SNiPAcard

Block annotations

Block info

genomic range	chr1:196,887,457-197,127,578
block size	240,122 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.197 [-1.519 – 0.704]	**gene(s) hit or close-by**	ASPM, CFHR2, CFHR4, ZBTB41
phastCons	μ = 0.001 [0 – 0.003]	**eQTL gene(s)**	–
GERP++	μ = 0.497 [-4.16 – 3.37]	**potentially regulated gene(s)**	–
CADD score	μ = 5.015 [0.698 – 8.621]	**disease gene(s)**	ASPM

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link	
ASPM	MICROCEPHALY 5, PRIMARY, AUTOSOMAL RECESSIVE	OMIM	MIM:608716	OMIM
ASPM	Primary Autosomal Recessive Microcephaly	DECIPHER	MIM:608716	OMIM
ASPM	Autosomal recessive primary microcephaly	OrphaNet	OrphaNet:2512	orphanet

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)
CFHR4	missense variant	ENST00000251424	NM_006684.4	ENSP00000251424	E/G	gAa/gGa	?	?	1
CFHR4	missense variant	ENST00000367416	NM_001201551.1, NM_001201550.2	ENSP00000356386	E/G	gAa/gGa	?	?	1
CFHR4	missense variant	ENST00000367418	?	ENSP00000356388	E/G	gAa/gGa	?	?	1
CFHR4	missense variant	ENST00000608469	?	ENSP00000477162	E/G	gAa/gGa	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ASPM	ENST00000612785	?	ENSP00000479244	1
ASPM	ENST00000367409	NM_018136.4	ENSP00000356379	1
ASPM	ENST00000294732	NM_001206846.1	ENSP00000294732	1
ASPM	ENST00000367408	?	ENSP00000356378	1
CFHR2	ENST00000367421	?	ENSP00000356391	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ZBTB41	ENST00000367405	NM_194314.2	ENSP00000356375	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ZBTB41	ENST00000467322	?	1