Locus 182 (rs17220241)

SNiPAcard

Block annotations

Block info

genomic range	chr6:32,809,582-32,861,183
block size	51,602 bp
variant count	14 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.257 [-2.534 – 0.965]	gene(s) hit or close-by	HLA-Z, PSMB8, PSMB8-AS1, PSMB9, TAP1, TAP2, XXbac-BPG246D15.9
phastCons	μ = 0.032 [0 – 0.164]	eQTL gene(s)	–
GERP++	μ = -0.736 [-4.68 – 3.04]	potentially regulated gene(s)	COL11A2, COL11A2, COL11A2, COL11A2, COL11A2, COL11A2, COL11A2, HLA-DOB, HLA-DOB, HLA-DOB, HLA-DOB, HLA-DOB, HLA-DOB, HLA-DPB1, HLA-DPB1, HLA-DPB1, HLA-DPB1, HLA-DPB1, HLA-DPB1, HLA-DPB1, HLA-DPB1, HLA-Z, HLA-Z, HLA-Z, HLA-Z, HLA-Z, HLA-Z, HLA-Z, HLA-Z, HLA-Z
CADD score	μ = 4.633 [0.832 – 13.03]	disease gene(s)	TAP2, TAP1, PSMB8, , , , , , COL11A2, , , , HLA-DPB1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
TAP2	BARE LYMPHOCYTE SYNDROME, TYPE I	OMIM	MIM:604571
TAP1	BARE LYMPHOCYTE SYNDROME, TYPE I	OMIM	MIM:604571
PSMB8	AUTOINFLAMMATION, LIPODYSTROPHY, AND DERMATOSIS SYNDROME	OMIM	MIM:256040
	WEISSENBACHER-ZWEYMULLER SYNDROME	OMIM	MIM:277610
	OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA	OMIM	MIM:215150
	DEAFNESS, AUTOSOMAL RECESSIVE 53	OMIM	MIM:609706
	FIBROCHONDROGENESIS 2	OMIM	MIM:614524
	DEAFNESS, AUTOSOMAL DOMINANT 13	OMIM	MIM:601868
	STICKLER SYNDROME, TYPE III	OMIM	MIM:184840
	WEISSENBACHER-ZWEYMULLER SYNDROME	OMIM	MIM:277610
	OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA	OMIM	MIM:215150
	DEAFNESS, AUTOSOMAL RECESSIVE 53	OMIM	MIM:609706
	FIBROCHONDROGENESIS 2	OMIM	MIM:614524
	DEAFNESS, AUTOSOMAL DOMINANT 13	OMIM	MIM:601868
	STICKLER SYNDROME, TYPE III	OMIM	MIM:184840
	WEISSENBACHER-ZWEYMULLER SYNDROME	OMIM	MIM:277610
	OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA	OMIM	MIM:215150
	DEAFNESS, AUTOSOMAL RECESSIVE 53	OMIM	MIM:609706
	FIBROCHONDROGENESIS 2	OMIM	MIM:614524
	DEAFNESS, AUTOSOMAL DOMINANT 13	OMIM	MIM:601868
	STICKLER SYNDROME, TYPE III	OMIM	MIM:184840
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	OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA	OMIM	MIM:215150
	DEAFNESS, AUTOSOMAL RECESSIVE 53	OMIM	MIM:609706
	FIBROCHONDROGENESIS 2	OMIM	MIM:614524
	DEAFNESS, AUTOSOMAL DOMINANT 13	OMIM	MIM:601868
	STICKLER SYNDROME, TYPE III	OMIM	MIM:184840
COL11A2	WEISSENBACHER-ZWEYMULLER SYNDROME	OMIM	MIM:277610
COL11A2	OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA	OMIM	MIM:215150

COL11A2	DEAFNESS, AUTOSOMAL RECESSIVE 53	OMIM	MIM:609706	OMIM
COL11A2	FIBROCHONDROGENESIS 2	OMIM	MIM:614524	OMIM
COL11A2	DEAFNESS, AUTOSOMAL DOMINANT 13	OMIM	MIM:601868	OMIM
COL11A2	STICKLER SYNDROME, TYPE III	OMIM	MIM:184840	OMIM
PSMB8	Nakajo syndrome	DECIPHER	MIM:256040	OMIM
	Weissenbacher-Zweymueller syndrome (WZS)	DECIPHER	MIM:277610	OMIM
	Stickler syndrome type 3 (STL3)	DECIPHER	MIM:184840	OMIM
	deafness autosomal dominant type 13 (DFNA13)	DECIPHER	MIM:601868	OMIM
	autosomal recessive otospondylomegaepiphyseal dysplasia (OSMED)	DECIPHER	MIM:215150	OMIM
	deafness autosomal recessive type 53 (DFNB53)	DECIPHER	MIM:609706	OMIM
	Weissenbacher-Zweymueller syndrome (WZS)	DECIPHER	MIM:277610	OMIM
	Stickler syndrome type 3 (STL3)	DECIPHER	MIM:184840	OMIM
	deafness autosomal dominant type 13 (DFNA13)	DECIPHER	MIM:601868	OMIM
	autosomal recessive otospondylomegaepiphyseal dysplasia (OSMED)	DECIPHER	MIM:215150	OMIM
	deafness autosomal recessive type 53 (DFNB53)	DECIPHER	MIM:609706	OMIM
	Weissenbacher-Zweymueller syndrome (WZS)	DECIPHER	MIM:277610	OMIM
	Stickler syndrome type 3 (STL3)	DECIPHER	MIM:184840	OMIM
	deafness autosomal dominant type 13 (DFNA13)	DECIPHER	MIM:601868	OMIM
	autosomal recessive otospondylomegaepiphyseal dysplasia (OSMED)	DECIPHER	MIM:215150	OMIM
	deafness autosomal recessive type 53 (DFNB53)	DECIPHER	MIM:609706	OMIM
	Weissenbacher-Zweymueller syndrome (WZS)	DECIPHER	MIM:277610	OMIM
	Stickler syndrome type 3 (STL3)	DECIPHER	MIM:184840	OMIM
	deafness autosomal dominant type 13 (DFNA13)	DECIPHER	MIM:601868	OMIM
	autosomal recessive otospondylomegaepiphyseal dysplasia (OSMED)	DECIPHER	MIM:215150	OMIM
	deafness autosomal recessive type 53 (DFNB53)	DECIPHER	MIM:609706	OMIM
	Weissenbacher-Zweymueller syndrome (WZS)	DECIPHER	MIM:277610	OMIM
	Stickler syndrome type 3 (STL3)	DECIPHER	MIM:184840	OMIM
	deafness autosomal dominant type 13 (DFNA13)	DECIPHER	MIM:601868	OMIM
	autosomal recessive otospondylomegaepiphyseal dysplasia (OSMED)	DECIPHER	MIM:215150	OMIM
	deafness autosomal recessive type 53 (DFNB53)	DECIPHER	MIM:609706	OMIM
	Weissenbacher-Zweymueller syndrome (WZS)	DECIPHER	MIM:277610	OMIM
	Stickler syndrome type 3 (STL3)	DECIPHER	MIM:184840	OMIM
	deafness autosomal dominant type 13 (DFNA13)	DECIPHER	MIM:601868	OMIM
	autosomal recessive otospondylomegaepiphyseal dysplasia (OSMED)	DECIPHER	MIM:215150	OMIM
	deafness autosomal recessive type 53 (DFNB53)	DECIPHER	MIM:609706	OMIM
COL11A2	Weissenbacher-Zweymueller syndrome (WZS)	DECIPHER	MIM:277610	OMIM
COL11A2	Stickler syndrome type 3 (STL3)	DECIPHER	MIM:184840	OMIM
COL11A2	deafness autosomal dominant type 13 (DFNA13)	DECIPHER	MIM:601868	OMIM
COL11A2	autosomal recessive otospondylomegaepiphyseal dysplasia (OSMED)	DECIPHER	MIM:215150	OMIM
COL11A2	deafness autosomal recessive type 53 (DFNB53)	DECIPHER	MIM:609706	OMIM
TAP2	Immunodeficiency by defective expression of HLA class 1	OrphaNet	OrphaNet:34592	orphanet
TAP1	Immunodeficiency by defective expression of HLA class 1	OrphaNet	OrphaNet:34592	orphanet
PSMB8	CANDLE syndrome	OrphaNet	OrphaNet:325004	orphanet
PSMB8	JMP syndrome	OrphaNet	OrphaNet:324999	orphanet
PSMB8	Nakajo-Nishimura syndrome	OrphaNet	OrphaNet:2615	orphanet
HLA-DPB1	Chronic berylliosis	OrphaNet	OrphaNet:133	orphanet
HLA-DPB1	Granulomatosis with polyangiitis	OrphaNet	OrphaNet:900	orphanet
COL11A2	Autosomal recessive nonsyndromic sensorineural deafness type DFNB	OrphaNet	OrphaNet:90636	orphanet
COL11A2	WEISSENBACHER-ZWEYMULLER SYNDROME	OrphaNet	OrphaNet:3450	orphanet
COL11A2	Autosomal dominant nonsyndromic sensorineural deafness type DFNA	OrphaNet	OrphaNet:90635	orphanet
COL11A2	OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA	OrphaNet	OrphaNet:1427	orphanet
COL11A2	Stickler syndrome, type 3	OrphaNet	OrphaNet:166100	orphanet
COL11A2	Fibrochondrogenesis	OrphaNet	OrphaNet:2021	orphanet

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000445615	1	ENCP00000048222	HLA-DOB HLA-DOB HLA-DOB HLA-DOB HLA-DOB HLA-DOB
ENCE00000445701	1	ENCP00000048255	HLA-DPB1 HLA-DPB1 HLA-DPB1 HLA-DPB1 HLA-DPB1 HLA-DPB1 HLA-DPB1 HLA-DPB1
		ENCP00000048262	COL11A2 COL11A2 COL11A2 COL11A2 COL11A2 COL11A2 COL11A2
		ENCP00000048235	HLA-Z HLA-Z HLA-Z HLA-Z HLA-Z HLA-Z HLA-Z HLA-Z

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000025365 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K27me3, PolII, Rad21, Nrsf, CTCF, TAF1, H3K4me2, H3K4me3
		HSMMtube	DNase1, H3K27me3, H2AZ, H3K4me2
		blood (K562)	PolII, Max, H3K4me1, Rad21, H3K9ac, Cmyc, H2AZ, Nrsf, CTCFL, H3K4me2, CTCF, H3K27me3, DNase1, H3K4me3
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H3K4me3, H3K4me2, DNase1, CTCF, H2AZ
		liver (HepG2)	H3K4me3, H3K27me3, DNase1, Nrsf, PolII, Rad21, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac, CTCF
		blood (GM12878)	Nrsf, PolII, H2AZ, DNase1, Yy1, Rad21, H3K79me2, H3K4me3, H3K27ac, H3K4me2, CTCF, H3K9ac, H3K36me3
		lung (IMR90)	CTCF, H3K9ac, DNase1, H3K27ac, H3K4me2, H3K4me3
		nervous (NH-A)	H3K9ac, CTCF, H3K4me2, H3K4me3, H3K27me3, DNase1
		skin (NHEK)	H3K4me2, H3K4me3, CTCF, H3K36me3, DNase1, H3K9ac, H3K27ac
		NHLF	DNase1, H3K4me3, CTCF, H3K4me2, H3K9ac, H3K27ac, H3K36me3
		Osteobl	H3K36me3, CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, CTCF, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	H3K27ac, H3K4me3, H3K9ac, H3K4me2, H3K36me3, CTCF
		cervix (HeLa-S3)	H3K9ac, Nrsf, H3K4me2, H3K27ac, TAF1, H3K4me3, PolII, CTCF, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3, H3K4me3, H3K9ac, DNase1, H3K4me1, CTCF, H3K4me2, H3K27ac
		endothelium (HUVEC)	H3K36me3, Max, H3K4me3, H3K4me2, PolII, CTCF, H3K27me3, DNase1
		A549	DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00000025366 (promoter flanking region)	2	embryonic stem cell (H1ESC)	PolII, Rad21, p300, CTCF, Tcf12, TAF1, SP1, DNase1
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	H3K4me2, CTCF
		blood (GM12878)	H3K36me3
		lung (IMR90)	CTCF
		nervous (NH-A)	CTCF

		skin (NHEK)	CTCF
		Osteobl	H3K36me3, CTCF
		blood (DND-41)	H3K36me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		A549	CTCF, H3K4me3
ENSR00001703138 (CTCF binding site)	1	endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		nervous (NH-A)	H3K27me3
		A549	H3K27me3
ENSR00001494626 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	H3K27me3, CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		blood (K562)	H3K27me3, Rad21, CTCF
		skin (NHDF-AD)	CTCF
		breast (HMEC)	CTCF
		muscle (HSMM)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF, H3K27me3
		liver (HepG2)	CTCF, Rad21, H3K27me3
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	CTCF
		A549	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
HLA-Z	upstream gene variant	3010	ENST00000434920	?	?	1
PSMB8-AS1	upstream gene variant, downstream gene variant	2333	ENST00000415067	?	?	2
PSMB8-AS1	upstream gene variant, downstream gene variant	2328	ENST00000413039	?	?	2
PSMB8-AS1	upstream gene variant, downstream gene variant	2303	ENST00000458296	?	?	2
PSMB8-AS1	upstream gene variant, downstream gene variant	2328	ENST00000429600	?	?	2
PSMB8-AS1	upstream gene variant, downstream gene variant	2281	ENST00000453426	?	?	3
PSMB8-AS1	upstream gene variant, downstream gene variant	2329	ENST00000412095	?	?	2
PSMB9	upstream gene variant, downstream gene variant	54	ENST00000467593	?	?	5
PSMB9	upstream gene variant, downstream gene variant	1006	ENST00000395330	?	ENSP00000378739	3
PSMB9	upstream gene variant, downstream gene variant	1691	ENST00000464863	?	?	4
PSMB9	upstream gene variant, downstream gene variant	1006	ENST00000374859	NM_002800.4	ENSP00000363993	4
PSMB9	upstream gene variant, downstream gene variant	339	ENST00000414474	?	ENSP00000394363	4
TAP1	downstream gene variant, upstream gene variant	489	ENST00000354258	NM_000593.5	ENSP00000346206	4
TAP1	downstream gene variant, upstream gene variant	2446	ENST00000487296	?	?	3
TAP1	downstream gene variant, upstream gene variant	534	ENST00000486332	?	?	4
TAP2	upstream gene variant	3035	ENST00000620123	NM_000544.3	ENSP00000481712	1
TAP2	upstream gene variant	3078	ENST00000374899	NM_018833.2	ENSP00000364034	1
TAP2	upstream gene variant	3025	ENST00000374897	NM_001290043.1	ENSP00000364032	1

XXbac-BPG246D15.9	upstream gene variant	2983	ENST00000452392	?	ENSP00000391806 1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PSMB8	ENST00000374881	NM_004159.4	ENSP00000364015	1
PSMB8	ENST00000484003	?	?	1
PSMB8	ENST00000395339	?	ENSP00000378748	1
PSMB8	ENST00000374882	NM_148919.3	ENSP00000364016	1
PSMB9	ENST00000414474	?	ENSP00000394363	4
PSMB9	ENST00000395330	?	ENSP00000378739	5
PSMB9	ENST00000374859	NM_002800.4	ENSP00000363993	3
PSMB9	ENST00000464863	?	?	3
PSMB9	ENST00000467593	?	?	1
TAP1	ENST00000354258	NM_000593.5	ENSP00000346206	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
PSMB8	ENST00000490613	?	1
PSMB9	ENST00000464863	?	2