Locus 51 (rs778986)

SNiPAcard

Block annotations

Block info

genomic range	chr19:5,844,537-5,844,649
block size	113 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 1.409 [-0.147 – 2.964]	gene(s) hit or close-by	AC024592.9, FUT3, FUT6
phastCons	μ = 0.498 [0 – 0.996]	eQTL gene(s)	–
GERP++	μ = 0.964 [-0.403 – 2.33]	potentially regulated gene(s)	–
CADD score	μ = 7.561 [3.992 – 11.13]	disease gene(s)	FUT6

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM970545	1
?	HGMD curated	HGMD	CM930259	1

Disease gene annotation

gene	trait	source DB	source entry/link
FUT6	FUCOSYLTRANSFERASE 6 DEFICIENCY	OMIM	MIM:613852

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
FUT3	missense variant	ENST00000589620	NM_001097639.1	ENSP00000465804	2	2	deleterious, tolerated		2
FUT3	missense variant	ENST00000589714	?	ENSP00000467081	2	2	deleterious, tolerated		2
FUT3	missense variant	ENST00000589918	NM_001097640.1	ENSP00000468123	2	2	deleterious, tolerated		2
FUT3	missense variant	ENST00000303225	NM_000149.3	ENSP00000305603	2	2	deleterious, tolerated		2
FUT3	missense variant	ENST00000458379	NM_001097641.1	ENSP00000416443	2	2	deleterious, tolerated		2
FUT3	missense variant	ENST00000587048	?	ENSP00000468515	R/W	Cgg/Tgg	tolerated	benign	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC024592.9	upstream gene variant	2829	ENST00000589276	?	?	2
FUT3	downstream gene variant	14	ENST00000587048	?	ENSP00000468515	1
FUT3	downstream gene variant	1927	ENST00000593144	?	?	2
FUT3	downstream gene variant	2169	ENST00000588539	?	?	2

FUT3	downstream gene variant	69	ENST00000585715	?	ENSP00000467633	2
FUT3	downstream gene variant	188	ENST00000587183	?	ENSP00000468795	2
FUT6	upstream gene variant	4809	ENST00000286955	NM_001040701.1	ENSP00000286955	2
FUT6	upstream gene variant	4804	ENST00000591079	?	?	2
FUT6	upstream gene variant	4834	ENST00000524754	?	ENSP00000431708	2
FUT6	upstream gene variant	4795	ENST00000318336	NM_000150.2	ENSP00000313398	2