SNiPAcard

Block annotations

Block info

genomic range	chr3:57,133,138-57,149,229
block size	16,092 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.135 [-1.696 – 0.831]	**gene(s) hit or close-by**	IL17RD
phastCons	μ = 0.112 [0 – 0.992]	**eQTL gene(s)**	ARHGEF3, FLNB
GERP++	μ = -0.423 [-4.73 – 2.58]	**potentially regulated gene(s)**	–
CADD score	μ = 6.125 [0.859 – 19.03]	**disease gene(s)**	FLNB, IL17RD

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
FLNB	SPONDYLOCARPOTARSAL SYNOSTOSIS SYNDROME (SCT;)	DECIPHER	MIM:272460
FLNB	atelosteogenesis type 1 (AO1)	DECIPHER	MIM:108720
FLNB	atelosteogenesis type 3 (AO3)	DECIPHER	MIM:108721
FLNB	boomerang dysplasia (BOOMD)	DECIPHER	MIM:112310
FLNB	autosomal dominant Larsen syndrome (LRS1)	DECIPHER	MIM:150250
FLNB	ATELOSTEOGENESIS, TYPE I	OMIM	MIM:108720
FLNB	SPONDYLOCARPOTARSAL SYNOSTOSIS SYNDROME	OMIM	MIM:272460
FLNB	LARSEN SYNDROME	OMIM	MIM:150250
FLNB	BOOMERANG DYSPLASIA	OMIM	MIM:112310
FLNB	ATELOSTEOGENESIS, TYPE III	OMIM	MIM:108721
IL17RD	HYPOGONADOTROPIC HYPOGONADISM 18 WITH OR WITHOUT ANOSMIA	OMIM	MIM:615267
FLNB	SPONDYLOCARPOTARSAL SYNOSTOSIS SYNDROME (SCT;)	OrphaNet	OrphaNet:3275
FLNB	ATELOSTEOGENESIS, TYPE II	OrphaNet	OrphaNet:1190
FLNB	BOOMERANG DYSPLASIA	OrphaNet	OrphaNet:1263
FLNB	ATELOSTEOGENESIS, TYPE III	OrphaNet	OrphaNet:56305
FLNB	Autosomal dominant Larsen syndrome	OrphaNet	OrphaNet:503
IL17RD	Kallmann Syndrome	OrphaNet	OrphaNet:478

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
IL17RD	missense variant	ENST00000463523	?	ENSP00000417516	T/M	aCg/aTg	tolerated	benign	1
IL17RD	missense variant	ENST00000320057	?	ENSP00000322250	T/M	aCg/aTg	tolerated	benign	1
IL17RD	missense variant	ENST00000296318	NM_017563.3	ENSP00000296318	T/M	aCg/aTg	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
FLNB	ENST00000484981	ILMN_1664922	monocyte	5.80×10-4 (p-value)	Fairfax et al.	1
FLNB	ENST00000295956					
FLNB	ENST00000429972					
FLNB	ENST00000481470					
FLNB	ENST00000358537					
FLNB	ENST00000419752					
ARHGEF3	?	ENSG00000163947	transformed fibroblasts	2.44×10-5 (p-value)	GTEx Portal V6	1

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
MYC, MAX	MA0059.1	6	yes	-0.063	1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001479531 (enhancer)	1	embryonic stem cell (H1ESC)	H3K36me3
		blood (K562)	H3K27me3
ENSR00001365042 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1, Rad21, CTCF
		HSMMtube	CTCF, DNase1
		blood (K562)	H3K27me3, Max, Rad21, CTCF
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		lung (IMR90)	DNase1, CTCF
		blood (GM12878)	Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	DNase1, Max, Cmyc, CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, DNase1, CTCF
		A549	CTCF
ENSR00001365044 (enhancer)	1	cervix (HeLa-S3)	DNase1
		endothelium (HUVEC)	H3K27me3
		embryonic stem cell (H1ESC)	H3K36me3, DNase1
		blood (K562)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
IL17RD	downstream gene variant	1561	ENST00000467210	?	ENSP00000418368	2
IL17RD	downstream gene variant	4774	ENST00000479825	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL17RD	ENST00000469841	?	?	8
IL17RD	ENST00000467210	?	ENSP00000418368	5

IL17RD	ENST00000320057	?	ENSP00000322250	8
IL17RD	ENST00000296318	NM_017563.3	ENSP00000296318	8
IL17RD	ENST00000463523	?	ENSP00000417516	8
IL17RD	ENST00000479825	?	?	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
IL17RD	ENST00000469841	?	1