SNiPAcard

Block annotations

Block info

genomic range	chr11:125,882,173-125,901,248
block size	19,076 bp
variant count	43 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.312 [-4.048 – 2.849]	**gene(s) hit or close-by**	CDON
phastCons	μ = 0.064 [0 – 1]	**eQTL gene(s)**	–
GERP++	μ = -0.344 [-8.68 – 4.03]	**potentially regulated gene(s)**	–
CADD score	μ = 4.002 [0.107 – 22.5]	**disease gene(s)**	CDON

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CDON	HOLOPROSENCEPHALY 11	OMIM	MIM:614226
CDON	Holoprosencephaly 11	DECIPHER	MIM:614226
CDON	Semilobar holoprosencephaly	OrphaNet	OrphaNet:220386
CDON	Alobar holoprosencephaly	OrphaNet	OrphaNet:93925
CDON	Microform holoprosencephaly	OrphaNet	OrphaNet:280200
CDON	Midline interhemispheric variant of holoprosencephaly	OrphaNet	OrphaNet:93926
CDON	Septopreoptic holoprosencephaly	OrphaNet	OrphaNet:280195
CDON	Lobar holoprosencephaly	OrphaNet	OrphaNet:93924

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)
CDON	missense variant	ENST00000392693	NM_001243597.1	ENSP00000376458	K/E	Aaa/Gaa	?	?	1
CDON	missense variant	ENST00000534661	?	ENSP00000436755	K/E	Aaa/Gaa	?	?	1
CDON	missense variant	ENST00000263577	NM_016952.4	ENSP00000263577	K/E	Aaa/Gaa	?	?	1
CDON	missense variant	ENST00000531586	?	ENSP00000434212	K/E	Aaa/Gaa	?	?	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001608006 (enhancer)	3	liver (HepG2)	H3K4me1
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CDON	downstream gene variant, upstream gene variant	339	ENST00000525625	?	?	18

CDON	upstream gene variant	244	ENST00000531830	?	ENSP00000432571	11
CDON	downstream gene variant	56	ENST00000527967	?	ENSP00000436940	17
CDON	downstream gene variant	1168	ENST00000534818	?	ENSP00000437176	16
CDON	downstream gene variant	45	ENST00000531586	?	ENSP00000434212	16
CDON	upstream gene variant	289	ENST00000534661	?	ENSP00000436755	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CDON	ENST00000531586	?	ENSP00000434212	16
CDON	ENST00000525625	?	?	22
CDON	ENST00000531830	?	ENSP00000432571	22
CDON	ENST00000534661	?	ENSP00000436755	31
CDON	ENST00000263577	NM_016952.4	ENSP00000263577	42
CDON	ENST00000527967	?	ENSP00000436940	16
CDON	ENST00000392693	NM_001243597.1	ENSP00000376458	42
CDON	ENST00000534818	?	ENSP00000437176	11

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CDON	ENST00000525625	?	4