Locus 273 (rs12121180)

SNiPAcard

Block annotations

Block info

genomic range	chr1:154,282,222-154,283,908
block size	1,687 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.457 [-0.764 – -0.15]	gene(s) hit or close-by	RP11-137P24.5
phastCons	μ = 0.006 [0.001 – 0.011]	eQTL gene(s)	HAX1, IL6R, S100A4, UBAP2L
GERP++	μ = 0.050 [-0.135 – 0.235]	potentially regulated gene(s)	–
CADD score	μ = 4.104 [3.269 – 4.939]	disease gene(s)	HAX1, IL6R

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
HAX1	NEUTROPENIA, SEVERE CONGENITAL, 3, AUTOSOMAL RECESSIVE	DECIPHER	MIM:610738
HAX1	NEUTROPENIA, SEVERE CONGENITAL, 3, AUTOSOMAL RECESSIVE	OMIM	MIM:610738
IL6R	INTERLEUKIN 6, SERUM LEVEL OF, QUANTITATIVE TRAIT LOCUS	OMIM	MIM:614752
IL6R	SOLUBLE INTERLEUKIN-6 RECEPTOR, SERUM LEVEL OF, QUANTITATIVE [...]	OMIM	MIM:614689
HAX1	Kostmann syndrome	OrphaNet	OrphaNet:99749

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
IL6R	ENST00000368485	ILMN_1696394	blood	1.01×10-4 (p-value)	Westra et al.	1
IL6R	ENST00000507256					
IL6R	ENST00000502679					
IL6R	ENST00000515190					
UBAP2L	ENST00000484819	ILMN_1814789	blood	9.02×10-5 (p-value)	Westra et al.	1
UBAP2L	ENST00000361546					
UBAP2L	ENST00000428595					
UBAP2L	ENST00000428931					
HAX1	ENST00000531435	ILMN_1750658	blood	6.09×10-5 (p-value)	Westra et al.	1
HAX1	ENST00000447768					
HAX1	ENST00000459914					
HAX1	ENST00000435087					
HAX1	ENST00000328703					
HAX1	ENST00000457918					
HAX1	ENST00000471326					
HAX1	ENST00000483970					
HAX1	ENST00000532105					
HAX1	ENST00000492550					
HAX1	ENST00000477780					
S100A4	?	ENSG00000196154	esophagus mucosa	5.27×10-6 (p-value)	GTEx Portal V6	2

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP11-137P24.5	upstream gene variant	1030	ENST00000612670	?	?	2