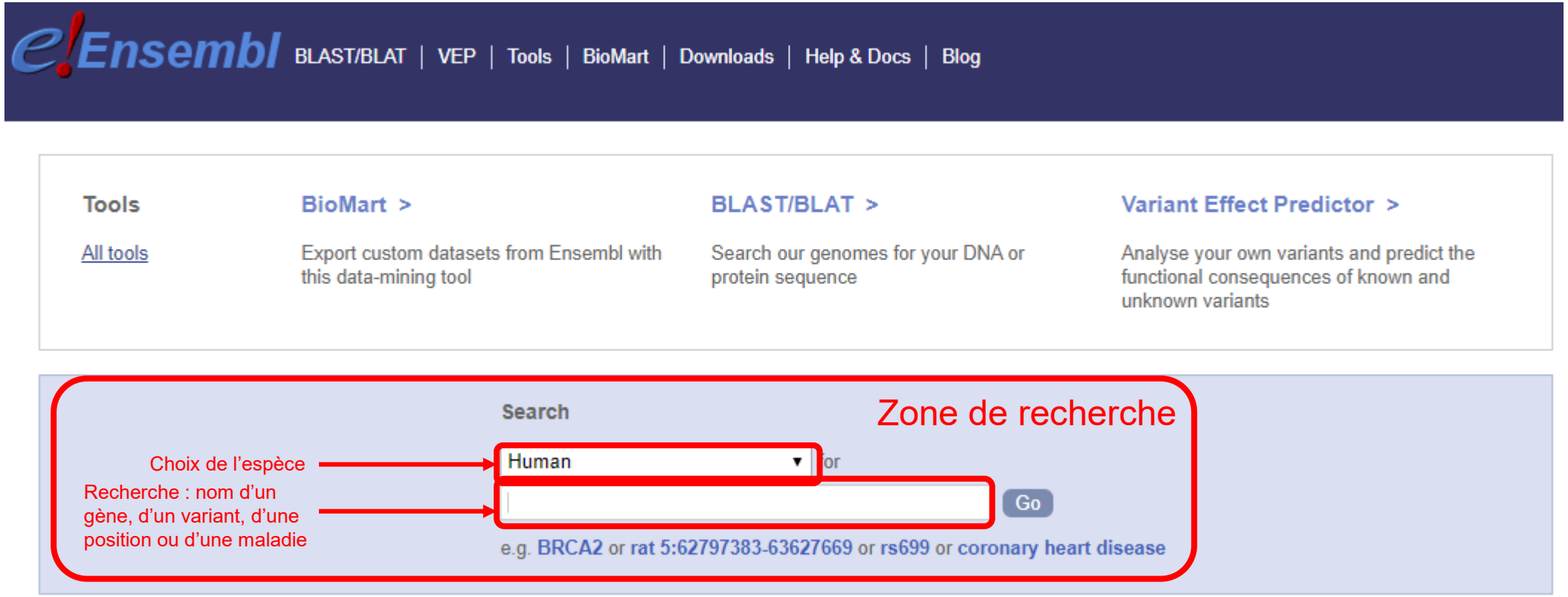
Fiche technique : comment interroger la base de données Ensembl ? http://www.ensembl.org/index.html

Ensembl est une base de données sur les séquences de génomes.



Location: 1:230,709,548-230,710,548 | Variant: rs699

Variant displays
- Explore this variant
- Genomic context
  - Genes and regulation
  - Flanking sequence
- Population genetics
- Phenotype data
- Sample genotypes
- Linkage disequilibrium
- Phylogenetic context
- Citations
- 3D Protein model

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rs699 SNP

Most severe consequence missense variant | See all predicted consequences

Alleles A/G | Ancestral: G | MAF: 0.29 (A) | Highest population MAF: 0.50

Change tolerance CADD: G:1.939 | GERP: -6.59

Location Chromosome 1:230710048 (forward strand) | VCF: `1 230710048 rs699 A G`

Co-located variants dbSNP rs1553314015 (A/-) ; HGMD-PUBLIC CM920010

Evidence status

Clinical significance

HGVS names This variant has 5 HGVS names - Show

Synonyms This variant has 13 synonyms - Show

Genotyping chips This variant has assays on 11 chips - Hide
- HumanCoreExome-12
- Illumina_Human610_Quad
- Illumina_ImmunoChip
- Illumina_Human660W-quad
- Illumina_HumanOmni2.5
- Illumina_ExomeChip
- Illumina_HumanHap650Y
- HumanOmniExpress
- Illumina_1M-duo
- Illumina_HumanOmni1-Quad
- Illumina_HumanHap550

Original source Variants (including SNPs and indels) imported from dbSNP (release 152) | View in dbSNP

About this variant This variant overlaps 1 transcript, has 2504 sample genotypes, is associated with 8 phenotypes and is mentioned in 188 citations.

Informations sur les allèles du variant

Localisation dans le génome

Puces à ADN permettant l'identification

Gene and Transcript consequences

Show/hide columns | Filter

Gene	Transcript (strand)	Allele (Tr. allele)	Consequence Type	Position in transcript	Position in CDS	Position in protein	AA	Codons	SIFT	PolyPhen	CADD	REVEL	MetaLR	Mutation Assessor	Detail
ENSG00000135744 HGNC: AGT	ENST00000366667.5 (-) biotype: protein_coding	G (C)	missense variant	843 (out of 2116)	803 (out of 1458)	268 (out of 485)	M/T	ATG/ACG	1	0	1	0.157	0	0.003	Show

No Gene expression correlations

Error loading eQTL data from REST

No overlap with Ensembl Regulatory features

No overlap with Ensembl Motif features

Conséquences sur l'expression du gène

Population genetics

1000 Genomes Project Phase 3 allele frequencies

ALL	AFR	AMR	EAS	EUR	SAS
C: 17% T: 83%	C: 7% T: 93%	C: 11% T: 89%	C: 23% T: 77%	C: 20% T: 80%	C: 24% T: 76%
	Sub-populations	Sub-populations	Sub-populations	Sub-populations	Sub-populations

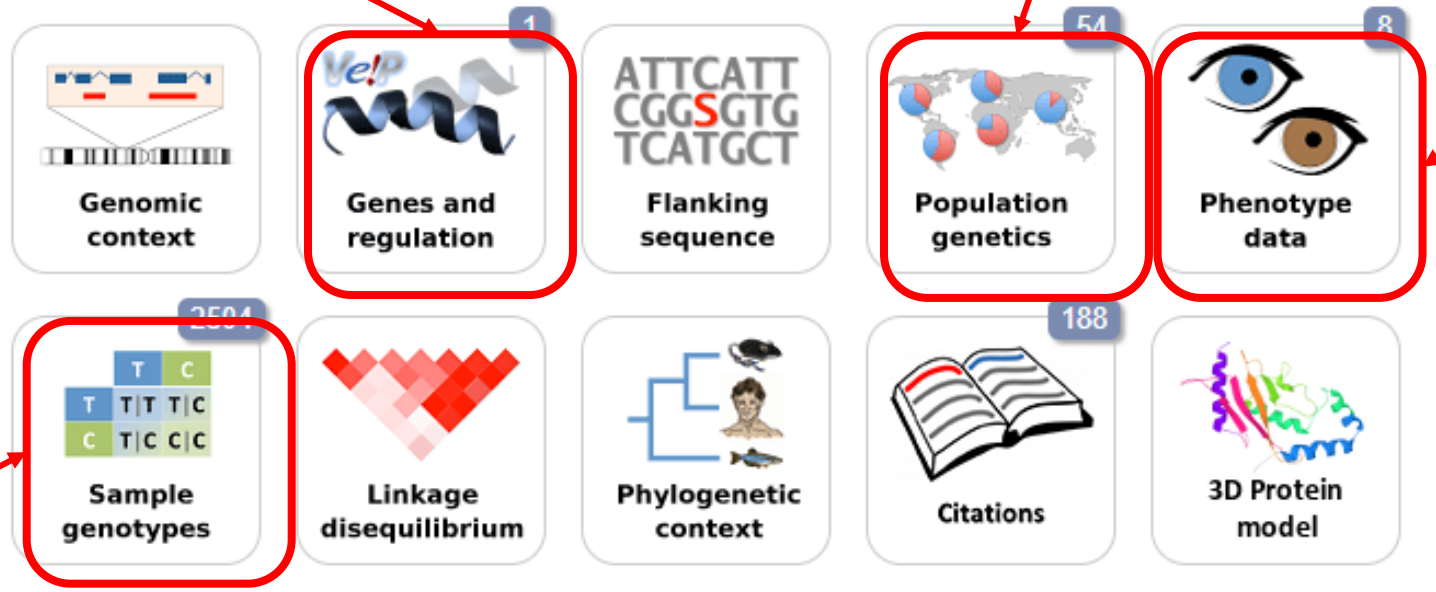
Jump to: 1000 Genomes Project Phase 3 (32) | gnomAD genomes (8) | TOPMed (1) | UK10K (2)

1000 Genomes Project Phase 3 (32)

Show All entries | Show/hide columns

Population	Allele: frequency (count)		Genotype: frequency (count)	
ALL	C: 0.168 (841)	T: 0.832 (4167)	C|C: 0.037 (92)	C|T: 0.262 (657)
AFR	C: 0.073 (96)	T: 0.927 (1226)	C|C: 0.009 (6)	C|T: 0.127 (84)
ACB	C: 0.089 (17)	T: 0.911 (175)	C|C: 0.021 (2)	C|T: 0.135 (13)
ASW	C: 0.098 (12)	T: 0.902 (110)	C|T: 0.197 (12)	T|T: 0.803 (49)

Fréquences des allèles par population



Phénotypes associés

Echantillons du génotype (populations et individus analysés)

Show All entries | Show/hide columns

Number of genotypes		Population	Description
2504	Show	ALL	All phase 3 individuals
96	Show	ACB	African Caribbean in Barbados
661	Show	AFR	African
347	Show	AMR	American
61	Show	ASW	African Ancestry in Southwest US
86	Show	BEB	Bengali in Bangladesh
93	Show	CDX	Chinese Dai in Xishuangbanna, China
99	Show	CEU	Utah residents with Northern and Western European ancestry
103	Show	CHB	Han Chinese in Bejing, China

Phenotype Data

Significant association(s)

Show All entries

Phenotype, disease and trait
ClinVar: phenotype not specified
Coronary Artery Disease
HYPERTENSION, ESSENTIAL, SUSCEPTIBILITY TO
Mean arterial pressure
Preeclampsia, susceptibility to
Renal dysplasia
Susceptibility to progression to renal failure in IgA nephropathy
Systolic blood pressure