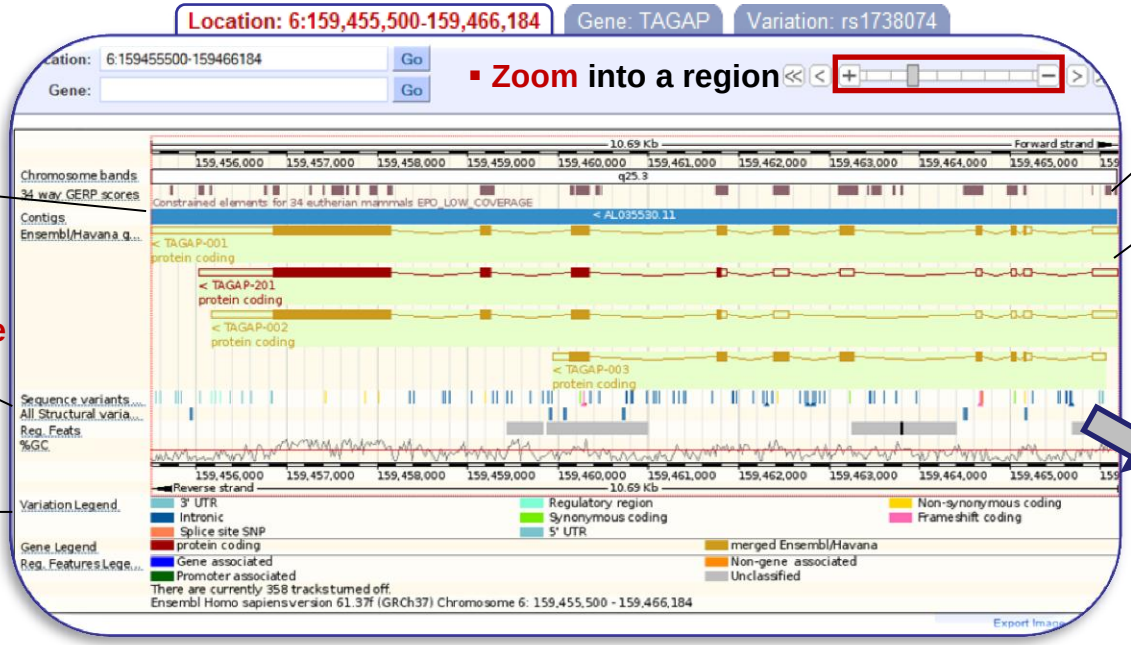
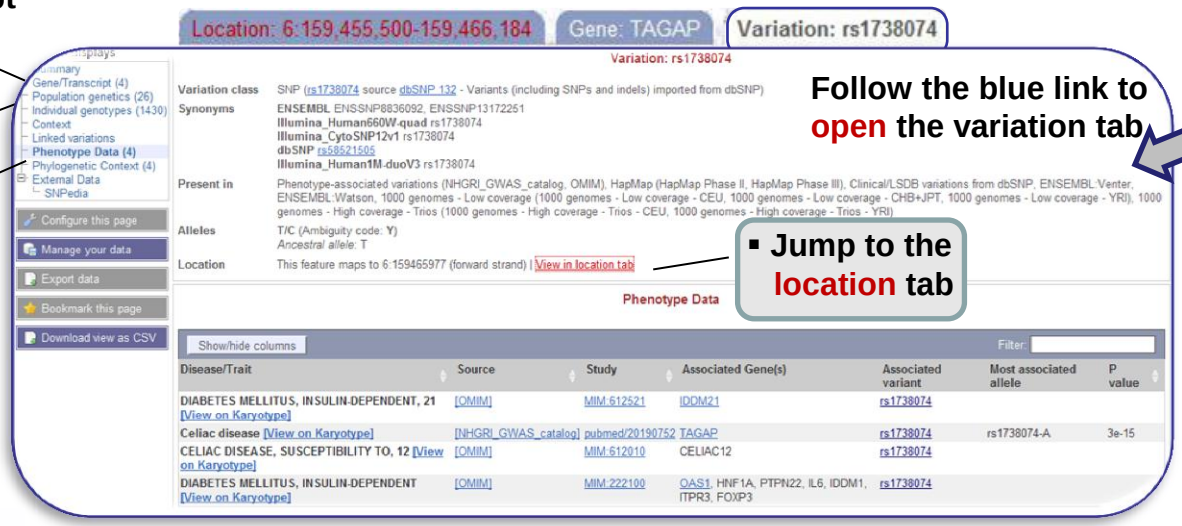
Ensembl variations quick reference card

EMBL-EBI wellcome trust sanger institute



- The **genome** is shown as a blue bar, labeled with contig names
- **Configure this page** to turn on variation tracks
- **Variations** are coloured according to their consequence type
- **Conserved** sequences
- **Transcripts** for the *TAGAP* gene are aligned to the genome

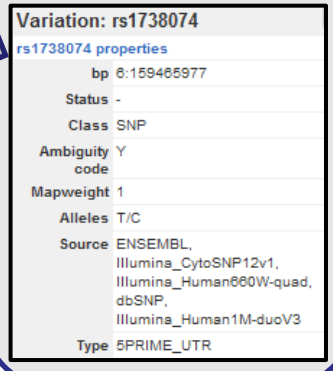


- **Gene** and transcript locations
- Allele **frequencies** for populations
- Associated **phenotypes**

Follow the blue link to **open** the variation tab

- Jump to the **location** tab

Click a variation for a detailed pop-up



www.ensembl.org

Contact us at helpdesk@ensembl.org

Reference: Flicek, P. et al. Ensembl 2011 (2011) Nucleic Acids Research 39:D800-D806
Reference: BMC Bioinformatics – Ensembl Update 2010 http://www.biomedcentral.com/series/ENSEMBL2010
Tutorials: http://www.ensembl.org/info/website/tutorials

Ensembl variations quick reference card

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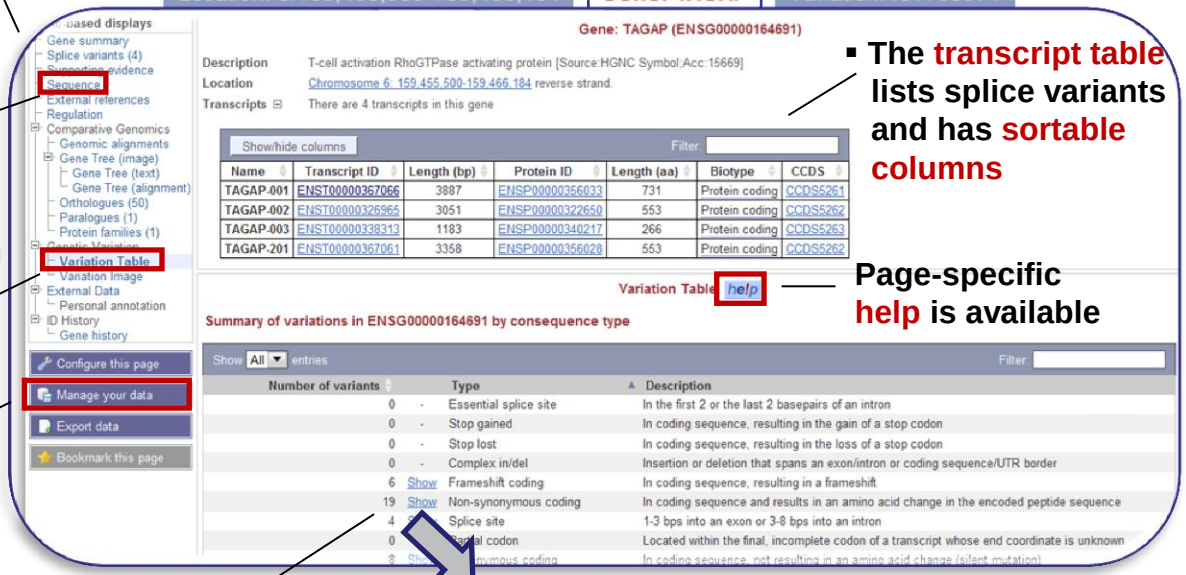
e!Ensembl BLAST/BLAT | BioMart | Tools

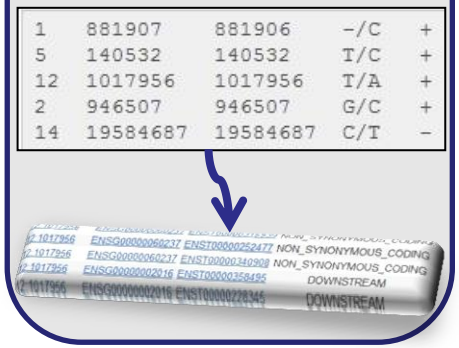
- Contextual navigation for a gene
- View variations in the genomic sequence
- The variation table shows variations for a gene
- Use the variant effect predictor
- Search for a gene

Location: 6:159,455,500-159,466,184 | Gene: TAGAP | Variation: rs1738074



- The transcript table lists splice variants and has sortable columns
- Page-specific help is available

Click 'Tools' for the Variant Effect Predictor



- Show a table for one variation type

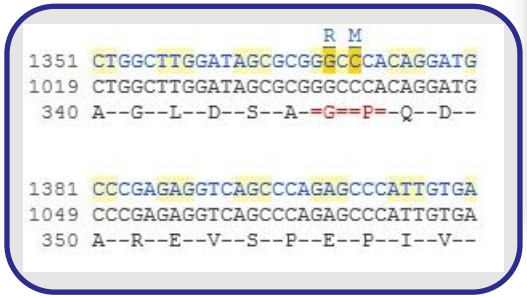
Non-synonymous coding variants [back to top]

Show All entries | Show/hide columns | Filter:

ID	Chr: bp	Alleles	Source	Amino Acid	AA co-ordinate	Transcript
rs13106561	4:123301312	A/C	dbSNP	T/P	30 (1)	ENST00000458011
rs13106730	4:123301310	C/G	dbSNP	A/G	29 (2)	ENST00000458011
rs13106366	4:123301235	A/C	dbSNP	N/T	4 (2)	ENST00000458011
rs13106561	4:123301312	A/C	dbSNP	T/P	30 (1)	ENST00000446706
rs13106730	4:123301310	C/G	dbSNP	A/G	29 (2)	ENST00000446706
rs13106366	4:123301235	A/C	dbSNP	N/T	4 (2)	ENST00000446706
rs13106561	4:123301312	A/C	dbSNP	T/P	30 (1)	ENST00000439307
rs13106730	4:123301310	C/G	dbSNP	A/G	29 (2)	ENST00000439307
rs13106366	4:123301235	A/C	dbSNP	N/T	4 (2)	ENST00000439307
rs13106561	4:123301312	A/C	dbSNP	T/P	12 (1)	ENST00000388725
rs13106730	4:123301310	C/G	dbSNP	A/G	11 (2)	ENST00000388725
rs13106561	4:123301312	A/C	dbSNP	T/P	30 (1)	ENST00000388724
rs13106730	4:123301310	C/G	dbSNP	A/G	29 (2)	ENST00000388724
rs13106366	4:123301235	A/C	dbSNP	N/T	4 (2)	ENST00000388724

- Ensembl transcripts (splice variants) are listed
- Click a variation ID to open the variation tab
- Possible amino acids at a position

- View variations on the cDNA sequence in the transcript tab



version 1.0