SNiPAcard

Block annotations

Block info

genomic range	chr1:154,631,123-154,652,119
block size	20,997 bp
variant count	11 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.331 [-0.913 – 1.261]	**gene(s) hit or close-by**	–
phastCons	μ = 0.126 [0 – 0.533]	**eQTL gene(s)**	IL6R, RUSC1-AS1
GERP++	μ = 0.161 [-0.815 – 1.54]	**potentially regulated gene(s)**	–
CADD score	μ = 4.915 [0.849 – 18.13]	**disease gene(s)**	IL6R

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link	
IL6R	INTERLEUKIN 6, SERUM LEVEL OF, QUANTITATIVE TRAIT LOCUS	OMIM	MIM:614752	OMIM
IL6R	SOLUBLE INTERLEUKIN-6 RECEPTOR, SERUM LEVEL OF, QUANTITATIVE [...]	OMIM	MIM:614689	OMIM

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RUSC1-AS1	ENST00000543656	ILMN_2114185	blood	4.94×10^{-5} (p-value)	MuTHER consortium	4
RUSC1-AS1	ENST00000446880					
RUSC1-AS1	ENST00000450199					
IL6R	ENST00000368485	ILMN_1696394	blood	5.45×10^{-6} (p-value)	Westra et al.	5
IL6R	ENST00000507256					
IL6R	ENST00000502679					
IL6R	ENST00000515190					