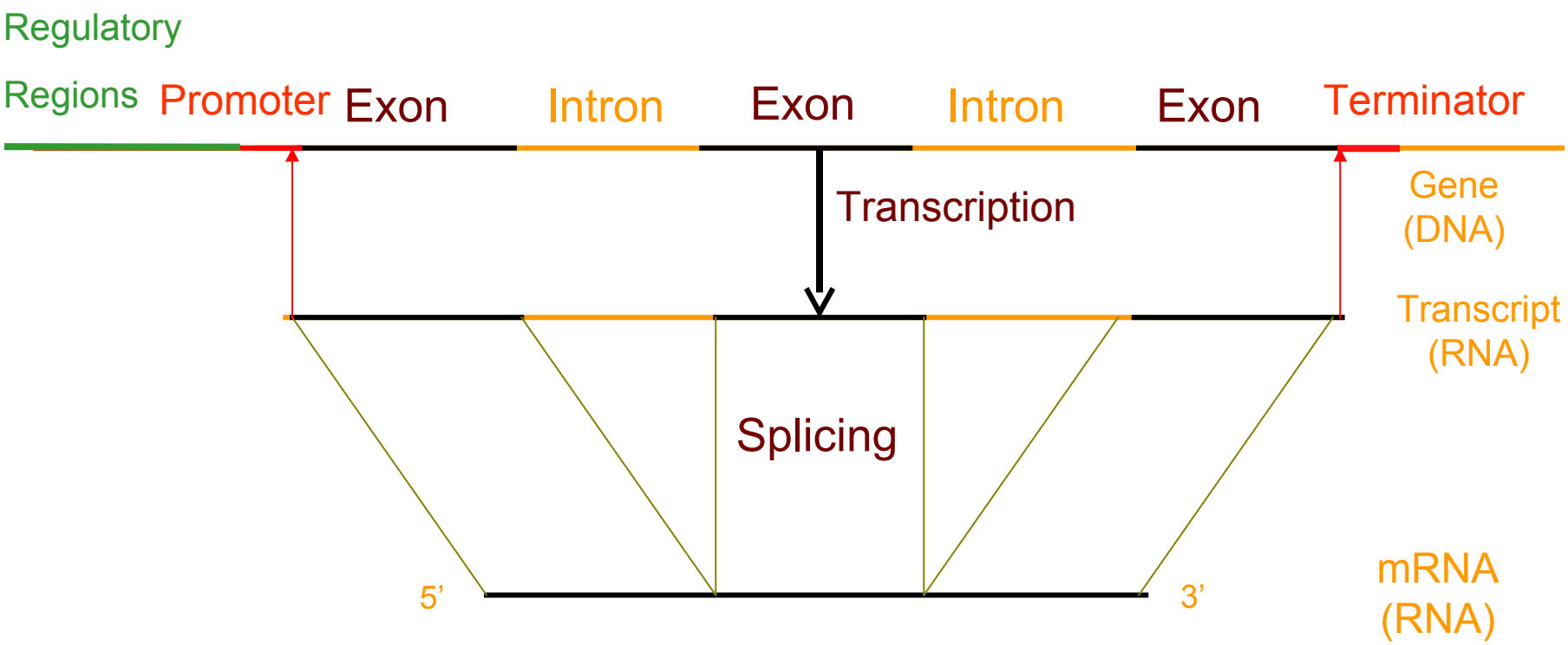
Multiple Enhancer Sequences



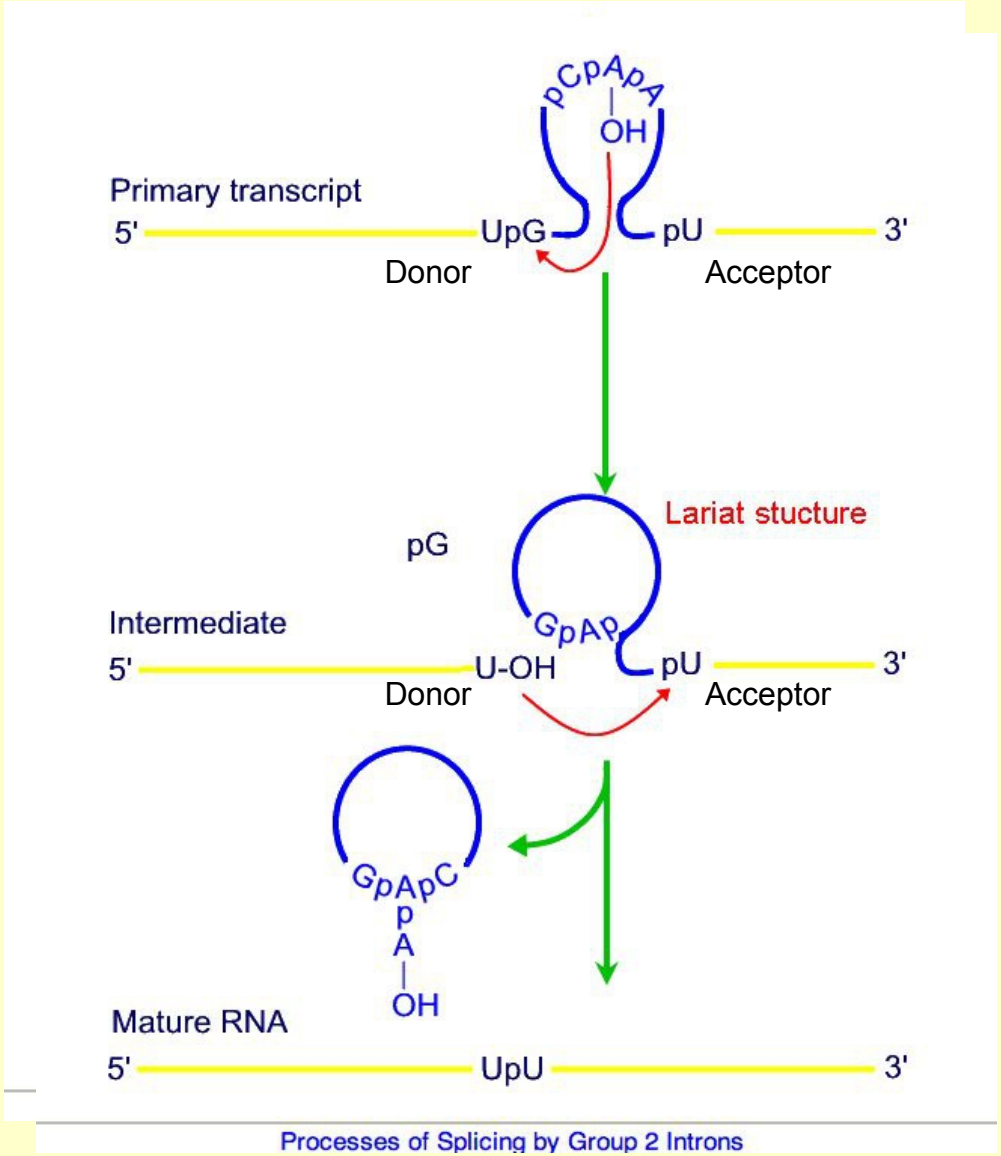
Active Genes are Transcribed into RNA



Splicing Transcript Yields Mature mRNA

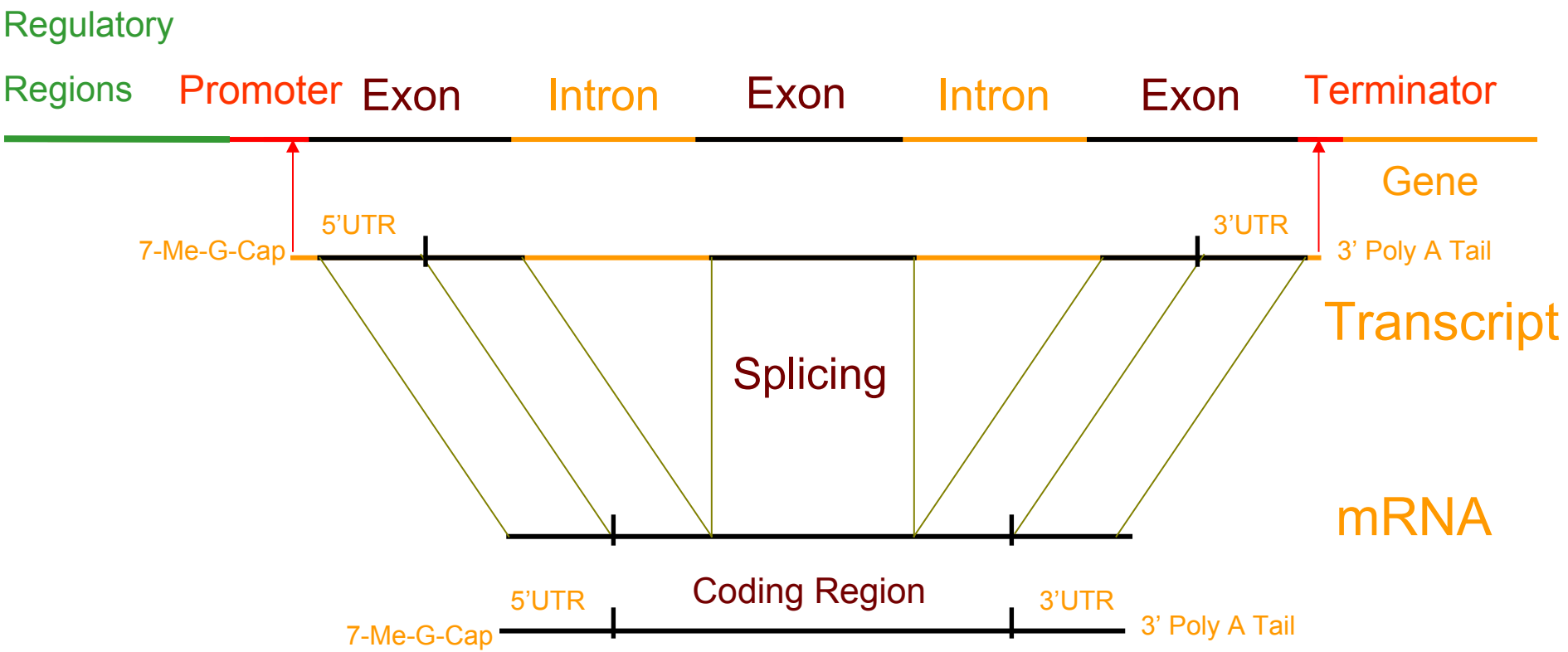


Intron Splicing Mechanism

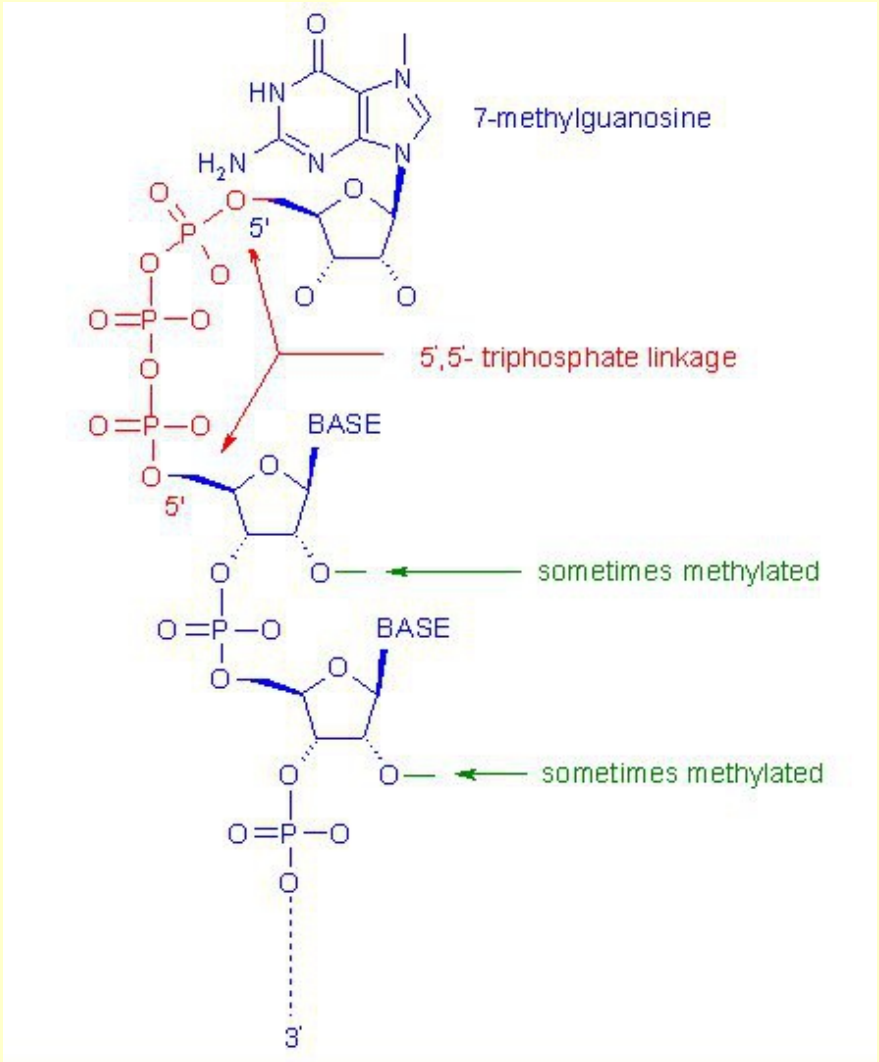


Processes of Splicing by Group 2 Introns

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

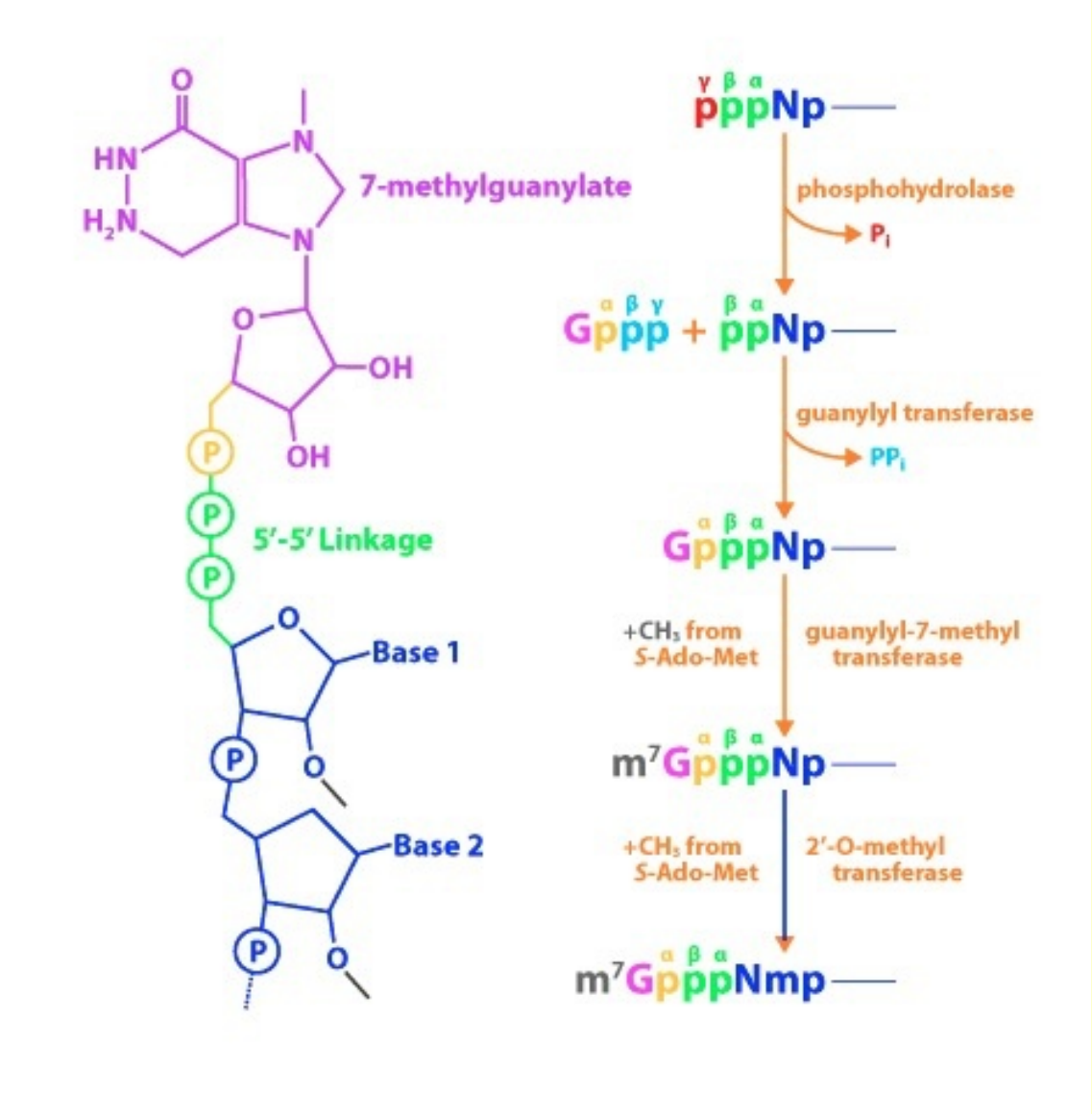


Structure of 5′ CAP

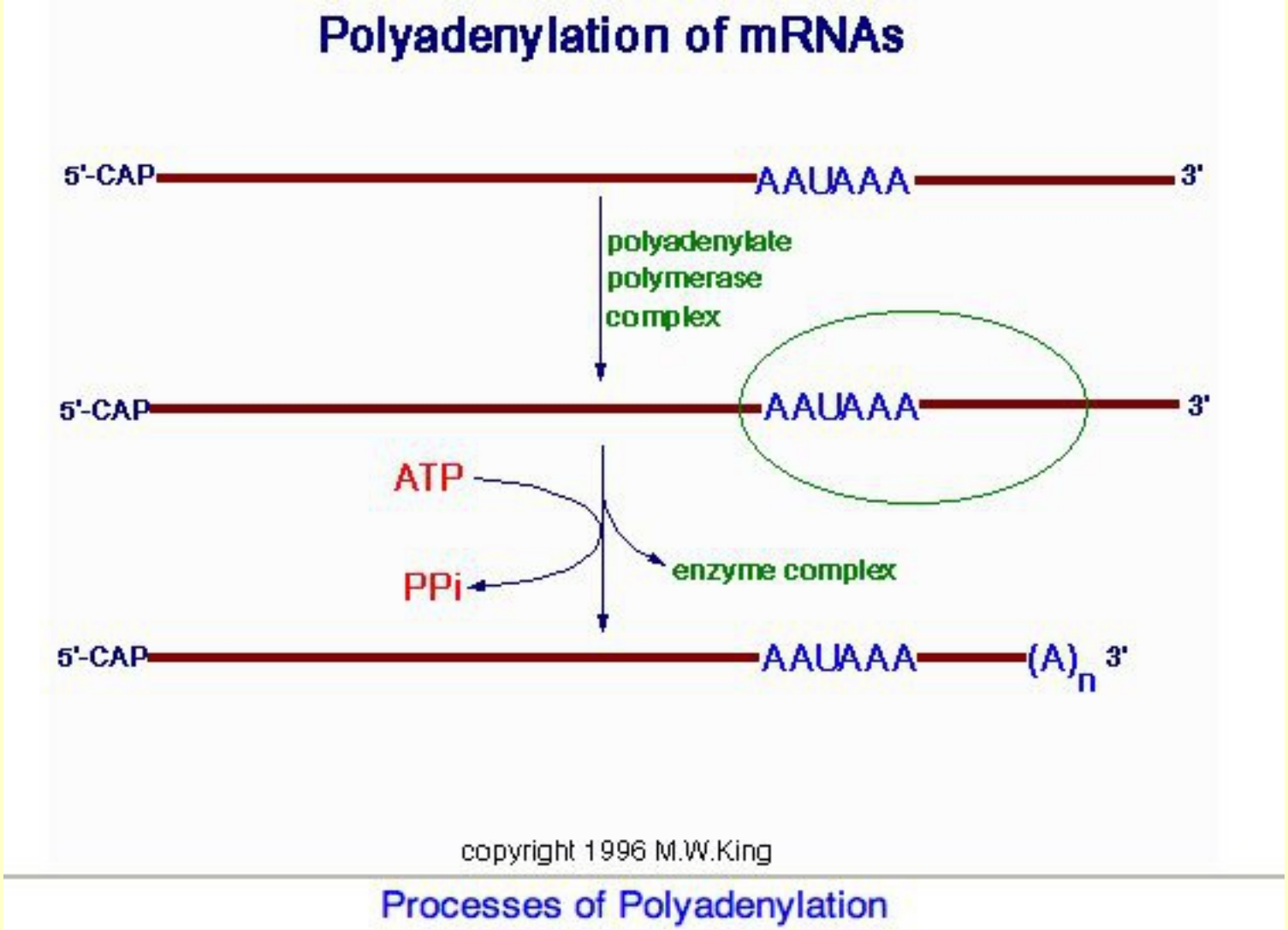


Structure of the 5'-Cap of Eukaryotic mRNAs

Mature mRNA contains 7-Methyl-Guanylate5′ Cap

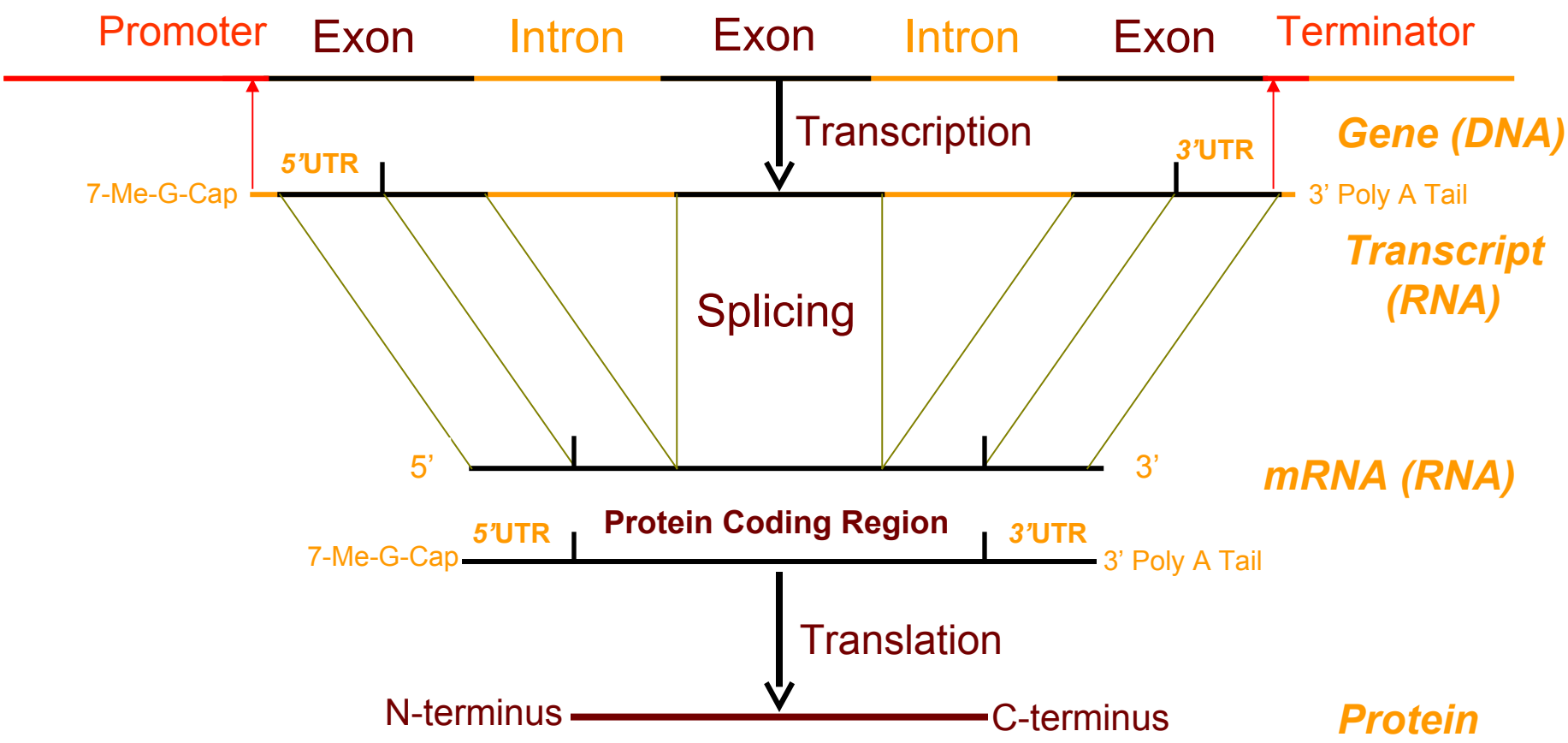


PolyAdenylation of mRNAs

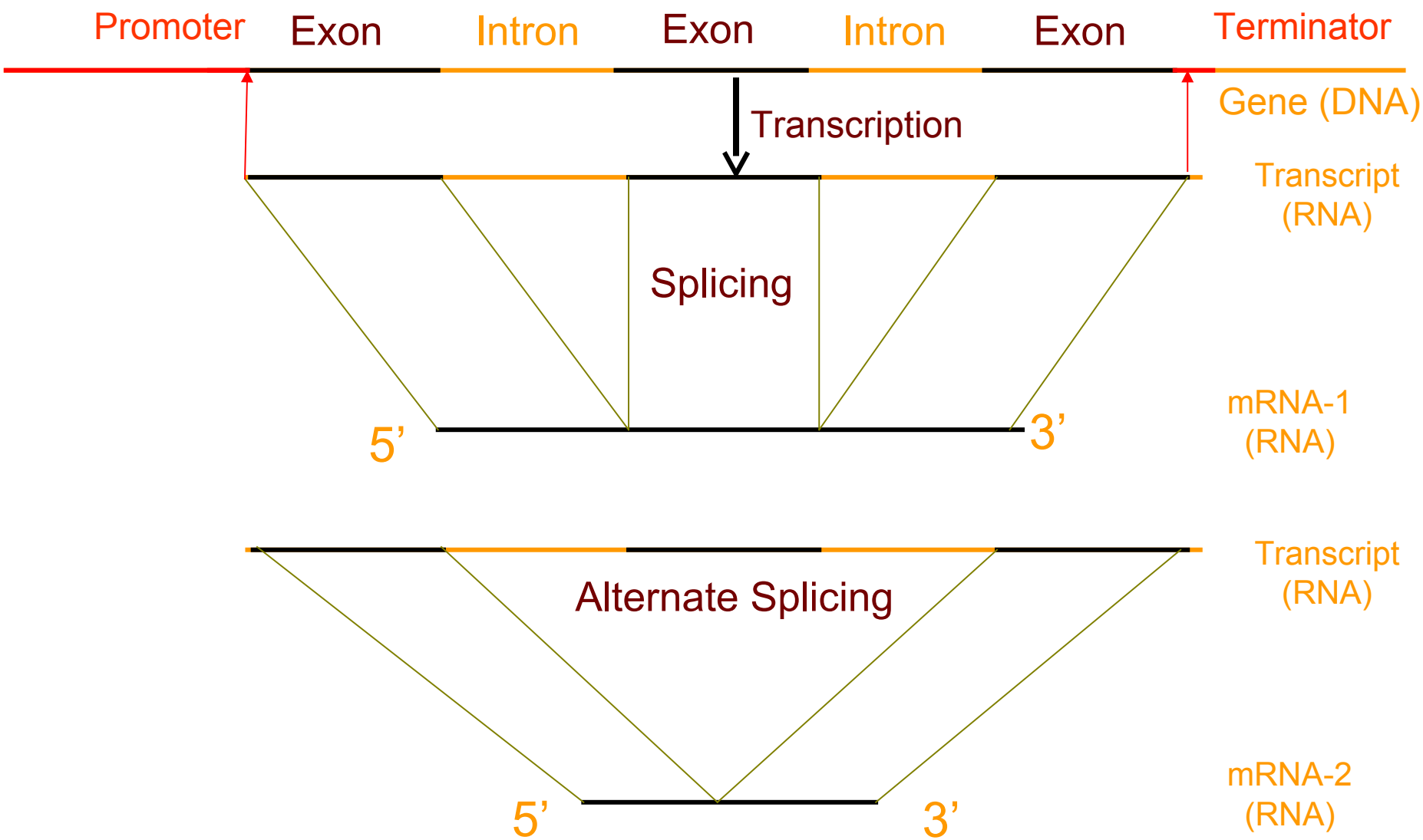


Processes of Polyadenylation

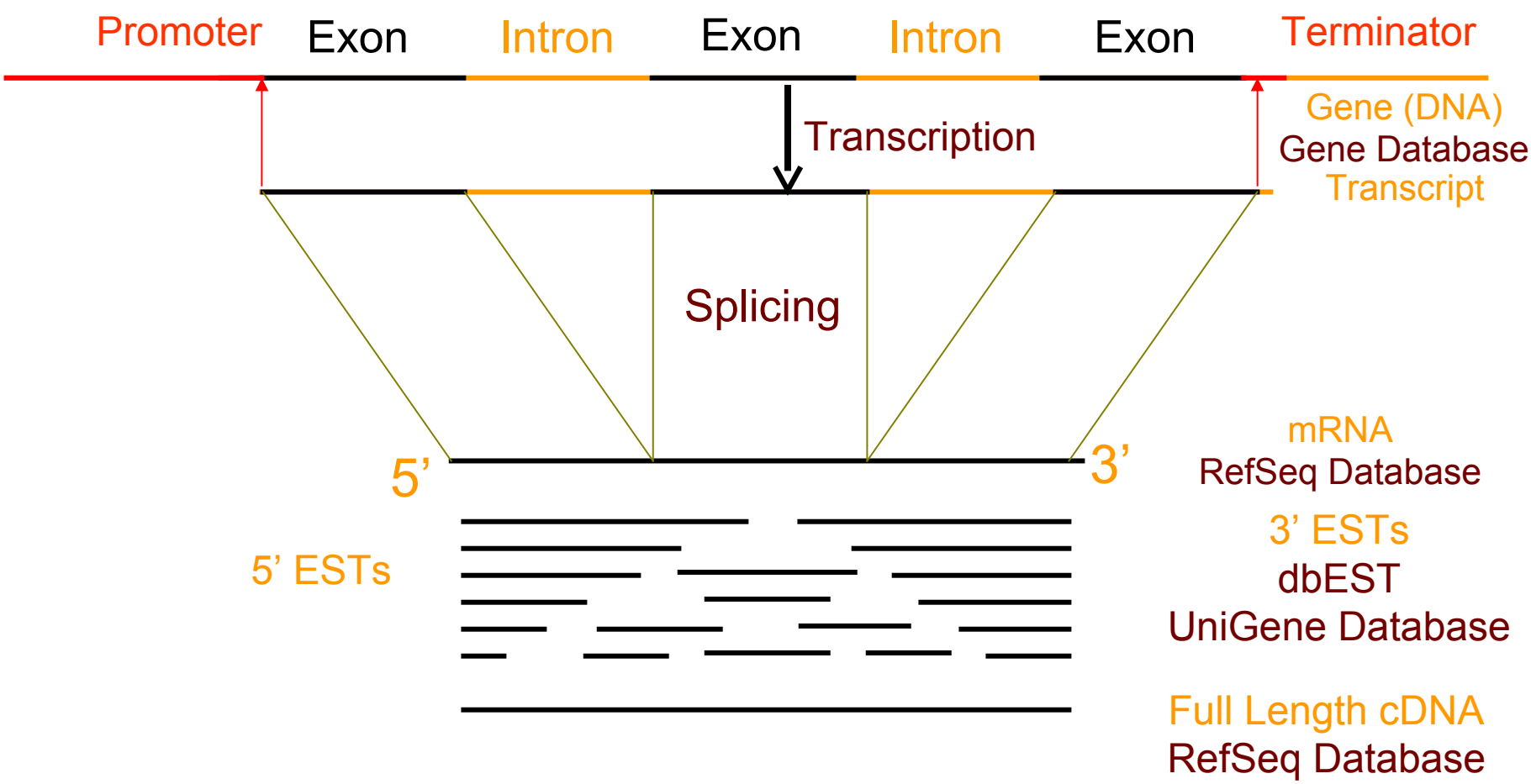
Mature mRNA contains Coding Region and 5′ and 3′ Untranslated Regions



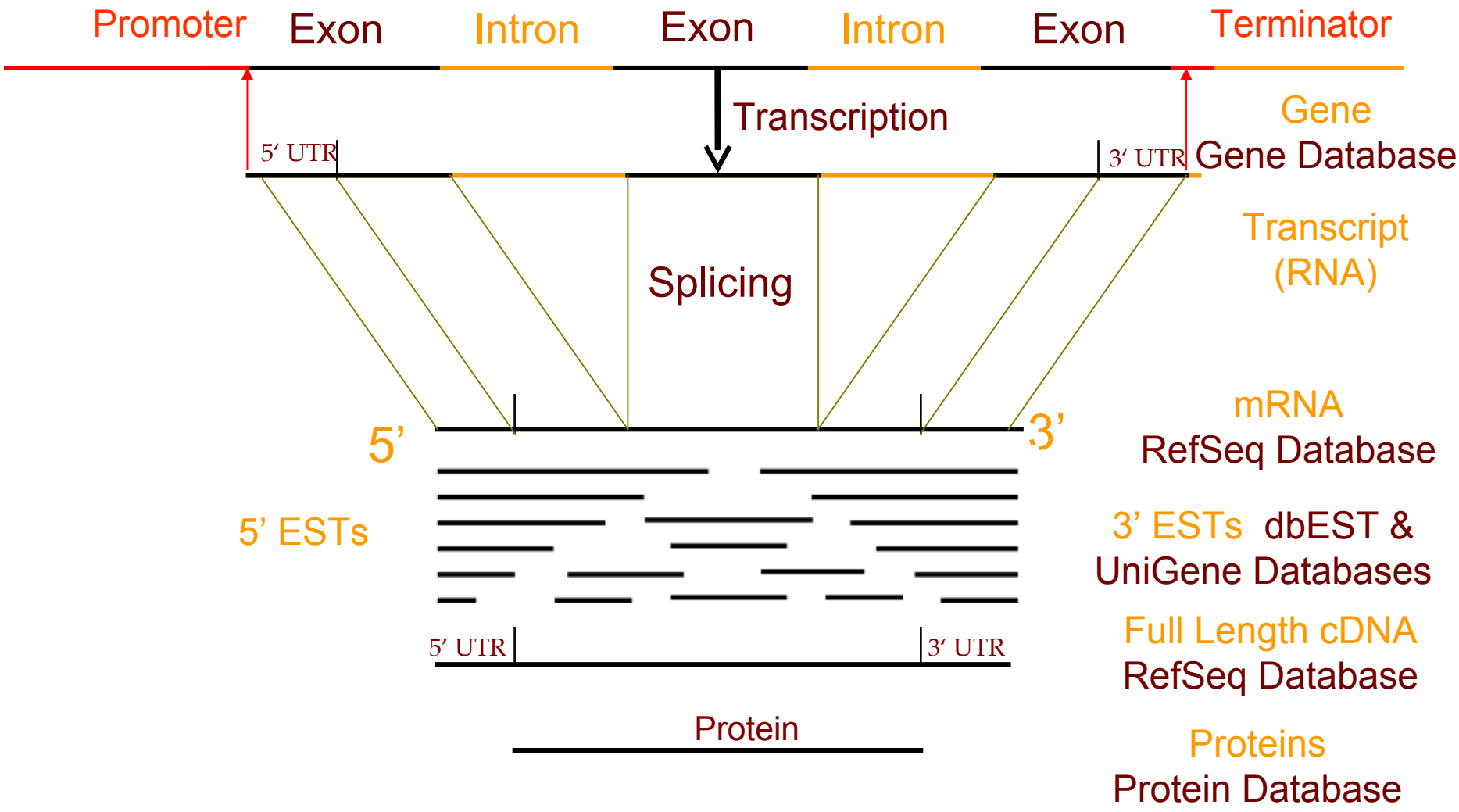
Alternative Splicing Generates Distinct Proteins in Different Tissues



Messenger RNA (mRNA) Databases
ESTs UniGene & Full Length cDNA RefSeq



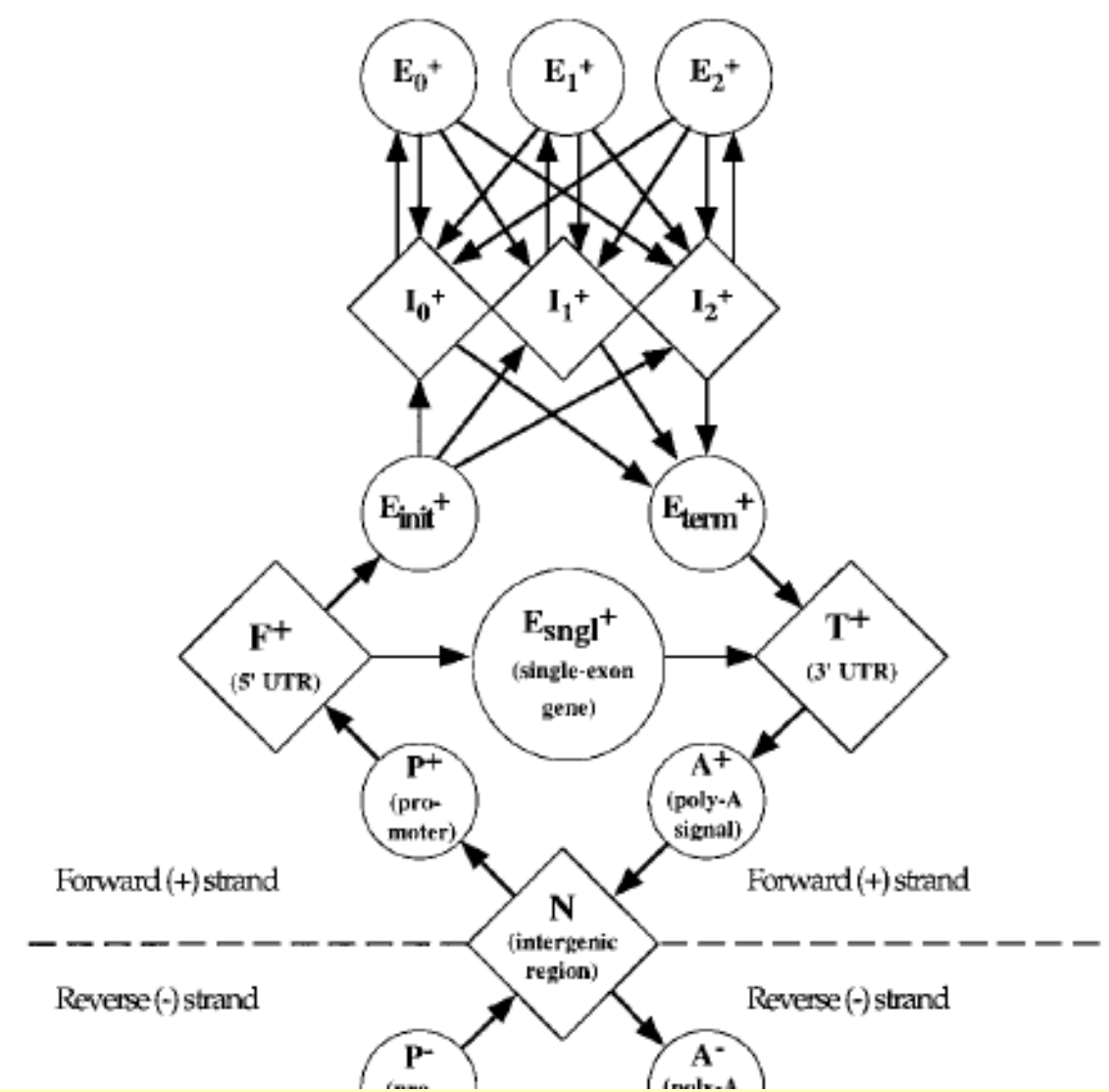
ESTs, Full Length cDNA
UniGene & RefSeq Databases



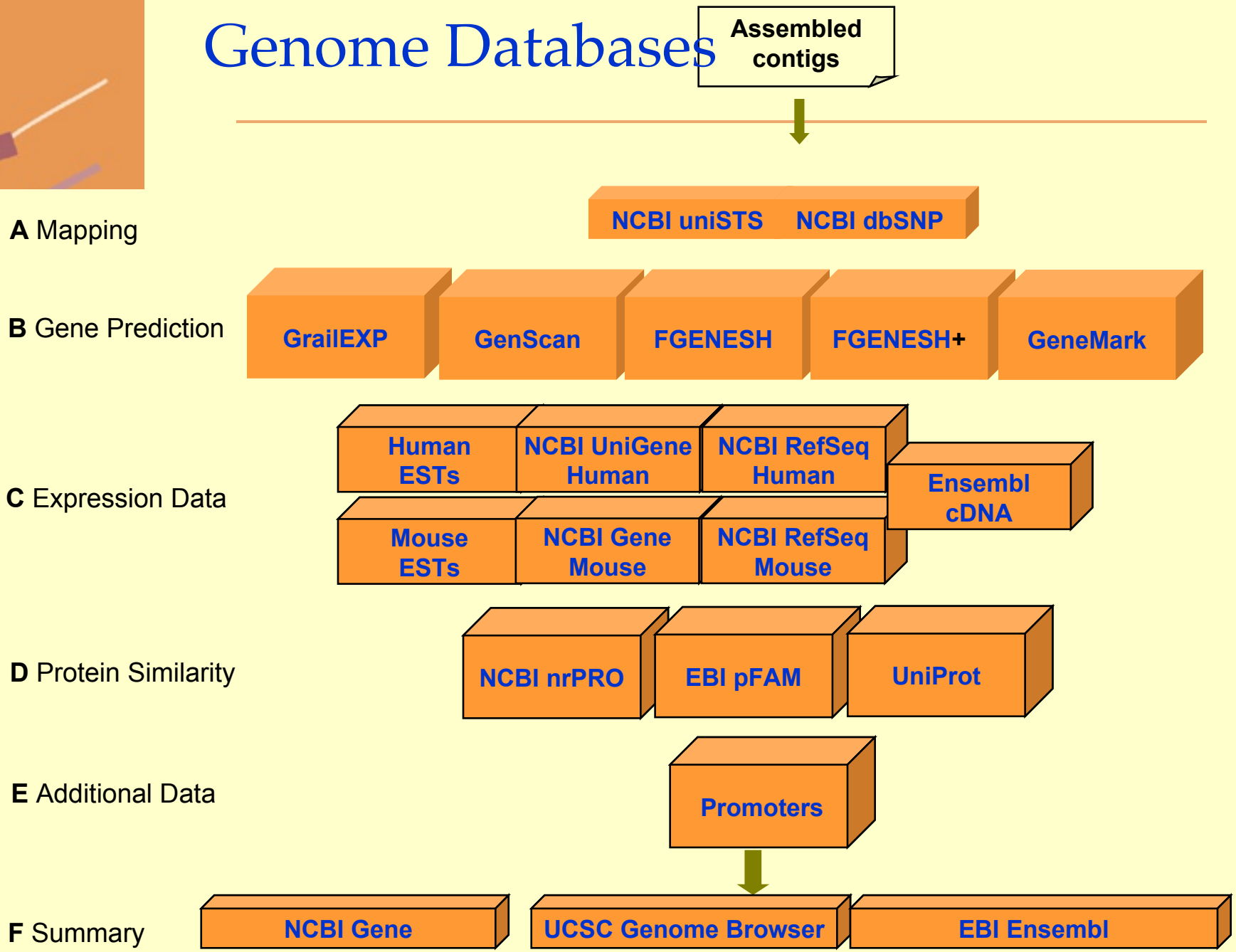
GENSCAN Gene Model

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

Hidden Markov models of gene structure

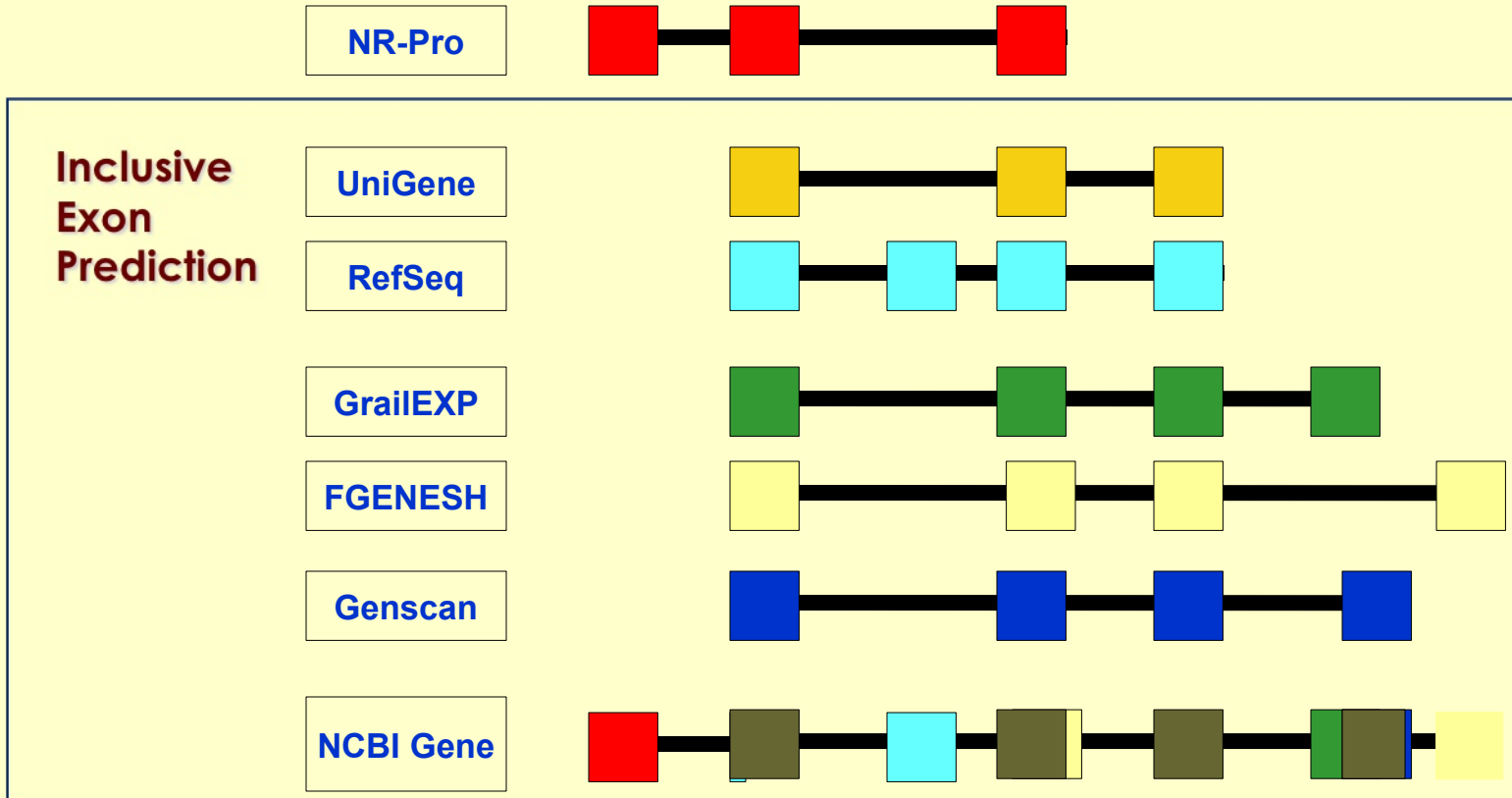


Genome Databases

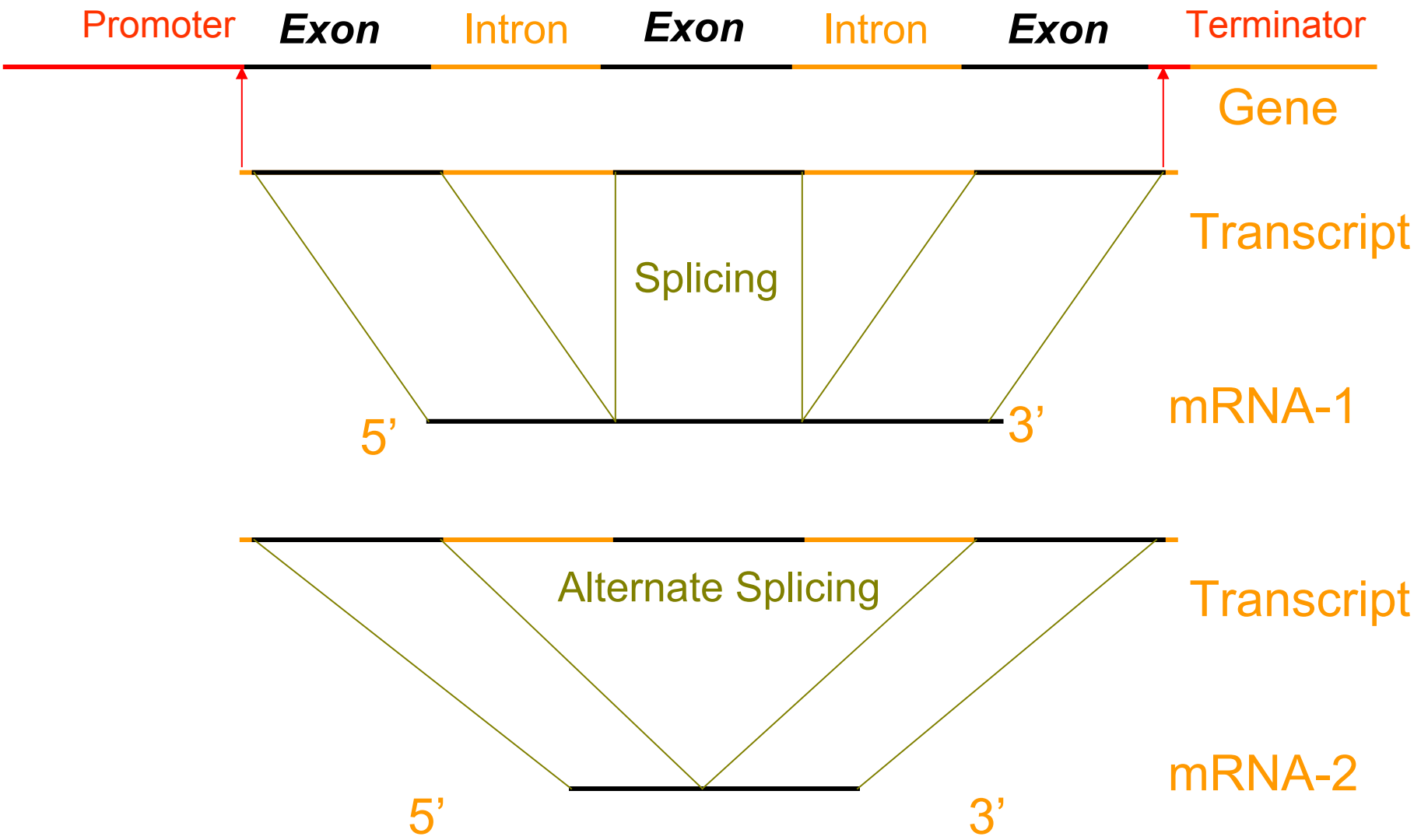


NCBIGene

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



Alternative Splicing Generates Distinct Proteins in Different Tissues



NCBI Genome

http://www.ncbi.nlm.nih.gov/genome/

NCBI Resources How To brutlag My NCBI Sign Out

Genome Genome dog Search

Limits Advanced Help

Genome

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

Using Genome

- Help
- Browse by Organism
- Download / FTP
- Submit a genome

Custom resources

- Human Genome
- Microbes
- Organelles
- Plants
- Viruses

Other Resources

- Assembly
- BioProject
- BioSample
- Map Viewer
- Protein Clusters

Genome Tools

- BLAST the Human Genome
- 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]

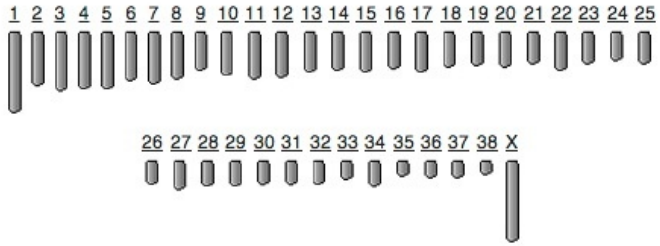
The dog, *Canis lupus familiaris*, is a useful model organism for medical research due to extensive genetic diversity and morphological variation within the species and to aggressive breeding practices that have resulted in inbred populations of dogs. Many breeds of dog are particularly susceptible to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

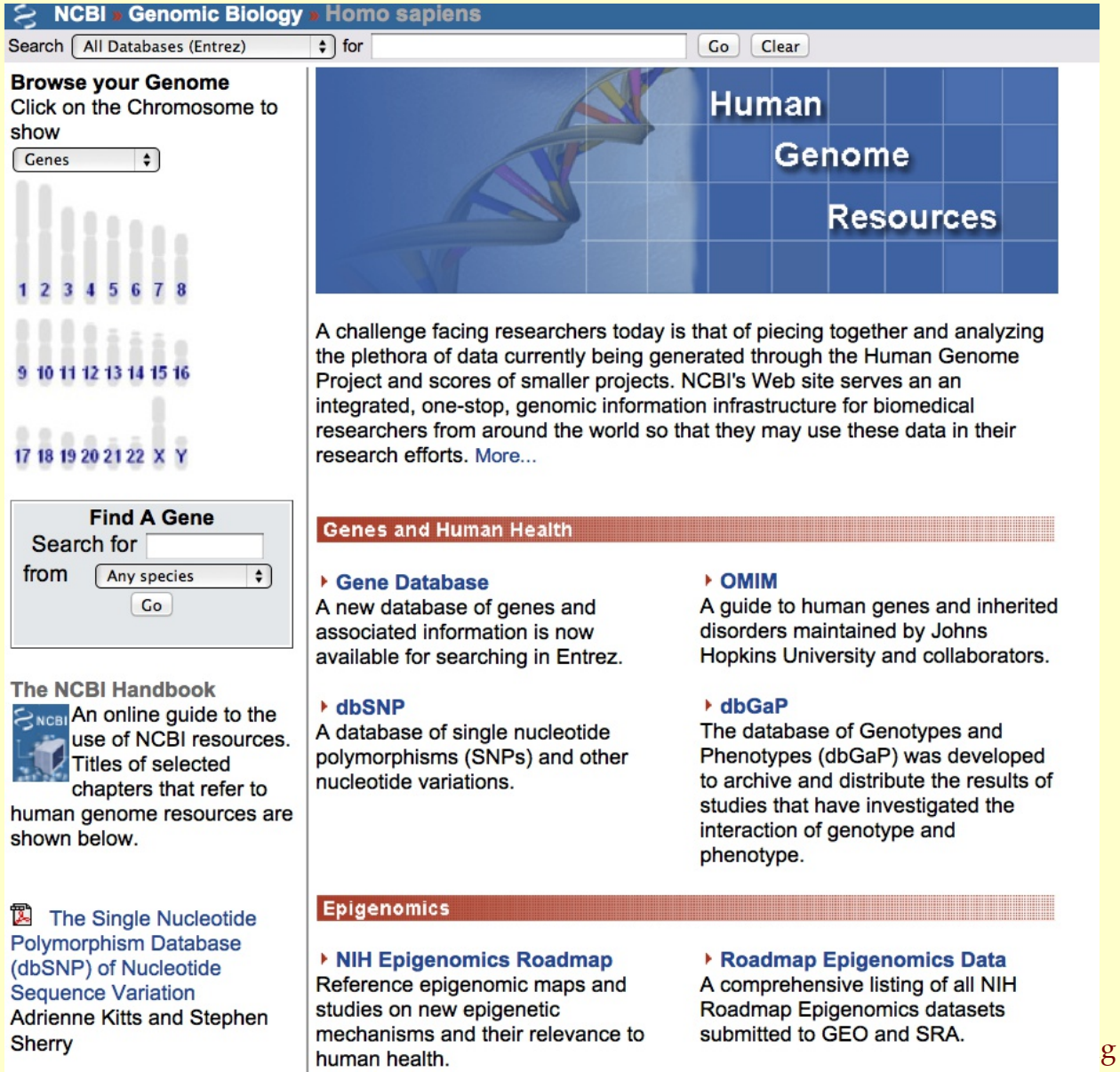
Chromosomes [1] Scaffolds or contigs [2] SRA or Traces [2]

Organism	BioProject	Assembly	Status	Chrs	Organelles	Size (Mb)	GC%	Gene	Protein
Canis lupus familiaris	PRJNA12384, PRJNA13179	CanFam3.1	●	39	1	2,410.98	35.5	28,864	42,303
Canis lupus familiaris	PRJNA10628	ASM18141v1	◑	-	-	1,517.48	41.1	-	
Canis lupus familiaris	PRJNA176193	Beagle	◑	-	-	2,254.63	40.7	-	
Canis lupus familiaris	PRJNA167192	-	◔	-	-	-	-	-	
Canis lupus familiaris	PRJNA186960	-	◔	-	-	-	-	-	

See more...

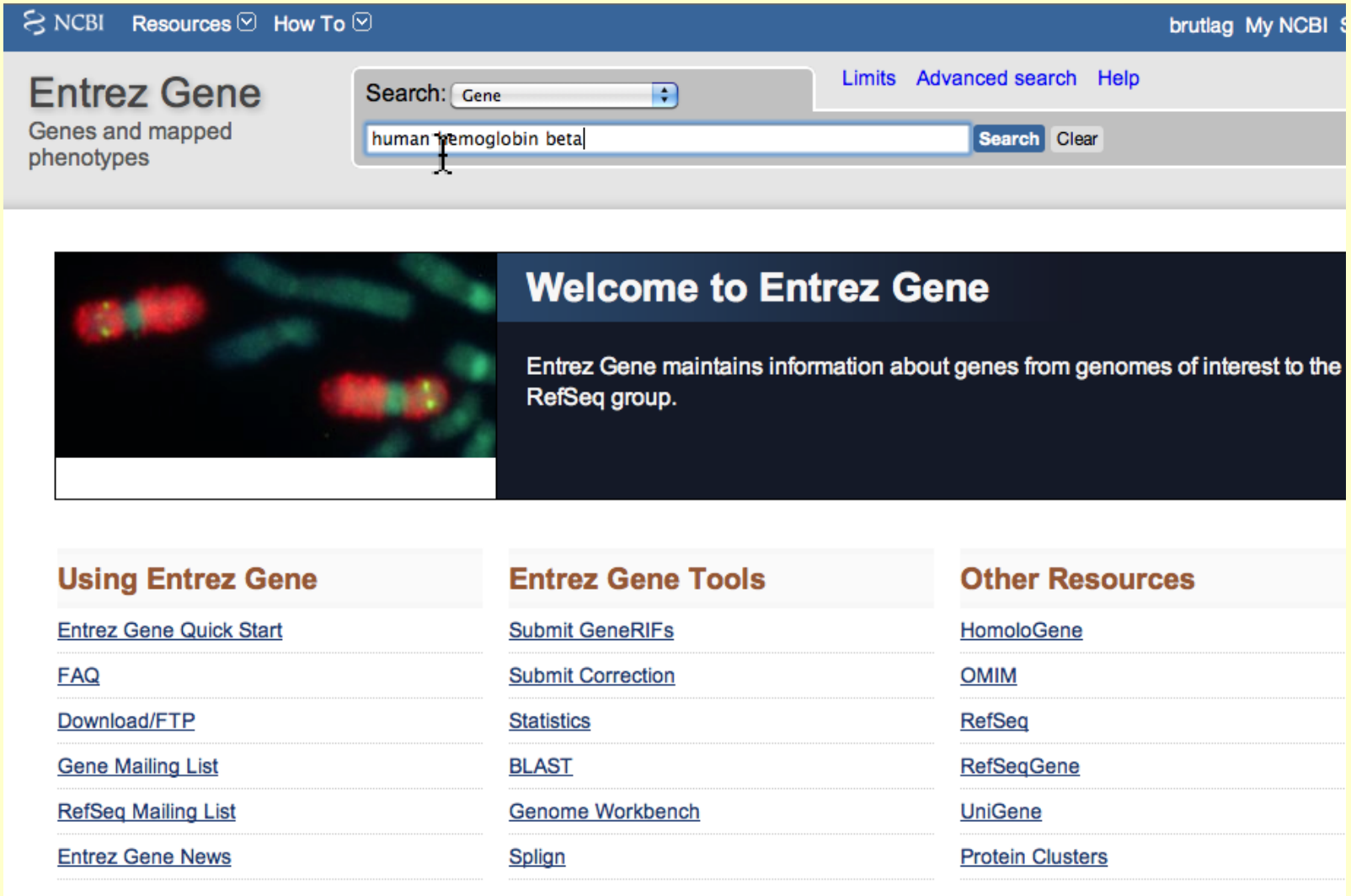
Human Genome Resources

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



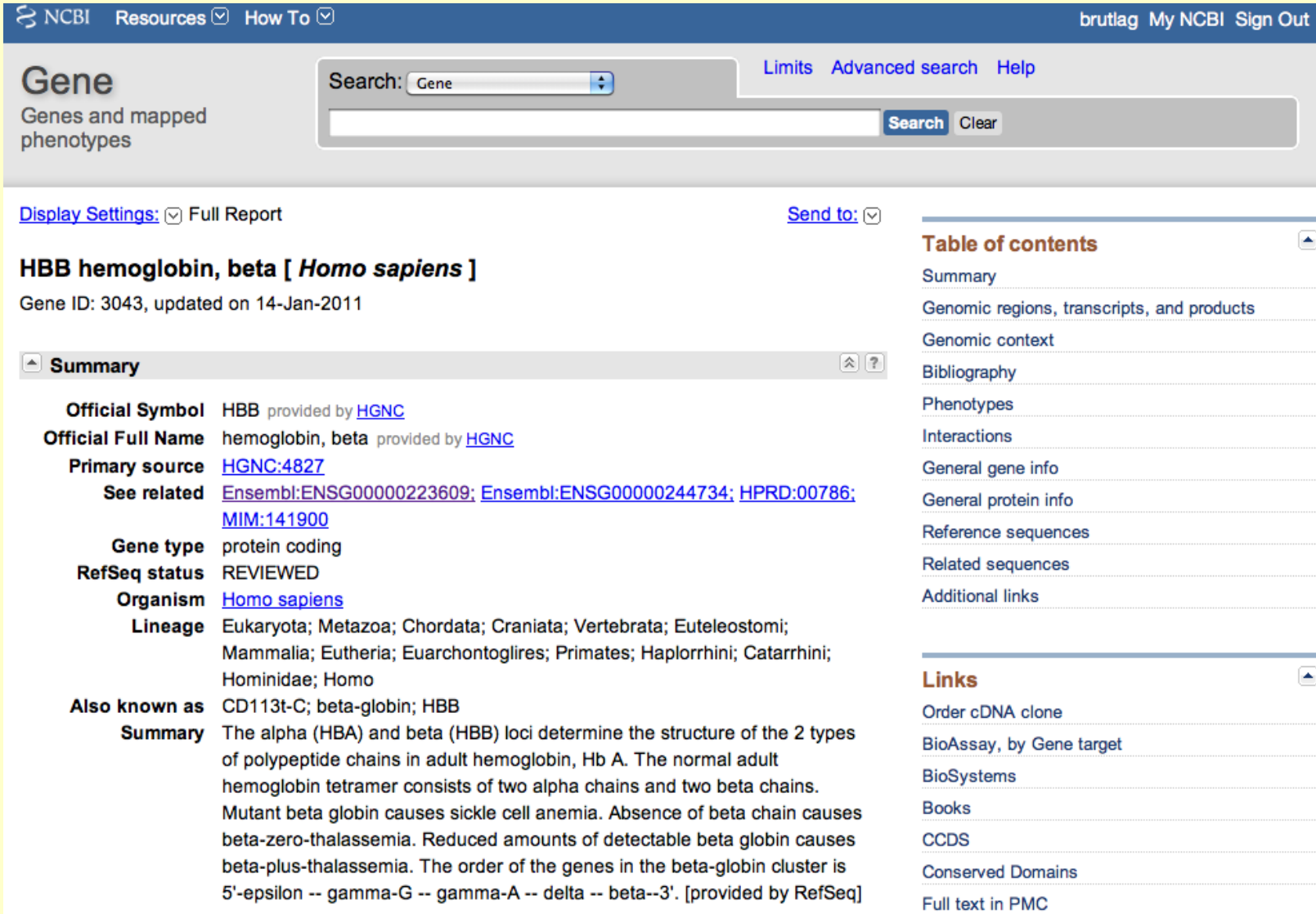
NCBI Entrez Gene

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



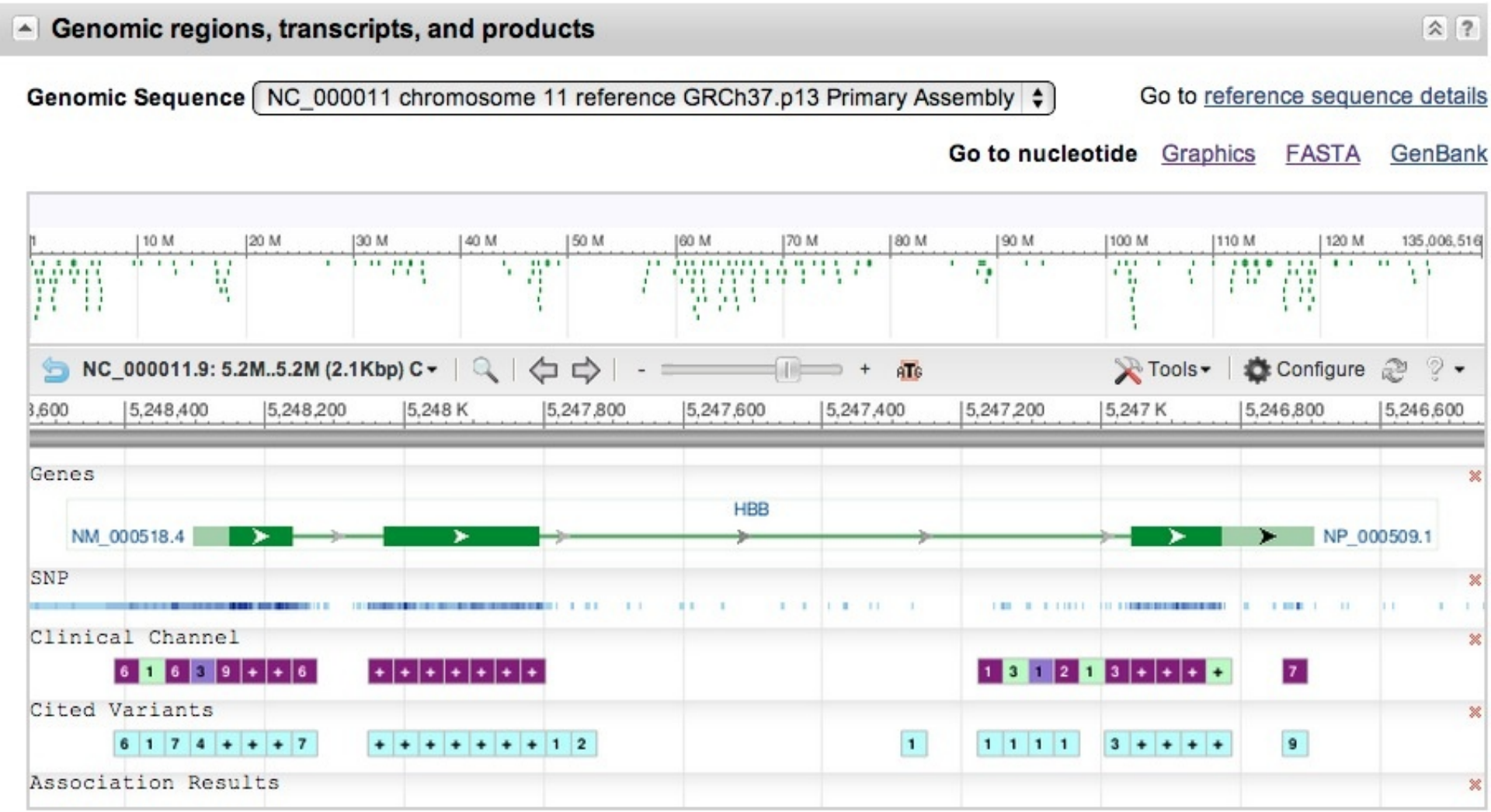
Human Beta-Hemoglobin Gene Entry

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



Human Beta-Hemoglobin Gene Entry

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



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 β-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 β-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 β-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) γ((A) γδβ)°-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
beta Thalassemia MedGen: C0005283, OMIM: 613985, GeneReviews: Beta-Thalassemia	Compare labs
Beta thalassemia, dominant inclusion body type MedGen: C1858990, OMIM: 603902, GeneReviews: Not available	Compare labs
Fetal hemoglobin quantitative trait locus 1 MedGen: C1841621, OMIM: 141749, GeneReviews: Not available	Compare labs
Hb SS disease MedGen: C0002895, OMIM: 603903, GeneReviews: Sickle Cell Disease	Compare labs
Heinz body anemias MedGen: C0700299, OMIM: 140700, GeneReviews: Not available	Compare labs
Susceptibility to malaria MedGen: C1970028, OMIM: 611162, GeneReviews: Not available	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

Human Beta-Hemoglobin Gene Entry

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

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 NC_000011 chromosome

Table of contents

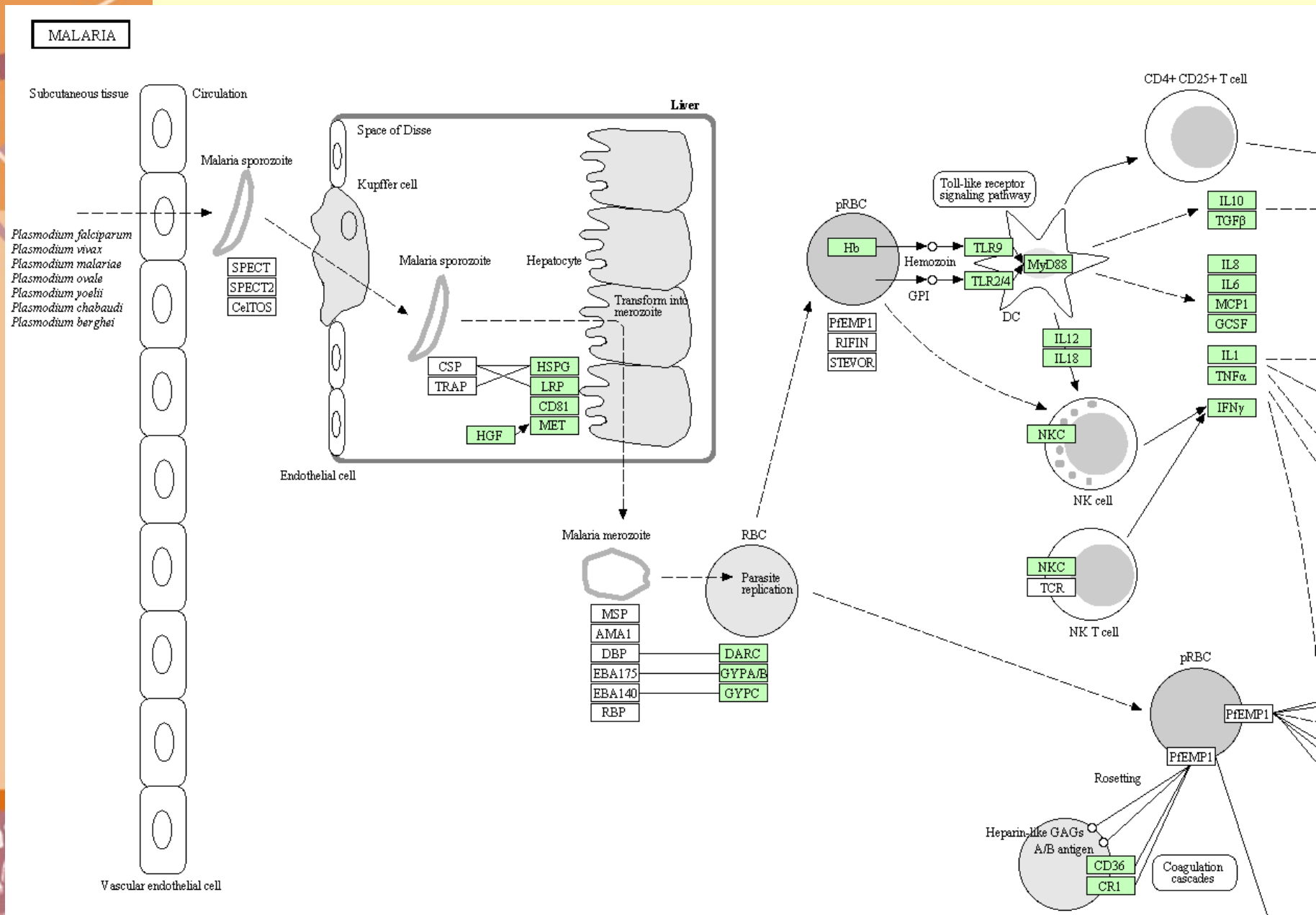
- Summary
- Genomic regions, transcripts, and products
- Genomic context
- Bibliography
- Phenotypes
- Interactions
- General gene info
- General protein info
- Reference sequences
- Related sequences
- Additional links

Links

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

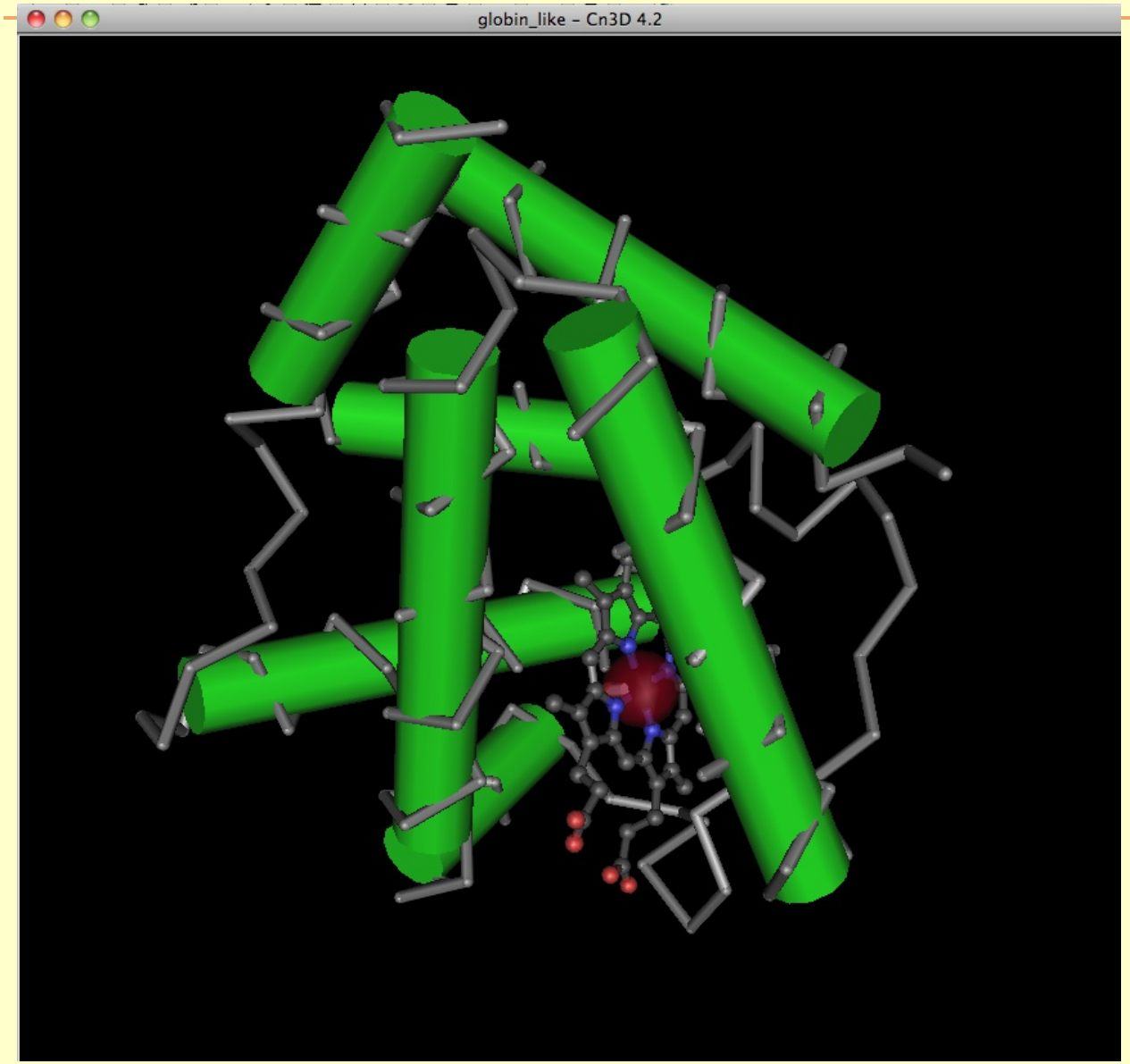
Human Beta-Hemoglobin Biosystems

http://www.ncbi.nlm.nih.gov/biosystems/152665



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

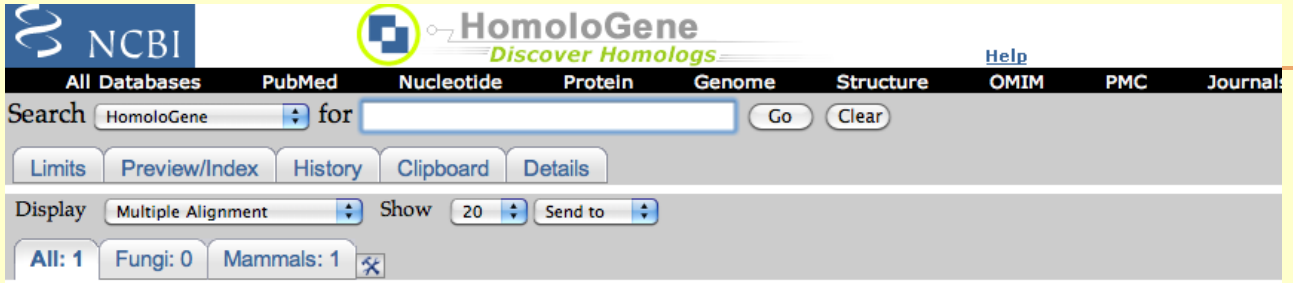
Display Settings: HomoloGene

HomoloGene:68066. Gene conserved in Eutheria

Genes	Proteins
Genes identified as putative homologs of one another during the construction of HomoloGene.	*Proteins used in sequence comparisons and their conserved domain architectures.*
HBB, *H.sapiens* hemoglobin, beta	NP_000509.1 147 aa
HBB, *P.troglodytes* hemoglobin, beta	XP_508242.1 147 aa
HBB, *M.mulatta* hemoglobin, beta	NP_001157900.1 147 aa
LOC609402, *C.lupus* hemoglobin subunit beta-like	XP_862442.1 162 aa
LOC480784, *C.lupus* hemoglobin subunit beta-like	XP_003433067.1 147 aa
HBD, *C.lupus* hemoglobin, delta	XP_534029.2 147 aa
LOC100337028, *B.taurus* hemoglobin fetal subunit beta-like	XP_002693271.1 145 aa
HBB, *B.taurus* hemoglobin, beta	NP_776342.1 145 aa
LOC781674, *B.taurus* hemoglobin fetal subunit beta-like	XP_003587018.1 145 aa
Beta-s, *M.musculus* hemoglobin subunit beta-1-like	NP_001188320.1 147 aa
Hbb-b1, *M.musculus* hemoglobin, beta adult major chain	NP_032246.2 147 aa
Hbb-b1, *R.norvegicus* hemoglobin, beta adult major chain	NP_942071.1 147 aa
LOC689064, *R.norvegicus* beta-globin	NP_001104739.1 147 aa
Hbb, *R.norvegicus* hemoglobin, beta	NP_150237.1 147 aa
LOC100134871, *R.norvegicus* beta globin minor gene	NP_001106694.1 147 aa

Human Beta-Hemoglobin Homologene

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



Multiple Sequence Alignment

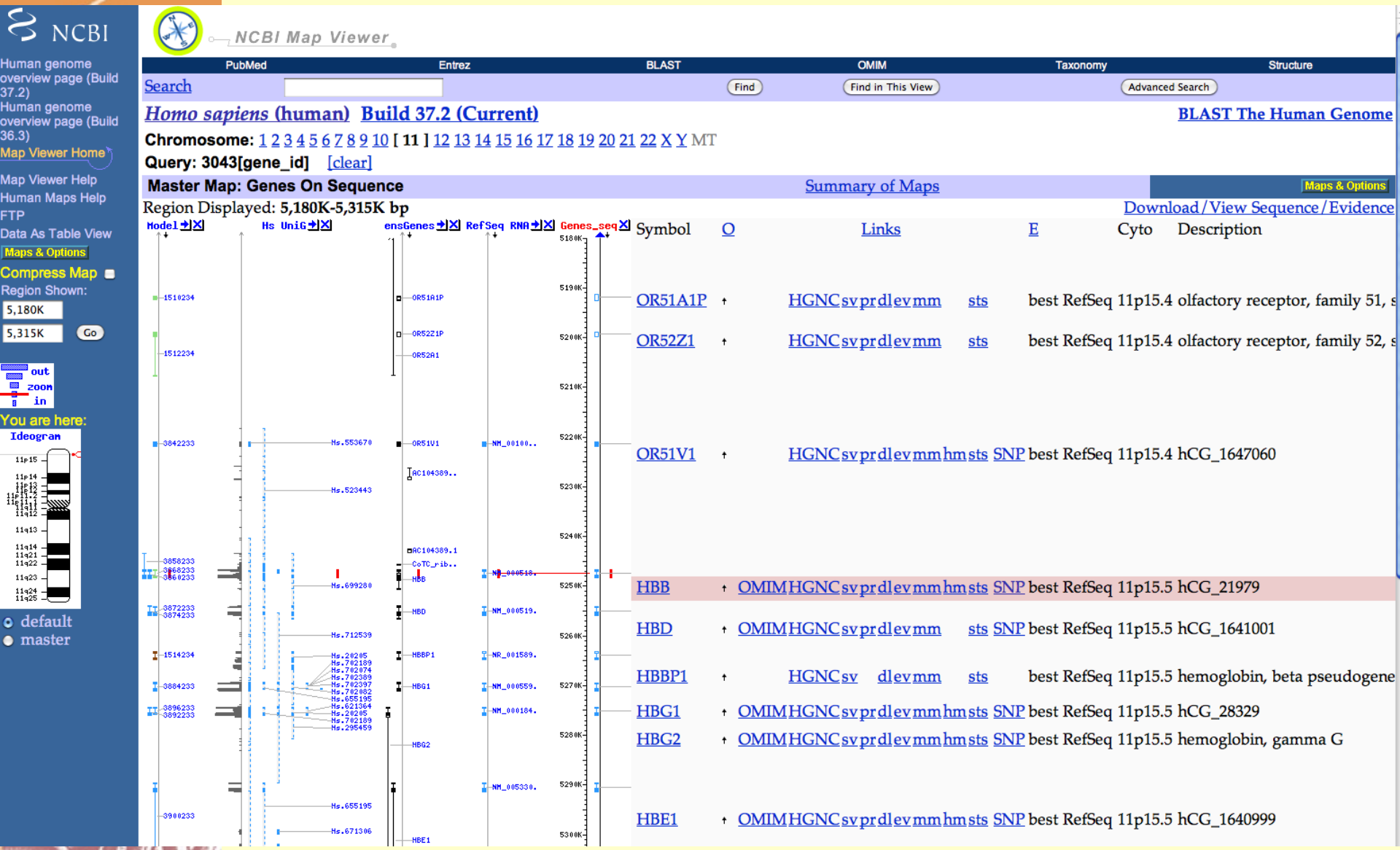
Generated by MUSCLE [see reference] version 3.6 (using option: -maxiters 2).

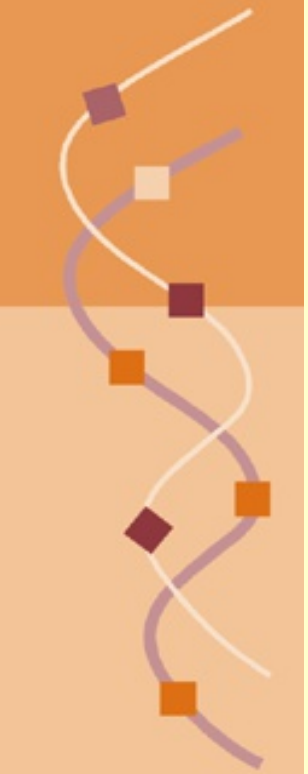
```
NP_000509.1          1   MVHLTPEEKSAVTALWGKVNVDEVGGEALGRLLVVYPWTQRFFESFGDLS   50
XP_508242.1          1   MVHLTPEEKSAVTALWGKVNVDEVGGEALGRLLVVYPWTQRFFESFGDLS   50
XP_850823.1          1   MVHLTAEEKSLVSGLWGKVNVDEVGGEALGRLLIVYPWTQRFFDSFGDLS   50
XP_534029.2          1   MVHLTAEEKSLISSMWGKVNVDEVGGEALGRLLIVYPWTQRFFDSFGDLS   50
XP_537902.1          1   MVHLTAEEKSLVSGLWGKVNVDEVGGEALGRLLIVYPWTQRFFDSFGDLS   50
XP_001249460.2       1   --MLSAEEKAAVTSLFAKVKVDEVGGEALGRLLVVYPWTQRFFESFGDLS   48
NP_001014902.1       1   --MLSAEEKAAVTSLFAKVKVDEVGGEALGRLLVVYPWTQRFFESFGDLS   48
XP_001252211.2       1   --MLSAEEKAAVTSLFAKVKVDEVGGEALGRLLVVYPWTQRFFESFGDLS   48
XP_001250142.2       1   --MLSAEEKAAVTSLFAKVKVDEVGGEALGRLLVVYPWTQRFFESFGDLS   48
NP_001103979.1       1   --MLSAEEKAAVTSLFAKVKVDEVGGEALGRLLVVYPWTQRFFESFGDLS   48
NP_776342.1          1   --MLTAEEKAAVTAFWGKVKVDEVGGEALGRLLVVYPWTQRFFESFGDLS   48
NP_032246.2          1   MVHLTDAEKAAVSGLWGKVNADEVGGEALGRLLVVYPWTQRYFDSFGDLS   50
NP_150237.1          1   MVHLTDAEKAAVNGLWGKVNPDDVGGEALGRLLVVYPWTQRYFDSFGDLS   50
XP_001069372.1       1   MVHLTDAEKATVSGLWGKVNADNVGAEALGRLLVVYPWTQRYFSKFGDLS   50
NP_942071.1          1   MVHLTDAEKATVNGLWGKVNPVEIGAESLASLLIVYPWTQRYFSKFGDLS   50

NP_000509.1         51   TPDAVMGNPKVKAHGKKVLGAFSDGLAHLDNLKGTFATLSELHCDKLHVD   100
XP_508242.1         51   TPDAVMGNPKVKAHGKKVLGAFSDGLAHLDNLKGTFATLSELHCDKLHVD   100
XP_850823.1         51   TPDAVMSNAKVKAHGKKVLNSFSDGLKNLDNLKGTFAKLSELHCDKLHVD   100
XP_534029.2         51   TPDAVMSNAKVKAHGKKVLNSFSDGLKNLDNLKGTFAKLSELHCDKLHVD   100
XP_537902.1         51   TPDAVMSNAKVKAHGKKVLNSFSDGLKNLDNLKGTFAKLSELHCDKLHVD   100
XP_001249460.2      49   SADAILGNPKVKAHGKKVLDSFCEGLKQLDDLKGAFASLSELHCDKLHVD   98
NP_001014902.1      49   SADAILGNPKVKAHGKKVLDSFCEGLKQLDDLKGAFASLSELHCDKLHVD   98
XP_001252211.2      49   SADAILGNPKVKAHGKKVLDSFCEGLKQLDDLKGAFASLSELHCDKLHVD   98
XP_001250142.2      49   SADAILGNPKVKAHGKKVLDSFCEGLKQLDDLKGAFASLSELHCDKLHVD   98
NP_001103979.1      49   SADAILGNPKVKAHGKKVLDSFCEGLKQLDDLKGAFASLSELHCDKLHVD   98
NP_776342.1         49   TADAVMNNPKVKAHGKKVLDSFSNGMKHLDDLKGTFAALSELHCDKLHVD   98
NP_032246.2         51   SASAIMGNAKVKAHGKKVITAFNDGLNHLDSLKGTFASLSELHCDKLHVD   100
NP_150237.1         51   SASAIMGNPKVKAHGKKVINAFNDGLKHLDNLKGTFAHLSELHCDKLHVD   100
XP_001069372.1      51   SASAIMGNPQVKAHGKKVINAFNDGLKHLDNLKGTFAHLSELHCDKLHVD   100
NP_942071.1         51   SVSAIMGNPQVKAHGEKVINAFDDGLKHLDNLKGTFASLSELHCDKLHVD   100
```

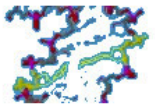
Human Beta-Hemoglobin Genome Map Viewer

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





Single Nucleotide Polymorphism



PubMed Nucleotide Protein Genome Structure PopSet Taxonomy OMIM Books SNP

Search for SNP on NCBI Reference Assembly

Search Entrez SNP for Go

BUILD 132

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

Go

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

Send rs# on all gene models to Batch Query Download all rs# to file. Genotype VarView

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 refresh

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

Region	Chr. position	mRNA pos	dbSNP rs# cluster id	Hetero-zygosity	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					

Variation Viewer for Human Beta-Hemoglobin

http://ww.ncbi.nlm.nih.gov/sites/varvu?gene=3043

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 *variation locations are based on these acc*
Links	HGMD, Panther, Gene, OMIM

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

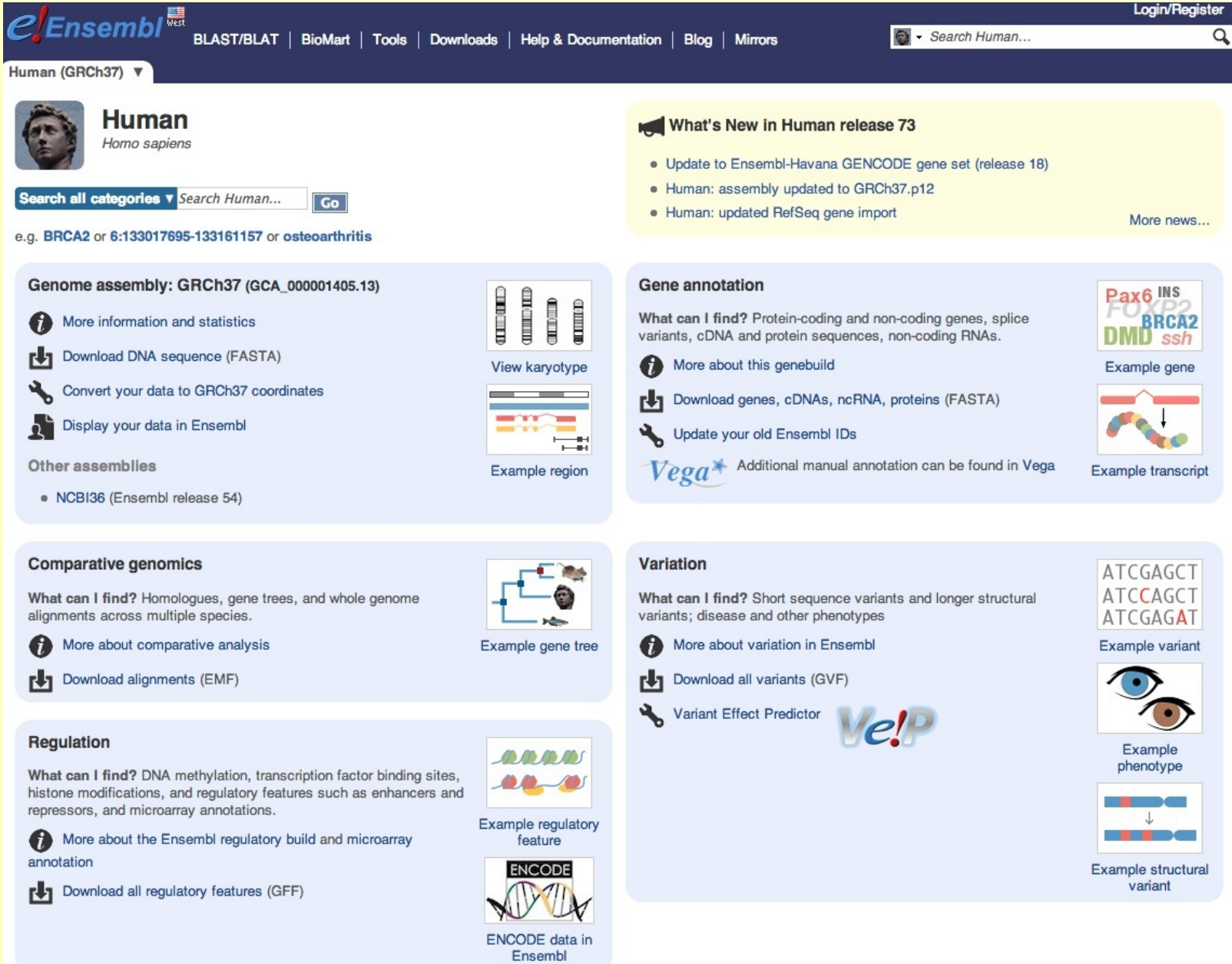
Var Class	Genomic	Transcript	Protein	Clinical interpretation	Test status	…	…	Freq	Pub…	MIM Al Var	Orig
SNC	g.70610C>G	c.16C>G	p.Pro6Ala			5	5			141900.0451	G
SNC	g.70611C>G	c.17C>G	p.Pro6Arg			3	3			141900.0296	G
DIP	g.70612delT	c.18delT	p.Pro6=fs			2	2				
DIP	g.70612delTins…	c.18delTinsCT	p.Pro6?fs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

Ensembl Home Page

http://www.ensembl.org/

Ensembl Human Genome

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



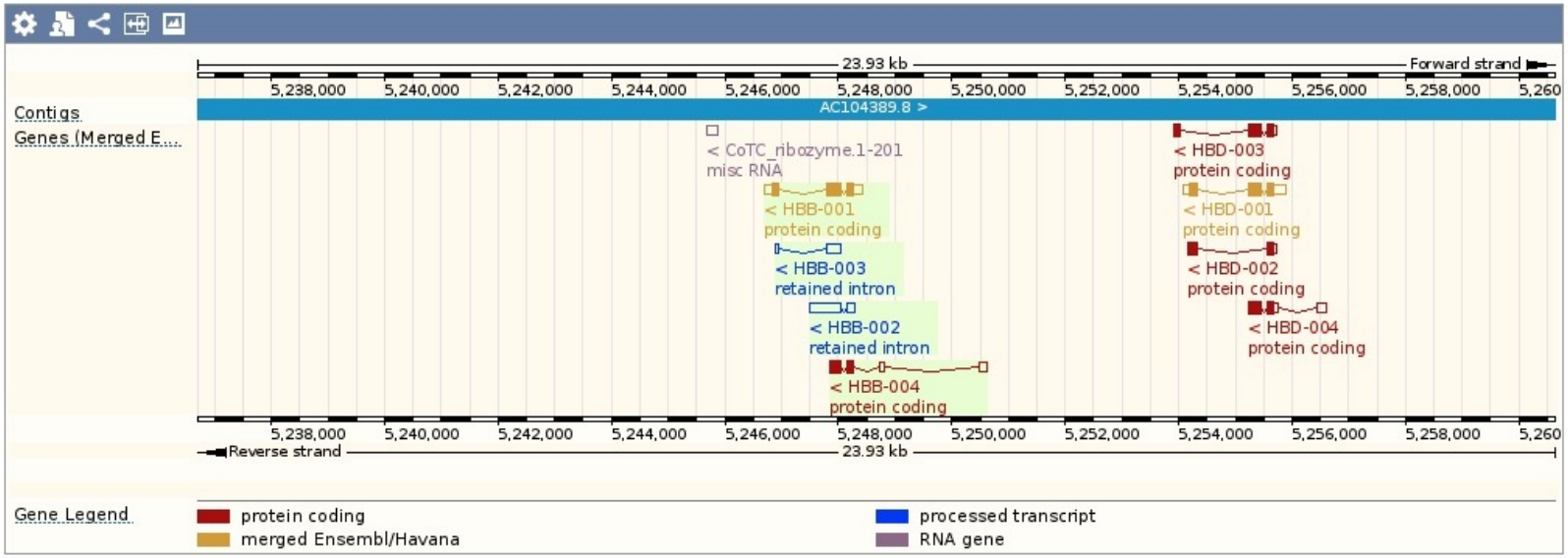
Gene: HBB ENSG00000244734

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

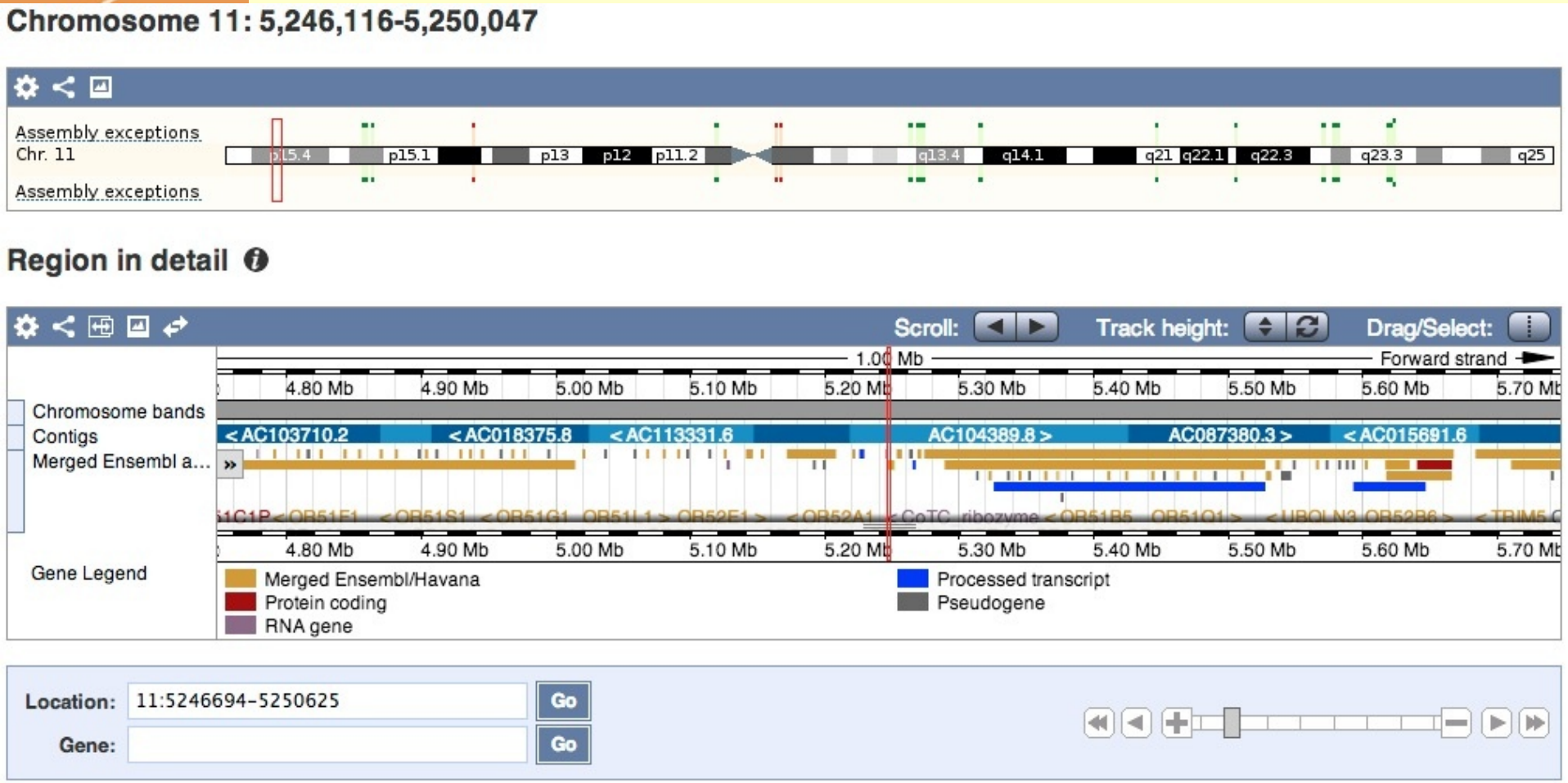
Name	HBB (HGNC Symbol)
Synonyms	beta-globin, CD113t-C, HBD [To view all Ensembl genes linked to the name click here.]
CCDS	This gene is a member of the Human CCDS set: CCDS7753
Ensembl version	ENSG00000244734.1
Gene type	Known protein coding
Prediction Method	Annotation for this gene includes both automatic annotation from Ensembl and Havana manual curation, see article.
Alternative genes	**This gene corresponds to the following database identifiers:** **Havana gene:** OTTHUMG00000066678 (version 2)

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



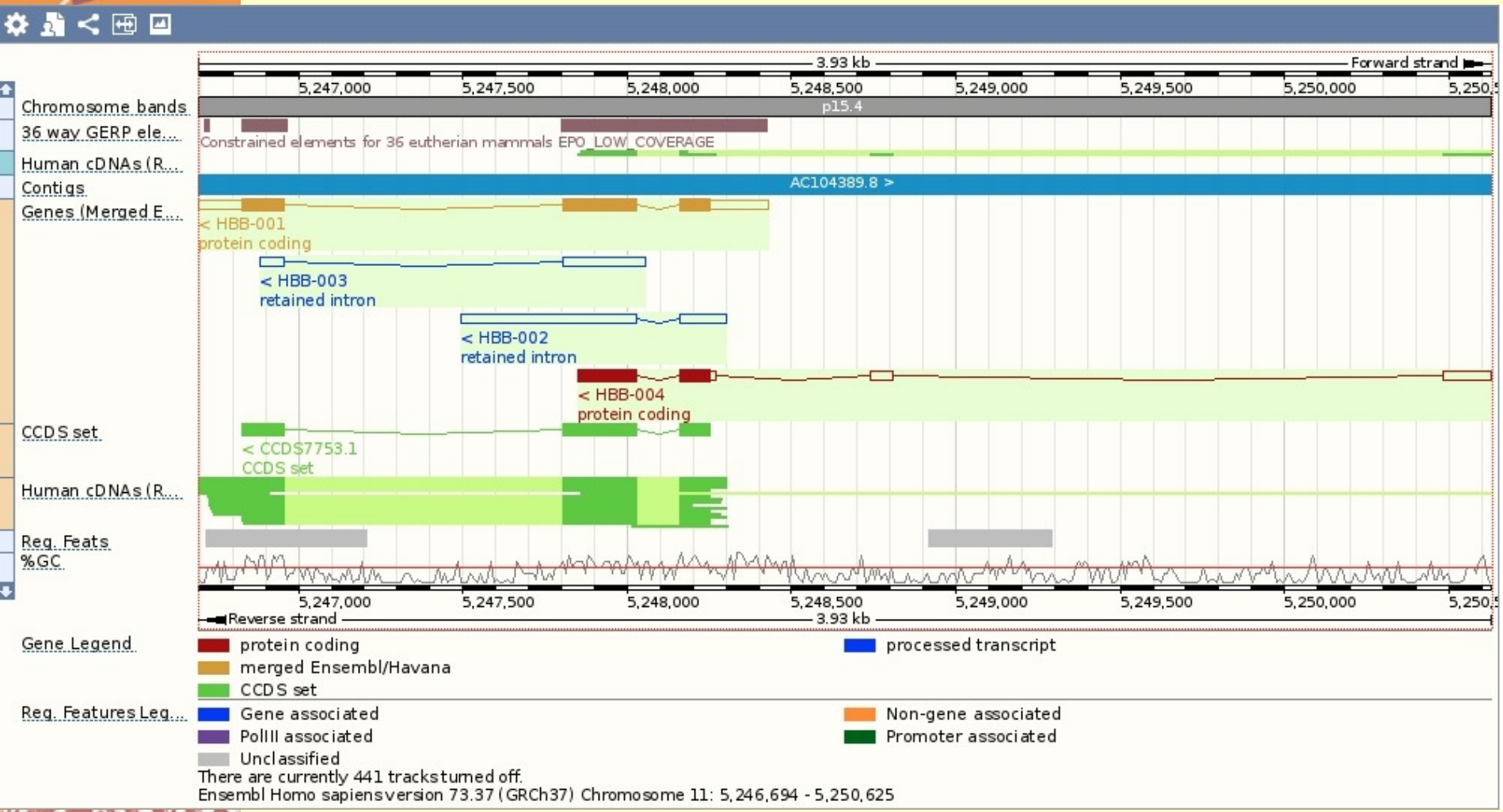
Ensembl Human HBB Gene

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



Ensembl Human HBB Gene Transcripts

http://uswest.ensembl.org/Homo_sapiens/Gene/Summary?db=core;g=ENSG00000244734;r=11:5246694-525062

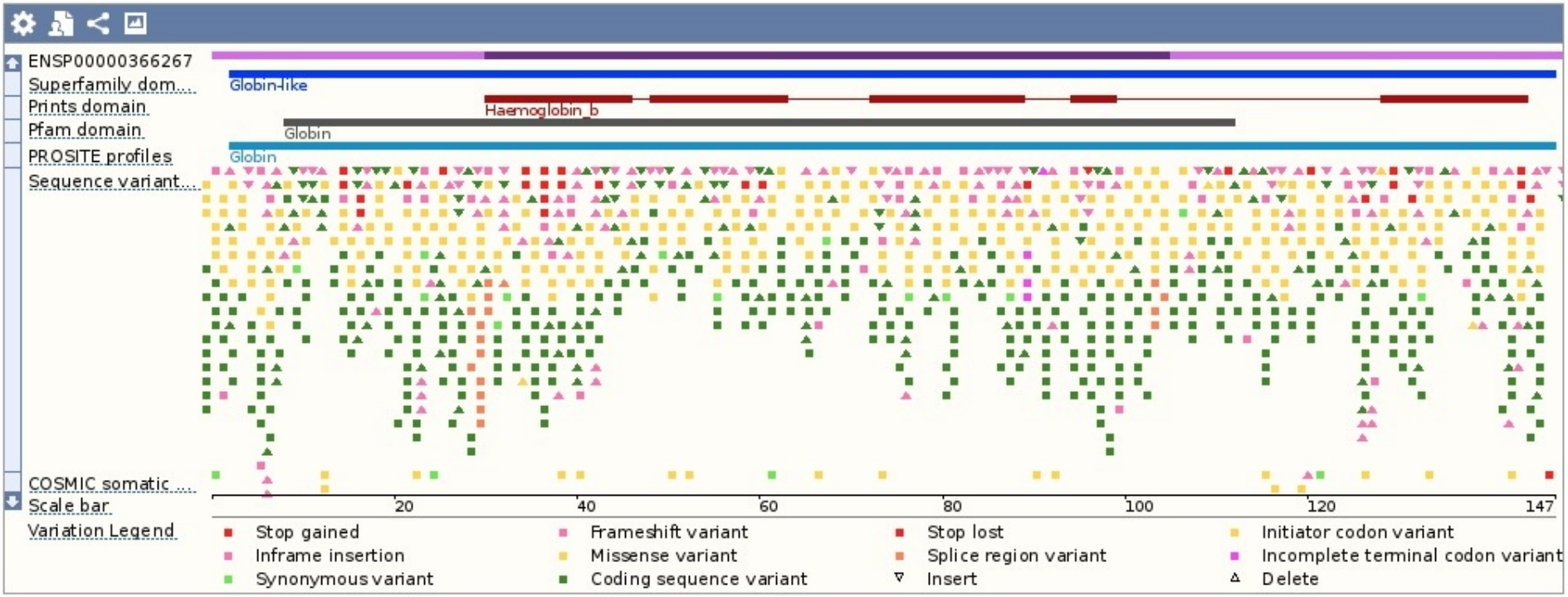


Ensembl Human HBB Protein Summary

http://uswest.ensembl.org/Homo_sapiens/Transcript/ProteinSummary?db=core;g=ENSG00000244734;r=11:524

Protein summary

Protein domains for ENSP00000333994.3

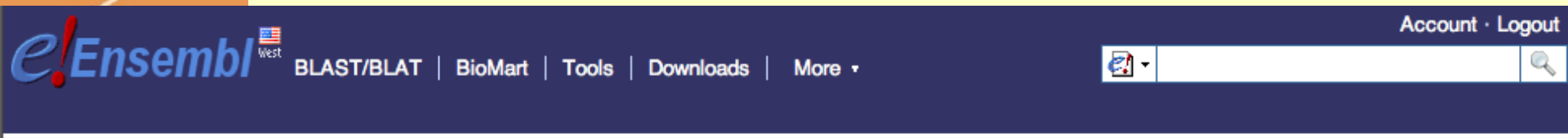


Statistics

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



Help & Documentation > Tutorials

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 Home Page

http://genome.ucsc.edu/

UCSC Genome Bioinformatics

Genomes - Blat - Tables - Gene Sorter - PCR - VisiGene - Session - FAQ - Help

Genome Browser

ENCODE

Neandertal

Blat

Table Browser

Gene Sorter

In Silico PCR

Genome Graphs

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.

UCSC Genome Browser

http://genome.ucsc.edu/cgi-bin/hgGateway

Genomes | Genome Browser | Tools | Mirrors | Downloads | My Data | About Us | Help

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.

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: submit

Click here to reset the browser user interface settings to their defaults.

track search | add custom tracks | track hubs | 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 Genome Browser

http://genome.ucsc.edu/cgi-bin/hgGateway

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 (uc010qav.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

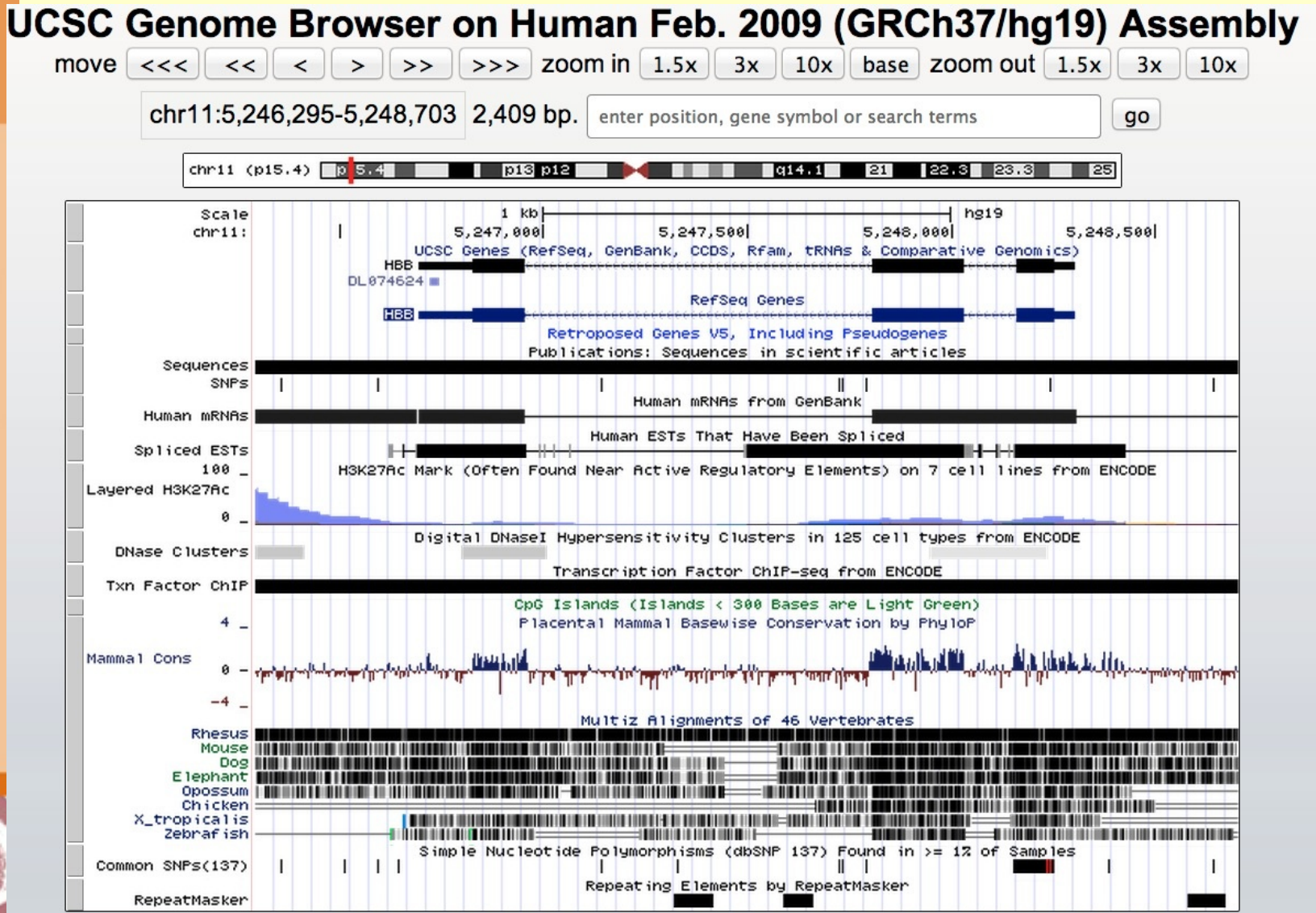
HBB at chr11:5246696-5248301 - (NM_000518) hemoglobin subunit beta
HBBP1 at chr11:5263185-5264822 - (NR_001589)

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

HBB Gene in the UCSC Genome Browser

http://genome.ucsc.edu/cgi-bin/hgTracks?position=chr11:5246696-5248301&hgsid=349247439&refGene=pack&



Mapping and Sequencing Tracks

refresh

Base Position: dense	Chromosome Band: hide	STS Markers: hide	FISH Clones: hide	Recomb Rate: hide	deCODE Recomb: hide
ENCODE Pilot: hide	Map Contigs: hide	Assembly: hide	GRC Map Contigs: hide	INSDC: hide	Gap: hide
BAC End Pairs: hide	Fosmid End Pairs: hide	GC Percent: hide	GRC Patch Release: hide	Hg18 Diff: hide	GRC Incident: hide
Hi Seq Depth: hide	Wiki Track: hide	BU ORChID: hide	Mapability: hide	Short Match: hide	Restr Enzymes: hide

Phenotype and Disease Associations

refresh

GAD View: hide	DECIPHER: hide	OMIM AV SNPs: hide	OMIM Genes: hide	OMIM Pheno Loci: hide	COSMIC: hide
LOVD Variants: hide	HGMD Variants: hide	UniProt Variants: hide	GWAS Catalog: hide	ISCA: hide	Coriell CNVs: hide
RGD Human QTL: hide	RGD Rat QTL: hide	MGI Mouse QTL: hide	GeneReviews: hide		

Genes and Gene Prediction Tracks

refresh

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

mRNA and EST Tracks

refresh

Human mRNAs: dense	Spliced ESTs: dense	Human ESTs: hide	Other mRNAs: hide	Other ESTs: hide	H-Inv: hide
UniGene: hide	Gene Bounds: hide	SIB Alt-Splicing: hide	Poly(A): hide	PolyA-Seq: hide	CGAP SAGE: hide
Human RNA Editing: hide					

Expression

refresh

Affy Exon Array: hide	Affy GNF1H: hide	Affy RNA Loc: hide	Affy U133: hide	Affy U133Plus2: hide	Affy U95: hide
Allen Brain: hide	Burge RNA-seq: hide	CSHL Small RNA-seq: hide	ENC Exon Array...: hide	ENC ProtGeno...: hide	ENC RNA-seq...: hide
GIS RNA PET: hide	GNF Atlas 2: hide	Illumina WG-6: hide	qPCR Primers: hide	RIKEN CAGE Loc: hide	Sestan Brain: hide

Regulation

refresh

ENCODE Regulation...: show	CD34 DnaseI: hide	CpG Islands: pack	ENC Chromatin...: hide	ENC DNA Methyl...: hide	ENC DNase/FAIRE...: hide
ENC Histone...: hide	ENC RNA Binding...: hide	ENC TF Binding...: hide	FSU Repli-chip: hide	ORegAnno: hide	Stanf Nucleosome: hide
SUNY SwitchGear: hide	SwitchGear TSS: hide	TFBS Conserved: hide	TS miRNA sites: hide	UMMS Brain Hist: hide	UW Repli-seq: hide
Vista Enhancers: hide	NKI Nuc Lamina...: hide	UCSF Brain Methyl: hide			

UCSC

Comparative Genomics

refresh

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

refresh

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

refresh

Variation and Repeats

refresh

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
GIS DNA PET: hide
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

UCSC Help File

http://genome.ucsc.edu/goldenPath/help/hgTracksHelp.html

UCSC Genome Bioinformatics

Home - Genomes - Blat - Tables - Gene Sorter - PCR - VisiGene - Proteome - Session - FAQ

Genome Browser User Guide

Table of Contents:

- What does the Genome Browser do?
- Getting started: Genome Browser gateways
- Fine-tuning the Genome Browser display
- Annotation track descriptions
- Using BLAT alignments
- Getting started on the Table Browser
- Getting started using Sessions
- Getting started on Genome Graphs
- Using the VisiGene Image Browser
- DNA text formatting
- Converting data between assemblies
- Downloading genome data
- Creating and Managing custom annotation tracks

Search the Genome Browser help pages: Submit

Search the Genome mailing list archives: Submit

See also the Open Helix tutorial and training materials.

Questions and feedback are welcome.

What does the Genome Browser do?

Ensembl Human Genome

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

UCSC Genome Home Page

http://genome.ucsc.edu/

UCSC Genome Bioinformatics

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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.

UCSC Genome Browser

http://genome.ucsc.edu/cgi-bin/hgGateway

Home | Genomes | Blat | Tables | Gene Sorter | PCR | Session | FAQ | Help

Human (*Homo sapiens*) Genome Browser Gateway

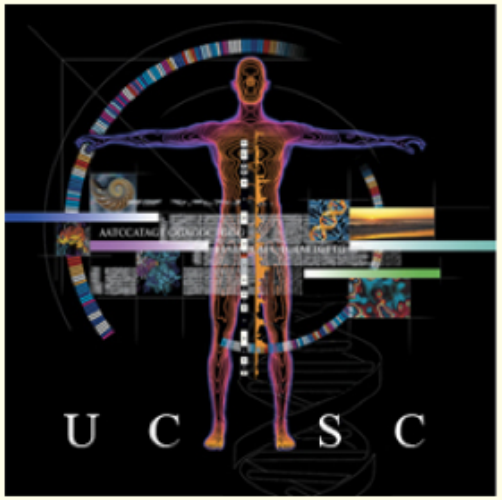
clade: Mammal | genome: Human | assembly: Feb. 2009 (GRCh37/hg19) | position or search term: opsin | gene | submit

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.



Homo sapiens
(Graphic courtesy of CBSE)

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.

UCSC

UCSC Genes

OPN4 (uc010qmk.1) at chr10:88414314-88426605 - opsin 4 isoform 2
OPN4 (uc009xsx.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 (uc010qde.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 (uc010qdf.1) at chr20:25052132-25062767 - visual system homeobox 1 isoform b
VSX1 (uc010qdd.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

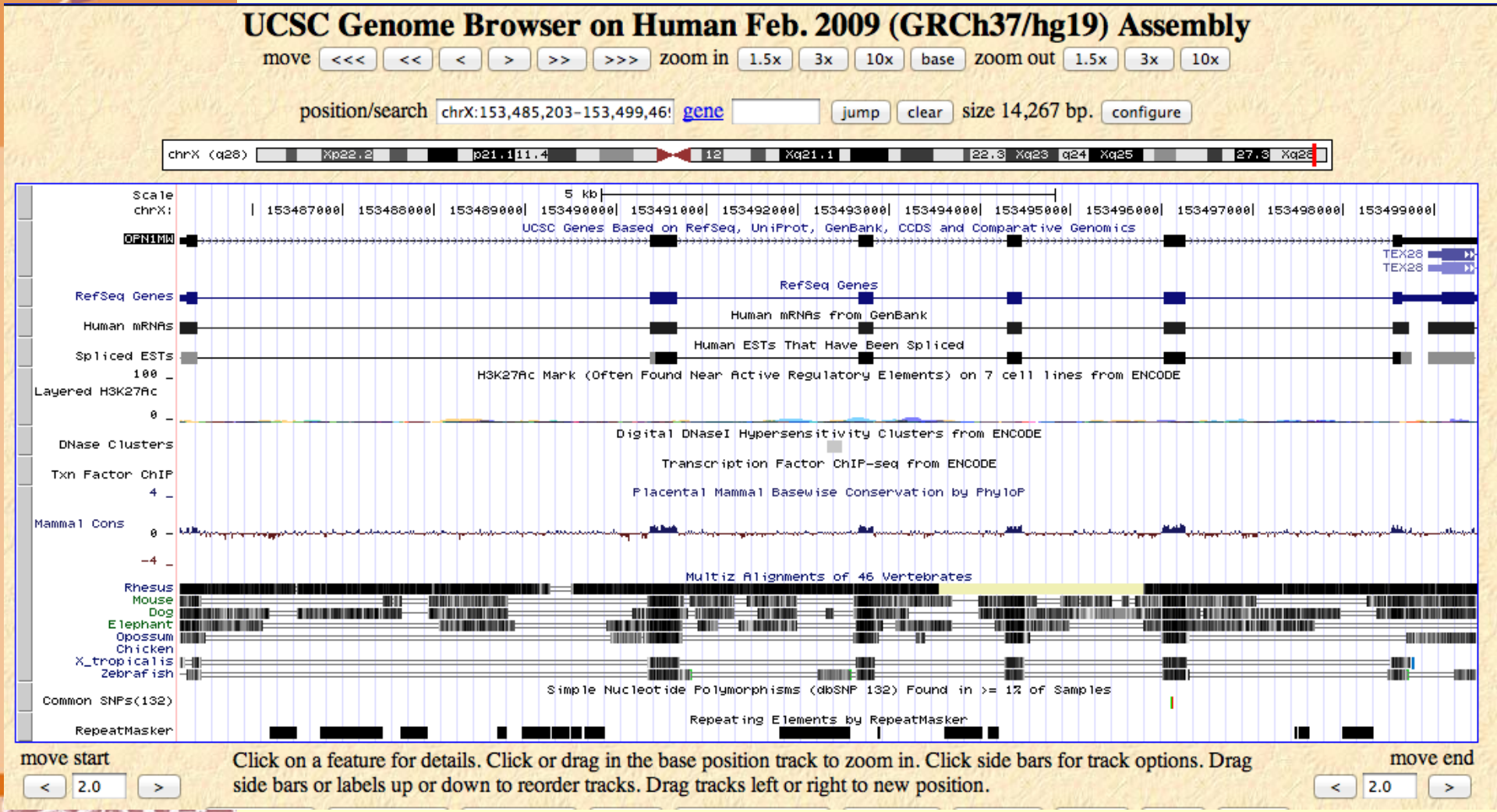
OPN1MW at chrX:153485203-153499470 - (NM_000513) medium-wave-sensitive opsin 1
OPN1MW at chrX:153448085-153462352 - (NM_000513) medium-wave-sensitive opsin 1
OPN5 at chr6:47749775-47794116 - (NM_181744) opsin-5
OPN1SW at chr7:128412543-128415844 - (NM_001708) short-wave-sensitive opsin 1
OPN1LW at chrX:153409725-153424507 - (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

http://genome.ucsc.edu/cgi-bin/hgTracks?position=chrX:153485203-153499469&hgsid=216983641&known



UCSC

http ...983641&known...

collapse 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.

expand all

Mapping and Sequencing Tracks

refresh

Base Position: dense	Chromosome Band: hide	STS Markers: hide	FISH Clones: hide	Recomb Rate: hide	Map Contigs: hide
Assembly: hide	GRC Map Contigs: hide	Gap: hide	BAC End Pairs: hide	Fosmid End Pairs: hide	GC Percent: hide
GRC Patch Release [No data-chrX]: hide	Hg18 Diff: hide	NCBI Incident: hide	Short Match: hide	Restr Enzymes: hide	Wiki Track: hide
BU ORChID: hide	Mapability: hide				

Phenotype and Disease Associations

refresh

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

Genes and Gene Prediction Tracks

refresh

UCSC Genes: pack	Alt Events: hide	GENCODE Genes V4: hide	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	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	H-Inv: hide
Gene Bounds: hide	SIB Alt-Splicing: hide	Poly(A): hide	CGAP SAGE: hide	Human RNA Editing: hide	

Expression

refresh

Affy Exon Array: hide	Affy GNF1H: hide	Affy RNA Loc: hide	Affy U133: hide	Affy U133Plus2: hide	Affy U95: hide
Allen Brain: hide	Burge RNA-seq: hide	ENC Exon Array...: hide	ENC ProtGeno...: hide	ENC RNA-seq...: hide	GNF Atlas 2: hide
Illumina WG-6	Sestan Brain				

UCSC

UCSC Help File

http://genome.ucsc.edu/goldenPath/help/hgTracksHelp.html

UCSC Genome Bioinformatics

Home - Genomes - Blat - Tables - Gene Sorter - PCR - VisiGene - Proteome - Session - FAQ

Genome Browser User Guide

Table of Contents:

- What does the Genome Browser do?
- Getting started: Genome Browser gateways
- Fine-tuning the Genome Browser display
- Annotation track descriptions
- Using BLAT alignments
- Getting started on the Table Browser
- Getting started using Sessions
- Getting started on Genome Graphs
- Using the VisiGene Image Browser
- DNA text formatting
- Converting data between assemblies
- Downloading genome data
- Creating and Managing custom annotation tracks

Search the Genome Browser help pages: Submit

Search the Genome mailing list archives: Submit

See also the Open Helix tutorial and training materials.

Questions and feedback are welcome.

What does the Genome Browser do?

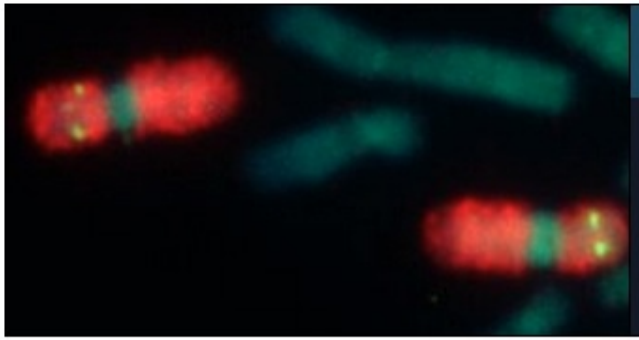
NCBI Gene

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

NCBI Resources How To brutlag My NCBI Sign Out

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

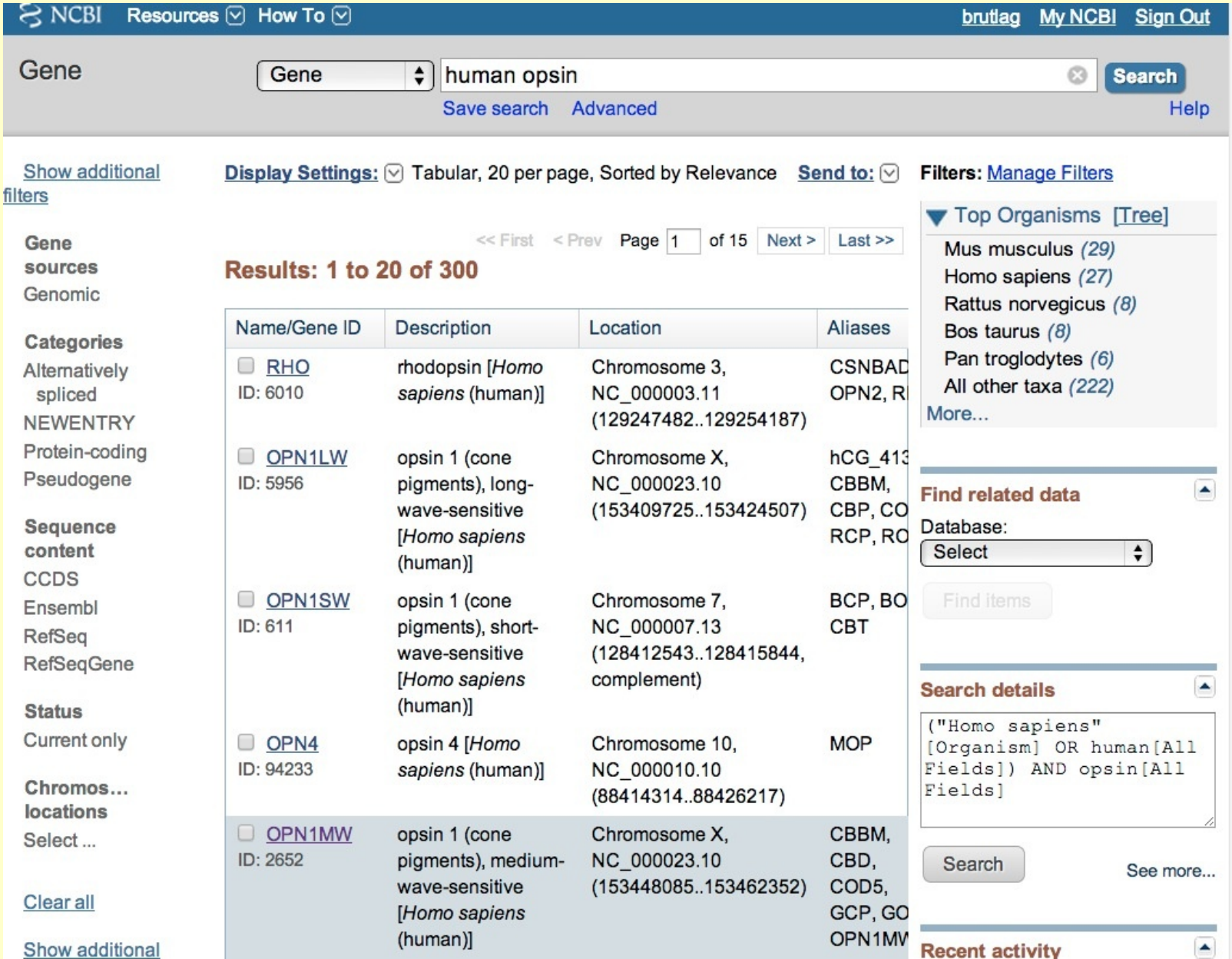
- Submit GeneRIFs
- Submit Correction
- Statistics
- BLAST
- Genome Workbench
- Splign

Other Resources

- HomoloGene
- OMIM
- RefSeq
- RefSeqGene
- UniGene
- Protein Clusters

NCBI Gene: Human Opsin

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



NCBI Gene: Human Opsin OPN1MW

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

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

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

Summary

Official Symbol	OPN1MW provided by HGNC
Official Full Name	opsin 1 (cone pigments), medium-wave-sensitive provided by HGNC
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

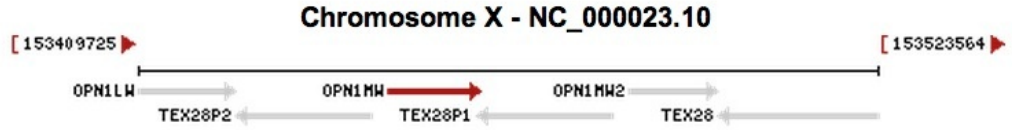
http://www.ncbi.nlm.nih.gov/gene/2652

Genomic context

Location: Xq28

Sequence: Chromosome: X; NC_000023.10 (153448085..153462352)

See OPN1MW in Epigenomics, MapViewer

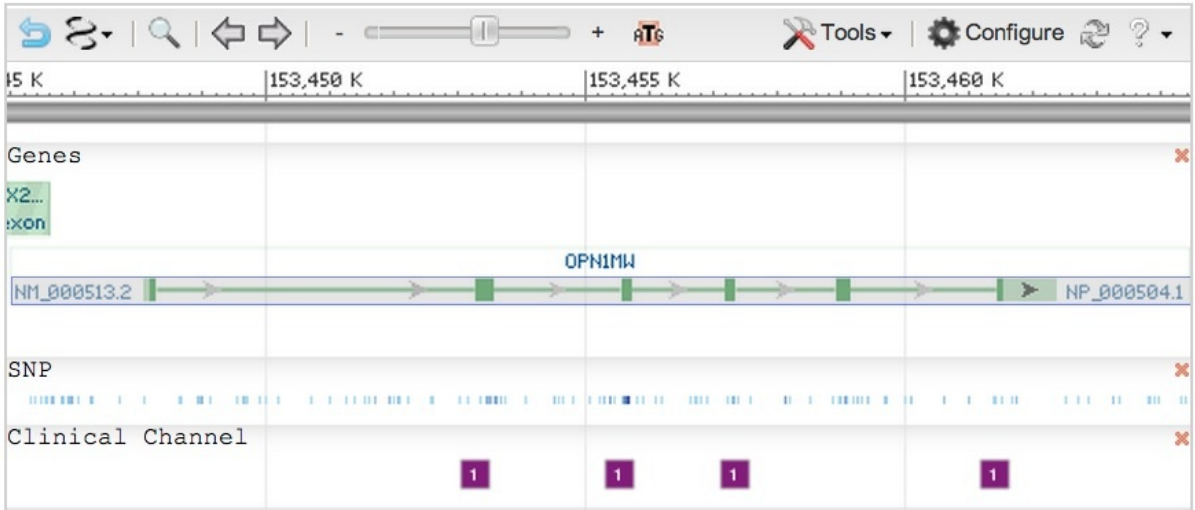


Genomic regions, transcripts, and products

Genomic Sequence NC_000023 chromosome X reference GRCh37.p13 Primary Assembly

Go to reference sequence details

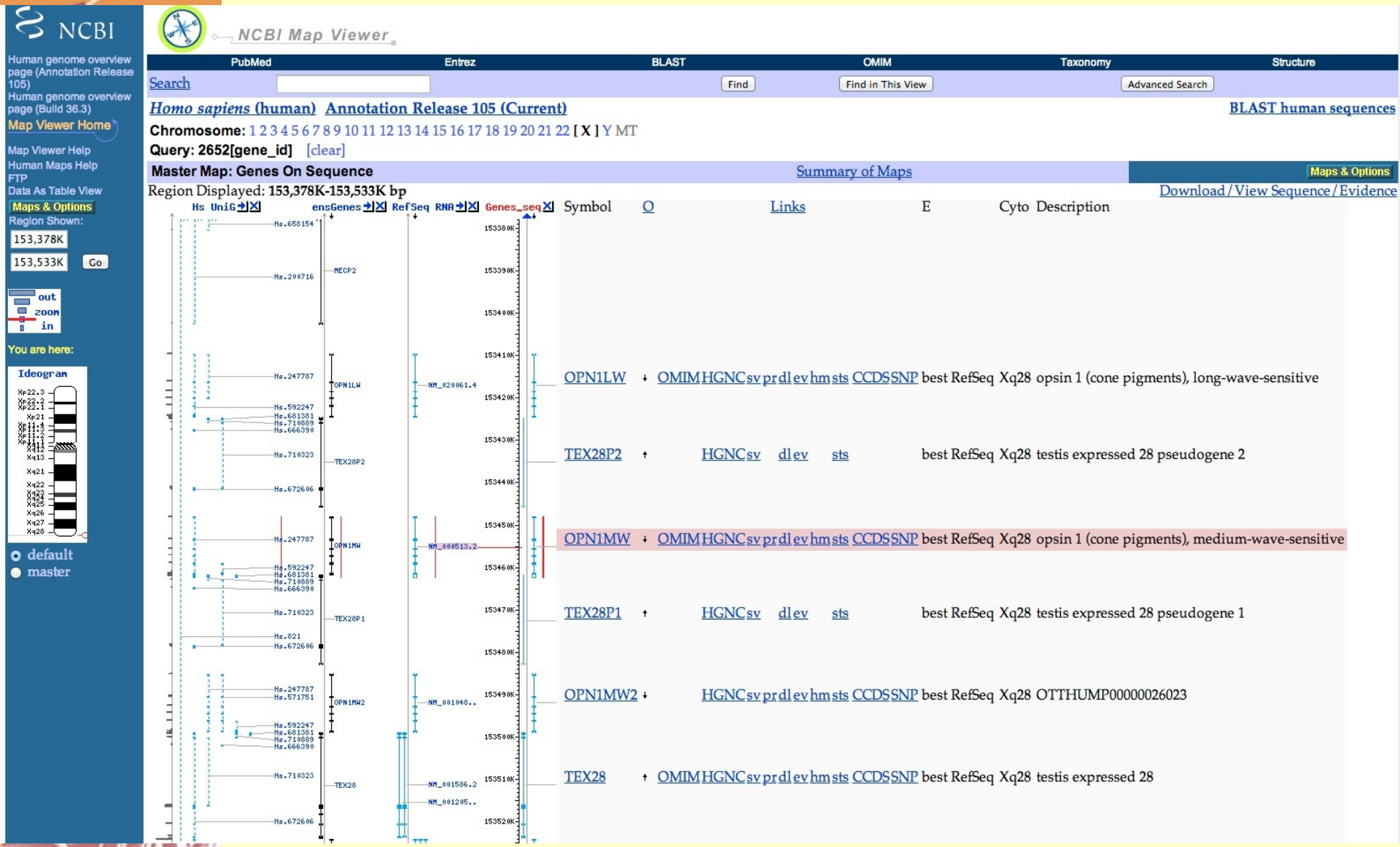
Go to nucleotide Graphics FASTA GenBank



- 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

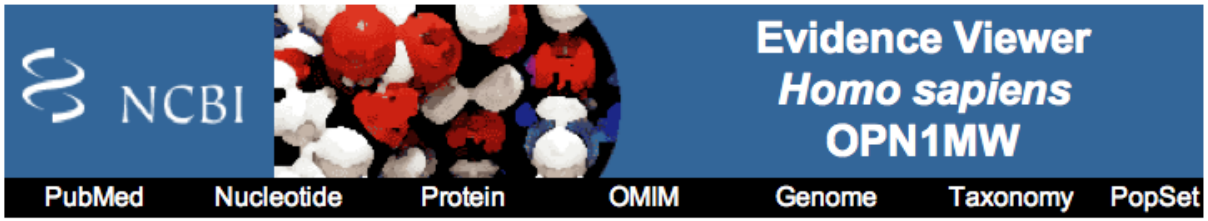
MapViewer: Human Opsin OPN1MW

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



Evidence Viewer for OPN1MW

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



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

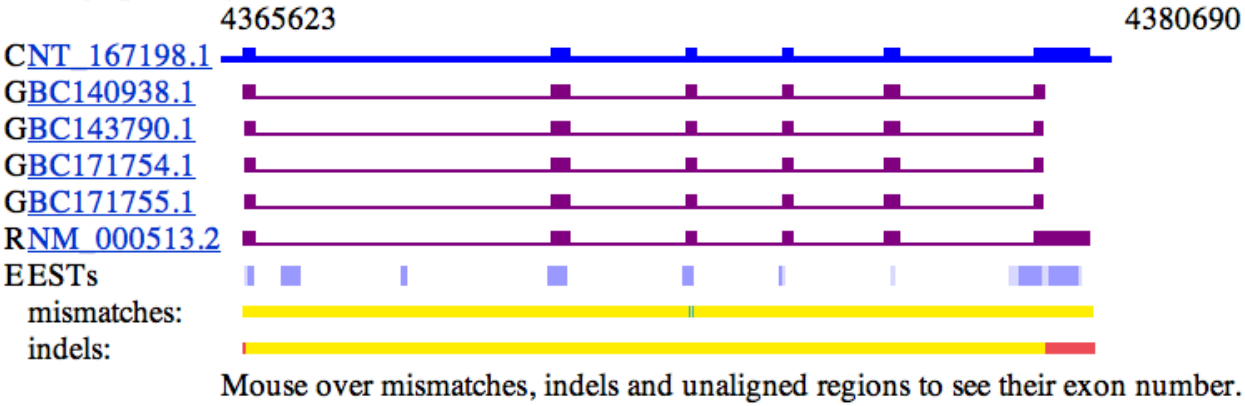
R = new since last genome build; R = updated since last genome build

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.

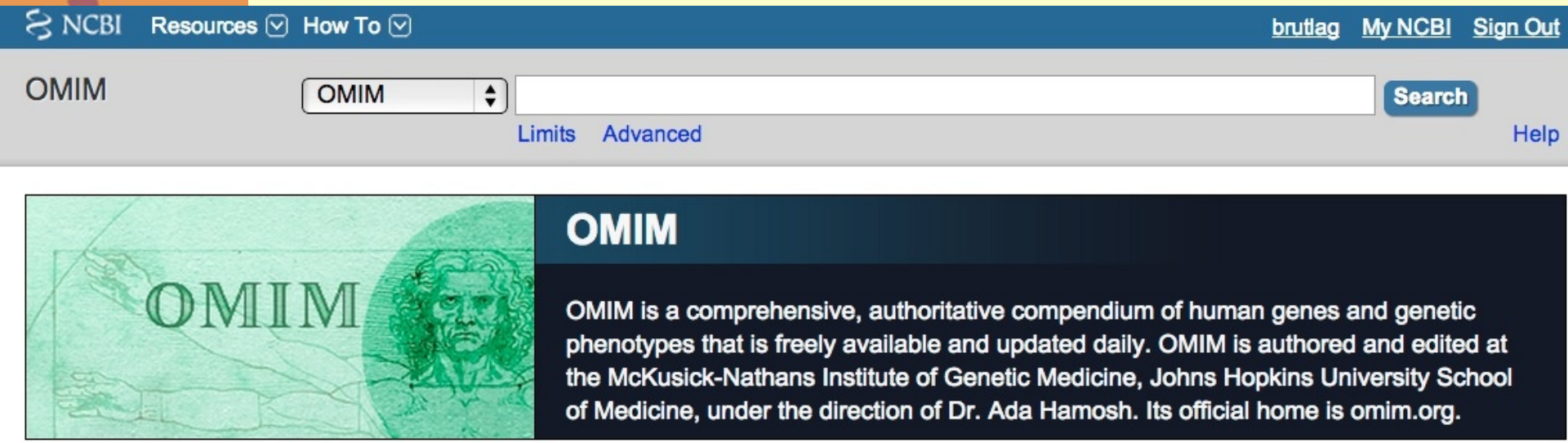
View graphic only



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

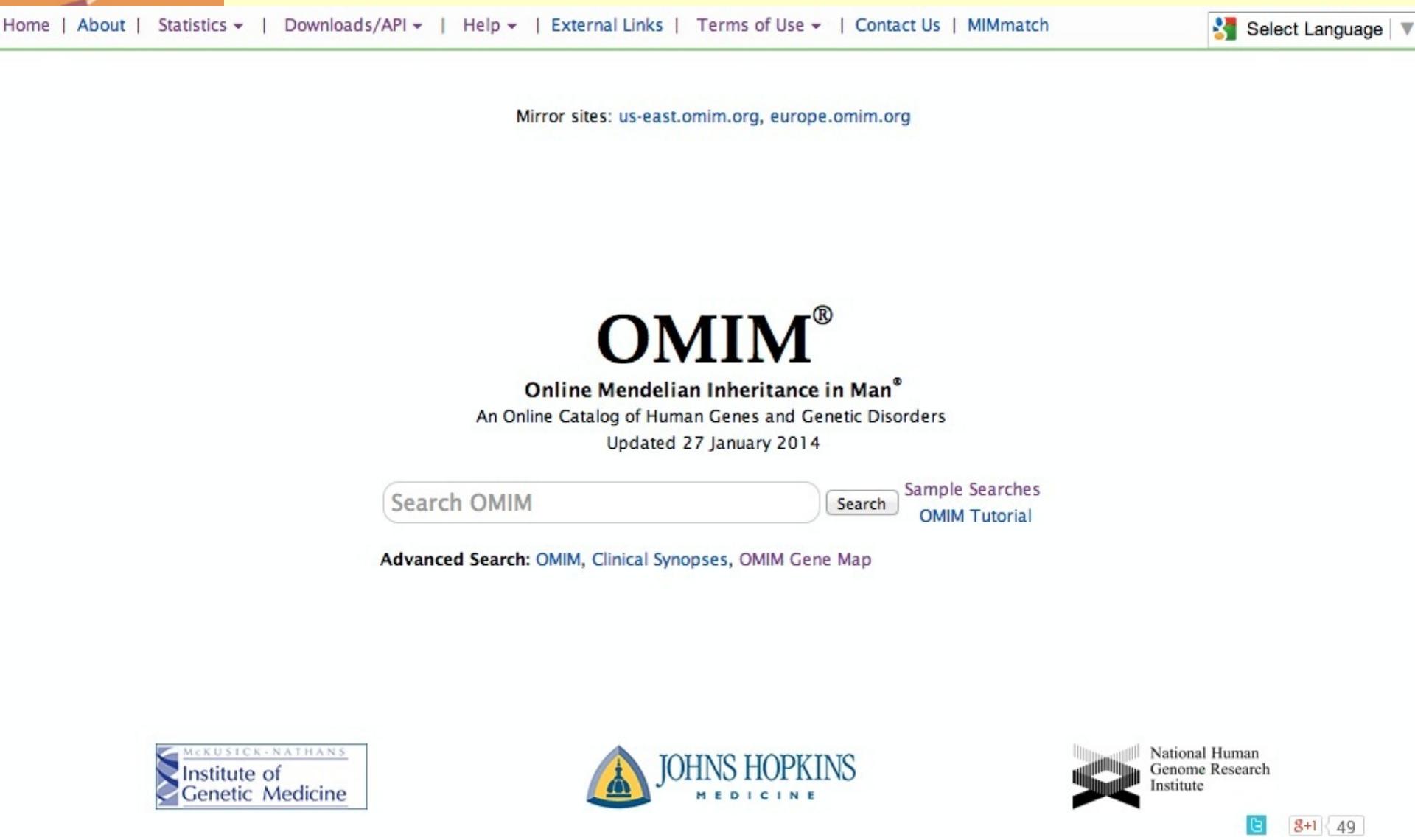
NCBI OMIM Home Page

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



OMIM Home Page

http://omim.org/



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

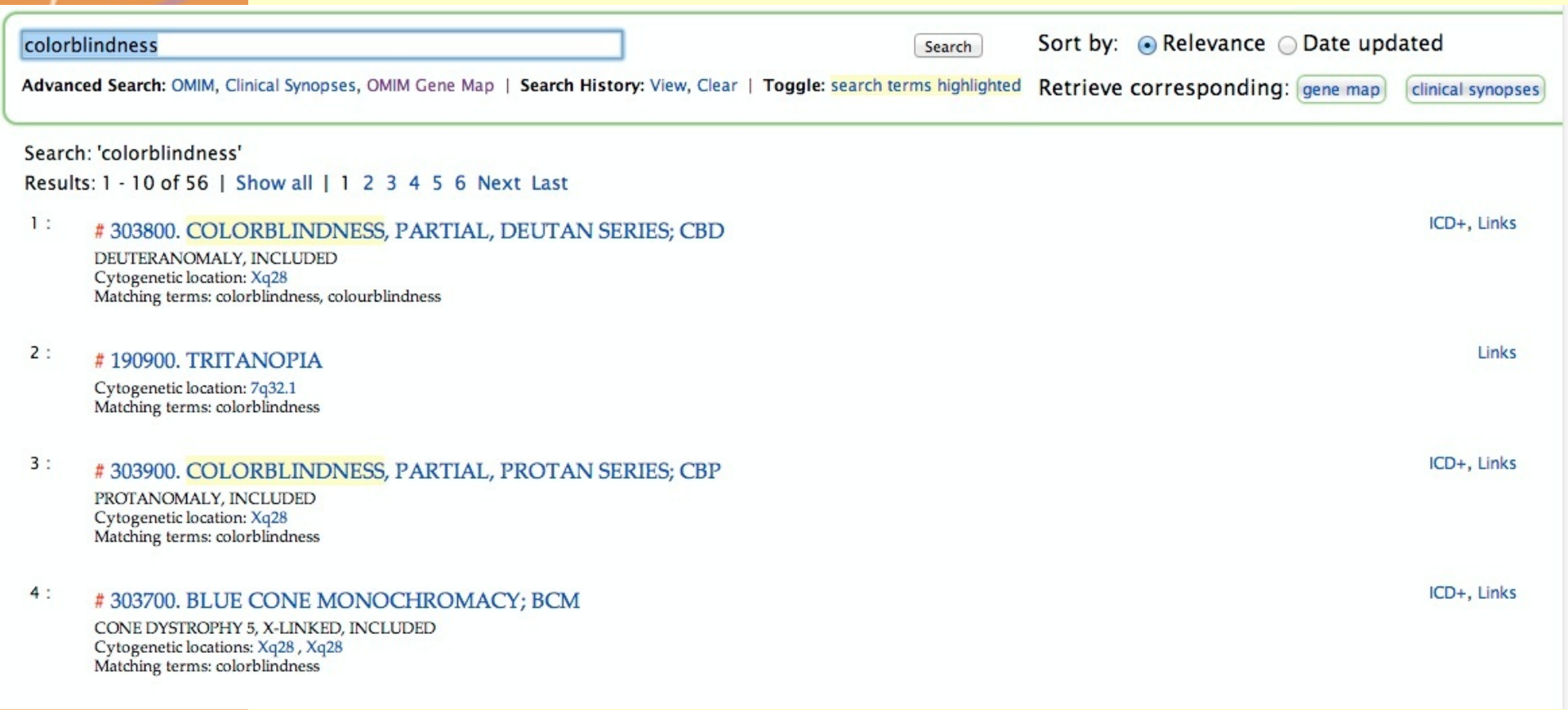
> 33% Phenotypes

OMIM Home Page

http://omim.org/

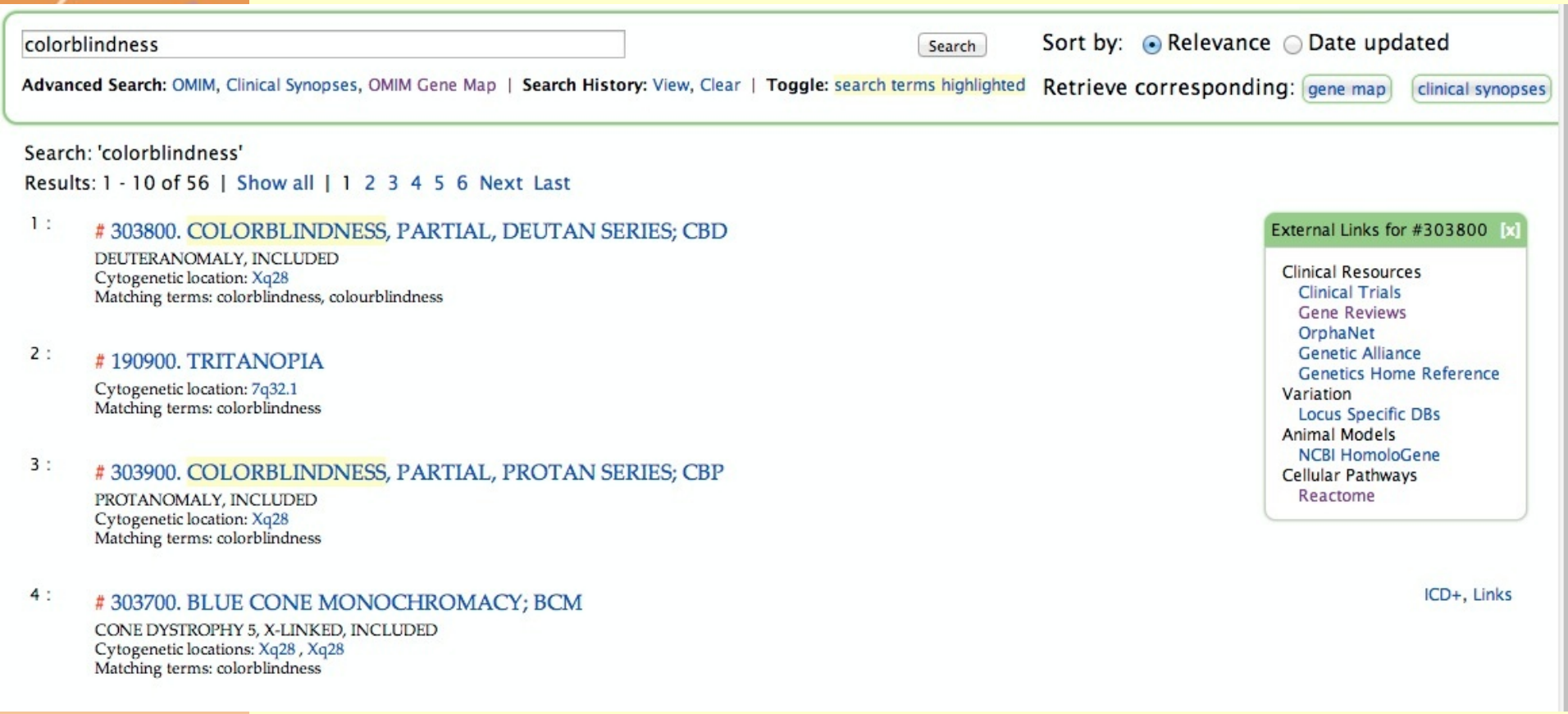
Colorblindness in OMIM

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



Colorblindness in OMIM

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



Colorblindness in OMIM

http://omim.org/entry/303800

OMIM®

#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

OMIM®

OMIM External Links

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.

OMIM®

OMIM External Links

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

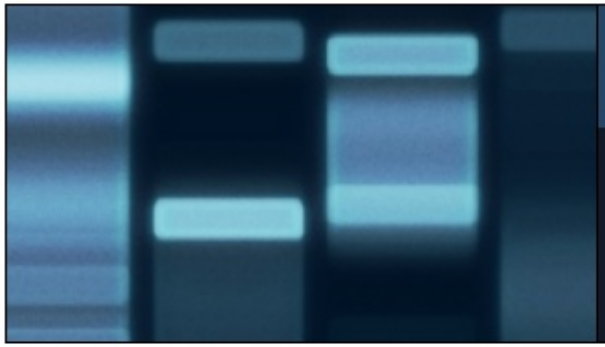
FlyBase	A Database of Drosophila Genes and Genomes.
NCBI HomoloGene	A system for automated detection of homologs among the annotated genes of several completely sequenced eukaryotic genomes.
MGI Mouse Phenotype	Phenotypes, alleles, and disease models from Mouse Genome Informatics.

RefSeq Gene

http://www.ncbi.nlm.nih.gov/refseq/rsg/

NCBI Resources How To brutlag My NCBI Sign Out

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)

Tools

- 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

NCBI UniGene

http://www.ncbi.nlm.nih.gov/unigene

NCBI Resources How To brutlag My NCBI Sign Out

UniGene UniGene Search

Limits Advanced Help

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

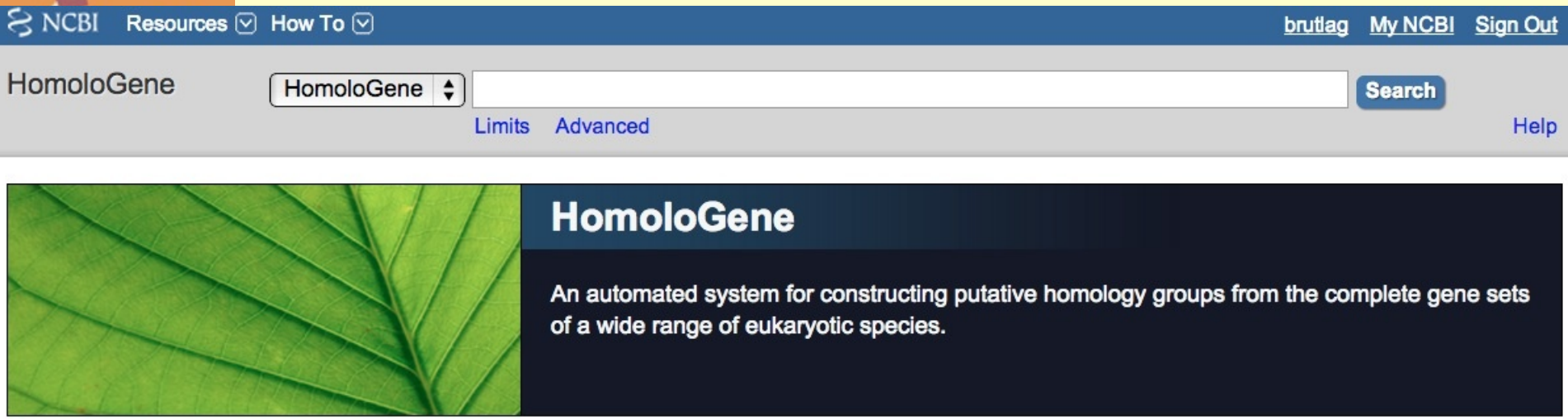
- UniGene Statistics
- Library browser
- Digital Differential Display (DDD)
- Downloads/FTP

Also of interest

- BioSample
- EST sequences
- Gene
- HomoloGene
- Map Viewer
- Short Read Archive (SRA)

NCBI Homologene Database

http://www.ncbi.nlm.nih.gov/homologene



NCBI

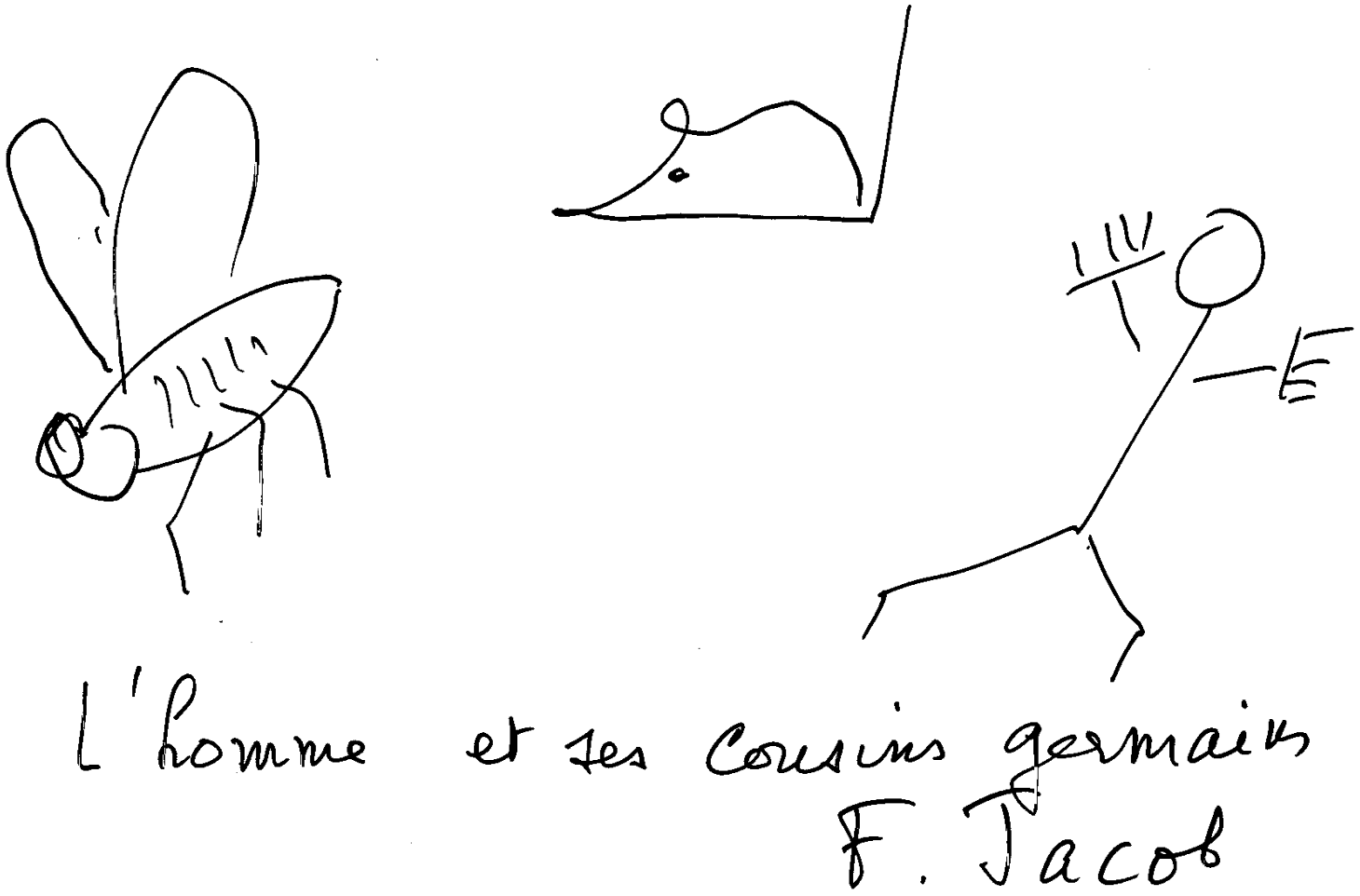
Release 67 Statistics

Release date:2012-12-12
Total species:20
Total HomoloGene groups:43074

Download

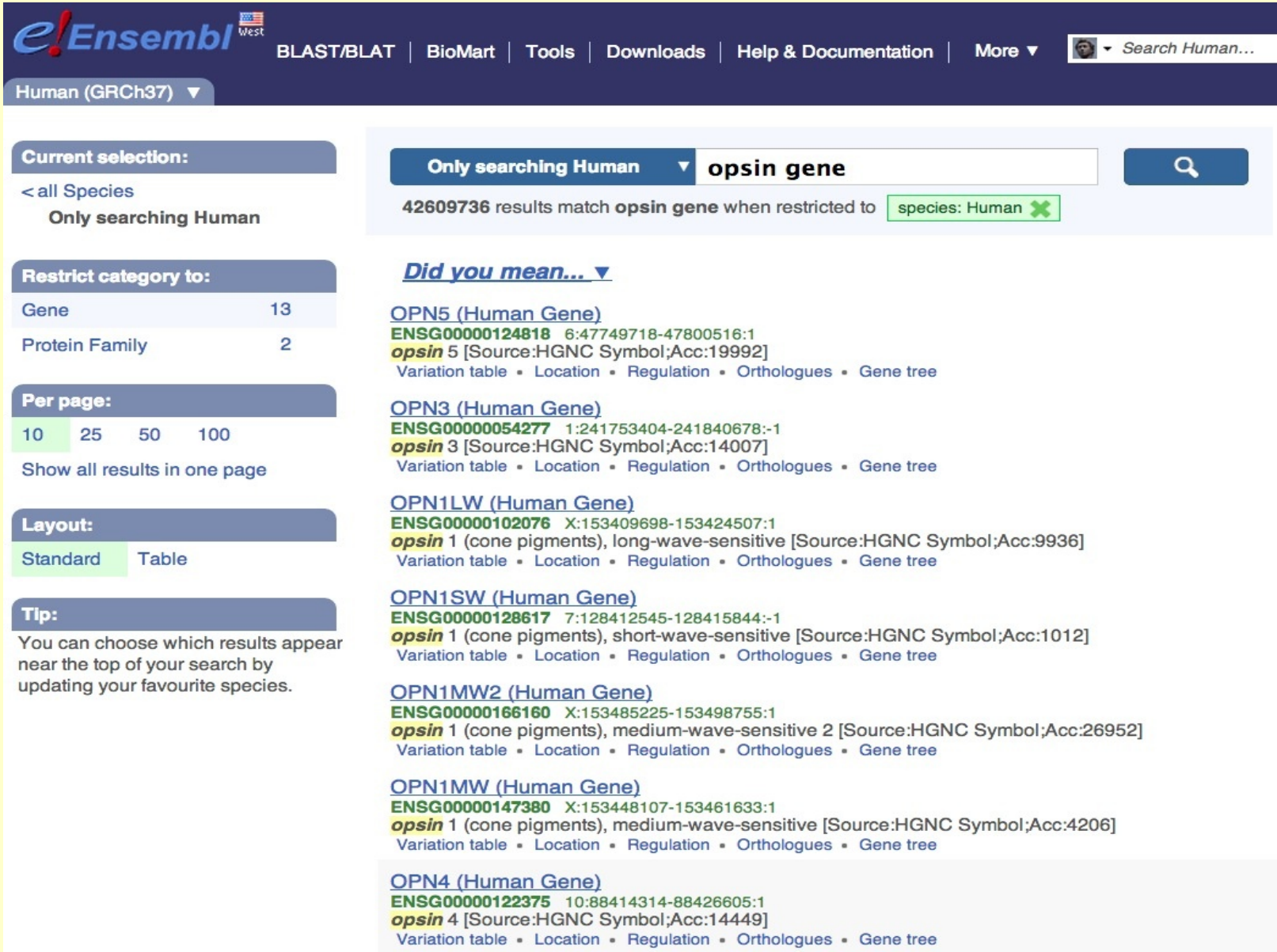
Scientific name	Common name	Genome release	Annotation release	Input genes	Grouped genes	Groups
Homo sapiens	human	GRCh37.p5	Build 37.3	19217	18867	18473
Pan troglodytes	chimpanzee	Pan_troglodytes-2.1.4	Build 3.1	19655	17367	17078
Macaca mulatta	Rhesus monkey	Mmul_051212	Build 1.2	22566	17151	16706
Canis 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



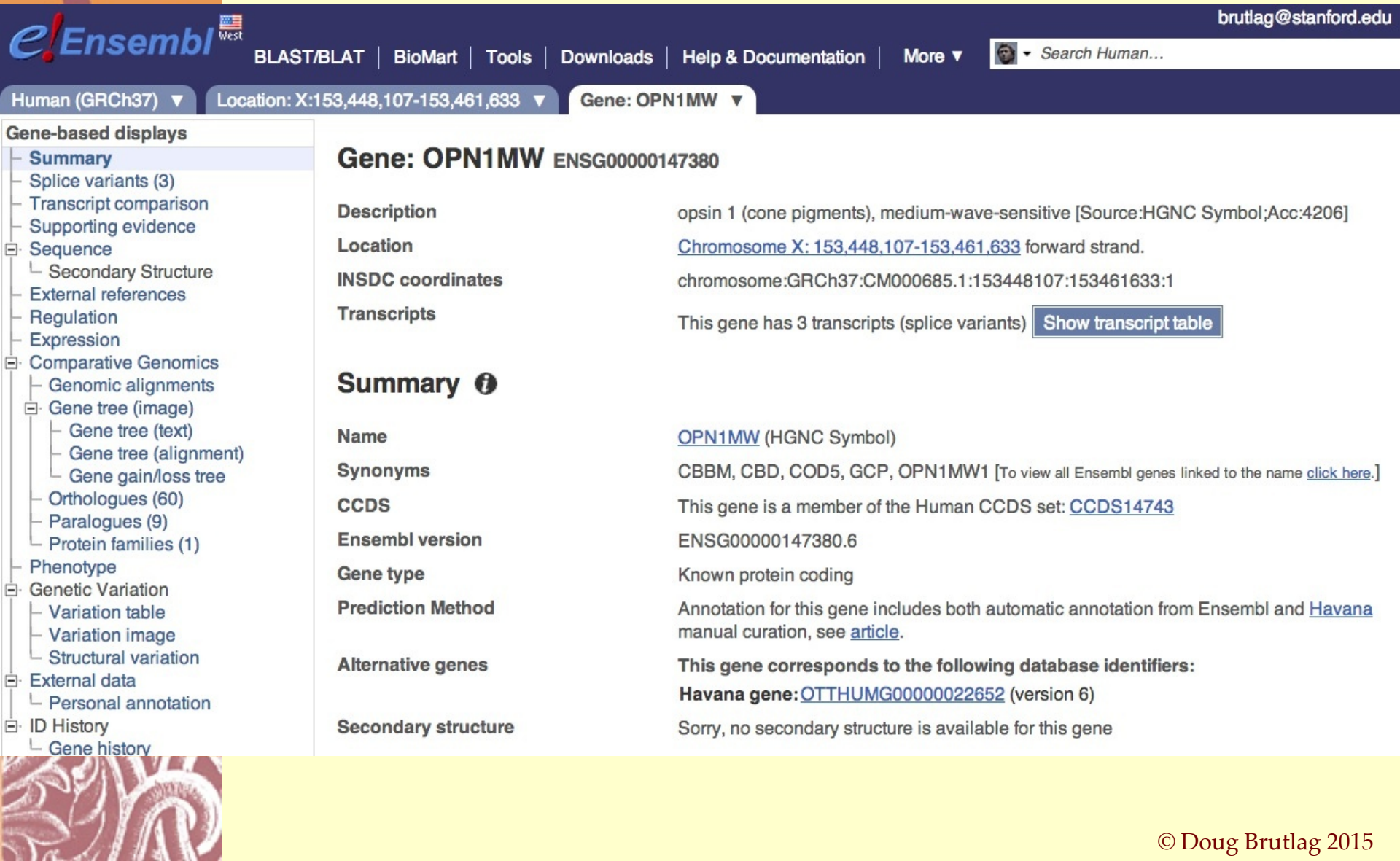
Ensembl Human Opsin Search

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



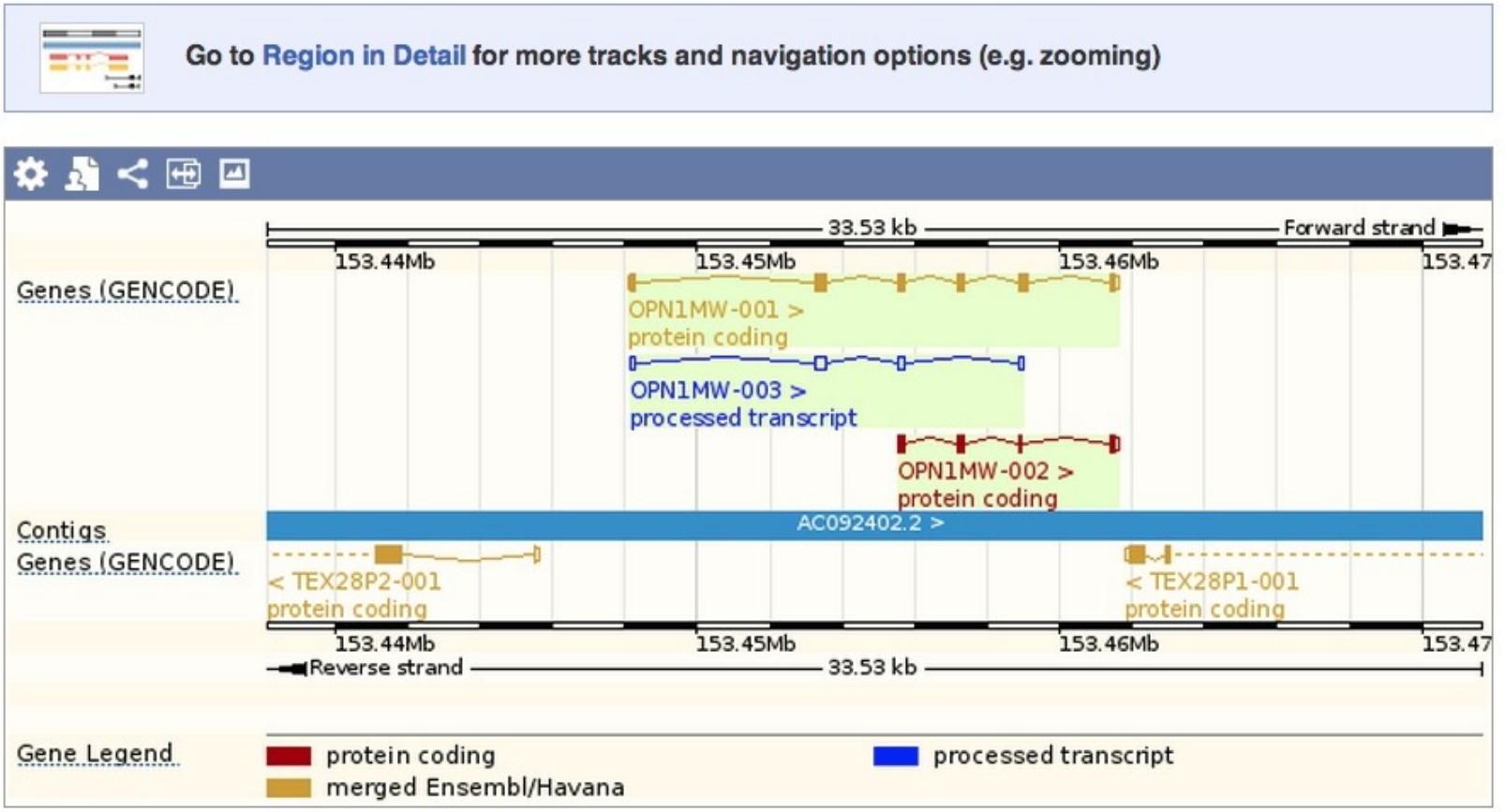
Ensembl Human OPN1MW Gene

http://uswest.ensembl.org/Homo_sapiens/Gene/Summary?g=ENSG00000147



Ensembl Opsin OPN1MW Gene Location

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

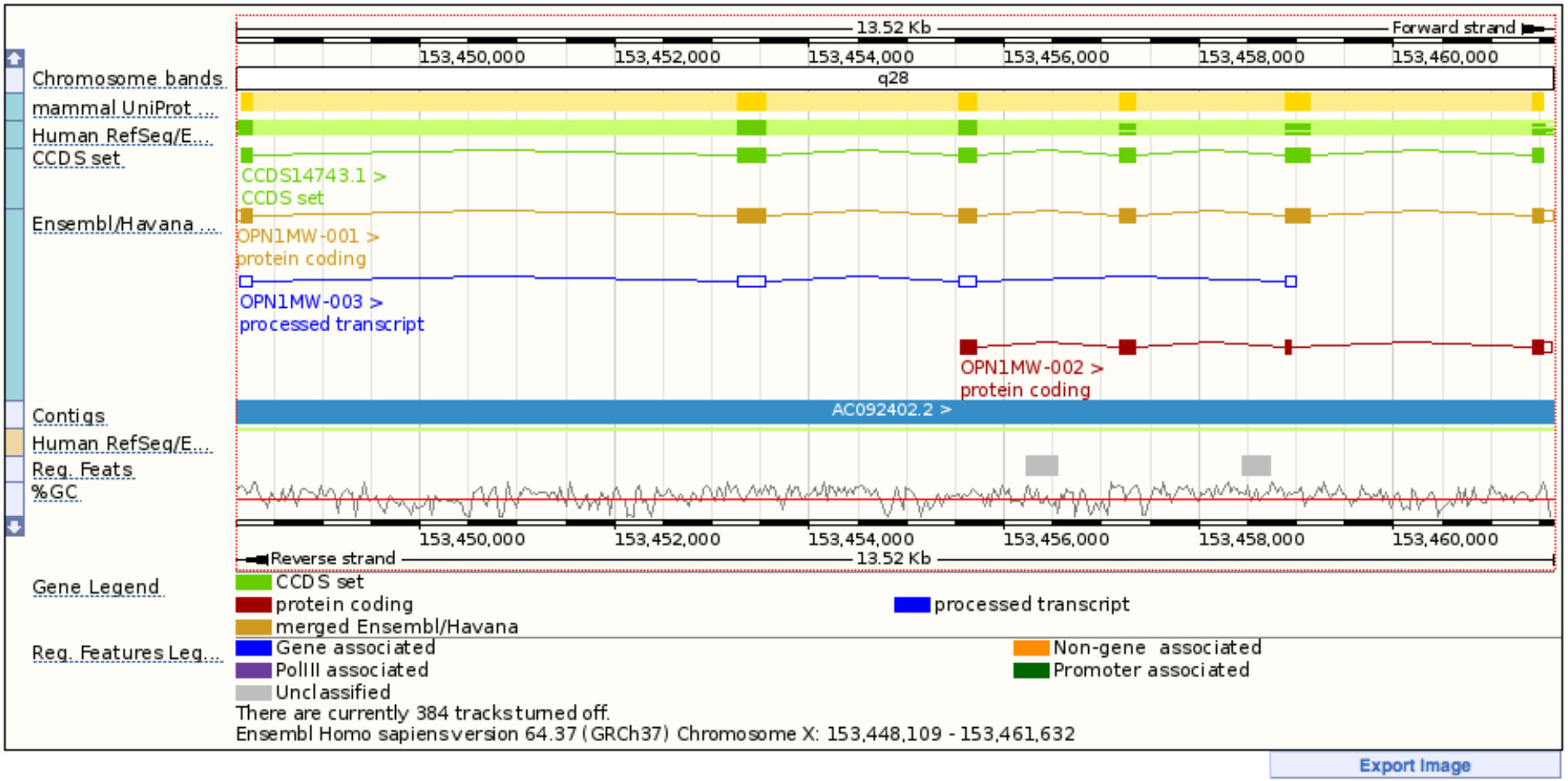


Configuring the display

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



Ensembl OPN1MW Opsin Protein

http://uswest.ensembl.org/Homo_sapiens/Transcript/ProteinSummary?db=core;g=ENSG0000

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

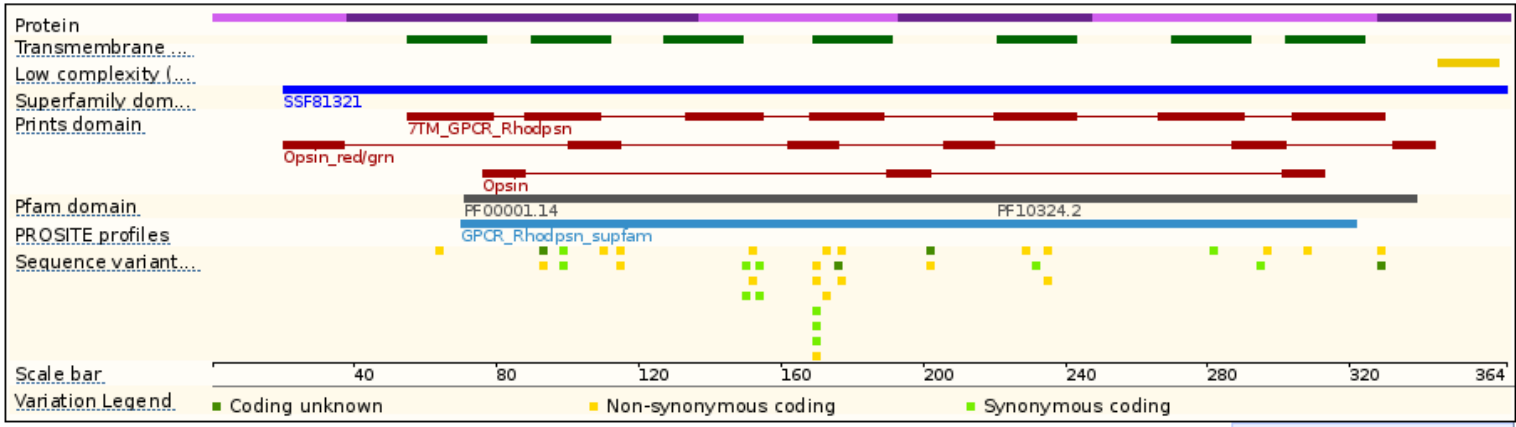
Show/hide columns | Filter

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



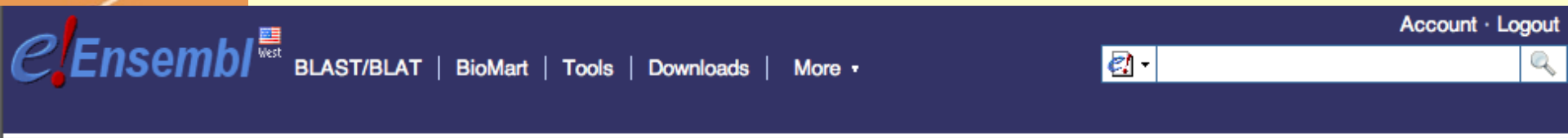
Export Image

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



Help & Documentation > Tutorials

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