SNiPAcard

Block annotations

Block info

genomic range	chr19:5,844,792-5,844,792
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	0.986	**gene(s) hit or close-by**	AC024592.9, FUT3
phastCons	0.002	**eQTL gene(s)**	–
GERP++	2.33	**potentially regulated gene(s)**	–
CADD score	8.418	**disease gene(s)**	–

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM930258	1
Le(-) PHENOTYPE	OMIM curated	OMIM	MIM:111100	1

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	ENST00000589918	NM_001097640.1	ENSP00000468123	R/L	cGg/cTg	?	?	1
FUT3	missense variant	ENST00000589620	NM_001097639.1	ENSP00000465804	R/L	cGg/cTg	?	?	1
FUT3	missense variant	ENST00000303225	NM_000149.3	ENSP00000305603	R/L	cGg/cTg	?	?	1
FUT3	missense variant	ENST00000587048	?	ENSP00000468515	R/L	cGg/cTg	?	?	1
FUT3	missense variant	ENST00000585715	?	ENSP00000467633	R/L	cGg/cTg	?	?	1
FUT3	missense variant	ENST00000458379	NM_001097641.1	ENSP00000416443	R/L	cGg/cTg	?	?	1
FUT3	missense variant	ENST00000589714	?	ENSP00000467081	R/L	cGg/cTg	?	?	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	2686	ENST00000589276	?	?	1
FUT3	downstream gene variant	1784	ENST00000593144	?	?	1
FUT3	downstream gene variant	2026	ENST00000588539	?	?	1
FUT3	downstream gene variant	45	ENST00000587183	?	ENSP00000468795	1