SNiPAcard

Block annotations

Block info

genomic range	chr3:142,602,666-142,631,909
block size	29,244 bp
variant count	13 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.009 [-0.853 – 1.31]	**gene(s) hit or close-by**	PCOLCE2
phastCons	μ = 0.019 [0 – 0.107]	**eQTL gene(s)**	PCOLCE2
GERP++	μ = 0.012 [-1.66 – 2.8]	**potentially regulated gene(s)**	ATR, PLS1, RP11-372E1.4, RP11-372E1.7, RP11-383G6.3, RP13-635I23.3, U2SURP
CADD score	μ = 5.080 [0.216 – 12.01]	**disease gene(s)**	ATR

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
1-oleoylglycerophosphoethanolamine	2.36×10^{-6}	Metabolomics GWAS Server	24816252	7

Disease gene annotation

gene	trait	source DB	source entry/link
ATR	SECKEL SYNDROME 1	OMIM	MIM:210600
ATR	CUTANEOUS TELANGIECTASIA AND CANCER SYNDROME, FAMILIAL	OMIM	MIM:614564
ATR	Seckel syndrome type 1 (SCKL1)	DECIPHER	MIM:210600
ATR	Seckel syndrome	OrphaNet	OrphaNet:808
ATR	Familial cutaneous telangiectasia and oropharyngeal predisposition cancer syndrome	OrphaNet	OrphaNet:313846

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PCOLCE2	ENST00000485766	ILMN_1746888	adipocyte	1.81×10^{-15} (p-value)	MuTHER consortium	7
PCOLCE2	ENST00000470795					
PCOLCE2	ENST00000295992					
PCOLCE2	?	ENSG00000163710	aorta	1.11×10^{-11} (p-value)	GTEx Portal V6	12
PCOLCE2	?	ENSG00000163710	left ventricle	5.95×10^{-6} (p-value)	GTEx Portal V6	7

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000357025	1	ENCP00000038521	RP11-372E1.4
		ENCP00000038539	RP13-635I23.3
ENCE00000356905	1	ENCP00000038522	RP11-372E1.7

	ENCP00000038513	PLS1
	ENCP00000038531	U2SURP
	ENCP00000038504	RP11-383G6.3 ATR
	ENCP00000038510	PLS1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001679457 (open chromatin region)	1	endothelium (HUVEC)	H3K27me3
		embryonic stem cell (H1ESC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
ENSR00001679459 (enhancer)	1	HSMMtube	H3K27me3
		blood (DND-41)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
PCOLCE2	downstream gene variant, upstream gene variant	512	ENST00000483454	?	?	7
PCOLCE2	upstream gene variant	2010	ENST00000495732	?	?	2
PCOLCE2	upstream gene variant	2172	ENST00000485766	?	ENSP00000419842	2
PCOLCE2	upstream gene variant	2326	ENST00000493733	?	?	2
PCOLCE2	upstream gene variant	2049	ENST00000461818	?	?	2
PCOLCE2	upstream gene variant	1868	ENST00000295992	NM_013363.3	ENSP00000295992	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PCOLCE2	ENST00000495732	?	?	5
PCOLCE2	ENST00000461818	?	?	5
PCOLCE2	ENST00000485766	?	ENSP00000419842	5
PCOLCE2	ENST00000493733	?	?	5
PCOLCE2	ENST00000295992	NM_013363.3	ENSP00000295992	5