

Bioinformatics I -- Lecture 26

- SNPs
 - t-SNPs
 - Applications of SNPs

SNPs = single nucleotide polymorphisms

What are polymorphisms?

- Genetic differences between individuals in a population.
- Changes related to alleles.
- Single nucleotide polymorphisms (one base substitution)
 - Noncoding
 - Coding
 - synonymous -- same amino acid, different codon
 - non-synonymous
 - missense -- change in amino acid
 - nonsense -- stop codon
- Frame-shifts
 - One or more base Insertion/deletion

NCBI : SNP database

How To: View all SNPs associated with a gene

Starting with...

a gene name

1. Search the Gene database with the gene name. If you know the gene symbol and species, enter them as follows: tpo[sym] AND human[orgn]
2. Click on the desired gene.
3. In the list of Links on the right, click "SNP:GeneView". If the link is not present, no SNPs are currently linked to this gene.

a nucleotide or protein accession number (e.g. NM_001126)

1. Search the Nucleotide or Protein database with the accession number.
2. In the Links menu in the upper right, click on "GeneView in dbSNP". If the link is not present, click on the "Gene" link in the same menu and continue at step 3 above under "a gene name".

a nucleotide sequence

1. Go to the BLAST home page and click "nucleotide blast" under Basic BLAST.
2. Paste the sequence in the query box.
3. Enter the name of the organism of interest in the "Organism" box. Click the BLAST button.
4. Click on the desired sequence from the results.
5. Continue at step 2 under "a nucleotide or protein accession number" above.

a protein sequence

1. Go to the BLAST home page and click "protein blast" under Basic BLAST.
2. Paste the sequence in the query box.
3. Enter the name of the organism of interest in the "Organism" box. Click the BLAST button.
4. Click on the desired sequence from the results.
5. Continue at step 2 under "**a nucleotide or protein accession number**" above.

GeneView page from dbSNP link

GeneView

GeneView via analysis of contig annotation: [TAP1](#) transporter 1, ATP-binding cassette, sub-family B (MDR/TAP)

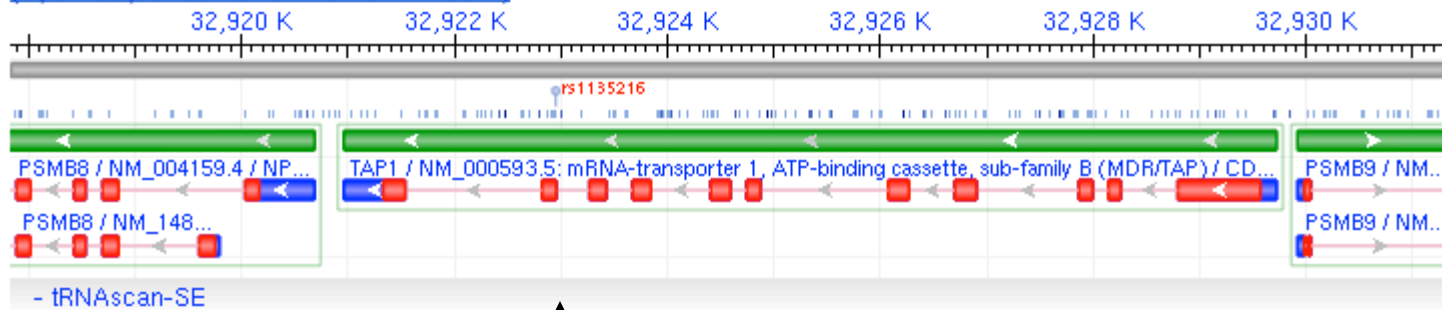
View more variation on this gene (click to hide).

Include clinically associated: in gene region cSNP has frequency double hit

Assembly	SNP to Chr	Chr	Chr position	Contig	Contig position	Allele
reference	-	6	32922953	NT_007592.14	23673225	T

Function	mRNA				Protein		
	mRNA to Chr	Accession	Position	Allele change	Accession	Position	Residue change
missense	-	NM_000593.5	2245	GAC ⇒ GGC	NP_000584.2	697	D [Asp] ⇒ G [Gly]

(Open sequence viewer in a new window.)



Goes to chromosome 6 navigator window.

Tagging SNPs, tSNPs

- SNPs that are highly correlated are redundant information
- **tSNPs** are selected as the minimal non-redundant set of SNPs in a population, such that the genotypes can be reconstructed from the tSNPs.
- tSNPs allow genotyping with fewer steps
 - PCR amplification experiments determine which base is present.
- **Block based tagging**

Block based tagging requires that haplotype "blocks" first be inferred. In the majority of cases when you are investigating association within a candidate gene you are likely to start of with a large number of potential SNPs to choose from, and using various measures of linkage disequilibrium and inferred haplotypes it is possible to define 'haplotype blocks' of markers that are in strong LD with each other, but not with those in other blocks. The exact definition of a haplotype block is open to interpretation, and there are a number of different methods for choosing your haplotype blocks ([Gabriel et al 2002](#),)

[\[Hide banner\]](#) [\[Bookmark this\]](#) [\[Link to Image\]](#) [\[SNP genotype data\]](#) [\[tag SNP Data\]](#) [\[HapMap LD Data\]](#) [\[Phased Haplotype Data\]](#) [\[High-res Image\]](#) [\[Help\]](#)
[\[Reset\]](#)

[Search](#)

[Help links:](#) [- LD -](#) [- tagSNPs -](#) [- Phased Haplotype -](#) [- Genotype data -](#) [- Frequency data -](#) [- Symbols and colours used -](#)

Landmark or Region :

TCF7L2

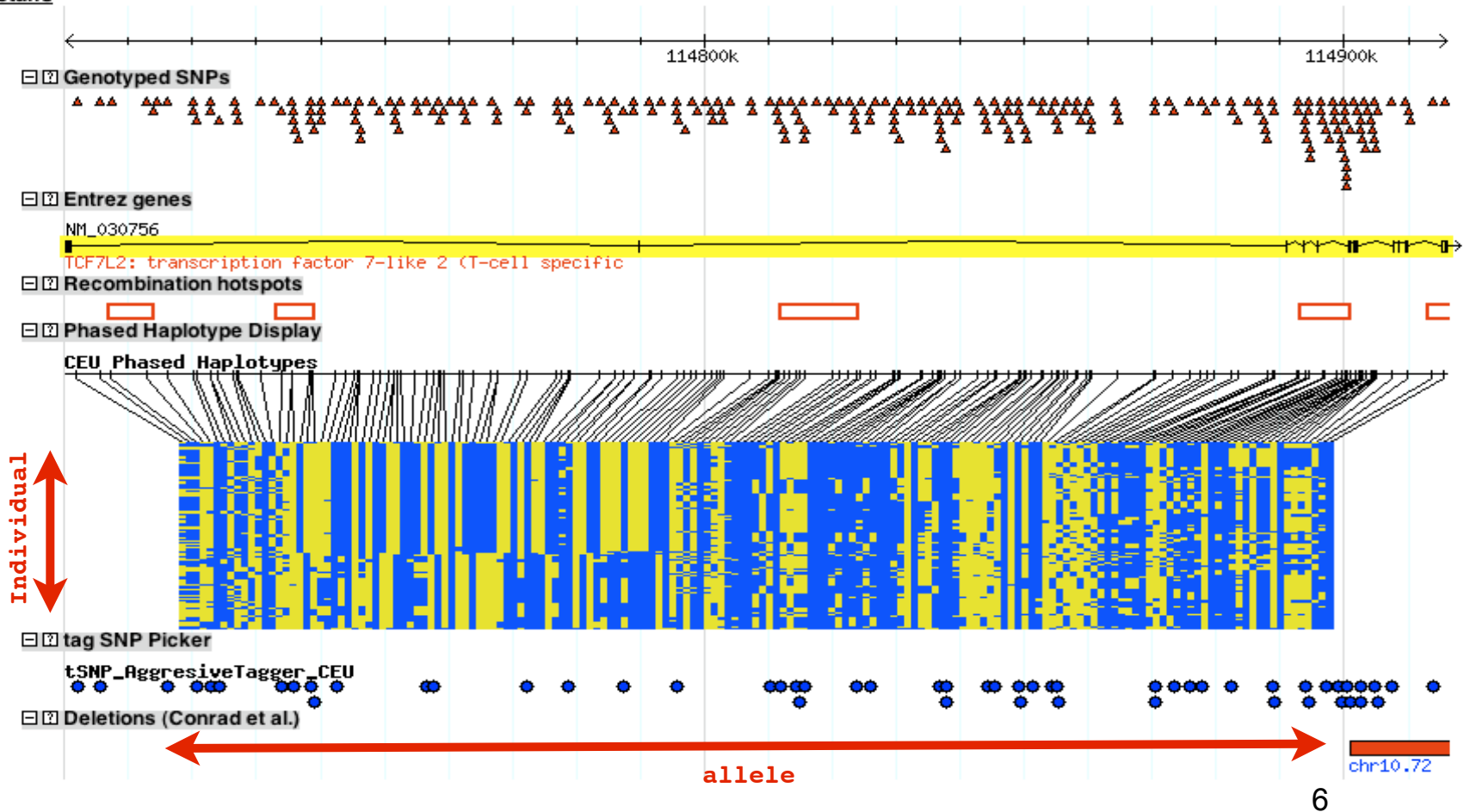
Reports & Analysis :

Data Source

Scroll/Zoom: Show 215.9 kbp Flip

Population descriptors: YRI: Yoruba in Ibadan, Nigeria, JPT: Japanese in Tokyo, Japan, CHB: Han Chinese in Beijing, China, CEU: CEPH (Utah residents with ancestry from northern and western Europe)

- [Overview](#)
- [Region](#)
- [Details](#)



Uses of SNPs

- Personalized medicine
 - Sensitivity to
 - diseases
 - drugs
 - chemicals
 - pathogens
 - vaccines
- Livestock breeding
- Human migrations