SNiPAcard

Block annotations

Block info

genomic range	chr11:102,719,534-102,777,303
block size	57,770 bp
variant count	20 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.157 [-1.126 – 1.467]	**gene(s) hit or close-by**	BOLA3P1, MMP12, MMP3, RNU7-159P
phastCons	μ = 0.143 [0 – 0.967]	**eQTL gene(s)**	–
GERP++	μ = -0.305 [-6.11 – 3.83]	**potentially regulated gene(s)**	AP000619.6, DYNC2H1, MMP20, MMP3
CADD score	μ = 4.600 [0.023 – 17.09]	**disease gene(s)**	MMP3, DYNC2H1, MMP20

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Local histogram emphysema pattern	<1.00×10-8	GWAS Catalog	25006744	1

Disease gene annotation

gene	trait	source DB	source entry/link
MMP3	CORONARY HEART DISEASE, SUSCEPTIBILITY TO, 6	OMIM	MIM:614466
DYNC2H1	SHORT-RIB THORACIC DYSPLASIA 3 WITH OR WITHOUT POLYDACTYLY	OMIM	MIM:613091
MMP20	AMELOGENESIS IMPERFECTA, HYPOMATURATION TYPE, IIA2	OMIM	MIM:612529
DYNC2H1	short rib-polydactyly syndrome type 3 (SRPS3)	DECIPHER	MIM:263510
DYNC2H1	asphyxiating thoracic dystrophy type 3 (ATD3)	DECIPHER	MIM:613091
DYNC2H1	Short rib-polydactyly syndrome, Verma-Naumoff type	OrphaNet	OrphaNet:93271
DYNC2H1	Short rib-polydactyly syndrome, Majewski type	OrphaNet	OrphaNet:93269
DYNC2H1	Jeune syndrome	OrphaNet	OrphaNet:474
MMP20	Hypomaturation amelogenesis imperfecta	OrphaNet	OrphaNet:100033

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000120359	1	ENCP00000014401	MMP3
		ENCP00000014410	DYNC2H1
ENCE00000120215	1	ENCP00000014398	AP000619.6
		ENCP00000014385	MMP20

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000568035 (promoter flanking region)	5	NHLF	DNase1
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
		breast (HMEC)	DNase1
		cervix (HeLa-S3)	DNase1
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	DNase1

		nervous (NH-A)	DNase1
		A549	H3K27me3
		skin (NHEK)	DNase1, H3K27ac
ENSR00001606530 (CTCF binding site)	1	NHLF	DNase1
		skin (NHDF-AD)	DNase1
		breast (HMEC)	DNase1
		muscle (HSMM)	DNase1
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	DNase1
		A549	H3K27me3
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1
ENSR00001606534 (enhancer)	1	HSMMtube	H3K27me3
		A549	H3K27me3
		blood (DND-41)	H3K27me3
ENSR00000568041 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
BOLA3P1	upstream gene variant	2527	ENST00000530713	?	?	1
MMP12	downstream gene variant, upstream gene variant	27	ENST00000571244	NM_002426.4	ENSP00000458585	4
MMP3	upstream gene variant	5000	ENST00000299855	NM_002422.3	ENSP00000299855	1
RNU7-159P	downstream gene variant, upstream gene variant	161	ENST00000459284	?	?	3

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
MMP12	ENST00000571244	NM_002426.4	ENSP00000458585	P	ccG/ccA	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
MMP12	ENST00000571244	NM_002426.4	ENSP00000458585	2