SNiPAcard

Block annotations

Block info

genomic range	chr13:113,830,329-113,911,651
block size	81,323 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.113 [-0.295 – 0.741]	**gene(s) hit or close-by**	CUL4A, PCID2, PROZ
phastCons	μ = 0.005 [0 – 0.022]	**eQTL gene(s)**	PROZ, RP11-120K24.4
GERP++	μ = -1.060 [-3.57 – 1.1]	**potentially regulated gene(s)**	–
CADD score	μ = 2.255 [1.398 – 4.143]	**disease gene(s)**	PROZ

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
PROZ	Cerebral sinovenous thrombosis	OrphaNet	OrphaNet:329217

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RP11-120K24.4	?	ENSG00000269356	sigmoid colon	3.87×10^{-7} (p-value)	GTEx Portal V6	3
PROZ	?	ENSG00000126231	liver	1.11×10^{-7} (p-value)	GTEx Portal V6	6
PROZ	ENST00000375547	ILMN_1664565	liver	1.02×10^{-14} (p-value)	Innocenti et al.	1
PROZ	ENST00000342783					

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CUL4A	upstream gene variant	2206	ENST00000375441	NM_001278513.1	ENSP00000364590	1
CUL4A	downstream gene variant	2562	ENST00000470067	?	?	1
CUL4A	upstream gene variant, downstream gene variant	3613	ENST00000488558	?	ENSP00000480367	2
CUL4A	upstream gene variant	2707	ENST00000326335	NM_001278514.1	ENSP00000322132	1
CUL4A	upstream gene variant	3896	ENST00000498562	?	?	1
CUL4A	upstream gene variant	3557	ENST00000375440	NM_001008895.2	ENSP00000364589	1
CUL4A	upstream gene variant	2785	ENST00000451881	NM_003589.2	ENSP00000389118	1
CUL4A	upstream gene variant, downstream gene variant	3846	ENST00000463426	?	?	2
CUL4A	upstream gene variant	2849	ENST00000617546	?	ENSP00000481782	1
CUL4A	upstream gene variant	3629	ENST00000474116	?	?	1

CUL4A	upstream gene variant	3643	ENST00000494985	?	?	1
PCID2	downstream gene variant	1597	ENST00000246505	?	ENSP00000246505	1
PCID2	downstream gene variant	1596	ENST00000375477	?	ENSP00000364626	1
PCID2	downstream gene variant	1596	ENST00000375479	NM_018386.3, NM_001127203.2	ENSP00000364628	1
PCID2	downstream gene variant	1602	ENST00000375457	NM_001258213.1	ENSP00000364606	1
PCID2	downstream gene variant	4146	ENST00000475433	?	?	1
PCID2	downstream gene variant	2257	ENST00000473462	?	?	1
PCID2	downstream gene variant	1597	ENST00000463102	?	?	1
PCID2	downstream gene variant	1598	ENST00000462653	?	?	1
PCID2	downstream gene variant	1597	ENST00000375459	?	ENSP00000364608	1
PCID2	downstream gene variant	4205	ENST00000480971	?	?	1
PCID2	downstream gene variant	1524	ENST00000622406	NM_001258212.1	ENSP00000479494	1
PCID2	downstream gene variant	1597	ENST00000493650	?	?	1
PCID2	downstream gene variant	1562	ENST00000337344	NM_001127202.2	ENSP00000337405	1
PROZ	downstream gene variant	3632	ENST00000375547	NM_003891.2	ENSP00000364697	1
PROZ	downstream gene variant	3635	ENST00000342783	NM_001256134.1	ENSP00000344458	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CUL4A	ENST00000472083	?	?	1
CUL4A	ENST00000451881	NM_003589.2	ENSP00000389118	3
CUL4A	ENST00000326335	NM_001278514.1	ENSP00000322132	3
CUL4A	ENST00000375441	NM_001278513.1	ENSP00000364590	3
CUL4A	ENST00000488558	?	ENSP00000480367	1
CUL4A	ENST00000498562	?	?	2
CUL4A	ENST00000375440	NM_001008895.2	ENSP00000364589	3
CUL4A	ENST00000463426	?	?	1
CUL4A	ENST00000617546	?	ENSP00000481782	3
CUL4A	ENST00000494985	?	?	1
PCID2	ENST00000337344	NM_001127202.2	ENSP00000337405	2
PCID2	ENST00000493650	?	?	1
PCID2	ENST00000491548	?	?	1
PCID2	ENST00000375479	NM_018386.3, NM_001127203.2	ENSP00000364628	2
PCID2	ENST00000622406	NM_001258212.1	ENSP00000479494	2
PCID2	ENST00000475433	?	?	2
PCID2	ENST00000473462	?	?	1
PCID2	ENST00000375459	?	ENSP00000364608	2

PCID2	ENST00000375477	?	ENSP00000364626	2
PCID2	ENST00000484641	?	?	2
PCID2	ENST00000480971	?	?	2
PCID2	ENST00000246505	?	ENSP00000246505	2
PCID2	ENST00000375457	NM_001258213.1	ENSP00000364606	2