SNiPAcard

Block annotations

Block info

genomic range	chr20:24,788,418-24,897,155
block size	108,738 bp
variant count	60 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.308 [-4.928 – 0.939]	**gene(s) hit or close-by**	–
phastCons	μ = 0.066 [0 – 0.743]	**eQTL gene(s)**	ABHD12
GERP++	μ = -0.366 [-3.16 – 0.823]	**potentially regulated gene(s)**	ACSS1, CST7, PYGB
CADD score	μ = 3.159 [0.037 – 15.94]	**disease gene(s)**	ABHD12

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
ABHD12	POLYNEUROPATHY, HEARING LOSS, ATAXIA, RETINITIS PIGMENTOSA, AND [...]	OMIM	MIM:612674
ABHD12	Polyneuropathy - hearing loss - ataxia - retinitis pigmentosa - cataract	OrphaNet	OrphaNet:171848

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ABHD12	ENST00000465694	ILMN_1745116	monocyte	6.09×10-4 (p-value)	Fairfax et al.	1
ABHD12	ENST00000376542					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000291041	1	ENCP00000031107	CST7
ENCE00000291312	1	ENCP00000031129	PYGB
ENCE00000291043	1	ENCP00000031112	ACSS1
		ENCP00000031111	ACSS1
		ENCP00000031107	CST7

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000399317 (CTCF binding site)	1	cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K27me3
		HSMMtube	H3K27me3
		liver (HepG2)	H3K27me3
		lung (IMR90)	H3K27me3
		Osteobl	H3K27me3
ENSR00000399318 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, DNase1
		A549	H3K4me3, H3K27me3