New variation resources at the UCSC Genome Browser

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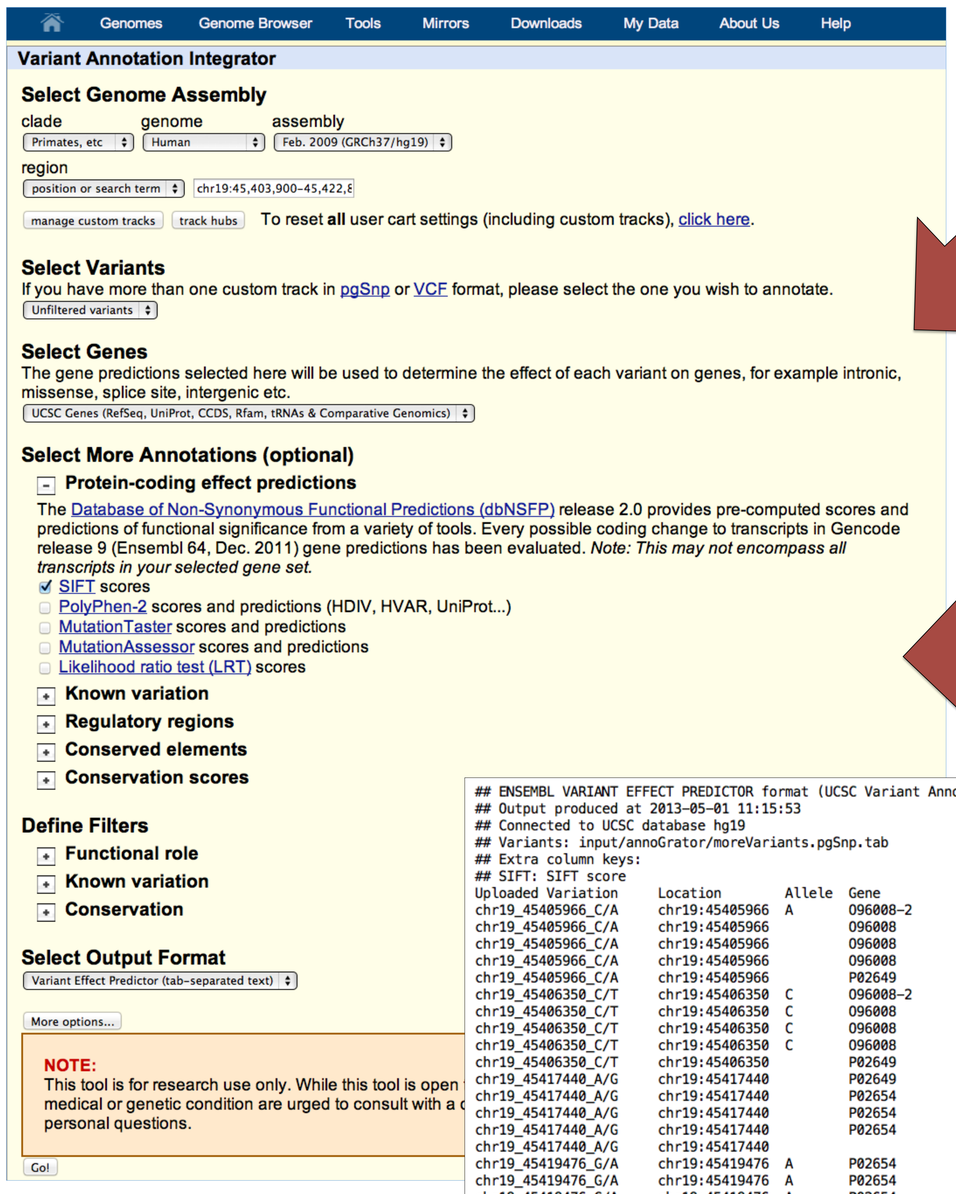
Annotate your variants:



Upload variants:

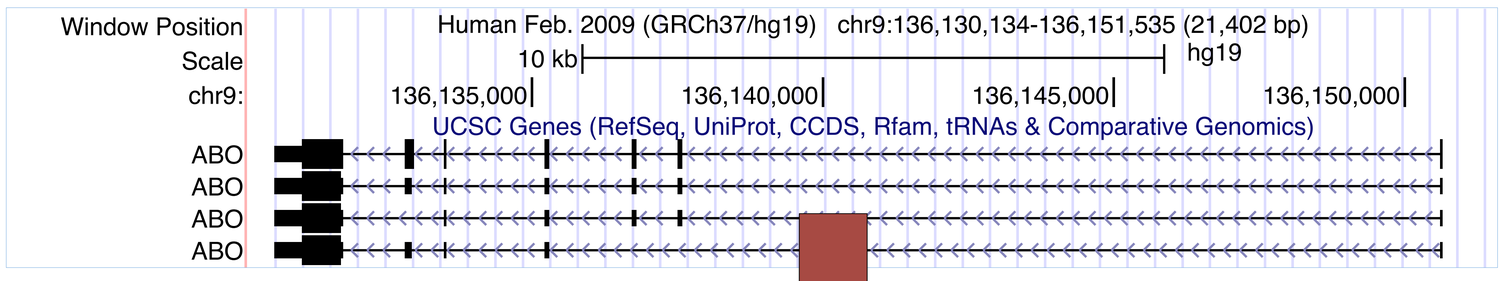
```
track type=pgSnp visibility=pack db=hg19 name=myVariants description="Unfiltered variants"
browser position chr19:45403900-45422800
chr19   45405965   45405966   C/A   2   0,0   0,0
chr19   45406349   45406350   C/T   2   0,0   0,0
chr19   45417439   45417440   A/G   2   0,0   0,0
chr19   45419475   45419476   G/A   2   0,0   0,0
```

Download variants with predicted functional effects and annotations from UCSC Genome Browser data tracks:

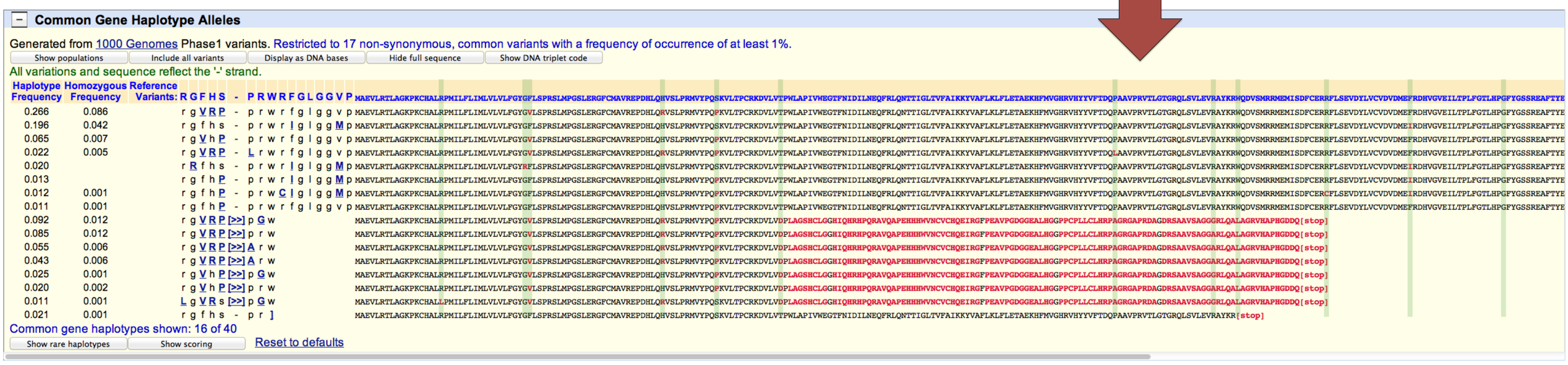
```
## ENSEMBL VARIANT EFFECT PREDICTOR format (UCSC Variant Annotation Int
## Output produced at 2013-05-01 11:15:53
## Connected to UCSC database hg19
## Variants: input/annoGrator/moreVariants.pgSnp.tab
## Extra column keys:
## SIFT: SIFT score
Uploaded Variation  Location        Allele  Gene        Feature     Feature type  Consequence          Position in cDNA  Position in CDS  Position in protein  Amino acid change  Codon change  Co-located Variation  Extra
chr19_45405966_C/A  chr19:45405966  A       O96008-2    uc002ozz.3  Transcript    3_prime_UTR_variant  2449              -                -                    -                  -             -                     EXON=9/9
chr19_45405966_C/A  chr19:45405966          O96008      uc002ozx.4  Transcript    intron_variant       -                 -                -                    -                  -             -                     INTRON=9/10
chr19_45405966_C/A  chr19:45405966          O96008      uc002ozy.4  Transcript    intron_variant       -                 -                -                    -                  -             -                     INTRON=9/10
chr19_45405966_C/A  chr19:45405966          O96008      uc002paa.4  Transcript    intron_variant       -                 -                -                    -                  -             -                     INTRON=8/9
chr19_45405966_C/A  chr19:45405966          P02649      uc002pab.3  Transcript    upstream_gene_variant -                -                -                    -                  -             -                     DISTANCE=3073
chr19_45406350_C/T  chr19:45406350  C       O96008-2    uc002ozz.3  Transcript    3_prime_UTR_variant  2833              -                -                    -                  -             -                     SIFT=0.010000;EXON=9/9
chr19_45406350_C/T  chr19:45406350  C       O96008      uc002ozx.4  Transcript    missense_variant     1111              1010             337                  L/P                CTG/CCG       -                     SIFT=0.010000;EXON=10/10
chr19_45406350_C/T  chr19:45406350  C       O96008      uc002ozy.4  Transcript    missense_variant     1114              1010             337                  L/P                CTG/CCG       -                     SIFT=0.010000;EXON=10/10
chr19_45406350_C/T  chr19:45406350  C       O96008      uc002paa.4  Transcript    missense_variant     1206              1010             337                  L/P                CTG/CCG       -                     SIFT=0.010000;EXON=9/9
chr19_45406350_C/T  chr19:45406350          P02649      uc002pab.3  Transcript    upstream_gene_variant -                -                -                    -                  -             -                     SIFT=0.010000;DISTANCE=2689
chr19_45417440_A/G  chr19:45417440          P02649      uc002pab.3  Transcript    downstream_gene_variant -              -                -                    -                  -             rs111575056           DISTANCE=4789
chr19_45417440_A/G  chr19:45417440          P02654      uc002pac.1  Transcript    upstream_gene_variant -                -                -                    -                  -             rs111575056           DISTANCE=137
chr19_45417440_A/G  chr19:45417440          P02654      uc002pad.1  Transcript    upstream_gene_variant -                -                -                    -                  -             rs111575056           DISTANCE=137
chr19_45417440_A/G  chr19:45417440          P02654      uc002pae.1  Transcript    upstream_gene_variant -                -                -                    -                  -             rs111575056           DISTANCE=481
chr19_45417440_A/G  chr19:45417440                      uc002paf.1  Transcript    upstream_gene_variant -                -                -                    -                  -             rs111575056           DISTANCE=1994
chr19_45419476_G/A  chr19:45419476  A       P02654      uc002pac.1  Transcript    missense_variant     340               88               30                   V/I                GTC/ATC       rs72654455            SIFT=0.840000;EXON=4/5
chr19_45419476_G/A  chr19:45419476  A       P02654      uc002pad.1  Transcript    missense_variant     278               88               30                   V/I                GTC/ATC       rs72654455            SIFT=0.840000;EXON=3/4
chr19_45419476_G/A  chr19:45419476  A       P02654      uc002pae.1  Transcript    missense_variant     178               88               30                   V/I                GTC/ATC       rs72654455            SIFT=0.840000;EXON=3/4
chr19_45419476_G/A  chr19:45419476  A                   uc002paf.1  Transcript    non_coding_exon_variant 43             -                -                    -                  -             rs72654455            SIFT=0.840000;EXON=1/2
```

View gene alleles (haplotypes) from 1000 Genomes data:

Click on a gene in the UCSC Genes track:



Scroll to the Gene Alleles section:



Haplotype and homozygosity frequencies.

Summary of variants in each haplotype allele.

Sequences with predicted effects. Variants are highlighted by vertical lines, and changes to the predicted amino acid sequence are highlighted in red. This gene (ABO) shows a frameshift mutation in several haplotypes.

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See our wiki for more details:

http://genomewiki.ucsc.edu/index.php/BoG2013VariationPoster