SNiPAcard

Block annotations

Block info

genomic range	chr1:160,834,616-160,899,100
block size	64,485 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.163 [-0.178 – 0.435]	**gene(s) hit or close-by**	CD244, ITLN1, RP11-312J18.6, RP11-544M22.1
phastCons	μ = 0.032 [0 – 0.099]	**eQTL gene(s)**	–
GERP++	μ = 0.255 [-0.854 – 1.97]	**potentially regulated gene(s)**	ARHGAP30, FCER1G, ITLN1, MPZ, MPZ, NIT1, PPOX, RP11-312J18.3, RP11-404F10.2, SLAMF1, SLAMF7, TOMM40L, USF1, USP21
CADD score	μ = 3.439 [1.402 – 9.687]	**disease gene(s)**	USF1, MPZ, PPOX

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
USF1	HYPERLIPIDEMIA, FAMILIAL COMBINED	OMIM	MIM:144250
USF1	HYPERLIPIDEMIA, COMBINED, 1	OMIM	MIM:602491
MPZ	ADIE PUPIL	OMIM	MIM:103100
MPZ	ROUSSY-LEVY HEREDITARY AREFLEXIC DYSTASIA	OMIM	MIM:180800
MPZ	CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1B	OMIM	MIM:118200
MPZ	NEUROPATHY, CONGENITAL HYPOMYELINATING OR AMYELINATING, [...]	OMIM	MIM:605253
MPZ	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2I	OMIM	MIM:607677
MPZ	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2J	OMIM	MIM:607736
MPZ	CHARCOT-MARIE-TOOTH DISEASE, DOMINANT INTERMEDIATE D	OMIM	MIM:607791
MPZ	HYPERTROPHIC NEUROPATHY OF DEJERINE-SOTTAS	OMIM	MIM:145900
PPOX	PORPHYRIA VARIEGATA	OMIM	MIM:176200
MPZ	Autosomal dominant Charcot-Marie-Tooth disease type 2J	OrphaNet	OrphaNet:99943
MPZ	Autosomal dominant intermediate Charcot-Marie-Tooth disease type D	OrphaNet	OrphaNet:100046
MPZ	Autosomal dominant intermediate Charcot-Marie-Tooth disease with neuropathic pain	OrphaNet	OrphaNet:324585
MPZ	ROUSSY-LEVY SYNDROME	OrphaNet	OrphaNet:3115
MPZ	Charcot-Marie-Tooth disease type 1B	OrphaNet	OrphaNet:101082
MPZ	Dejerine-Sottas syndrome	OrphaNet	OrphaNet:64748
MPZ	Autosomal dominant Charcot-Marie-Tooth disease type 2I	OrphaNet	OrphaNet:99942
PPOX	PORPHYRIA VARIEGATA	OrphaNet	OrphaNet:79473

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000040974	1	ENCP00000005175	NIT1
		ENCP00000005210	MPZ MPZ
		ENCP00000005140	RP11-404F10.2
		ENCP00000005151	ITLN1
		ENCP00000005186	PPOX

		ENCP00000005149	RP11-312J18.3
		ENCP00000005138	SLAMF1
		ENCP00000005204	TOMM40L
		ENCP00000005169	ARHGAP30
		ENCP00000005163	USF1
		ENCP00000005143	SLAMF7
		ENCP00000005168	ARHGAP30
		ENCP00000005142	SLAMF7
		ENCP00000005201	FCER1G
		ENCP00000005185	PPOX
ENCE00000041210	1	ENCP00000005182	USP21

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001587177 (open chromatin region)	1	blood (K562)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CD244	upstream gene variant	2126	ENST00000492063	?	ENSP00000432636	1
CD244	upstream gene variant	2126	ENST00000322302	NM_001166664.1	ENSP00000313619	1
CD244	upstream gene variant	2066	ENST00000368033	NM_001166663.1	ENSP00000357012	1
CD244	upstream gene variant	1971	ENST00000368034	NM_016382.3	ENSP00000357013	1
ITLN1	downstream gene variant	693	ENST00000326245	NM_017625.2	ENSP00000323587	1
ITLN1	downstream gene variant	3466	ENST00000487531	?	?	1
RP11-312J18.6	downstream gene variant	2429	ENST00000427339	?	?	1
RP11-544M22.1	upstream gene variant	3155	ENST00000356006	?	?	1