SNiPAcard

Block annotations

Block info

genomic range	chr19:49,504,260-49,524,220
block size	19,961 bp
variant count	12 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.621 [-5.773 – 4.333]	**gene(s) hit or close-by**	CGB, CTB-60B18.10, CTB-60B18.6, LHB, MIR6798, NTF6A, RUVBL2
phastCons	μ = 0.089 [0 – 1]	**eQTL gene(s)**	CGB, CTB-60B18.10, RUVBL2
GERP++	μ = -1.376 [-8.36 – 3.27]	**potentially regulated gene(s)**	NUCB1
CADD score	μ = 4.534 [0.004 – 14.25]	**disease gene(s)**	LHB

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
LHB	LUTEINIZING HORMONE, BETA POLYPEPTIDE	OMIM	MIM:152780
LHB	Leydig cell hypoplasia due to LHB deficiency	OrphaNet	OrphaNet:325448

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CGB	?	ENSG00000104827	pancreas	3.53×10^{-11} (p-value)	GTEx Portal V6	12
CTB-60B18.10	?	ENSG00000268655	testis	3.15×10^{-8} (p-value)	GTEx Portal V6	12
RUVBL2	?	ENSG00000183207	tibial artery	7.13×10^{-7} (p-value)	GTEx Portal V6	9

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000231691	1	ENCP00000024577	NUCB1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000217422 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K27me3, DNase1
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K27me3
		lung (IMR90)	H3K27me3
		skin (NHEK)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CGB	downstream gene variant	1906	ENST00000357383	NM_000737.3	ENSP00000349954	1
CTB-60B18.10	downstream gene variant, upstream gene variant	928	ENST00000600007	?	?	6
CTB-60B18.6	downstream gene variant	2183	ENST00000604577	?	ENSP00000474022	1
CTB-60B18.6	downstream gene variant	1990	ENST00000591656	?	ENSP00000466140	1
LHB	downstream gene variant, upstream gene variant	46	ENST00000391869	?	ENSP00000375742	7
LHB	downstream gene variant, upstream gene variant	7	ENST00000221421	NM_000894.2	ENSP00000221421	7
MIR6798	upstream gene variant, downstream gene variant	787	ENST00000612887	?	?	5
NTF6A	downstream gene variant	4498	ENST00000591175	?	?	1
RUVBL2	downstream gene variant	419	ENST00000594017	?	?	5
RUVBL2	upstream gene variant, downstream gene variant	24	ENST00000595002	?	?	9
RUVBL2	upstream gene variant, downstream gene variant	323	ENST00000627972	?	ENSP00000486242	5
RUVBL2	downstream gene variant	877	ENST00000593570	?	ENSP00000469488	4
RUVBL2	downstream gene variant	79	ENST00000594338	?	?	4
RUVBL2	downstream gene variant	81	ENST00000601968	?	ENSP00000471524	4
RUVBL2	downstream gene variant	1441	ENST00000596837	?	ENSP00000469189	4
RUVBL2	downstream gene variant	603	ENST00000595090	NM_006666.1	ENSP00000473172	4
RUVBL2	downstream gene variant	44	ENST00000596247	?	ENSP00000471538	4
RUVBL2	downstream gene variant	4427	ENST00000595811	?	ENSP00000469760	1
RUVBL2	downstream gene variant	47	ENST00000221413	?	ENSP00000221413	4
RUVBL2	downstream gene variant	1639	ENST00000598768	?	?	1

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
LHB	ENST00000391869	?	ENSP00000375742	P	ccA/ccC	1
LHB	ENST00000221421	NM_000894.2	ENSP00000221421	P	ccA/ccC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LHB	ENST00000391869	?	ENSP00000375742	1
LHB	ENST00000221421	NM_000894.2	ENSP00000221421	1
RUVBL2	ENST00000594017	?	?	2
RUVBL2	ENST00000594338	?	?	7
RUVBL2	ENST00000596837	?	ENSP00000469189	1
RUVBL2	ENST00000596247	?	ENSP00000471538	7
RUVBL2	ENST00000601968	?	ENSP00000471524	7
RUVBL2	ENST00000595811	?	ENSP00000469760	1
RUVBL2	ENST00000221413	?	ENSP00000221413	7
RUVBL2	ENST00000627972	?	ENSP00000486242	6
RUVBL2	ENST00000595090	NM_006666.1	ENSP00000473172	7

RUVBL2	ENST00000593570	?	ENSP00000469488	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
RUVBL2	ENST00000595090	NM_006666.1	ENSP00000473172	1