Locus 145 (rs3947)

SNiPAcard

Block annotations

Block info

genomic range	chr8:11,685,453-11,712,639
block size	27,187 bp
variant count	38 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.616 [-3.186 – 2.014]	gene(s) hit or close-by	CTSB, FDFT1, RP11-589N15.2
phastCons	μ = 0.019 [0 – 0.216]	eQTL gene(s)	AF131215.9, ANTXR1, CTSB, FAM167A, FAM167A-AS1, FDFT1, OR7E158P, RP11-148O21.3, RP11-148O21.6, RP11-419I17.1
GERP++	μ = -0.897 [-6.68 – 4.09]	potentially regulated gene(s)	BLK, CTSB, DEFB130, DEFB130, FAM167A, FAM66A, FDFT1, RP11-589N15.1, ZNF705C
CADD score	μ = 4.194 [0.347 – 11.93]	disease gene(s)	ANTXR1, BLK

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Parkinson's disease	<1.00×10-6	GWAS Catalog	22451204	1

Disease gene annotation

gene	trait	source DB	source entry/link
ANTXR1	GAPO SYNDROME	OMIM	MIM:230740
ANTXR1	HEMANGIOMA, CAPILLARY INFANTILE	OMIM	MIM:602089
BLK	MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 11	OMIM	MIM:613375
ANTXR1	GAPO syndrome	OrphaNet	OrphaNet:2067
ANTXR1	Familial capillary hemangioma	OrphaNet	OrphaNet:91415
BLK	MODY syndrome	OrphaNet	OrphaNet:552

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CTSB	?	ENSG00000164733	pancreas	3.36×10-9 (p-value)	GTEx Portal V6	37
FAM167A-AS1	?	ENSG00000184608	pancreas	3.84×10-6 (p-value)	GTEx Portal V6	1
CTSB	?	ENSG00000164733	caudate basal ganglia	1.97×10-8 (p-value)	GTEx Portal V6	38
CTSB	?	ENSG00000164733	muscularis mucosae	1.44×10-21 (p-value)	GTEx Portal V6	38
CTSB	?	ENSG00000164733	spleen	1.03×10-7 (p-value)	GTEx Portal V6	31
CTSB	?	ENSG00000164733	tibial nerve	9.07×10-16 (p-value)	GTEx Portal V6	38
CTSB	?	ENSG00000164733	atrial appendage	1.32×10-11 (p-value)	GTEx Portal V6	38
CTSB	?	ENSG00000164733	transformed fibroblasts	4.17×10-10 (p-value)	GTEx Portal V6	38
RP11-148O21.3	?	ENSG00000254774	transformed fibroblasts	4.47×10-7 (p-value)	GTEx Portal V6	10
RP11-148O21.6	?	ENSG00000269954	transformed fibroblasts	3.03×10-8 (p-value)	GTEx Portal V6	10

CTSB	?	ENSG00000164733	tibial artery	4.71×10-28 (p-value)	GTEx Portal V6	38
CTSB	?	ENSG00000164733	breast	4.68×10-11 (p-value)	GTEx Portal V6	35
CTSB	?	ENSG00000164733	gastroesophageal junction	3.17×10-7 (p-value)	GTEx Portal V6	29
CTSB	?	ENSG00000164733	putamen	3.94×10-8 (p-value)	GTEx Portal V6	31
CTSB	?	ENSG00000164733	sun exposed skin	6.70×10-19 (p-value)	GTEx Portal V6	38
OR7E158P	?	ENSG00000254948	sun exposed skin	6.03×10-6 (p-value)	GTEx Portal V6	1
CTSB	?	ENSG00000164733	aorta	3.92×10-14 (p-value)	GTEx Portal V6	38
CTSB	?	ENSG00000164733	subcutaneous adipocytes	9.65×10-10 (p-value)	GTEx Portal V6	38
CTSB	ENST00000353047	ILMN_1696360	blood	2.56×10-9 (p-value)	MuTHER consortium	13
CTSB	ENST00000531551		adipocyte	2.14×10-8 (p-value)	MuTHER consortium	13
CTSB	ENST00000420692					
CTSB	ENST00000353047	ILMN_2359742	blood	1.38×10-8 (p-value)	MuTHER consortium	12
CTSB	ENST00000531551		adipocyte	1.04×10-6 (p-value)	MuTHER consortium	12
FDFT1	ENST00000618539	ILMN_2144088	adipocyte	3.12×10-6 (p-value)	MuTHER consortium	13
FDFT1	ENST00000615631					
FDFT1	ENST00000622850					
FDFT1	ENST00000528643					
FDFT1	ENST00000525954					
FDFT1	ENST00000443614					
FDFT1	ENST00000538689					
FDFT1	ENST00000528812					
FDFT1	ENST00000220584					
FDFT1	ENST00000525607					
FDFT1	ENST00000623368					
FDFT1	ENST00000530664					
FDFT1	ENST00000525900					
FDFT1	ENST00000446331					
FAM167A	ENST00000284486	ILMN_1687213	blood	8.36×10-8 (p-value)	MuTHER consortium	13
CTSB	?	ENSG00000164733	unexposed skin	1.38×10-6 (p-value)	GTEx Portal V6	23
CTSB	ENST00000353047	ILMN_1696360	monocyte	6.80×10-25 (p-value)	Zeller et al.	6
CTSB	ENST00000531551					
CTSB	ENST00000420692					
CTSB	ENST00000353047	ILMN_2359742	monocyte	4.67×10-16 (p-value)	Zeller et al.	4
CTSB	ENST00000531551					
FDFT1	ENST00000618539	ILMN_2144088	monocyte	3.93×10-19 (p-value)	Zeller et al.	6
FDFT1	ENST00000615631					
FDFT1	ENST00000622850					
FDFT1	ENST00000528643					
FDFT1	ENST00000525954					
FDFT1	ENST00000443614					
FDFT1	ENST00000538689					
FDFT1	ENST00000528812					
FDFT1	ENST00000220584					
FDFT1	ENST00000525607					
FDFT1	ENST00000623368					
FDFT1	ENST00000530664					

FDFT1	ENST00000525900						
FDFT1	ENST00000446331						
CTSB	?	ENSG00000164733	blood	4.99×10^{-8} (p-value)	GTEx Portal V6	30	
CTSB	?	ENSG00000164733	lung	1.65×10^{-6} (p-value)	GTEx Portal V6	19	
AF131215.9	?	ENSG00000269918	lung	4.39×10^{-7} (p-value)	GTEx Portal V6	4	
CTSB	?	ENSG00000164733	thyroid	3.73×10^{-6} (p-value)	GTEx Portal V6	16	
AF131215.9	?	ENSG00000269918	thyroid	2.78×10^{-6} (p-value)	GTEx Portal V6	1	
CTSB	?	ENSG00000164733	skeletal muscle	2.41×10^{-6} (p-value)	GTEx Portal V6	21	
CTSB	?	ENSG00000164733	sigmoid colon	1.52×10^{-6} (p-value)	GTEx Portal V6	17	
CTSB	ENST00000353047	ILMN_1696360	monocyte	9.33×10^{-6} (p-value)	Fairfax et al.	2	
CTSB	ENST00000531551		b-cell	2.08×10^{-5} (p-value)	Fairfax et al.	2	
CTSB	ENST00000420692						
FDFT1	ENST00000618539	ILMN_1741096	b-cell	1.27×10^{-9} (p-value)	Fairfax et al.	2	
FDFT1	ENST00000622850						
FDFT1	ENST00000615631						
FDFT1	ENST00000528643						
FDFT1	ENST00000525954						
FDFT1	ENST00000538689						
FDFT1	ENST00000443614						
FDFT1	ENST00000528812						
FDFT1	ENST00000220584						
FDFT1	ENST00000525900						
FDFT1	ENST00000530664						
FDFT1	ENST00000525607						
FDFT1	ENST00000623368						
FDFT1	ENST00000446331						
FAM167A	ENST00000284486	ILMN_1687213	monocyte	2.26×10^{-4} (p-value)	Fairfax et al.	2	
			b-cell	2.00×10^{-7} (p-value)	Fairfax et al.	2	
CTSB	?	ENSG00000164733	left ventricle	4.25×10^{-6} (p-value)	GTEx Portal V6	3	
CTSB	?	ENSG00000164733	nucleus accumbens	2.42×10^{-7} (p-value)	GTEx Portal V6	3	
RP11-419I17.1	?	ENSG00000270154	cortex	5.96×10^{-7} (p-value)	GTEx Portal V6	2	

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
ANTXR1	ENST00000409829	ILMN_1811877	chr2	monocyte	3.17×10^{-13} (p-value)	Zeller 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)
CTCF	MA0139.1	2	no	0.000	1

ENCODE promoter-associated distal DHS (Enhancer)

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

ENCE00000507012	1	ENCP00000054977	FDFT1
		ENCP00000054989	RP11-589N15.1
ENCE00000506878	1	ENCP00000054958	BLK
ENCE00000506825	2	ENCP00000054951	FAM167A
		ENCP00000054980	CTSB
ENCE00000506836	1	ENCP00000054985	CTSB
		ENCP00000054953	FAM167A
		ENCP00000054979	CTSB
		ENCP00000055004	ZNF705C DEFB130 FAM66A DEFB130

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001386037 (open chromatin region)	2	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		blood (K562)	PolII, H3K36me3
		muscle (HSMM)	H3K36me3
		breast (HMEC)	H3K36me3
		endothelium (HUVEC)	H3K36me3, PolII
		blood (GM12878)	PolII, H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001719967 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3
		blood (K562)	PolII, H3K36me3
		muscle (HSMM)	H3K36me3
		endothelium (HUVEC)	H3K36me3, PolII
		lung (IMR90)	H3K36me3
		blood (GM12878)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001386041 (CTCF binding site)	4	embryonic stem cell (H1ESC)	H3K36me3, PolII, Rad21, CTCF, DNase1
		HSMMtube	H3K36me3, DNase1
		blood (K562)	PolII, H3K36me3
		skin (NHDF-AD)	H3K36me3, DNase1
		muscle (HSMM)	H3K36me3
		liver (HepG2)	PolII, H3K36me3
		lung (IMR90)	H3K36me3
		blood (GM12878)	PolII, H3K36me3
		nervous (NH-A)	H3K36me3
		skin (NHEK)	CTCF, DNase1, H3K4me1, H3K36me3
		NHLF	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		breast (HMEC)	CTCF, H3K36me3
		cervix (HeLa-S3)	CTCF, H3K36me3
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3, PolII
		A549	H3K36me3
ENSR00001386042 (open chromatin region)	3	embryonic stem cell (H1ESC)	H3K36me3, PolII
		blood (K562)	PolII, H3K36me3
		breast (HMEC)	H3K36me3, H3K4me1
		muscle (HSMM)	H3K36me3
		endothelium (HUVEC)	H3K36me3, PolII
		blood (GM12878)	PolII, H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	DNase1, H3K36me3, H3K4me1
ENSR00001386043 (TF binding site)	1	embryonic stem cell (H1ESC)	H3K36me3, PolII, DNase1
		blood (K562)	PolII
		muscle (HSMM)	H3K36me3

		breast (HMEC)	H3K4me1
		endothelium (HUVEC)	H3K36me3, PolII
		blood (GM12878)	H3K36me3, PolII
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K4me1, H3K36me3
ENSR00001719968 (TF binding site)	1	embryonic stem cell (H1ESC)	H3K36me3, PolII, DNase1
		blood (K562)	PolII
		breast (HMEC)	H3K4me1
		endothelium (HUVEC)	H3K36me3, PolII
		blood (GM12878)	PolII, H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	DNase1, H3K4me1, H3K36me3

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
FDFT1	7	ENST00000220584 ENST00000443614 ENST00000446331 ENST00000525607 ENST00000525777 ENST00000525900 ENST00000525954 ENST00000528643 ENST00000528812 ENST00000530664 ENST00000538689 ENST00000615631 ENST00000618539 ENST00000622850 ENST00000623368	hsa-miR-216b-5p hsa-miR-433-3p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTSB	downstream gene variant	555	ENST00000531551	?	ENSP00000436456	8
CTSB	downstream gene variant, upstream gene variant	205	ENST00000524654	?	ENSP00000432077	5
CTSB	downstream gene variant	1289	ENST00000534382	?	ENSP00000435260	5
CTSB	downstream gene variant	1289	ENST00000532656	?	ENSP00000431143	5
CTSB	downstream gene variant	1296	ENST00000527215	?	ENSP00000433379	5
CTSB	downstream gene variant, upstream gene variant	45	ENST00000526481	?	ENSP00000473301	18
CTSB	downstream gene variant	45	ENST00000353047	NM_001908.3	ENSP00000345672	6
CTSB	downstream gene variant	1335	ENST00000528965	?	ENSP00000433929	5
CTSB	downstream gene variant	1252	ENST00000531502	?	ENSP00000435886	11
CTSB	downstream gene variant	551	ENST00000453527	NM_147783.2	ENSP00000409917	8
CTSB	downstream gene variant	1224	ENST00000505496	?	ENSP00000435650	12
CTSB	downstream gene variant	1232	ENST00000524500	?	ENSP00000436074	12
CTSB	downstream gene variant	571	ENST00000345125	NM_147781.2	ENSP00000342070	8
CTSB	downstream gene variant, upstream gene variant	950	ENST00000525315	?	?	13
CTSB	downstream gene variant	284	ENST00000530640	?	ENSP00000435105	8
CTSB	downstream gene variant, upstream gene variant	1200	ENST00000532370	?	?	13
CTSB	downstream gene variant	601	ENST00000534510	?	ENSP00000434217	8

CTSB	downstream gene variant	586	ENST00000533455	NM_147780.2, NM_147782.2	ENSP00000432244	8
CTSB	downstream gene variant	700	ENST00000527243	?	ENSP00000434725	18
CTSB	downstream gene variant	701	ENST00000534149	?	ENSP00000436122	18
CTSB	downstream gene variant, upstream gene variant	251	ENST00000420692	?	?	11
CTSB	downstream gene variant	685	ENST00000526195	?	ENSP00000436627	18
CTSB	downstream gene variant, upstream gene variant	122	ENST00000530624	?	?	17
CTSB	downstream gene variant	325	ENST00000531089	?	ENSP00000433215	8
CTSB	downstream gene variant	150	ENST00000533572	?	ENSP00000433995	20
CTSB	downstream gene variant	723	ENST00000526645	?	ENSP00000431518	18
CTSB	downstream gene variant	135	ENST00000534636	?	ENSP00000436159	20
CTSB	downstream gene variant, upstream gene variant	291	ENST00000533110	?	?	21
CTSB	downstream gene variant	126	ENST00000525076	?	?	21
CTSB	downstream gene variant, upstream gene variant	87	ENST00000532409	?	?	22
CTSB	downstream gene variant	165	ENST00000530296	?	ENSP00000435074	20
CTSB	downstream gene variant	2072	ENST00000532392	?	ENSP00000432408	3
CTSB	downstream gene variant, upstream gene variant	328	ENST00000530290	?	?	22
FDFT1	downstream gene variant	1094	ENST00000525607	?	ENSP00000432551	8
FDFT1	downstream gene variant	966	ENST00000525954	?	?	8
FDFT1	downstream gene variant	819	ENST00000528643	NM_001287751.1	ENSP00000431649	8
FDFT1	downstream gene variant	1825	ENST00000529464	?	ENSP00000434770	4
FDFT1	downstream gene variant	973	ENST00000528812	NM_001287747.1	ENSP00000431749	8
FDFT1	downstream gene variant	723	ENST00000446331	?	?	8
FDFT1	downstream gene variant	1150	ENST00000443614	?	ENSP00000390367	8
FDFT1	downstream gene variant	629	ENST00000538689	NM_001287750.1, NM_001287744.1	ENSP00000444248	8
FDFT1	downstream gene variant	624	ENST00000220584	NM_004462.4	ENSP00000220584	8
FDFT1	downstream gene variant	1093	ENST00000530664	NM_001287749.1	ENSP00000432331	8
FDFT1	downstream gene variant	629	ENST00000623368	NM_001287745.1	ENSP00000485229	8
FDFT1	downstream gene variant	629	ENST00000618539	NM_001287742.1	ENSP00000480828	8
FDFT1	downstream gene variant	629	ENST00000622850	NM_001287748.1	ENSP00000484122	8
FDFT1	downstream gene variant	629	ENST00000615631	NM_001287743.1	ENSP00000481481	8
FDFT1	downstream gene variant	1810	ENST00000528729	?	?	4
FDFT1	downstream gene variant	1028	ENST00000525900	?	ENSP00000434714	8
FDFT1	downstream gene variant	1739	ENST00000525283	?	ENSP00000433985	4
FDFT1	downstream gene variant	3131	ENST00000532266	?	ENSP00000435900	2
FDFT1	upstream gene variant, downstream gene variant	1612	ENST00000529521	?	?	5
FDFT1	downstream gene variant	1848	ENST00000531249	?	?	1
FDFT1	downstream gene variant	1320	ENST00000525777	?	ENSP00000436069	6

RP11-589N15.2	downstream gene variant, upstream gene variant	4	ENST00000602711	?	?	27

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTSB	ENST00000530640	?	ENSP00000435105	21
CTSB	ENST00000531089	?	ENSP00000433215	21
CTSB	ENST00000534382	?	ENSP00000435260	1
CTSB	ENST00000532656	?	ENSP00000431143	1
CTSB	ENST00000527215	?	ENSP00000433379	1
CTSB	ENST00000532392	?	ENSP00000432408	1
CTSB	ENST00000528965	?	ENSP00000433929	1
CTSB	ENST00000505496	?	ENSP00000435650	3
CTSB	ENST00000531502	?	ENSP00000435886	3
CTSB	ENST00000524500	?	ENSP00000436074	3
CTSB	ENST00000532370	?	?	2
CTSB	ENST00000525315	?	?	2
CTSB	ENST00000526195	?	ENSP00000436627	4
CTSB	ENST00000527243	?	ENSP00000434725	4
CTSB	ENST00000525076	?	?	4
CTSB	ENST00000534149	?	ENSP00000436122	4
CTSB	ENST00000526645	?	ENSP00000431518	4
CTSB	ENST00000533572	?	ENSP00000433995	5
CTSB	ENST00000530296	?	ENSP00000435074	5
CTSB	ENST00000345125	NM_147781.2	ENSP00000342070	21
CTSB	ENST00000353047	NM_001908.3	ENSP00000345672	24
CTSB	ENST00000453527	NM_147783.2	ENSP00000409917	21
CTSB	ENST00000526481	?	ENSP00000473301	9
CTSB	ENST00000420692	?	?	17
CTSB	ENST00000534636	?	ENSP00000436159	5
CTSB	ENST00000534510	?	ENSP00000434217	21
CTSB	ENST00000531551	?	ENSP00000436456	21
CTSB	ENST00000533455	NM_147780.2, NM_147782.2	ENSP00000432244	21
CTSB	ENST00000530624	?	?	11
CTSB	ENST00000530290	?	?	4
CTSB	ENST00000532409	?	?	4
CTSB	ENST00000533110	?	?	3
FDFT1	ENST00000525900	?	ENSP00000434714	8
FDFT1	ENST00000525777	?	ENSP00000436069	8
FDFT1	ENST00000528812	NM_001287747.1	ENSP00000431749	8
FDFT1	ENST00000443614	?	ENSP00000390367	8

FDFT1	ENST00000443614	?	ENSP00000390367	8
FDFT1	ENST00000538689	NM_001287750.1, NM_001287744.1	ENSP00000444248	8
FDFT1	ENST00000446331	?	?	8
FDFT1	ENST00000528729	?	?	1
FDFT1	ENST00000525607	?	ENSP00000432551	8
FDFT1	ENST00000530664	NM_001287749.1	ENSP00000432331	8
FDFT1	ENST00000623368	NM_001287745.1	ENSP00000485229	8
FDFT1	ENST00000220584	NM_004462.4	ENSP00000220584	8
FDFT1	ENST00000529464	?	ENSP00000434770	1
FDFT1	ENST00000525283	?	ENSP00000433985	1
FDFT1	ENST00000525954	?	?	8
FDFT1	ENST00000532266	?	ENSP00000435900	1
FDFT1	ENST00000528643	NM_001287751.1	ENSP00000431649	8
FDFT1	ENST00000622850	NM_001287748.1	ENSP00000484122	8
FDFT1	ENST00000618539	NM_001287742.1	ENSP00000480828	8
FDFT1	ENST00000615631	NM_001287743.1	ENSP00000481481	8

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTSB	ENST00000530640	?	ENSP00000435105	5
CTSB	ENST00000531089	?	ENSP00000433215	5
CTSB	ENST00000345125	NM_147781.2	ENSP00000342070	5
CTSB	ENST00000353047	NM_001908.3	ENSP00000345672	8
CTSB	ENST00000453527	NM_147783.2	ENSP00000409917	5
CTSB	ENST00000526481	?	ENSP00000473301	8
CTSB	ENST00000534510	?	ENSP00000434217	5
CTSB	ENST00000531551	?	ENSP00000436456	5
CTSB	ENST00000533455	NM_147780.2, NM_147782.2	ENSP00000432244	5

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CTSB	ENST00000420692	?	6