Locus 113 (rs2545801)

SNiPAcard

Block annotations

Block info

genomic range	chr5:176,836,532-176,842,474
block size	5,943 bp
variant count	5 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.148 [-0.974 – 1.747]	gene(s) hit or close-by	F12, GRK6
phastCons	μ = 0.095 [0.001 – 0.375]	eQTL gene(s)	F12, GRIN2B, MXD3, RAB24, RGS14
GERP++	μ = 1.385 [-0.529 – 4.44]	potentially regulated gene(s)	F12, F12
CADD score	μ = 7.601 [4.209 – 16.43]	disease gene(s)	F12, GRIN2B

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Blood metabolite ratios	$<6.00\times10^{-26}$	GWAS Catalog	24816252	1
Circulating vasoactive peptide levels	$<1.00\times10^{-67}$	GWAS Catalog	23381795	1
Metabolite levels	$<3.00\times10^{-14}$	GWAS Catalog	22916037	1
Activated partial thromboplastin time	$<6.00\times10^{-88}$	GWAS Catalog	22703881	1
Metabolite levels	$<9.00\times10^{-11}$	GWAS Catalog	22286219	1
Platelet function and related traits	$<1.00\times10^{-6}$	GWAS Catalog	21546496	1
Activated partial thromboplastin time	$<2.00\times10^{-30}$	GWAS Catalog	20303064	1
bradykinin, des-arg(9)	2.76×10^{-23}	Metabolomics GWAS Server	24816252	3

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Factor xii polymorphism	benign	ClinVar	RCV000001226.2	1
?	HGMD curated	HGMD	CR982412	1
FACTOR XII POLYMORPHISM	OMIM curated	OMIM	MIM:610619	1

Disease gene annotation

gene	trait	source DB	source entry/link	
F12	FACTOR XII DEFICIENCY	OMIM	MIM:234000	
F12	ANGIOEDEMA, HEREDITARY, TYPE III	OMIM	MIM:610618	
GRIN2B	MENTAL RETARDATION, AUTOSOMAL DOMINANT 6	OMIM	MIM:613970	
F12	Hereditary angioedema type 3	OrphaNet	OrphaNet:100054	
F12	Congenital factor XII deficiency	OrphaNet	OrphaNet:330	
GRIN2B	Mental retardation, autosomal dominant 6	DECIPHER	MIM:613970	

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RAB24	ENST00000478234	ILMN_1714393	blood	1.49×10^{-5} (p-value)	MuTHER consortium	3

RAB24	ENST00000393610		adipocyte	4.14×10-5 (p-value)	MuTHER consortium	3
RAB24	ENST00000471466					
RAB24	ENST00000303270					
MXD3	ENST00000503473					
RAB24	ENST00000303251					
RAB24	ENST00000393611					
RAB24	ENST00000495458					
F12	ENST00000253496	ILMN_1671766	blood	1.14×10-6 (p-value)	Westra et al.	3
F12	ENST00000504406					
F12	ENST00000514943					
F12	ENST00000502854					
F12	ENST00000510358					
RGS14	ENST00000506944	ILMN_1696828	blood	1.13×10-6 (p-value)	Westra et al.	3
RGS14	ENST00000514102					
RGS14	ENST00000509289					
RGS14	ENST00000511890					
RGS14	ENST00000503044					
RGS14	ENST00000502731					
RGS14	ENST00000425155					
RGS14	ENST00000408923					
MXD3	?	ENSG00000213347	muscularis mucosae	7.40×10-7 (p-value)	GTEx Portal V6	5
F12	?	ENSG00000131187	liver	3.72×10-13 (p-value)	GTEx Portal V6	5
F12	ENST00000504406	ILMN_1671766	liver	<2.2×10-16 (p-value)	Innocenti et al.	1
F12	ENST00000253496					
F12	ENST00000502854					
F12	ENST00000514943					
F12	ENST00000510358					

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
GRIN2B	ENST00000609686	ILMN_1884244	chr12	blood	2.71×10-7 (p-value)	Westra et al.	1

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000046906		F12

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001297853 (promoter flanking region)	2	endothelium (HUVEC)	H3K27me3
		embryonic stem cell (H1ESC)	H3K27me3
		HSMMtube	H3K27me3
		blood (GM12878)	PolII, H2AZ, H3K4me1, DNase1, H3K27ac
		Osteobl	H3K27me3
		lung (IMR90)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
F12	upstream gene variant	4550	ENST00000503736	?	?	1
F12	upstream gene variant	3313	ENST00000253496	NM_000505.3	ENSP00000253496	3

F12	upstream gene variant	3313	ENST00000253496	NM_000505.3	ENSP00000253496	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GRK6	ENST00000502598	?	ENSP00000422873	5
GRK6	ENST00000506296	?	ENSP00000421055	5

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
F12	ENST00000253496	NM_000505.3	ENSP00000253496	1