

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: position or search term: 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	pos1	pos2	ref	alt	freq	info	freq
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,
chr19	45419475	45419476	G/A		2	0,0	0,0

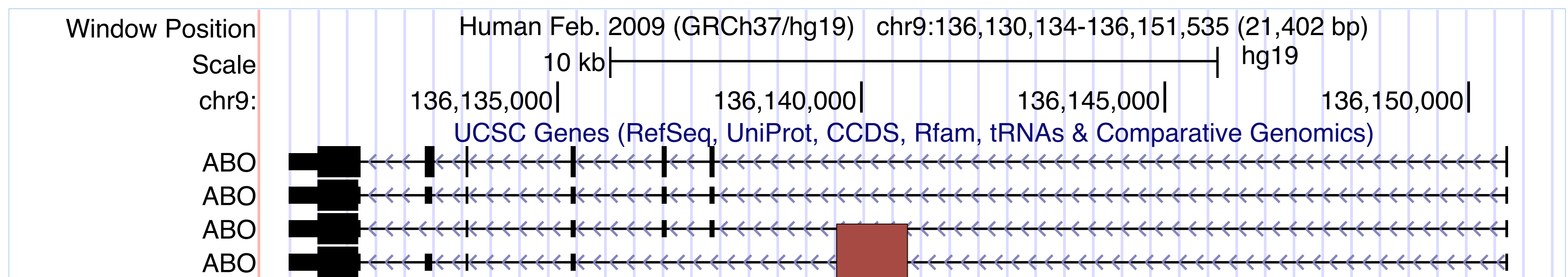
Download variants with predicted functional effects and annotations from UCSC Genome Browser data tracks:

```
## 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
```

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	A	096008	uc002ozx.4	Transcript	intron_variant	-	-	-	-	-	-	INTRON=9/10
chr19_45405966_C/A	chr19:45405966	A	096008	uc002zozy.4	Transcript	intron_variant	-	-	-	-	-	-	INTRON=9/10
chr19_45405966_C/A	chr19:45405966	A	096008	uc002paa.4	Transcript	intron_variant	-	-	-	-	-	-	INTRON=8/9
chr19_45405966_C/A	chr19:45405966	A	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	C	P02649	uc002pab.3	Transcript	upstream_gene_variant	-	-	-	-	-	-	SIFT=0.010000;DISTANCE=2689
chr19_45417440_A/G	chr19:45417440	A	P02649	uc002pab.3	Transcript	downstream_gene_variant	-	-	-	-	-	-	DISTANCE=4789
chr19_45417440_A/G	chr19:45417440	A	P02654	uc002pac.1	Transcript	upstream_gene_variant	-	-	-	-	-	-	DISTANCE=137
chr19_45417440_A/G	chr19:45417440	A	P02654	uc002pad.1	Transcript	upstream_gene_variant	-	-	-	-	-	-	DISTANCE=137
chr19_45417440_A/G	chr19:45417440	A	P02654	uc002pae.1	Transcript	upstream_gene_variant	-	-	-	-	-	-	DISTANCE=481
chr19_45417440_A/G	chr19:45417440	A	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	r572654455	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	r572654455	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	r572654455	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 Frequency Variants: R G F H S - P R W R F G L G G V P

Frequency	Frequency	Variants: R G F H S - P R W R F G L G G V P
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]

Haplotype and homozygosity frequencies. 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.

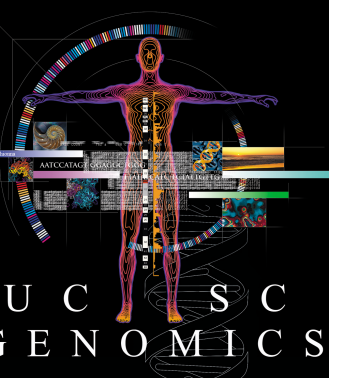
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See our wiki for more details:

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

