

New variation resources at the UCSC Genome Browser

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Annotate your variants:

Variant Annotation Integrator

Select Genome Assembly
clade: Primates, etc. genome: Human assembly: Feb. 2009 (GRCh37/hg19)

region: chr19:45,403,900-45,422,800

Select Variants
If you have more than one custom track in pgSnp or VCF format, please select the one you wish to annotate.
Unfiltered variants

Select Genes
The gene predictions selected here will be used to determine the effect of each variant on genes, for example intronic, missense, splice site, intergenic etc.
UCSC Genes (RefSeq, UniProt, CCDS, Rfam, tRNAs & Comparative Genomics)

Select More Annotations (optional)
 Protein-coding effect predictions
The Database of Non-Synonymous Functional Predictions (dbNSFP) release 2.0 provides pre-computed scores and predictions of functional significance from a variety of tools. Every possible coding change to transcripts in Gencode release 9 (Ensembl 64, Dec. 2011) gene predictions has been evaluated. Note: This may not encompass all transcripts in your selected gene set.
 SIFT scores
 PolyPhen-2 scores and predictions (HDIV, HVAR, UniProt...)
 MutationTaster scores and predictions
 MutationAssessor scores and predictions
 Likelihood ratio test (LRT) scores

Known variation
Regulatory regions
Conserved elements
Conservation scores

Define Filters
 Functional role
 Known variation
 Conservation

Select Output Format
Variant Effect Predictor (tab-separated text)

NOTE:
This tool is for research use only. While this tool is open medical or genetic condition are urged to consult with a personal questions.

Upload variants:

```
track type=pgSnp visibility=pack db=hg19 name=myVariants description="Unfiltered variants"
browser position chr19:45403900-45422800
```

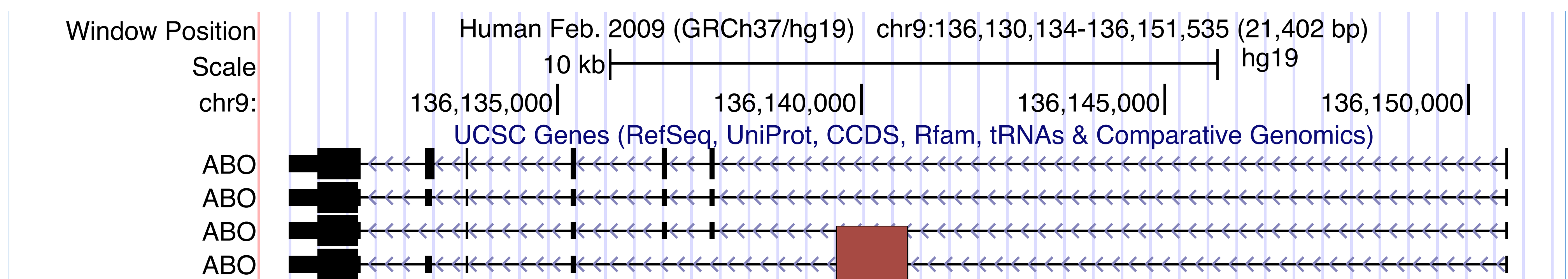
chr	pos	ref	alt	type	score	info
chr19	45405965	A	C/A	2	0,0	0,0
chr19	45406349	A	C/T	2	0,0	0,0
chr19	45417439	A	A/G	2	0,0	0,0
chr19	45419475	A	G/A	2	0,0	0,0

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 096008-2 uc002oz2.3 Transcript 3_prime_UTR_variant 2449 - - - - - - EXON=9/9
chr19_45405966_C/A chr19:45405966 096008 uc002ozx.4 Transcript intron_variant - - - - - - INTRON=9/10
chr19_45405966_C/A chr19:45405966 096008 uc002zozy.4 Transcript intron_variant - - - - - - INTRON=9/10
chr19_45405966_C/A chr19:45405966 096008 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 096008-2 uc002oz2.3 Transcript 3_prime_UTR_variant 2833 - - - - - - SIFT=0.010000; EXON=9/9
chr19_45406350_C/T chr19:45406350 C 096008 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 096008 uc002zozy.4 Transcript missense_variant 1114 1010 337 L/P CTG/CCG SIFT=0.010000; EXON=10/10
chr19_45406350_C/T chr19:45406350 C 096008 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 - - - - - - DISTANCE=4789
chr19_45417440_A/G chr19:45417440 P02654 uc002pac.1 Transcript upstream_gene_variant - - - - - - DISTANCE=137
chr19_45417440_A/G chr19:45417440 P02654 uc002pad.1 Transcript upstream_gene_variant - - - - - - DISTANCE=137
chr19_45417440_A/G chr19:45417440 P02654 uc002pae.1 Transcript upstream_gene_variant - - - - - - DISTANCE=481
chr19_45417440_A/G chr19:45417440 P02654 uc002paf.1 Transcript upstream_gene_variant - - - - - - DISTANCE=1994
chr19_45419476_G/A chr19:45419476 A P02654 uc002pac.1 Transcript missense_variant 340 88 30 V/I GTC/ATC SIFT=0.040000; EXON=4/5
chr19_45419476_G/A chr19:45419476 A P02654 uc002pad.1 Transcript missense_variant 278 88 30 V/I GTC/ATC SIFT=0.040000; EXON=3/4
chr19_45419476_G/A chr19:45419476 A P02654 uc002pae.1 Transcript missense_variant 178 88 30 V/I GTC/ATC SIFT=0.040000; EXON=3/4
chr19_45419476_G/A chr19:45419476 A P02654 uc002paf.1 Transcript non_coding_exon_variant 43 - - - - - - SIFT=0.040000; 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:

Common Gene Haplotype Alleles

Generated from 1000 Genomes Phase1 variants. Restricted to 17 non-synonymous, common variants with a frequency of occurrence of at least 1%.

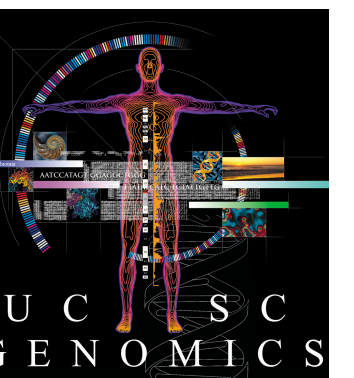
Frequency Variants: R G F H S

Frequency	Variants: R G F H S
0.266	0.086 r g V R P - p r w r f g l g g v
0.196	0.042 r g f h s - p r w r l g l g g v
0.065	0.007 r g V h P - p r w r f g l g g v
0.022	0.005 r g V R P - L r w r f g l g g v
0.020	r R f h s - p r w r l g l g g v
0.013	r g f h P - p r w r l g l g g v
0.012	0.001 r g f h P - p r w C l g l g g v
0.011	0.001 r g f h P - p r w r f g l g g v
0.092	0.012 r g V R P >>> p g w
0.085	0.012 r g V R P >>> p r w
0.055	0.006 r g V R P >>> A r w
0.043	0.006 r g V R P >>> A r w
0.025	0.001 r g V h P >>> p g w
0.020	0.002 r g V h P >>> p r w
0.011	0.001 L g V R s >>> p g w
0.021	0.001 r g f h s - p r]

Common gene haplotypes shown: 16 of 40

Haplotype and Summary of variants in each haplotype allele. Variants 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>

