Locus 352 (rs17412738)

SNiPAcard

Block annotations

Block info

genomic range	chr3:132,185,652-132,410,214
block size	224,563 bp
variant count	68 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.141 [-4.075 – 3.286]	gene(s) hit or close-by	ACAD11, ACKR4, DNAJC13, HSPA8P19, NPHP3, NPHP3-ACAD11, UBA5
phastCons	μ = 0.087 [0 – 1]	eQTL gene(s)	DNAJC13, NPHP3, NPHP3-AS1
GERP++	μ = -0.410 [-10.8 – 5.69]	potentially regulated gene(s)	ACAD11, DNAJC13
CADD score	μ = 3.787 [0.022 – 14.29]	disease gene(s)	NPHP3, DNAJC13

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
methionine	2.75×10^{-6}	Metabolomics GWAS Server	24816252	9
Coronary artery disease	5.43×10^{-5}	dbGaP	pha003031	1

Disease gene annotation

gene	trait	source DB	source entry/link
NPHP3	RENAL-HEPATIC-PANCREATIC DYSPLASIA 1	OMIM	MIM:208540
NPHP3	MECKEL SYNDROME, TYPE 7	OMIM	MIM:267010
NPHP3	NEPHRONOPHTHISIS 3	OMIM	MIM:604387
NPHP3	Meckel syndrome type 7 (MKS7)	DECIPHER	MIM:267010
NPHP3	nephronophthisis type 3 (NPHP3)	DECIPHER	MIM:604387
NPHP3	renal-hepatic-pancreatic dysplasia (RHPD)	DECIPHER	MIM:208540
NPHP3	Late-onset autosomal recessive medullary cystic kidney disease	OrphaNet	OrphaNet:93589
NPHP3	Senior-Loken syndrome	OrphaNet	OrphaNet:3156
NPHP3	NPHP3-related Meckel-like syndrome	OrphaNet	OrphaNet:3032
NPHP3	RENAL-HEPATIC-PANCREATIC DYSPLASIA	OrphaNet	OrphaNet:294415
DNAJC13	Young adult-onset Parkinsonism	OrphaNet	OrphaNet:2828

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)
ACAD11	missense variant	ENST00000264990	NM_032169.4	ENSP00000264990	T/K	aCg/aAg	?	?	1
ACAD11	missense variant	ENST00000481970	?	ENSP00000420907	T/K	aCg/aAg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
NPHP3	?	ENSG00000113971	muscularis mucosae	2.27×10-7 (p-value)	GTEx Portal V6	63
DNAJC13	?	ENSG00000138246	muscularis mucosae	3.42×10-6 (p-value)	GTEx Portal V6	3
NPHP3-AS1	?	ENSG00000248724	transformed fibroblasts	2.27×10-9 (p-value)	GTEx Portal V6	66
NPHP3	?	ENSG00000113971	transformed fibroblasts	1.28×10-13 (p-value)	GTEx Portal V6	66
DNAJC13	?	ENSG00000138246	transformed fibroblasts	5.95×10-6 (p-value)	GTEx Portal V6	37
NPHP3	?	ENSG00000113971	tibial artery	1.54×10-9 (p-value)	GTEx Portal V6	66
NPHP3	?	ENSG00000113971	left ventricle	8.29×10-8 (p-value)	GTEx Portal V6	62
NPHP3	?	ENSG00000113971	blood	1.65×10-9 (p-value)	GTEx Portal V6	66
NPHP3	?	ENSG00000113971	subcutaneous adipocytes	1.71×10-10 (p-value)	GTEx Portal V6	66
DNAJC13	ENST00000509279	ILMN_1752281	blood	1.07×10-5 (p-value)	Westra et al.	6
DNAJC13	ENST00000260818					
NPHP3	?	ENSG00000113971	lung	2.22×10-6 (p-value)	GTEx Portal V6	28
NPHP3	?	ENSG00000113971	aorta	3.78×10-7 (p-value)	GTEx Portal V6	30
NPHP3-AS1	?	ENSG00000248724	tibial nerve	1.81×10-5 (p-value)	GTEx Portal V6	3
NPHP3	?	ENSG00000113971	skeletal muscle	1.16×10-5 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000038218		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000354562	1	ENCP00000038224	ACAD11

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001483665 (promoter flanking region)	1	NHLF	H3K36me3, H3K27ac, DNase1
		embryonic stem cell (H1ESC)	DNase1
		Osteobl	H3K27ac, H3K4me2, H3K36me3
		blood (K562)	DNase1
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	H3K4me1, H3K4me3, DNase1, H3K27ac
		muscle (HSMM)	H3K36me3, DNase1
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K36me3
		endothelium (HUVEC)	Cjun, DNase1
		liver (HepG2)	DNase1
		lung (IMR90)	DNase1, H2BK12ac, H3K18ac, H3K36me3
		A549	H3K36me3
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2
		skin (NHEK)	H3K36me3, H3K4me1
ENSR00001374079 (promoter flanking region)	2	NHLF	DNase1
		blood (K562)	DNase1, H2AZ
		blood (DND-41)	H3K27ac, H3K36me3, H3K4me1

		skin (NHDF-AD)	H3K4me1, DNase1
		endothelium (HUVEC)	H3K4me1
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me1
ENSR00001374085 (promoter flanking region)	1	NHLF	H3K27ac, DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		Osteobl	H3K4me2, H3K27ac
		skin (NHDF-AD)	H3K4me1, H3K4me3, DNase1, H3K4me2, H3K27ac
		breast (HMEC)	H3K27ac
		muscle (HSMM)	DNase1
		endothelium (HUVEC)	Cjun, DNase1
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H2BK12ac, H3K18ac, H3K27ac, H3K4me2, H4K5ac, H3K4ac, H2BK120ac, H2BK20ac, H4K91ac, H4K8ac, DNase1
		nervous (NH-A)	H3K4me2, DNase1
		skin (NHEK)	H3K27ac, H3K4me1, DNase1
ENSR00001678918 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		endothelium (HUVEC)	H3K36me3
		blood (GM12878)	Yy1
		blood (K562)	H3K27me3
ENSR00001678920 (CTCF binding site)	1	Osteobl	H3K36me3
		blood (DND-41)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ACAD11	upstream gene variant	1185	ENST00000264990	NM_032169.4	ENSP00000264990	1
ACAD11	upstream gene variant	3987	ENST00000507705	?	?	1
ACAD11	upstream gene variant	2259	ENST00000496418	?	?	1
ACAD11	downstream gene variant, upstream gene variant	1588	ENST00000489991	?	?	3
ACAD11	upstream gene variant	1842	ENST00000485198	?	ENSP00000419973	1
ACAD11	downstream gene variant, upstream gene variant	2064	ENST00000481970	?	ENSP00000420907	2
ACAD11	upstream gene variant	1826	ENST00000469042	?	?	1
ACAD11	downstream gene variant, upstream gene variant	781	ENST00000510100	?	ENSP00000423796	2
ACAD11	upstream gene variant	1783	ENST00000487024	?	?	2
ACKR4	upstream gene variant, downstream gene variant	535	ENST00000509820	?	ENSP00000425557	2
ACKR4	upstream gene variant	4933	ENST00000249887	NM_178445.2, NM_016557.3	ENSP00000249887	1
DNAJC13	upstream gene variant	626	ENST00000464766	?	?	5
DNAJC13	upstream gene variant, downstream gene variant	319	ENST00000513822	?	?	4
DNAJC13	downstream gene variant	85	ENST00000260818	NM_015268.3	ENSP00000260818	2
DNAJC13	upstream gene variant	307	ENST00000463038	?	?	4
DNAJC13	downstream gene variant	67	ENST00000509279	?	ENSP00000426240	3
DNAJC13	upstream gene variant, downstream gene variant	1710	ENST00000506813	?	ENSP00000421735	10
DNAJC13	downstream gene variant	2468	ENST00000486798	?	?	4
HSPA8P19	downstream gene variant	887	ENST00000505359	?	?	1
NPHP3	downstream gene variant	2677	ENST00000465756	?	ENSP00000419907	1
NPHP3	downstream gene variant	473	ENST00000515289	?	?	2

NPHP3	downstream gene variant	1661	ENST00000474871	?	?	1
NPHP3	downstream gene variant, upstream gene variant	2334	ENST00000493732	?	?	2
NPHP3	downstream gene variant	2680	ENST00000337331	NM_153240.4	ENSP00000338766	1
NPHP3	downstream gene variant	2483	ENST00000490993	?	?	1
NPHP3	downstream gene variant, upstream gene variant	2680	ENST00000512094	?	ENSP00000427666	2
UBA5	downstream gene variant	1759	ENST00000493720	?	ENSP00000417879	1
UBA5	downstream gene variant	2195	ENST00000473651	?	ENSP00000424984	1
UBA5	downstream gene variant	963	ENST00000480955	?	?	1
UBA5	downstream gene variant	1004	ENST00000356232	NM_024818.3	ENSP00000348565	2
UBA5	upstream gene variant, downstream gene variant	851	ENST00000264991	NM_198329.2	ENSP00000264991	3
UBA5	downstream gene variant	1166	ENST00000494238	?	ENSP00000418807	2
UBA5	upstream gene variant	3831	ENST00000489361	?	ENSP00000417905	1
UBA5	downstream gene variant	2407	ENST00000494112	?	?	1
UBA5	downstream gene variant	1166	ENST00000468227	?	?	2

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
ACAD11	ENST00000264990	NM_032169.4	ENSP00000264990	I	atT/atC	1

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
DNAJC13	ENST00000464766	?	?	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ACAD11	ENST00000489991	?	?	8
ACAD11	ENST00000481970	?	ENSP00000420907	12
ACAD11	ENST00000485198	?	ENSP00000419973	22
ACAD11	ENST00000264990	NM_032169.4	ENSP00000264990	21
ACAD11	ENST00000510100	?	ENSP00000423796	8
ACAD11	ENST00000487024	?	?	11
ACAD11	ENST00000477604	?	?	4
ACAD11	ENST00000496418	?	?	22
ACAD11	ENST00000469042	?	?	22
ACAD11	ENST00000507705	?	?	9
ACKR4	ENST00000249887	NM_178445.2, NM_016557.3	ENSP00000249887	1
ACKR4	ENST00000509820	?	ENSP00000425557	4
DNAJC13	ENST00000509279	?	ENSP00000426240	3
DNAJC13	ENST00000506813	?	ENSP00000421735	2
DNAJC13	ENST00000464766	?	?	11

DNAJC13	ENST00000464766	?	?	11
DNAJC13	ENST00000486798	?	?	1
DNAJC13	ENST00000260818	NM_015268.3	ENSP00000260818	32
HSPA8P19	ENST00000505359	?	?	3
NPHP3	ENST00000515289	?	?	2
NPHP3	ENST00000490993	?	?	5
NPHP3	ENST00000474871	?	?	5
NPHP3	ENST00000465756	?	ENSP00000419907	5
NPHP3	ENST00000337331	NM_153240.4	ENSP00000338766	5
NPHP3	ENST00000512094	?	ENSP00000427666	1
NPHP3-ACAD11	ENST00000471702	?	ENSP00000419763	29
UBA5	ENST00000494238	?	ENSP00000418807	1
UBA5	ENST00000264991	NM_198329.2	ENSP00000264991	1
UBA5	ENST00000473651	?	ENSP00000424984	1
UBA5	ENST00000505777	?	ENSP00000427233	1
UBA5	ENST00000493720	?	ENSP00000417879	1
UBA5	ENST00000356232	NM_024818.3	ENSP00000348565	1
UBA5	ENST00000464068	?	ENSP00000420055	1
UBA5	ENST00000468022	?	ENSP00000418569	1
UBA5	ENST00000464101	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ACAD11	ENST00000481970	?	ENSP00000420907	1
ACAD11	ENST00000485198	?	ENSP00000419973	3
DNAJC13	ENST00000260818	NM_015268.3	ENSP00000260818	1
NPHP3-ACAD11	ENST00000471702	?	ENSP00000419763	3

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ACAD11	ENST00000487024	?	1
ACAD11	ENST00000496418	?	3
ACAD11	ENST00000469042	?	3
ACAD11	ENST00000507705	?	1
HSPA8P19	ENST00000505359	?	1
NPHP3	ENST00000493732	?	1