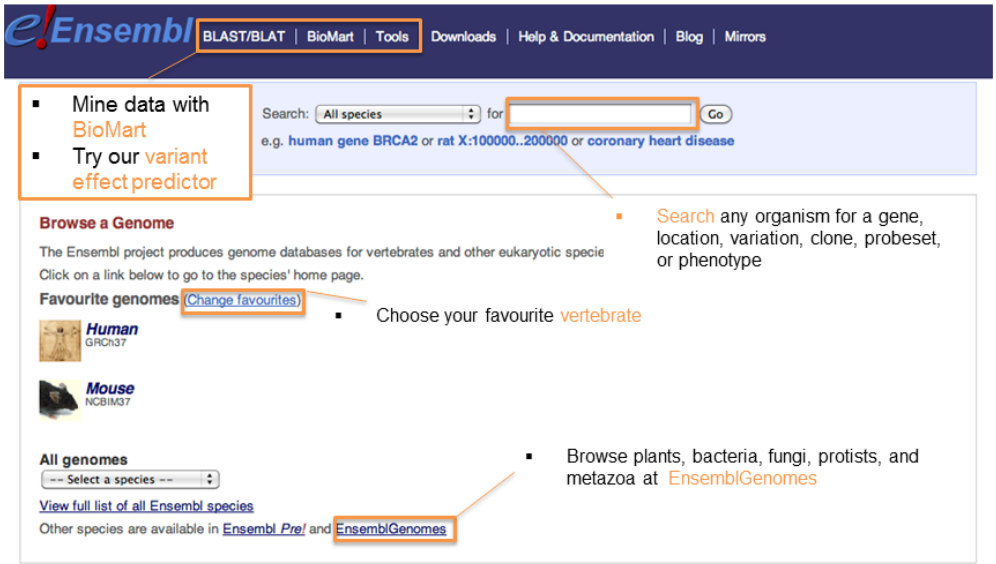
Getting started with Ensembl
www.ensembl.org

Ensembl provides genes and other **annotation** such as regulatory regions, conserved base pairs across species, and sequence variations. The Ensembl gene set is based on protein and mRNA evidence in **UniProtKB** and **NCBI RefSeq** databases, along with manual annotation from the **VEGA/Havana** group. All the data are freely available and can accessed via the web browser at www.ensembl.org. Perl programmers can directly access Ensembl databases through an Application Programming Interface (**Perl API**). Gene sequences can be downloaded from the Ensembl browser itself, or through the use of the **BioMart** web interface, which can extract information from the Ensembl databases without the need for programming knowledge by the user!



You will learn about

- Why do we need genome browsers?
- An introduction to Ensembl
- How information can be obtained from the site
- An overview of Ensembl tools

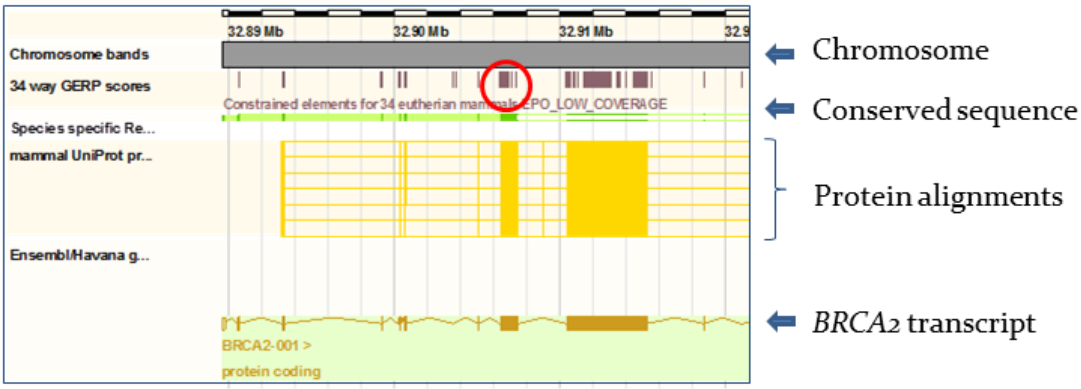
Tired of reading?

Check our video tutorial instead!
http://www.youtube.com/user/EnsemblHelpdesk

The Ensembl Genome browser
Introduction to BioMart

Introduction to Ensembl

Ensembl is a joint project between the EBI (European Bioinformatics Institute) and the Wellcome Trust Sanger Institute that annotates **chordate** genomes (i.e. vertebrates and closely related invertebrates with a notochord such as sea squirt). Gene sets from model organisms such as yeast and worm are also imported for comparative analysis by the Ensembl 'compara' team. Most annotation is updated every two months, leading to increasing Ensembl versions (such as version 62), however the gene sets are determined less frequently. A sister browser at www.ensemblgenomes.org is set up to access non-chordates, namely bacteria, plants, fungi, metazoa, and protists.



The Region in Detail view

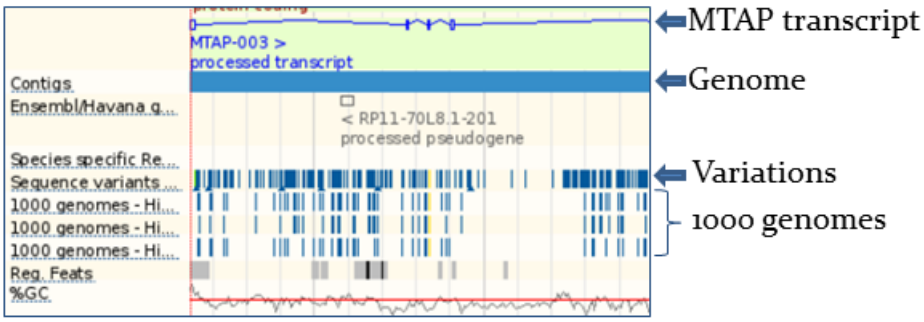
The vast amount of information associated with the genomic sequence demands a way to organise and access that information. This is where genome browsers come in. Ensembl strives to display many layers of genome annotation into a simplified view for the ease of the user. The picture above shows the '**Region in Detail**' page for the BRCA2 gene in human. The example shows blocks of conserved sequence reflecting conservation scores of sequence identity on a base pair level across 34 species. Conserved regions are displayed as dark blocks that represent local regions of alignment. One of the blocks is circled in red. You would only have to click on this block to see more details.

Can't see Uniprot?
To add more tracks just click:

And select mammals: Uniprot under protein alignments

Also in this figure are proteins from the UniProtKB aligned to the same genomic region. Filled yellow blocks show where these UniProtKB proteins align to the genome, and gaps in the alignment are shown as empty yellow blocks. Note, in this case, the UniProtKB proteins support most of the exons shown in the Ensembl BRCA2-001 transcript (in gold).

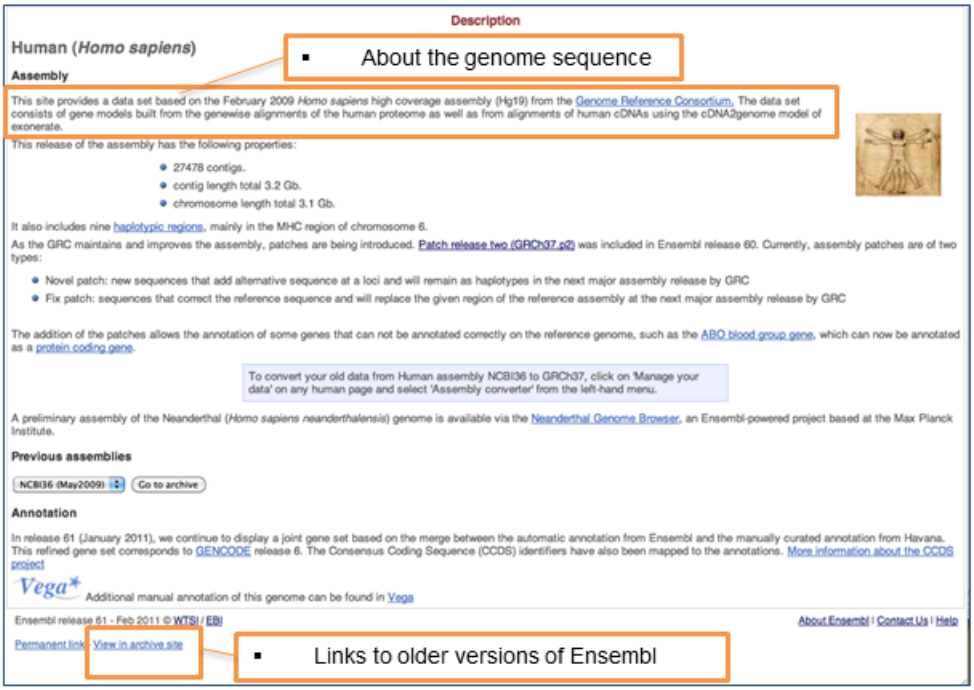
Both **Ensembl** and **Vega (Havana)** transcripts are portrayed as exons (boxes) and introns (connecting lines). In fact, filled boxes show coding sequence, and empty boxes reflect UnTranslated Regions (**UTRs**). This 'Region in Detail' view is useful for comparing Ensembl gene models with current proteins and mRNAs in other databases like **NCBI RefSeq**, **EMBL-Bank**, and, in the example above, UniProtKB. Everything in this view is aligned to the genome.

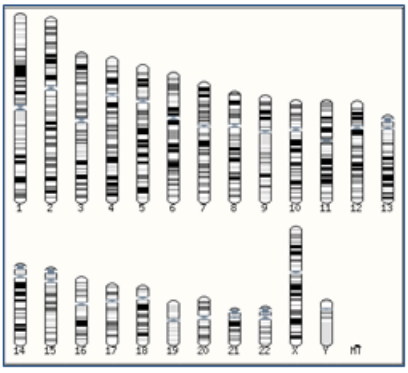


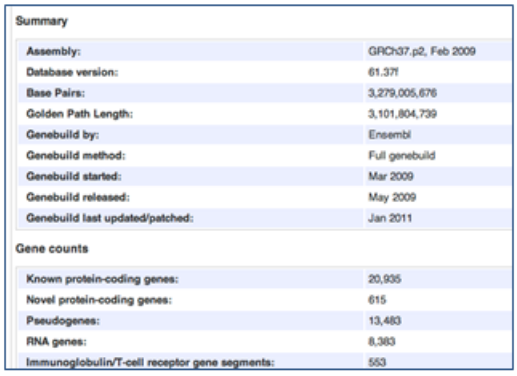
The Region in Detail view: 1000 genomes tracks

The region in detail view can be configured (using the *Configure this page* tool button) to show regulatory features, sequence variation, and more! Click on any vertical line in the variation track for a menu about the SNP (single nucleotide polymorphism) or InDel (insertion deletion mutation). Clicking on 'Variation properties' in the pop-up box will bring you to an information page for the genetic variation, including links to population frequencies, if known. You can do the same for any regulatory feature.

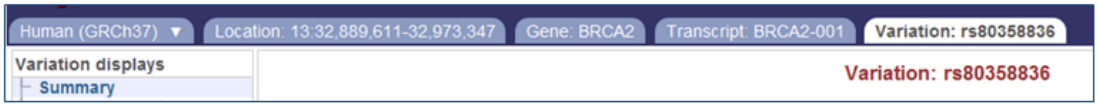
An index page is provided for each species with information about the source of the genomic sequence assembly, a karyotype (if available), and a link to past or archive sites. The picture below shows the Ensembl homepage for human. Links to the human karyotype, a summary of gene and genome information, and the most common InterPro domains in the genome are found at the left of this index page.







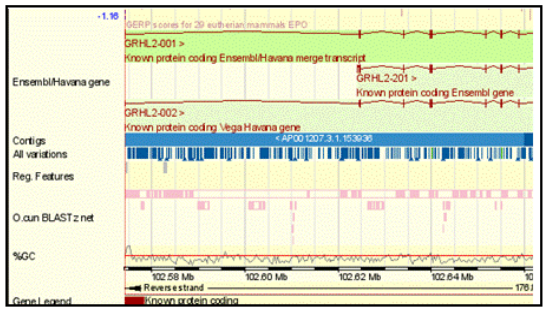
Ensembl devotes separate pages and views in the browser to display a variety of information types, using a tabbed structure.



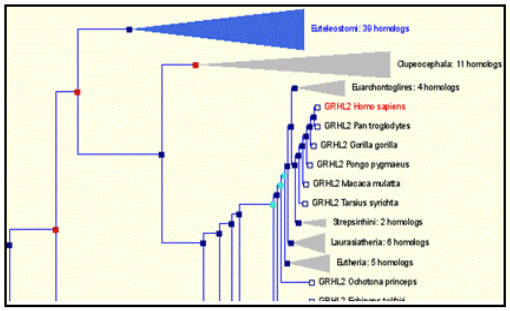
View genotype information in the variation tab, gene trees in the gene tab, a chromosomal region in the location tab, and cDNA sequence alongside the protein translation in the **transcript** pages. Compare conserved regions with the position of genes and population variation in the **Region in Detail** view. See homology relationships in the **gene** page, or perform a **BLAST** or **BLAT** search against any species in Ensembl.

Transcript Sequence w/Variations

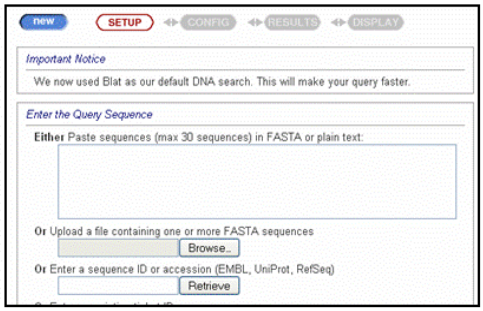
Genes, SNPs, and Conserved Regions



Homologues in Gene Trees



BLAST and BLAT aligners



Retrieving Data from Ensembl

BioMart is a very popular web-interface that can extract information from the Ensembl databases and present the user with a table of information without the need for programming. It can be used to output sequences or tables of genes along with gene positions (chromosome and base pair locations), single nucleotide polymorphisms (SNPs), homologues, and other annotation in HTML, text, or Microsoft Excel format. BioMart can also translate one type of ID to another, identify genes associated with an **InterPro** domain or gene ontology (**GO**) term, export gene expression data and lots more.

Ensembl uses MySQL relational databases to store its information. A comprehensive set of Application Programme Interfaces (APIs) serve as a middle-layer between underlying database schemes and more specific application programmes. The API aims to encapsulate the database layout by providing efficient high-level access to data tables and isolate applications from data layout changes.

Synopsis- What can I do with Ensembl?

- View genes along with other annotation along the chromosome
- View alternative transcripts (including splice variants) for a gene
- Explore homologues and phylogenetic trees across more than 50 species for any gene
- Compare whole genome alignments and conserved regions across species
- View microarray sequences that match to Ensembl genes
- View ESTs, clones, mRNA and proteins for any chromosomal region
- Examine single nucleotide polymorphisms (SNPs) for a gene or chromosomal region
- View SNPs across strains (rat, mouse), populations (human), or even breeds (dog)
- View positions and sequence of mRNA and protein that align with an Ensembl gene
- Upload your own data
- Use BLAST, or BLAT, a similar sequence alignment search tool, against any Ensembl genome
- Export sequence, or create a table of gene information with BioMart
- Use the Variant Effect Predictor

Need more help?

- Check Ensembl documentation
- Watch video tutorials on YouTube
- View the FAQs
- Try some exercises
- Read some publications

Stay in touch!

- Email the team with comments or questions at helpdesk@ensembl.org
- Follow the Ensembl blog
- Sign up to a mailing list

Further reading

Flicek, P. *et al.*
Ensembl 2011
Nucleic Acids Res. Advanced Access *(Database Issue)*
http://nar.oxfordjournals.org/content/early/2010/11/02/nar.gkq1064.full

Ensembl Methods Series
http://www.biomedcentral.com/series/ENSEMBL2010

Giulietta M Spudich and Xosé M Fernández-Suárez
Disease and Phenotype Data at Ensembl UNIT 6.11 in *Current Protocols in Human Genetics*, Apr 2011.

Xosé M. Fernández-Suárez and Michael K. Schuster
Using the Ensembl Genome Server to Browse Genomic Sequence Data.
UNIT 1.15 in *Current Protocols in Bioinformatics*, Jun 2010.

Giulietta M Spudich and Xosé M Fernández-Suárez
Touring Ensembl: A practical guide to genome browsing
BMC Genomics 2010, 11:295 (11 May 2010)

Vilella, A.J. *et. al*
EnsemblCompara GeneTrees: Analysis of complete, duplication aware phylogenetic trees in vertebrates.
Genome Res. 2009 Feb 19(2):327-35

Smedley, D. *et al*
BioMart – biological queries made easy
BMC Genomics 2009 Jan 14;10:22