Locus 121 (rs4150579)

SNiPAcard

Block annotations

Block info

genomic range	chr11:18,309,700-18,392,983
block size	83,284 bp
variant count	48 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.408 [-3.606 – 2.283]	gene(s) hit or close-by	GTF2H1, HPS5
phastCons	μ = 0.075 [0 – 1]	eQTL gene(s)	GTF2H1, HPS5, LDHA, LDHC, SAA1
GERP++	μ = -0.080 [-4.3 – 3.95]	potentially regulated gene(s)	AC084117.3, GTF2H1, HPS5, LDHA, RP11-113D6.3, RP11-613F22.6, TSG101, UEVLD
CADD score	μ = 5.040 [0.089 – 14.37]	disease gene(s)	LDHA, HPS5, SAA1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
PANCREATIC CANCER	<9.00×10-6	GWAS Catalog	23180869	1
Amyloid A serum levels	<2.00×10-51	GWAS Catalog	21124955	1
alpha-hydroxyisovalerate	1.08×10-23	Metabolomics GWAS Server	24816252	25

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Clinvar: phenotype not specified	benign	ClinVar	RCV000150827.1	1

Disease gene annotation

gene	trait	source DB	source entry/link
LDHA	GLYCOGEN STORAGE DISEASE XI	OMIM	MIM:612933
HPS5	HERMANSKY-PUDLAK SYNDROME 5	OMIM	MIM:614074
LDHA	Glycogen storage disease due to lactate dehydrogenase M-subunit deficiency	OrphaNet	OrphaNet:284426
SAA1	Secondary amyloidosis	OrphaNet	OrphaNet:85445
HPS5	Hermansky-Pudlak syndrome without pulmonary fibrosis	OrphaNet	OrphaNet:231512

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SAA1	ENST00000356524	ILMN_2304512	adipocyte	2.33×10-5 (p-value)	MuTHER consortium	25
SAA1	ENST00000405158					
SAA1	ENST00000532858					
GTF2H1	?	ENSG00000110768	muscularis mucosae	1.75×10-20 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	lung	8.65×10-14 (p-value)	GTEx Portal V6	44
HPS5	?	ENSG00000110756	lung	1.37×10-9 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	atrial appendage	7.22×10-9 (p-value)	GTEx Portal V6	44

GTF2H1	?	ENSG00000110768	transformed fibroblasts	2.71×10-28 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	breast	8.81×10-13 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	tibial artery	4.70×10-15 (p-value)	GTEx Portal V6	44
LDHC	?	ENSG00000166796	tibial artery	1.90×10-5 (p-value)	GTEx Portal V6	1
GTF2H1	?	ENSG00000110768	thyroid	7.29×10-14 (p-value)	GTEx Portal V6	44
LDHA	?	ENSG00000134333	skeletal muscle	1.73×10-8 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	skeletal muscle	2.31×10-7 (p-value)	GTEx Portal V6	40
GTF2H1	?	ENSG00000110768	EBV lymphocytes	5.82×10-7 (p-value)	GTEx Portal V6	25
GTF2H1	?	ENSG00000110768	transverse colon	1.75×10-14 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	liver	2.96×10-8 (p-value)	GTEx Portal V6	43
GTF2H1	?	ENSG00000110768	sun exposed skin	7.28×10-21 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	unexposed skin	7.19×10-20 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	aorta	6.66×10-16 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	left ventricle	1.11×10-9 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	subcutaneous adipocytes	2.66×10-27 (p-value)	GTEx Portal V6	44
GTF2H1	ENST00000534641	ILMN_1726693	monocyte	1.30×10-20 (p-value)	Zeller et al.	7
GTF2H1	ENST00000530496					
GTF2H1	ENST00000265963					
GTF2H1	ENST00000453096					
HPS5	ENST00000438420	ILMN_1655312	monocyte	1.77×10-14 (p-value)	Zeller et al.	7
HPS5	ENST00000349215					
HPS5	ENST00000396253					
HPS5	ENST00000438420	ILMN_2411731	monocyte	1.18×10-12 (p-value)	Zeller et al.	3
HPS5	ENST00000349215					
HPS5	ENST00000396253					
GTF2H1	?	ENSG00000110768	caudate basal ganglia	1.61×10-10 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	stomach	7.82×10-18 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	visceral adipocytes	5.18×10-18 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	cortex	2.04×10-9 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	spleen	4.46×10-10 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	sigmoid colon	1.48×10-6 (p-value)	GTEx Portal V6	14
GTF2H1	?	ENSG00000110768	tibial nerve	2.83×10-15 (p-value)	GTEx Portal V6	44
SAA1	?	ENSG00000173432	tibial nerve	1.62×10-5 (p-value)	GTEx Portal V6	8
GTF2H1	?	ENSG00000110768	esophagus mucosa	8.35×10-37 (p-value)	GTEx Portal V6	44
GTF2H1	ENST00000534641	ILMN_2157957	blood	1.42×10-5 (p-value)	Westra et al.	25
GTF2H1	ENST00000265963					
HPS5	ENST00000438420	ILMN_1655312	blood	7.84×10-7 (p-value)	Westra et al.	1
HPS5	ENST00000349215					
HPS5	ENST00000396253					

HPS5	ENST00000396253					
GTF2H1	?	ENSG00000110768	adrenal gland	1.02×10-19 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	testis	1.27×10-24 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	cerebellum	5.54×10-7 (p-value)	GTEx Portal V6	35
GTF2H1	?	ENSG00000110768	frontal cortex	1.14×10-8 (p-value)	GTEx Portal V6	44
GTF2H1	?	ENSG00000110768	blood	2.20×10-2 (q-value)	SeeQTL DB (HapMap)	9
GTF2H1	?	ENSG00000110768	terminal ileum	6.14×10-8 (p-value)	GTEx Portal V6	22
GTF2H1	?	ENSG00000110768	hypothalamus	1.22×10-7 (p-value)	GTEx Portal V6	33
GTF2H1	?	ENSG00000110768	blood	4.85×10-6 (p-value)	GTEx Portal V6	9
GTF2H1	?	ENSG00000110768	nucleus accumbens	1.06×10-6 (p-value)	GTEx Portal V6	15
?	?	ILMN_1726693	liver	6.66×10-15 (p-value)	Innocenti et al.	1

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
MYC, MAX	MA0059.1	10	no	-0.060	1

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000011407		
ENCP00000011408		HPS5

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000100096	1	ENCP00000011410	GTF2H1
		ENCP00000011419	LDHA
		ENCP00000011415	LDHA
		ENCP00000011420	AC084117.3
		ENCP00000011418	LDHA
ENCE00000100028	1	ENCP00000011426	TSG101 RP11-613F22.6
		ENCP00000011396	RP11-113D6.3
		ENCP00000011430	UEVLD
		ENCP00000011429	UEVLD

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000559412 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		HSMMtube	DNase1, H3K36me3, H3K27ac
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K27ac, H3K36me3, DNase1
		liver (HepG2)	H3K36me3
		blood (GM12878)	Egr1, H3K79me2, H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K36me3, H3K4me2
		skin (NHEK)	DNase1, H3K36me3
		NHLF	DNase1
		Osteobl	H3K27ac, H3K4me2, H3K36me3

		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K36me3, H3K4me2, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, H3K27ac, H3K4me1, H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K27ac, H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
ENSR00001568729 (promoter)	3	embryonic stem cell (H1ESC)	TAF1, H3K4me2, USF1, H3K9ac, H3K4me3, Yy1, ATF3, H4K5ac, H3K36me3, H3K27me3, PolII, Gabp, TAF7, Sin3Ak20, H3K27ac, DNase1
		HSMMtube	DNase1, H3K36me3, H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K79me2
		blood (K562)	Nfe2, E2F6, H3K9ac, Gabp, USF1, H3K79me2, HEY1, H2AZ, TAF1, H3K27ac, Yy1, ELF1, Jund, Max, Cfos, Cmyc, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	ATF3, H3K4me3, H3K27me3, H3K36me3, Cmyc, H3K27ac, H3K9ac, Yy1, PolII, H3K79me2, TAF1, ELF1, USF1, H3K4me1, H3K4me2, Gabp, DNase1
		blood (GM12878)	ATF3, PU1, Max, PolII, H2AZ, DNase1, Yy1, Gabp, ELF1, H3K79me2, H3K4me3, USF1, H3K27ac, Cfos, H3K4me2, H3K9ac
		lung (IMR90)	DNase1, H4K8ac, H3K79me2, H3K27ac, H3K18ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K9ac
		nervous (NH-A)	DNase1, H3K36me3, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, H3K36me3, H3K4me3, H3K27ac, H3K9ac, H3K4me2
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac, H3K36me3
		Osteobl	H3K36me3, H3K4me2, H2AZ, H3K27ac, H3K4me3
		blood (DND-41)	H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3, H3K4me1
		breast (HMEC)	H3K4me2, H3K9ac, H3K4me3, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, PolII, H3K36me3, Gabp, H3K79me2, BAF170, H3K9ac, H3K4me2, H3K27ac, TAF1, Max, H3K4me3
		monocytes (Monocytes-CD14+)	DNase1, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, PolII, H3K27ac, H3K9ac, H3K4me3, H3K4me2, Max, Cjun, DNase1, Cmyc
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00000684954 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	DNase1, Nfe2, H3K79me2, H3K36me3
		breast (HMEC)	H3K36me3, H3K4me2, H3K27ac
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	DNase1, H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3
		blood (GM12878)	H3K79me2, H3K27ac, BATF, H3K4me3
		A549	H3K36me3
		skin (NHEK)	H3K4me2, H3K36me3
ENSR00000559415 (enhancer)	2	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	H3K36me3
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001568731 (open chromatin region)	1	A549	H3K36me3
		blood (K562)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
GTF2H1	upstream gene variant, downstream gene variant	152	ENST00000532227	?	?	12
GTF2H1	upstream gene variant, downstream gene variant	20	ENST00000453096	NM_001142307.1	ENSP00000393638	7

GTF2H1	upstream gene variant, downstream gene variant	646	ENST00000526282	?	?	5
GTF2H1	upstream gene variant, downstream gene variant	2380	ENST00000526630	?	ENSP00000439774	2
GTF2H1	upstream gene variant, downstream gene variant	226	ENST00000418116	?	?	11
GTF2H1	upstream gene variant, downstream gene variant	236	ENST00000534641	?	ENSP00000435375	9
GTF2H1	upstream gene variant, downstream gene variant	216	ENST00000531757	?	?	8
GTF2H1	upstream gene variant, downstream gene variant	238	ENST00000525831	?	ENSP00000431481	8
GTF2H1	upstream gene variant, downstream gene variant	265	ENST00000265963	NM_005316.3	ENSP00000265963	9
GTF2H1	upstream gene variant, downstream gene variant	237	ENST00000526461	?	?	11
GTF2H1	upstream gene variant, downstream gene variant	190	ENST00000528427	?	?	5
GTF2H1	upstream gene variant, downstream gene variant	548	ENST00000607664	?	?	7
GTF2H1	upstream gene variant, downstream gene variant	147	ENST00000534213	?	?	13
GTF2H1	upstream gene variant, downstream gene variant	361	ENST00000524753	?	ENSP00000436575	11
GTF2H1	upstream gene variant, downstream gene variant	2941	ENST00000530496	?	ENSP00000433133	7
GTF2H1	upstream gene variant, downstream gene variant	898	ENST00000543932	?	?	7
HPS5	downstream gene variant, upstream gene variant	97	ENST00000399287	?	?	5
HPS5	upstream gene variant	101	ENST00000438420	NM_181508.1	ENSP00000399590	4
HPS5	upstream gene variant	101	ENST00000349215	NM_181507.1	ENSP00000265967	4
HPS5	downstream gene variant, upstream gene variant	187	ENST00000589545	?	?	4
HPS5	upstream gene variant	101	ENST00000396253	NM_007216.3	ENSP00000379552	4
HPS5	upstream gene variant	1239	ENST00000537258	?	ENSP00000437437	1
HPS5	upstream gene variant	422	ENST00000545561	?	?	2
HPS5	downstream gene variant, upstream gene variant	77	ENST00000531848	?	ENSP00000431758	6

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
HPS5	ENST00000531848	?	ENSP00000431758	L	ctC/ctA	1
HPS5	ENST00000396253	NM_007216.3	ENSP00000379552	L	ctC/ctA	1
HPS5	ENST00000349215	NM_181507.1	ENSP00000265967	L	ctC/ctA	1
HPS5	ENST00000438420	NM_181508.1	ENSP00000399590	L	ctC/ctA	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GTF2H1	ENST00000265963	NM_005316.3	ENSP00000265963	30
GTF2H1	ENST00000531757	?	?	9
GTF2H1	ENST00000528427	?	?	6
GTF2H1	ENST00000526630	?	ENSP00000439774	13
GTF2H1	ENST00000526282	?	?	7
GTF2H1	ENST00000530496	?	ENSP00000433133	14
GTF2H1	ENST00000524753	?	ENSP00000436575	7
GTF2H1	ENST00000543932	?	?	7

GTF2H1	ENST00000534213	?	?	3
GTF2H1	ENST00000526461	?	?	5
GTF2H1	ENST00000525831	?	ENSP00000431481	9
GTF2H1	ENST00000453096	NM_001142307.1	ENSP00000393638	30
GTF2H1	ENST00000418116	?	?	26
GTF2H1	ENST00000532227	?	?	5
GTF2H1	ENST00000534641	?	ENSP00000435375	30
HPS5	ENST00000399287	?	?	6
HPS5	ENST00000589545	?	?	6
HPS5	ENST00000531848	?	ENSP00000431758	10
HPS5	ENST00000544218	?	ENSP00000441781	3
HPS5	ENST00000396253	NM_007216.3	ENSP00000379552	13
HPS5	ENST00000543728	?	?	3
HPS5	ENST00000349215	NM_181507.1	ENSP00000265967	13
HPS5	ENST00000352460	?	?	3
HPS5	ENST00000545561	?	?	1
HPS5	ENST00000438420	NM_181508.1	ENSP00000399590	13

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
GTF2H1	ENST00000453096	NM_001142307.1	ENSP00000393638	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
GTF2H1	ENST00000528427	?	1
GTF2H1	ENST00000526461	?	2
GTF2H1	ENST00000532227	?	2