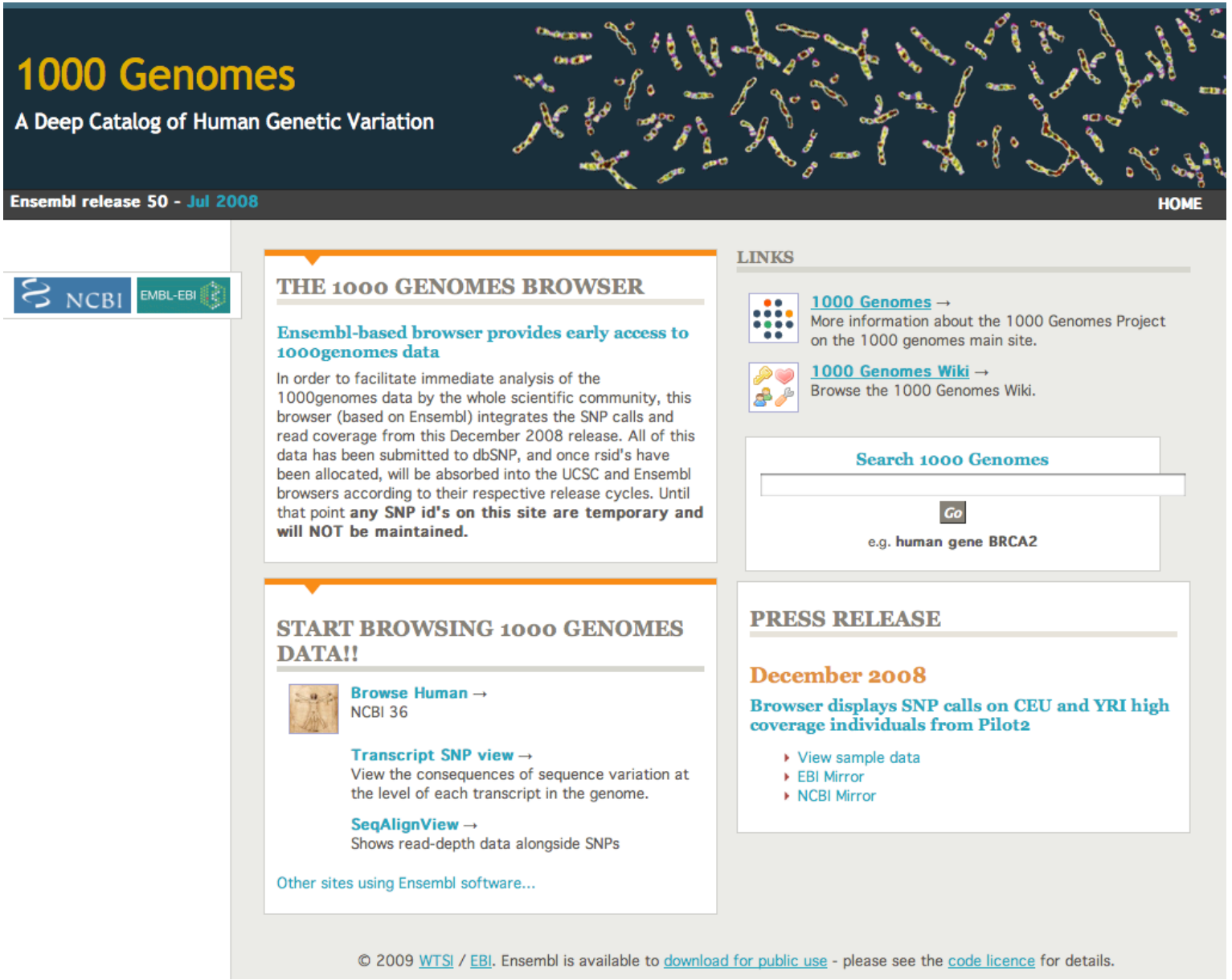
1000 Genomes Browser
Quick start guide

http://browser.1000genomes.org

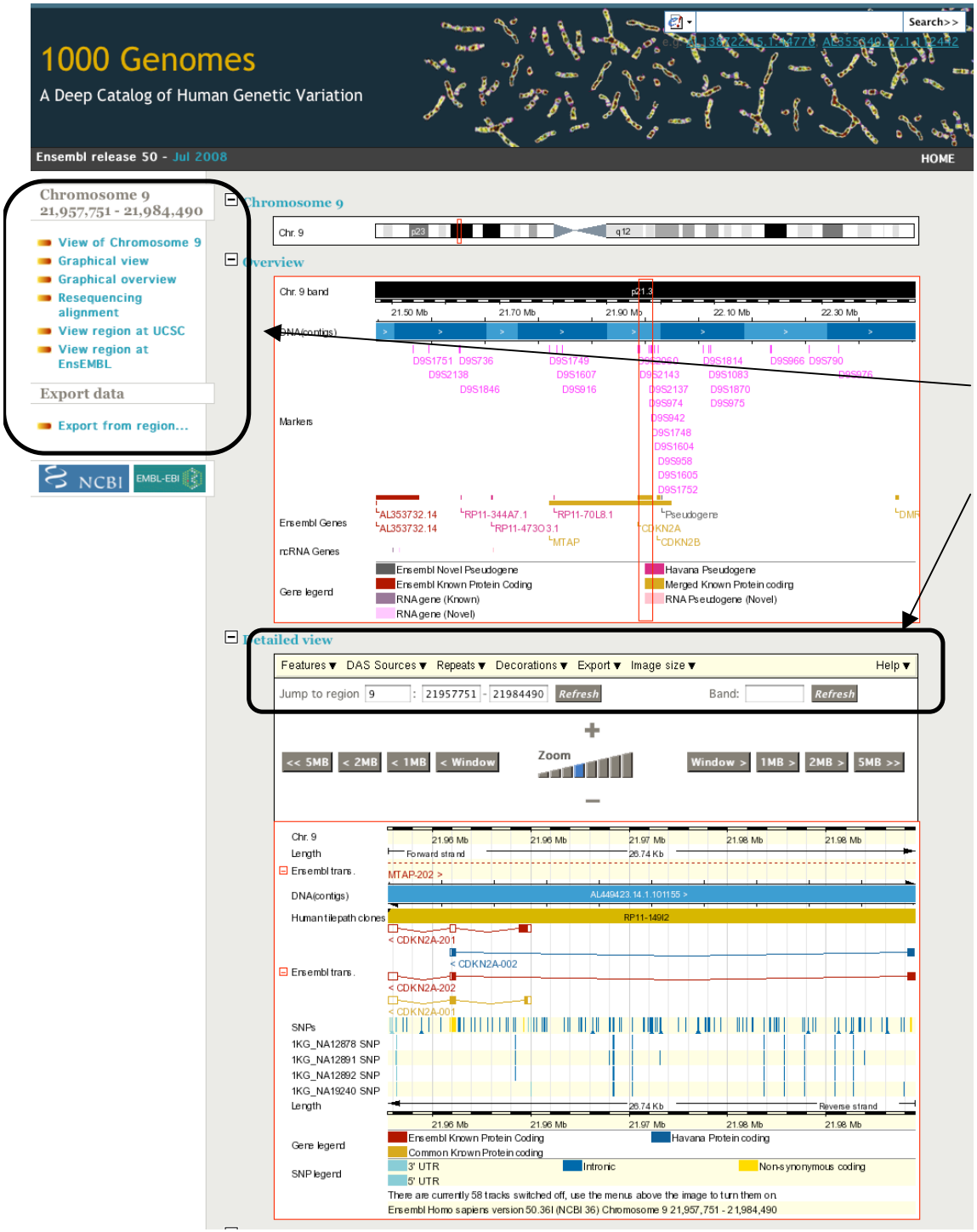
6 January 2008

Overview

- Based on version 50 (“old version”) of Ensembl code
- Contains all of the gene information normally present in Ensembl
  - Gene and transcript annotation, external references, sequence data
- There are things that don’t work and we have not transferred to “production” web hardware
  - Please send questions, problems or apparent errors to flicek@ebi.ac.uk



1000 Genomes Browser Home Page

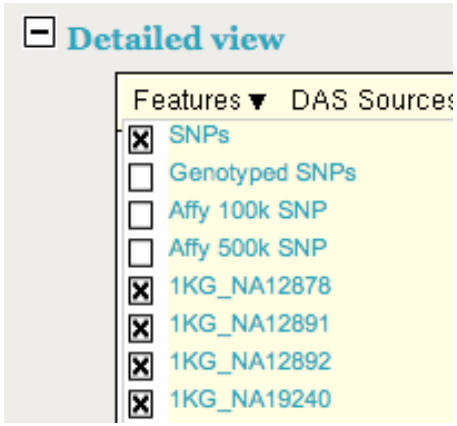


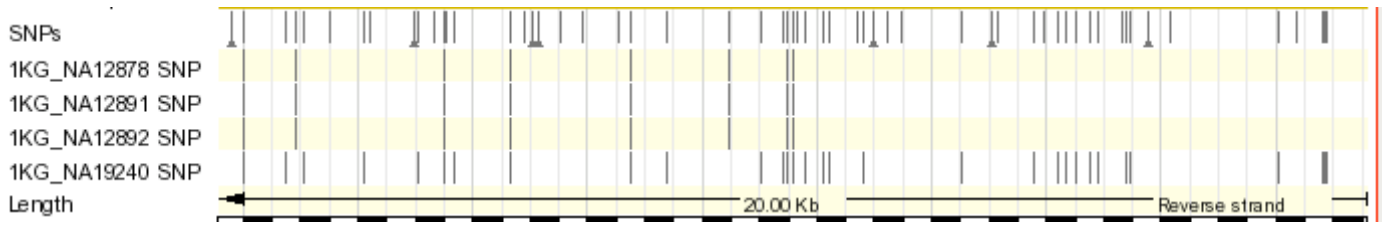
Main view

- Built on Ensembl
- Navigation is on the left hand side
- Options are drop down menus on the tops of the windows
- Includes only human data in current release
  - Comparative genomics information will be available in a future release
  - All appropriate pages have links to current versions of Ensembl and UCSC

Individual-specific SNPs

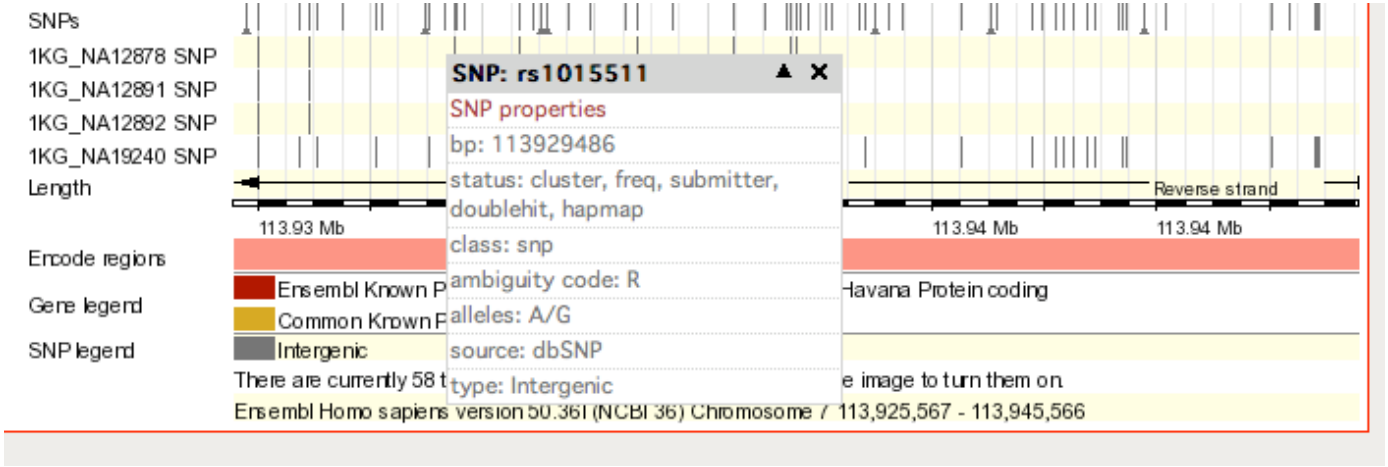
- The 1KG individuals can be viewed on the graphical view (contig view) pages
- These are selected from the “Features” menu and appear as tracks near the bottom of the display
- Tracks for all SNPs and the SNPs on selected Affy arrays are also available

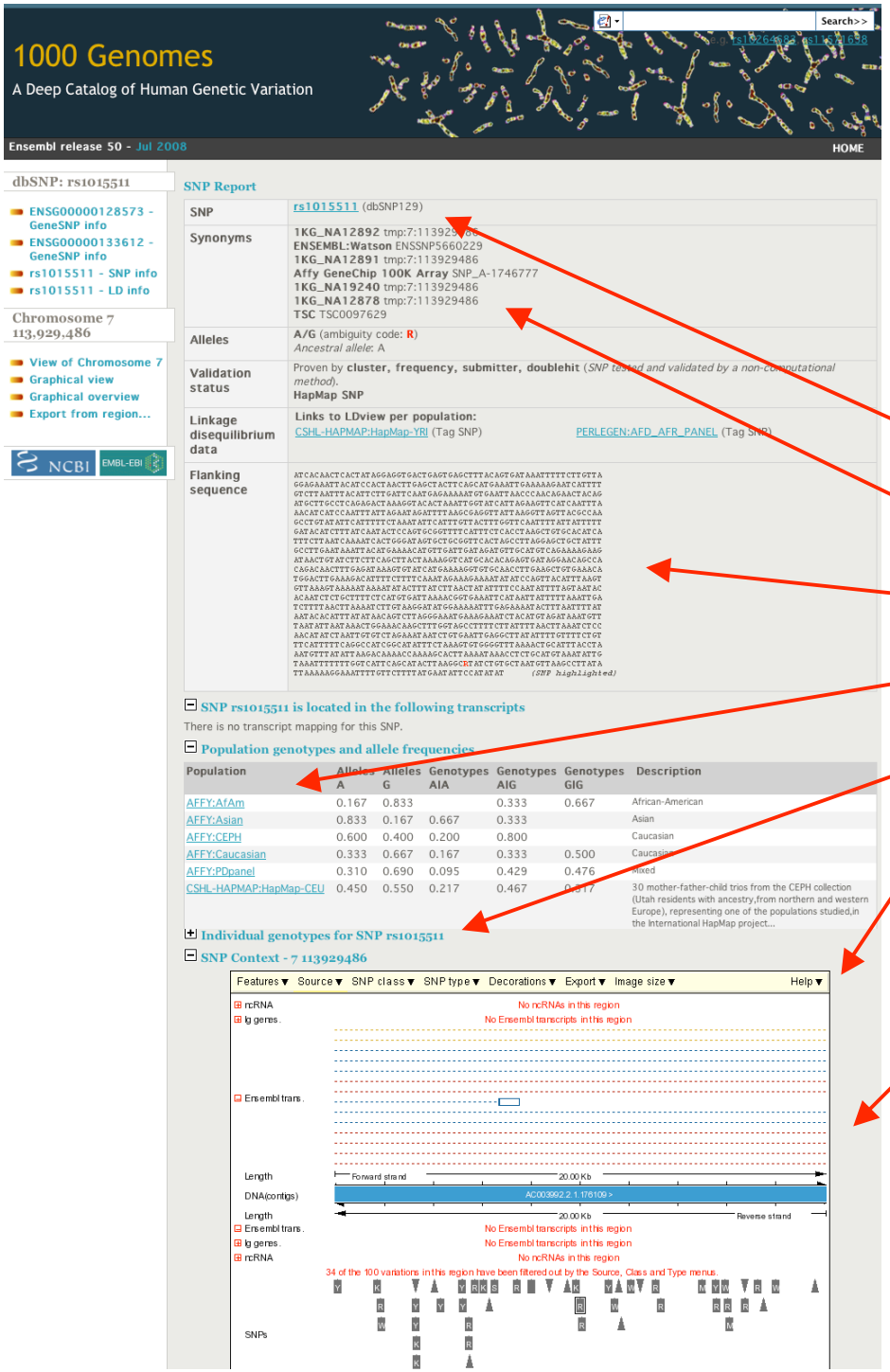




SNP Information

- SNPs are clickable which brings up a small window with basic information
- The “SNP properties” link leads to a dedicated page for the SNP with detailed information (mostly imported from dbSNP) about population frequencies, identifiers, individual genotypes and other information



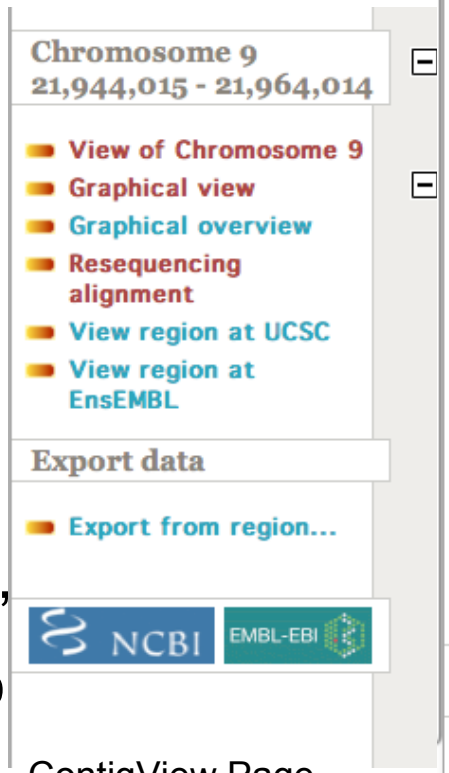


SNPView

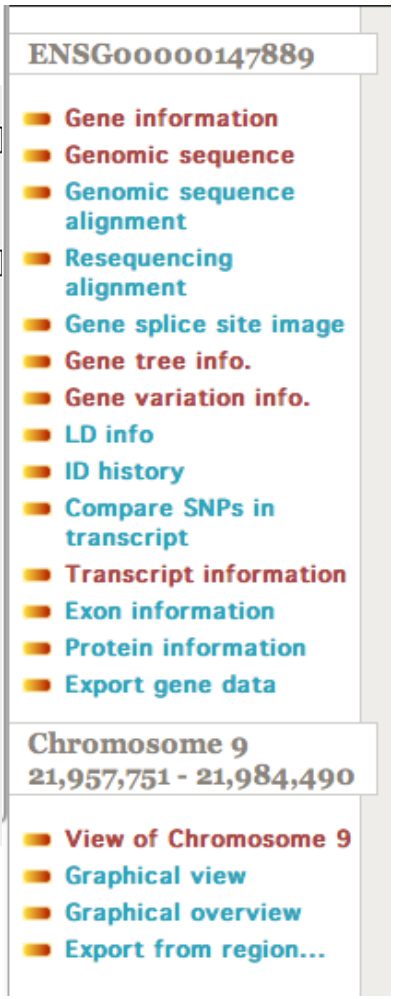
- Temporary rsName
- Identifiers
- Flanking sequence
- Population frequency
- Individual genotypes
- Change display options (individuals, SNP type, etc.)
- Location information

Resequencing alignment

- View any region of the genome in alignment with reference, 4 1KG individuals, Watson, Venter
- Assumption made that if there is sequence coverage and not a SNP called, the base is the same as the reference
- Use “Resequencing alignment” link on the left side of pages to access view

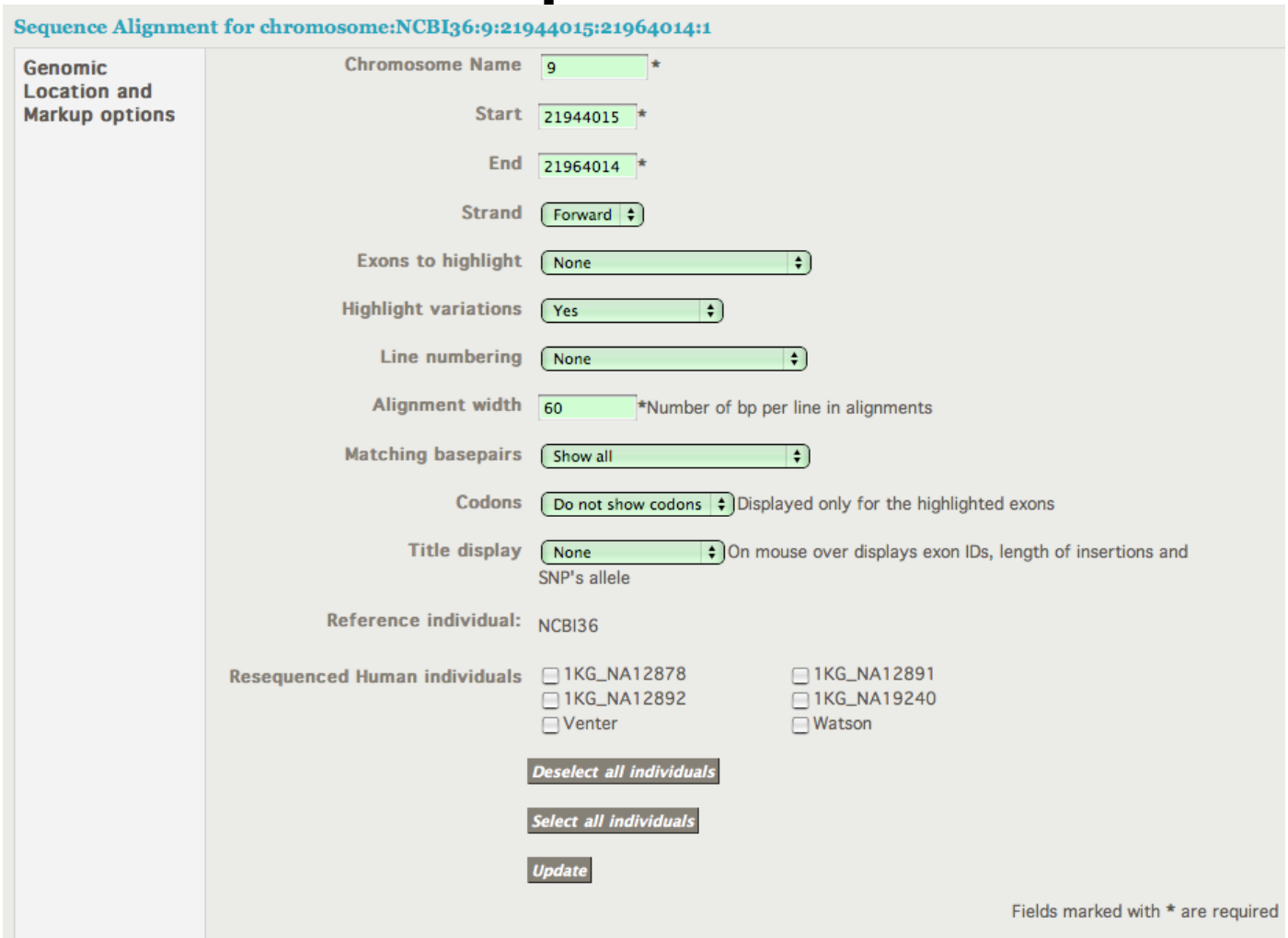


ContigView Page



GeneView Page

Resequencing alignment options

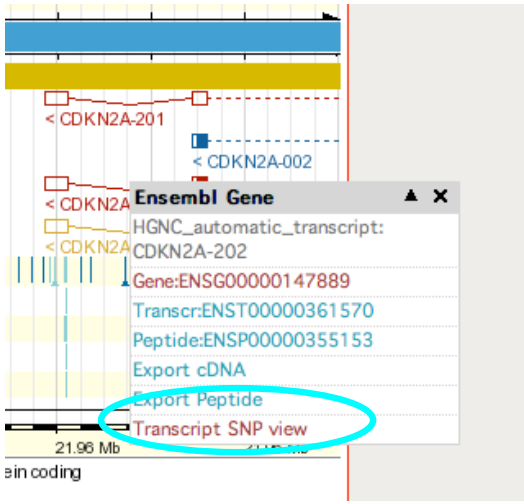


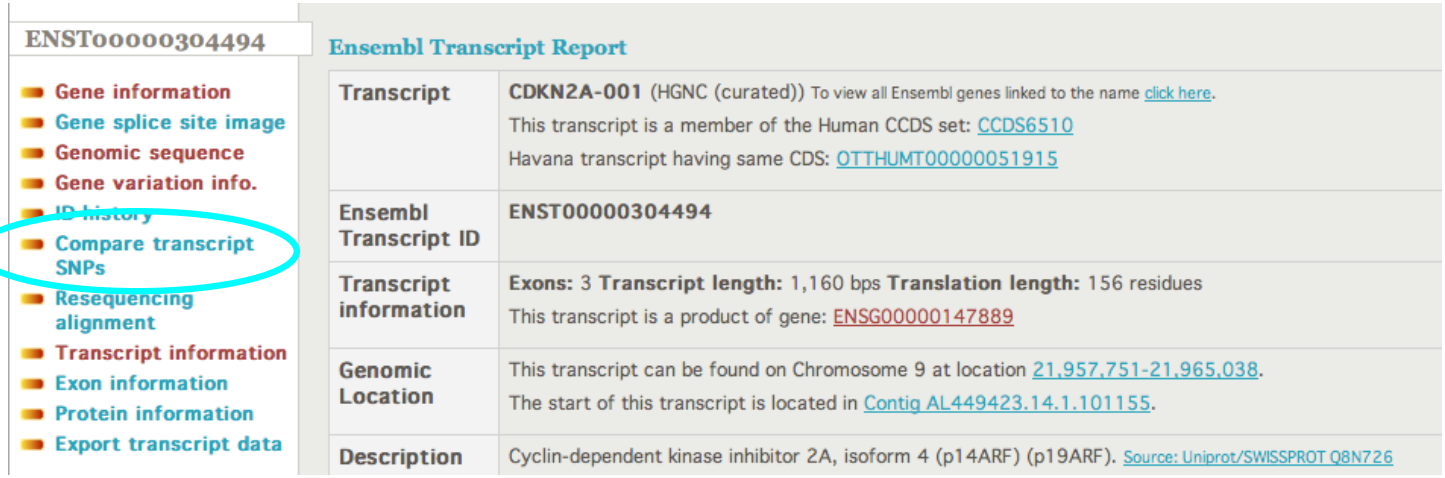
Resequencing alignment output

- Hets
- SNPs
- No coverage

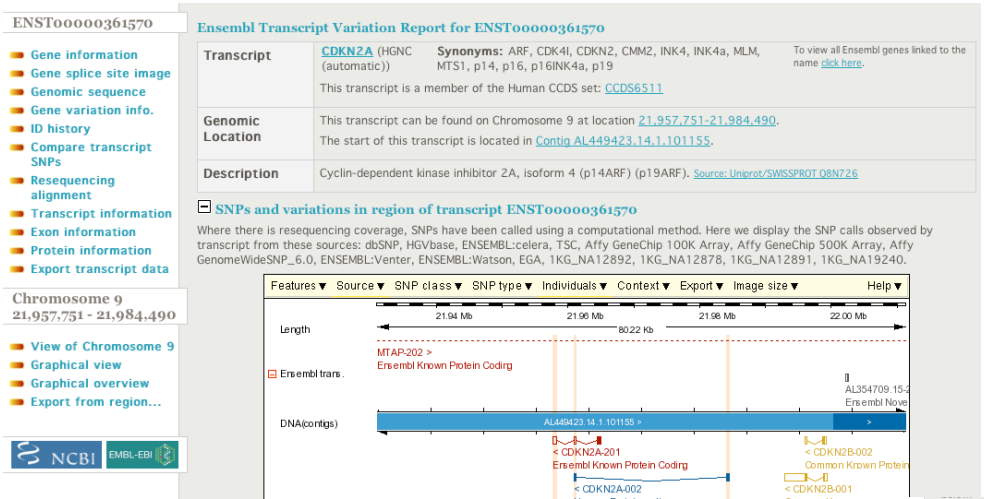
Individual SNP consequences

- TranscriptSNPView
  - Linked off all Ensembl Genes
  - From transcript pages in left hand menu
  - Color-coded display of how SNPs affect transcripts

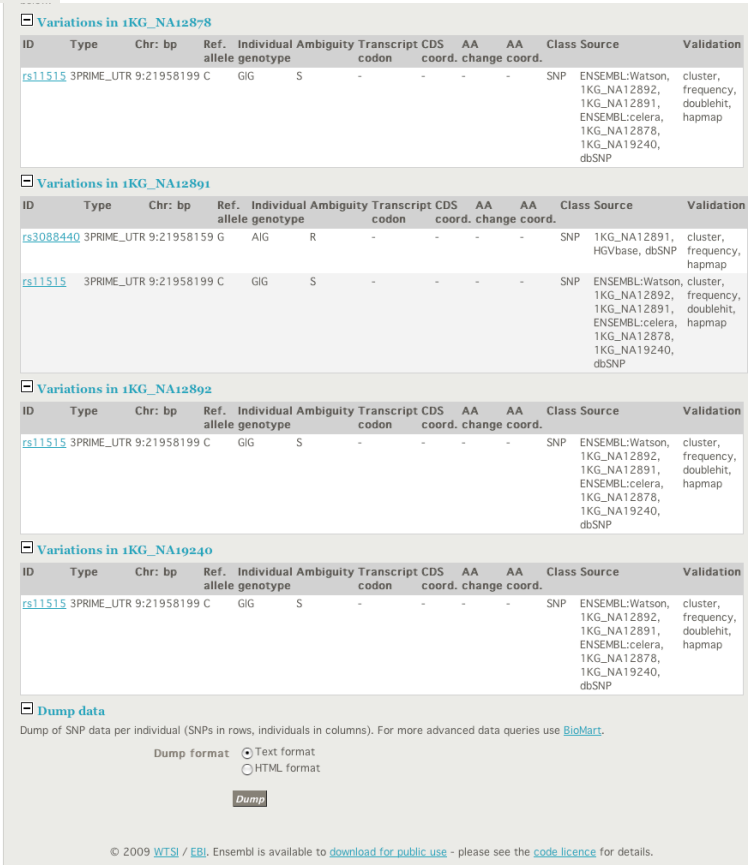




Main display



Summary table

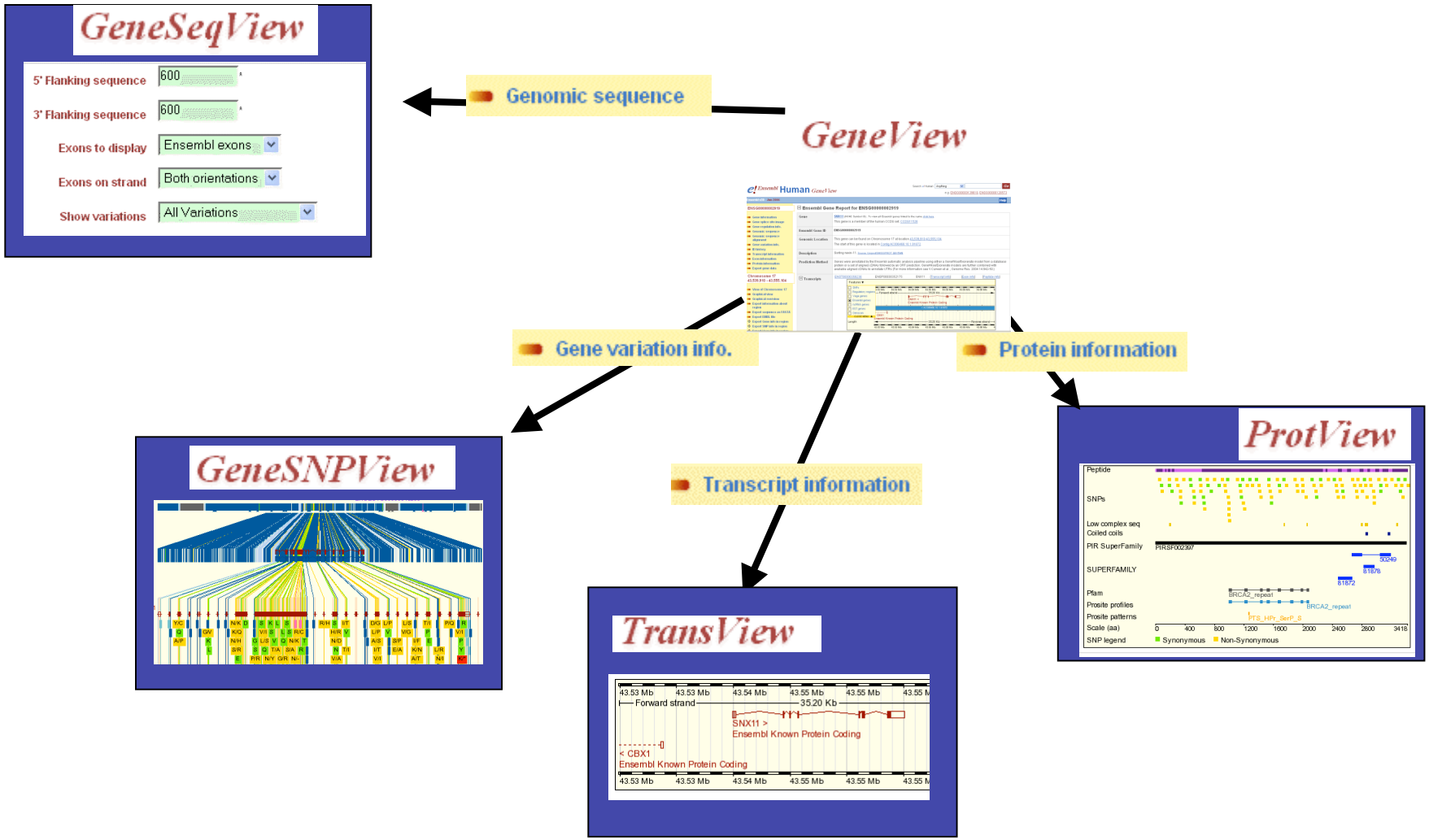


- Individual
- Coverage
- SNPs
- Legend
- SNP summary
  - Green
    - same as reference
  - Purple:
    - different
  - Hatched
    - heterozygous

LDView

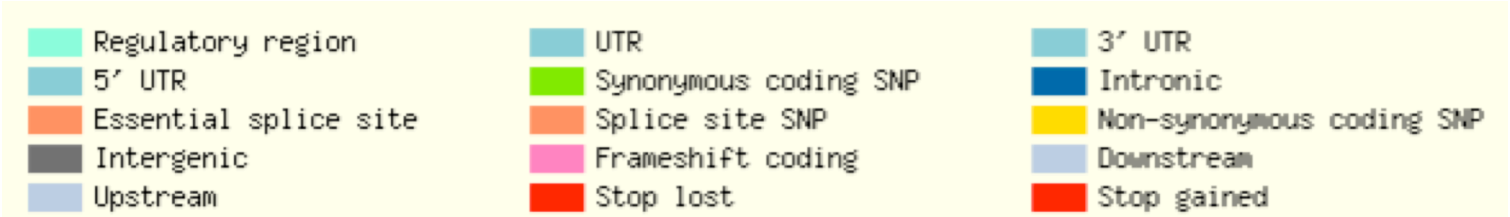
- Based on data from HapMap and Perlegen populations
- Populations selectable from drop down tab

More SNPs displays



SNP types

Non-synonymous	In coding sequence, resulting in an aa change
Synonymous	In coding sequence, not resulting in an aa change
Frameshift	In coding sequence, resulting in a frameshift
Stop lost	In coding sequence, resulting in the loss of a stop codon
Stop gained	In coding sequence, resulting in the gain of a stop codon
Essential splice site	In the first 2 or the last 2 basepairs of an intron
Splice site	1-3 bps into an exon or 3-8 bps into an intron
Upstream	Within 5 kb upstream of the 5'-end of a transcript
Regulatory region	In regulatory region annotated by Ensembl
5' UTR	In 5' UTR
Intronic	In intron
3' UTR	In 3' UTR
Downstream	Within 5 kb downstream of the 3'-end of a transcript
Intergenic	More than 5 kb away from a transcript



Credits

- Eugene Kulesha, Stephen Keenan
- Yuan Chen, Fiona Cunningham
- Laura Clarke, Zam Iqbal

- 1000 Genome Data providers
- Entire Ensembl Team