Locus 450 (rs9462846)

SNiPAcard

Block annotations

Block info

genomic range	chr6:42,824,756-42,892,495
block size	67,740 bp
variant count	71 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.793 [-7.379 – 3.635]	gene(s) hit or close-by	C6orf226, CNPY3, GLTSCR1L, PTCRA, RPL7L1
phastCons	μ = 0.074 [0 – 1]	eQTL gene(s)	C6orf226, PEX6, RPL7L1, RPL7P21
GERP++	μ = -0.003 [-4.25 – 4.82]	potentially regulated gene(s)	CUL9, PRPH2, PTCRA, PTK7, RP11-480N24.4
CADD score	μ = 4.608 [0.014 – 25.7]	disease gene(s)	PEX6, PRPH2

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
PEX6	PEROXISOME BIOGENESIS DISORDER 4A (ZELLWEGER)	OMIM	MIM:614862
PEX6	PEROXISOME BIOGENESIS DISORDER 4B	OMIM	MIM:614863
PEX6	peroxisome biogenesis disorder complementation group 4 (PBD-CG4)	DECIPHER	MIM:601498
PEX6	Zellweger syndrome (ZWS)	DECIPHER	MIM:214100
PEX6	NEONATAL ADRENOLEUKODYSTROPHY	OrphaNet	OrphaNet:44
PEX6	Infantile Refsum disease	OrphaNet	OrphaNet:772
PEX6	ZELLWEGER SYNDROME	OrphaNet	OrphaNet:912
PRPH2	Butterfly-shaped pigment dystrophy	OrphaNet	OrphaNet:99001
PRPH2	Retinitis pigmentosa	OrphaNet	OrphaNet:791
PRPH2	Central areolar choroidal dystrophy	OrphaNet	OrphaNet:75377
PRPH2	Adult-onset foveomacular vitelliform dystrophy	OrphaNet	OrphaNet:99000
PRPH2	RETINITIS PUNCTATA ALBESCENS	OrphaNet	OrphaNet:52427
PRPH2	FUNDUS ALBIPUNCTATUS	OrphaNet	OrphaNet:227796
PRPH2	Cone-rod dystrophy	OrphaNet	OrphaNet:1872

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)
PTCRA	missense variant	ENST00000616441	NM_001243168.1	ENSP00000477815	I/V	Atc/Gtc	?	?	1
PTCRA	missense variant	ENST00000304672	NM_138296.2	ENSP00000304447	I/V	Atc/Gtc	?	?	1
PTCRA	missense variant	ENST00000441198	NM_001243169.1	ENSP00000409550	I/V	Atc/Gtc	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RPL7L1	?	ENSG00000146223	muscularis mucosae	2.73×10-10 (p-value)	GTEx Portal V6	66
PEX6	?	ENSG00000124587	muscularis mucosae	7.85×10-6 (p-value)	GTEx Portal V6	5

RPL7L1	?	ENSG00000146223	lung	1.53×10-8 (p-value)	GTEx Portal V6	42
PEX6	?	ENSG00000124587	lung	2.54×10-12 (p-value)	GTEx Portal V6	66
RPL7L1	?	ENSG00000146223	transformed fibroblasts	4.14×10-24 (p-value)	GTEx Portal V6	66
PEX6	?	ENSG00000124587	transformed fibroblasts	2.36×10-10 (p-value)	GTEx Portal V6	61
RPL7L1	?	ENSG00000146223	tibial artery	9.04×10-17 (p-value)	GTEx Portal V6	66
PEX6	?	ENSG00000124587	tibial artery	5.99×10-9 (p-value)	GTEx Portal V6	33
RPL7L1	?	ENSG00000146223	breast	1.18×10-13 (p-value)	GTEx Portal V6	65
PEX6	?	ENSG00000124587	breast	2.13×10-7 (p-value)	GTEx Portal V6	23
RPL7L1	?	ENSG00000146223	blood	3.26×10-9 (p-value)	GTEx Portal V6	62
PEX6	?	ENSG00000124587	blood	2.64×10-9 (p-value)	GTEx Portal V6	33
RPL7L1	?	ENSG00000146223	thyroid	2.79×10-8 (p-value)	GTEx Portal V6	41
PEX6	?	ENSG00000124587	thyroid	8.77×10-9 (p-value)	GTEx Portal V6	33
PEX6	ENST00000304611	ILMN_1683279	monocyte	2.16×10-14 (p-value)	Fairfax et al.	9
PEX6	ENST00000244546		b-cell	9.71×10-13 (p-value)	Fairfax et al.	9
RPL7L1	?	ENSG00000146223	skeletal muscle	1.23×10-19 (p-value)	GTEx Portal V6	66
PEX6	?	ENSG00000124587	skeletal muscle	4.96×10-10 (p-value)	GTEx Portal V6	50
PEX6	?	ENSG00000124587	blood	5.06×10-6 (q-value)	SeeQTL DB (HapMap)	12
RPL7L1	?	ENSG00000146223	transverse colon	5.22×10-11 (p-value)	GTEx Portal V6	65
PEX6	?	ENSG00000124587	transverse colon	3.92×10-6 (p-value)	GTEx Portal V6	7
RPL7L1	?	ENSG00000146223	unexposed skin	5.02×10-10 (p-value)	GTEx Portal V6	55
PEX6	?	ENSG00000124587	unexposed skin	8.55×10-9 (p-value)	GTEx Portal V6	55
RPL7L1	?	ENSG00000146223	sun exposed skin	7.59×10-18 (p-value)	GTEx Portal V6	66
PEX6	?	ENSG00000124587	sun exposed skin	1.57×10-11 (p-value)	GTEx Portal V6	64
C6orf226	?	ENSG00000221821	sun exposed skin	8.24×10-6 (p-value)	GTEx Portal V6	5
RPL7L1	?	ENSG00000146223	aorta	3.25×10-12 (p-value)	GTEx Portal V6	66
PEX6	?	ENSG00000124587	aorta	8.93×10-7 (p-value)	GTEx Portal V6	5
RPL7L1	?	ENSG00000146223	left ventricle	3.73×10-12 (p-value)	GTEx Portal V6	66
PEX6	?	ENSG00000124587	left ventricle	5.40×10-7 (p-value)	GTEx Portal V6	6
RPL7L1	?	ENSG00000146223	subcutaneous adipocytes	6.97×10-16 (p-value)	GTEx Portal V6	66
PEX6	?	ENSG00000124587	subcutaneous adipocytes	4.18×10-10 (p-value)	GTEx Portal V6	65
RPL7L1	?	ENSG00000146223	tibial nerve	2.06×10-11 (p-value)	GTEx Portal V6	66
PEX6	?	ENSG00000124587	tibial nerve	5.09×10-12 (p-value)	GTEx Portal V6	65
C6orf226	?	ENSG00000221821	tibial nerve	2.55×10-6 (p-value)	GTEx Portal V6	6
RPL7L1	?	ENSG00000146223	esophagus mucosa	9.20×10-13 (p-value)	GTEx Portal V6	46
PEX6	?	ENSG00000124587	esophagus mucosa	1.28×10-9 (p-value)	GTEx Portal V6	64

RPL7L1	?	ENSG00000146223	gastroesophageal junction	3.82×10-8 (p-value)	GTEx Portal V6	58
RPL7L1	?	ENSG00000146223	testis	3.65×10-13 (p-value)	GTEx Portal V6	66
PEX6	?	ENSG00000124587	testis	1.26×10-8 (p-value)	GTEx Portal V6	64
PEX6	ENST00000304611	ILMN_1683279	skin	1.78×10-13 (p-value)	MuTHER consortium	22
PEX6	ENST00000244546		blood	6.63×10-18 (p-value)	MuTHER consortium	22
			adipocyte	2.33×10-16 (p-value)	MuTHER consortium	22
PEX6	ENST00000304611	ILMN_1683279	monocyte	2.55×10-60 (p-value)	Zeller et al.	6
PEX6	ENST00000244546					
?	?	ILMN_1736238	monocyte	2.91×10-21 (p-value)	Zeller et al.	6
RPL7L1	ENST00000493763	ILMN_1705908	monocyte	6.06×10-13 (p-value)	Zeller et al.	2
RPL7P21	ENST00000478287					
RPL7L1	ENST00000397415					
RPL7L1	ENST00000462348					
RPL7L1	?	ENSG00000146223	nucleus accumbens	4.29×10-7 (p-value)	GTEx Portal V6	28
PEX6	?	ENSG00000124587	atrial appendage	1.62×10-6 (p-value)	GTEx Portal V6	10
RPL7L1	?	ENSG00000146223	cerebellar hemisphere	7.46×10-11 (p-value)	GTEx Portal V6	60
RPL7L1	?	ENSG00000146223	pancreas	2.81×10-6 (p-value)	GTEx Portal V6	29
RPL7L1	?	ENSG00000146223	frontal cortex	1.58×10-6 (p-value)	GTEx Portal V6	4
RPL7L1	?	ENSG00000146223	visceral adipocytes	4.28×10-6 (p-value)	GTEx Portal V6	13
PEX6	?	ENSG00000124587	stomach	1.53×10-6 (p-value)	GTEx Portal V6	10
RPL7L1	?	ENSG00000146223	cerebellum	7.31×10-6 (p-value)	GTEx Portal V6	1
PEX6	?	ENSG00000124587	brain	2.62×10-5 (q-value)	SeeQTL DB (Myers et al.)	2
PEX6	ENST00000304611	204545_at	blood	6.70×10-8 (p-value)	Dixon et al.	1
PEX6	ENST00000244546					
PEX6	ENST00000304611	320_at	blood	2.60×10-8 (p-value)	Dixon et al.	1
PEX6	ENST00000244546					

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
PEX6	?	ENSG00000124587	chr6	brain	3.58×10-3 (q-value)	SeeQTL DB (Myers et al.)	2

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
EGR1	MA0341.1	2	yes	0.000	1
EGR1	MA0366.1	2	yes	0.000	1
EGR1	MA0162.2	11	yes	0.000	1

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000746055	1	ENST00000304672, ENST00000441198	PTCRA

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000048629		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000449464	1	ENCP00000048621	PRPH2
		ENCP00000048658	CUL9
		ENCP00000048660	CUL9
ENCE00000449466	1	ENCP00000048621	PRPH2
		ENCP00000048658	CUL9
		ENCP00000048660	CUL9
ENCE00000449798	1	ENCP00000048674	RP11-480N24.4
ENCE00000449690	1	ENCP00000048649	PTK7
ENCE00000449547	1	ENCP00000048629	PTCRA
		ENCP00000048651	PTK7

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001216922 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H4K5ac, H3K36me3, H3K27me3, PolII, Jund, TAF7, p300, H3K27ac, Yy1, TAF1, H3K4me2, H3K9ac, H3K4me3
		HSMMtube	H3K36me3, DNase1, H2AZ, H3K4me3, H3K27ac, H3K9ac, H3K4me2
		blood (K562)	PolII, H3K36me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	DNase1, TAF1, ELF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K36me3, PolII, Jund, Gabp
		blood (GM12878)	DNase1, Gabp, ELF1, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, H2AZ, PolII
		lung (IMR90)	H3K36me3, H3K4me3, H3K9ac, H3K4me2, H3K27ac, DNase1
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac, H3K36me3
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, H3K36me3, DNase1
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H2AZ, H3K36me3
		blood (DND-41)	H3K4me1, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K36me3
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, Gabp, PolII, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	Cmyc, H3K36me3, DNase1, Max, H3K4me3, PolII
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00001216923 (promoter flanking region)	6	monocytes (Monocytes-CD14+)	H3K4me1
		embryonic stem cell (H1ESC)	DNase1
		liver (HepG2)	H3K4me3, H3K4me1, H3K4me2
		blood (GM12878)	H2AZ, PU1
		blood (K562)	DNase1, Max, PU1
ENSR00001703966 (promoter flanking region)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF
		blood (K562)	DNase1, Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		lung (IMR90)	CTCF
		blood (GM12878)	DNase1, Rad21, CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF

		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF
ENSR00001216924 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1, CTCF, Rad21
		HSMMtube	CTCF
		blood (K562)	DNase1, Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		lung (IMR90)	CTCF
		blood (GM12878)	DNase1, Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF
ENSR00001495377 (enhancer)	1	Osteobl	H3K27me3, CTCF
		blood (DND-41)	CTCF
		blood (K562)	DNase1, CTCF
		muscle (HSMM)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		liver (HepG2)	CTCF
		blood (GM12878)	Rad21, CTCF
		A549	CTCF
		skin (NHEK)	CTCF
ENSR00001216925 (CTCF binding site)	2	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF
		blood (K562)	DNase1, H2AZ, Rad21, CTCF
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF
		liver (HepG2)	H3K4me3, Rad21, H2AZ, H3K4me2, CTCF, DNase1
		lung (IMR90)	DNase1, CTCF
		blood (GM12878)	H2AZ, Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	H3K4me2, CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF, H3K4me2
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	DNase1, CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF, DNase1, H3K27me3
ENSR00001703967 (promoter flanking region)	4	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF
		blood (K562)	H2AZ, Rad21, CTCF, DNase1, PU1
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF
		liver (HepG2)	H3K4me3, PolII, Rad21, H3K4me1, H2AZ, H3K4me2, CTCF, DNase1
		blood (GM12878)	H2AZ, Rad21, CTCF
		lung (IMR90)	DNase1, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	H3K4me2, CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, CTCF
		monocytes (Monocytes-CD14+)	CTCF, DNase1

		endothelium (HUVEC)	H3K36me3, CTCF
		A549	H3K27me3, DNase1, CTCF
ENSR00001216929 (promoter flanking region)	1	cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		embryonic stem cell (H1ESC)	DNase1, H3K9ac, PolII, Rad21, Egr1, CTCF, Yy1, HDAC2, Tcf12, TAF1, H3K4me2, H3K4me3
		endothelium (HUVEC)	H3K36me3, H3K27me3
		liver (HepG2)	DNase1, H3K4me3, H3K4me2
		Osteobl	H3K4me2
		nervous (NH-A)	DNase1
ENSR00001216930 (open chromatin region)	1	lung (IMR90)	H3K27me3
		blood (GM12878)	DNase1, H2AZ
		blood (K562)	H3K27me3
ENSR00001216931 (enhancer)	1	liver (HepG2)	H3K4me1
ENSR00001216932 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K4me1
		blood (K562)	DNase1, H3K36me3
		muscle (HSMM)	DNase1

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
RPL7L1	1	ENST00000304734 ENST00000397415 ENST00000459829 ENST00000483998 ENST00000493763	hsa-miR-1271-5p hsa-miR-96-5p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
C6orf226	downstream gene variant, upstream gene variant	176	ENST00000408925	NM_001008739.1	ENSP00000386146	19
CNPY3	upstream gene variant	4443	ENST00000372836	NM_006586.3	ENSP00000361926	1
CNPY3	upstream gene variant	4435	ENST00000394142	?	ENSP00000377698	1
GLTSCR1L	downstream gene variant	2764	ENST00000314073	?	ENSP00000313933	4
GLTSCR1L	downstream gene variant	2934	ENST00000614467	?	ENSP00000482211	3
GLTSCR1L	downstream gene variant	2766	ENST00000394168	NM_015349.1	ENSP00000377723	4
PTCRA	upstream gene variant	58	ENST00000446507	NM_001243170.1	ENSP00000392288	4
PTCRA	upstream gene variant	58	ENST00000304672	NM_138296.2	ENSP00000304447	4
PTCRA	upstream gene variant	58	ENST00000441198	NM_001243169.1	ENSP00000409550	4
PTCRA	upstream gene variant	58	ENST00000616441	NM_001243168.1	ENSP00000477815	4
RPL7L1	downstream gene variant	1568	ENST00000487619	?	?	5
RPL7L1	downstream gene variant	173	ENST00000497417	?	?	6
RPL7L1	downstream gene variant	446	ENST00000492836	?	?	8
RPL7L1	downstream gene variant	164	ENST00000304734	?	ENSP00000346063	11
RPL7L1	downstream gene variant	614	ENST00000602561	?	ENSP00000473356	5
RPL7L1	downstream gene variant	1460	ENST00000483998	?	?	7
RPL7L1	downstream gene variant	1751	ENST00000397415	?	?	7
RPL7L1	downstream gene variant	193	ENST00000493763	NM_198486.2	ENSP00000418221	11
RPL7L1	upstream gene variant, downstream gene variant	130	ENST00000459829	?	?	15
RPL7L1	downstream gene variant	509	ENST00000462348	?	?	5

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GLTSCR1L	ENST00000314073	?	ENSP00000313933	5
GLTSCR1L	ENST00000614467	?	ENSP00000482211	5
GLTSCR1L	ENST00000394168	NM_015349.1	ENSP00000377723	5
PTCRA	ENST00000441198	NM_001243169.1	ENSP00000409550	7
PTCRA	ENST00000446507	NM_001243170.1	ENSP00000392288	8
PTCRA	ENST00000304672	NM_138296.2	ENSP00000304447	7
PTCRA	ENST00000616441	NM_001243168.1	ENSP00000477815	7
RPL7L1	ENST00000304734	?	ENSP00000346063	9
RPL7L1	ENST00000602561	?	ENSP00000473356	8
RPL7L1	ENST00000462348	?	?	8
RPL7L1	ENST00000459829	?	?	1
RPL7L1	ENST00000397415	?	?	9
RPL7L1	ENST00000483998	?	?	9
RPL7L1	ENST00000493763	NM_198486.2	ENSP00000418221	9
RPL7L1	ENST00000487619	?	?	6

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
RPL7L1	ENST00000304734	?	ENSP00000346063	4
RPL7L1	ENST00000493763	NM_198486.2	ENSP00000418221	4

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RPL7L1	ENST00000497417	?	3
RPL7L1	ENST00000397415	?	6
RPL7L1	ENST00000487619	?	5