SNiPAcard

Block annotations

Block info

genomic range	chr6:29,613,377-29,671,356
block size	57,980 bp
variant count	27 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.431 [-3.465 – 1.743]	**gene(s) hit or close-by**	MOG, ZDHHC20P1, ZFP57
phastCons	μ = 0.044 [0 – 0.4]	**eQTL gene(s)**	–
GERP++	μ = 0.146 [-1.75 – 3.54]	**potentially regulated gene(s)**	GABBR1, GABBR1, GABBR1, GABBR1, GABBR1, GABBR1, GABBR1, HCG4P5, HCG4P5, HCG4P5, HCG4P5, HCG4P5, HCG4P5, HCG4P5, HLA-A, HLA-A, HLA-A, HLA-A, HLA-A, HLA-A, HLA-A, HLA-A, HLA-F, HLA-F, HLA-F, HLA-F, HLA-F, HLA-F, HLA-F, HLA-H, HLA-H, HLA-H, HLA-H, HLA-H, HLA-H, HLA-H, HLA-H, MOG, MOG, MOG, MOG, MOG, MOG, MOG, MOG, PPP1R11, PPP1R11, PPP1R11, PPP1R11, PPP1R11, PPP1R11, PPP1R11, PPP1R11, ZNRD1-AS1
CADD score	μ = 2.954 [0.116 – 10.4]	**disease gene(s)**	HLA-A, MOG, ZFP57, , , , , , , ,

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link	
HLA-A	Birdshot chorioretinopathy	OrphaNet	OrphaNet:179	orphanet
MOG	Narcolepsy-cataplexy	OrphaNet	OrphaNet:2073	orphanet
ZFP57	Transient neonatal diabetes mellitus	OrphaNet	OrphaNet:99886	orphanet
MOG	NARCOLEPSY 7	OMIM	MIM:614250	OMIM
	NARCOLEPSY 7	OMIM	MIM:614250	OMIM
	NARCOLEPSY 7	OMIM	MIM:614250	OMIM
	NARCOLEPSY 7	OMIM	MIM:614250	OMIM
ZFP57	DIABETES MELLITUS, TRANSIENT NEONATAL, 1	OMIM	MIM:601410	OMIM
	NARCOLEPSY 7	OMIM	MIM:614250	OMIM
	NARCOLEPSY 7	OMIM	MIM:614250	OMIM
	NARCOLEPSY 7	OMIM	MIM:614250	OMIM
	NARCOLEPSY 7	OMIM	MIM:614250	OMIM
ZFP57	Diabetes Mellitus, 6q24-Related Transient Neonatal	DECIPHER	MIM:601410	OMIM

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000735239	1	ENST00000376898, ENST00000494692, ENST00000376917, ENST00000490427, ENST00000376888, ENST00000396704, ENST00000416766, ENST00000431798, ENST00000483013, ENST00000376894, ENST00000396701	MOG

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000047827		MOG
		MOG

			MOG
			MOG
			MOG
			MOG
			MOG

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444016	1	ENCP00000047860	HLA-A HLA-A HLA-A HLA-A HLA-A HLA-A HLA-A HLA-A
		ENCP00000047832	HLA-F HLA-F HLA-F HLA-F HLA-F HLA-F HLA-F
		ENCP00000047875	PPP1R11 PPP1R11 PPP1R11 PPP1R11 PPP1R11 PPP1R11 PPP1R11 PPP1R11
		ENCP00000047871	ZNRD1-AS1
		ENCP00000047861	HCG4P5 HCG4P5 HCG4P5 HCG4P5 HCG4P5 HCG4P5 HCG4P5 HCG4P5
		ENCP00000047851	HLA-H HLA-H HLA-H HLA-H HLA-H HLA-H HLA-H HLA-H
		ENCP00000047876	PPP1R11 PPP1R11 PPP1R11 PPP1R11 PPP1R11 PPP1R11 PPP1R11 PPP1R11
		ENCP00000047824	GABBR1 GABBR1 GABBR1 GABBR1 GABBR1 GABBR1 GABBR1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000487538	2	embryonic stem cell (H1ESC)	PolII, Rad21, CTCF, Yy1, H3K4me2, H3K4me3, DNase1

(CTCF binding site)		HSMMtube	H2AZ, H3K4me2, CTCF, DNase1
		blood (K562)	PolII, H3K4me2, CTCF, DNase1, H3K4me3, H2AZ, THAP1, H3K27me3, Yy1
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, CTCF, H3K4me3, H3K4me2, DNase1
		liver (HepG2)	H3K4me3, ELF1, H2AZ, H3K4me2, CTCF, DNase1
		blood (GM12878)	H3K4me3, H3K27ac, H3K4me2, CTCF, PolII, H2AZ, DNase1, Yy1, ELF1
		lung (IMR90)	DNase1, H3K4me2, H3K4me3, CTCF
		nervous (NH-A)	H3K9ac, CTCF, DNase1, H3K4me2
		skin (NHEK)	H3K4me2, CTCF, DNase1
		NHLF	DNase1, CTCF
		Osteobl	CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	CTCF, H3K27ac, H3K4me3
		breast (HMEC)	CTCF, H3K4me2, H3K4me3, DNase1
		cervix (HeLa-S3)	H3K4me2, PolII, CTCF, DNase1
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, DNase1, CTCF, H3K4me2, PolII
		A549	DNase1, CTCF, H3K4me2, H3K4me3
ENSR00001494451 e! (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H3K4me1, H3K27me3, PolII, Rad21, CTCF, Yy1, H3K4me2, H3K4me3
		HSMMtube	DNase1, H2AZ, H3K4me2, CTCF
		blood (K562)	Yy1, THAP1, H2AZ, PolII, H3K4me2, CTCF, DNase1, H3K4me3
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, CTCF, H3K4me3, H3K4me2, DNase1
		liver (HepG2)	H3K4me3, ELF1, H2AZ, H3K4me2, CTCF, DNase1
		lung (IMR90)	DNase1, H3K4me2, H3K4me3, CTCF
		blood (GM12878)	PolII, H2AZ, DNase1, Yy1, ELF1, H3K4me3, H3K27ac, H3K4me2, CTCF
		nervous (NH-A)	DNase1, H3K4me2, CTCF, H3K9ac
		skin (NHEK)	CTCF, DNase1, H3K4me2
		NHLF	DNase1, CTCF
		Osteobl	CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	CTCF, H3K27ac, H3K4me3
		breast (HMEC)	DNase1, H3K4me3, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, H3K4me2, PolII, CTCF
		monocytes (Monocytes-CD14+)	H3K27ac, H3K9ac, H3K4me3, H3K4me2, DNase1, CTCF
		endothelium (HUVEC)	H3K36me3, H3K4me2, PolII, CTCF, DNase1
		A549	DNase1, CTCF, H3K4me2, H3K4me3
ENSR00000487541 e! (enhancer)	1	monocytes (Monocytes-CD14+)	H3K27me3
		embryonic stem cell (H1ESC)	H4K5ac, H3K27ac, DNase1
		endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
ENSR00000487543 e! (TF binding site)	5	endothelium (HUVEC)	H3K27me3
		embryonic stem cell (H1ESC)	DNase1, H3K27ac, CTCF, PolII
		blood (K562)	H3K27me3
		A549	H3K27me3
ENSR00000487548 e! (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3, Rad21, CTCF
		blood (DND-41)	H3K27me3
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF, H3K27me3
		skin (NHEK)	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MOG e!	upstream gene variant, downstream gene variant	1672	ENST00000416766 e!	?	ENSP00000409394 e!	4
MOG e!	upstream gene variant, downstream gene variant	1672	ENST00000376898 e!	NM_002433.4	ENSP00000366095 e!	4
MOG e!	upstream gene variant, downstream gene variant	1672	ENST00000490427 e!	NM_001170418.1	ENSP00000420350 e!	4
MOG e!	upstream gene variant, downstream gene variant	1266	ENST00000485885 e!	?	?	12
MOG e!	upstream gene variant, downstream gene variant	1672	ENST00000396704 e!	NM_001008228.2	ENSP00000379932 e!	4

MOG	upstream gene variant, downstream gene variant	1443	ENST00000376917	NM_206809.3	ENSP00000366115	4
MOG	upstream gene variant, downstream gene variant	259	ENST00000376889	?	ENSP00000366086	6
MOG	upstream gene variant, downstream gene variant	1554	ENST00000376894	?	ENSP00000366091	4
MOG	upstream gene variant, downstream gene variant	259	ENST00000485211	?	ENSP00000418872	6
MOG	upstream gene variant, downstream gene variant	1338	ENST00000476244	?	?	10
MOG	upstream gene variant, downstream gene variant	1672	ENST00000483013	?	ENSP00000418090	4
MOG	upstream gene variant, downstream gene variant	1592	ENST00000469353	?	?	2
MOG	upstream gene variant, downstream gene variant	921	ENST00000376903	?	ENSP00000366101	9
MOG	upstream gene variant, downstream gene variant	1465	ENST00000376888	NM_206814.5	ENSP00000366085	4
MOG	upstream gene variant, downstream gene variant	1672	ENST00000376891	NM_001008229.2	ENSP00000366088	4
MOG	upstream gene variant, downstream gene variant	1672	ENST00000431798	NM_206812.3	ENSP00000410866	4
MOG	upstream gene variant, downstream gene variant	598	ENST00000469603	?	?	10
MOG	upstream gene variant, downstream gene variant	1672	ENST00000396701	NM_206810.3	ENSP00000379929	4
MOG	upstream gene variant, downstream gene variant	1672	ENST00000494692	NM_206811.3	ENSP00000417405	4
ZDHHC20P1	downstream gene variant	4546	ENST00000435787	?	?	2
ZFP57	downstream gene variant, upstream gene variant	3541	ENST00000488757	NM_001109809.2	ENSP00000418259	8
ZFP57	downstream gene variant, upstream gene variant	3649	ENST00000376881	?	ENSP00000366078	4
ZFP57	downstream gene variant, upstream gene variant	288	ENST00000376883	?	ENSP00000366080	5

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
MOG	ENST00000396701	NM_206810.3	ENSP00000379929	12
MOG	ENST00000490427	NM_001170418.1	ENSP00000420350	12
MOG	ENST00000376888	NM_206814.5	ENSP00000366085	12
MOG	ENST00000396704	NM_001008228.2	ENSP00000379932	12
MOG	ENST00000494692	NM_206811.3	ENSP00000417405	12
MOG	ENST00000483013	?	ENSP00000418090	12
MOG	ENST00000431798	NM_206812.3	ENSP00000410866	12
MOG	ENST00000376903	?	ENSP00000366101	4
MOG	ENST00000485211	?	ENSP00000418872	10
MOG	ENST00000376891	NM_001008229.2	ENSP00000366088	12
MOG	ENST00000469353	?	?	2
MOG	ENST00000469603	?	?	3
MOG	ENST00000416766	?	ENSP00000409394	12
MOG	ENST00000376898	NM_002433.4	ENSP00000366095	12
MOG	ENST00000376894	?	ENSP00000366091	12
MOG	ENST00000476244	?	?	3
MOG	ENST00000376889	?	ENSP00000366086	10
MOG	ENST00000376917	NM_206809.3	ENSP00000366115	12
ZFP57	ENST00000376883	?	ENSP00000366080	3

ZFP57	ENST00000488757	NM_001109809.2	ENSP00000418259	3
ZFP57	ENST00000376881	?	ENSP00000366078	3