SNiPAcard

Block annotations

Block info

genomic range	chr5:135,362,550-135,400,035
block size	37,486 bp
variant count	26 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.648 [-4.435 – 1]	**gene(s) hit or close-by**	TGFBI
phastCons	μ = 0.003 [0 – 0.03]	**eQTL gene(s)**	AC009014.3, SMAD5, TGFBI, TRPC7
GERP++	μ = -2.821 [-9.26 – 3.43]	**potentially regulated gene(s)**	CTC-321K16.4, TGFBI
CADD score	μ = 3.253 [0.008 – 10.2]	**disease gene(s)**	TGFBI

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Response to cytidine analogues (gemcitabine)	$<4.00\times10^{-6}$	GWAS Catalog	24483146	1

Disease gene annotation

gene	trait	source DB	source entry/link
TGFBI	CORNEAL DYSTROPHY, AVELLINO TYPE	OMIM	MIM:607541
TGFBI	CORNEAL DYSTROPHY OF BOWMAN LAYER, TYPE II	OMIM	MIM:602082
TGFBI	CORNEAL DYSTROPHY, LATTICE TYPE IIIA	OMIM	MIM:608471
TGFBI	CORNEAL DYSTROPHY, GROENOUW TYPE I	OMIM	MIM:121900
TGFBI	CORNEAL DYSTROPHY OF BOWMAN LAYER, TYPE I	OMIM	MIM:608470
TGFBI	CORNEAL DYSTROPHY, LATTICE TYPE I	OMIM	MIM:122200
TGFBI	CORNEAL DYSTROPHY, EPITHELIAL BASEMENT MEMBRANE	OMIM	MIM:121820
TGFBI	Granular corneal dystrophy type I	OrphaNet	OrphaNet:98962
TGFBI	Lattice corneal dystrophy Type I	OrphaNet	OrphaNet:98964
TGFBI	Microcystic corneal dystrophy	OrphaNet	OrphaNet:98956
TGFBI	Granular corneal dystrophy type II	OrphaNet	OrphaNet:98963
TGFBI	Thiel-Behnke corneal dystrophy	OrphaNet	OrphaNet:98960
TGFBI	Reis-Bucklers corneal dystrophy	OrphaNet	OrphaNet:98961

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TGFBI	ENST00000506699	ILMN_1663866	blood	7.17×10^{-7} (p-value)	Westra et al.	17
TGFBI	ENST00000508076					
TGFBI	ENST00000504411					
TGFBI	ENST00000514554					
TGFBI	ENST00000442011					
TGFBI	ENST00000507018					
SMAD5	ENST00000545620	ILMN_2371984	blood	7.87×10^{-5} (p-value)	Westra et al.	2
SMAD5	ENST00000545279					
SMAD5	ENST00000545620	ILMN_1674551	monocyte	1.36×10^{-5} (p-value)	Fairfax et al.	2

SMAD5	ENST00000545279		b-cell	3.19×10-4 (p-value)	Fairfax et al.	2
TRPC7	ENST00000378459	ILMN_1683909	b-cell	3.95×10-4 (p-value)	Fairfax et al.	2
TRPC7	ENST00000509288					
TRPC7	ENST00000513104					
TRPC7	ENST00000502753					
TRPC7	ENST00000352189					
TRPC7	ENST00000514963					
TRPC7	ENST00000503275					
AC009014.3	?	ENSG00000271824	testis	7.88×10-6 (p-value)	GTEx Portal V6	1
SMAD5	ENST00000545279	ILMN_1674551	monocyte	4.13×10-14 (p-value)	Zeller et al.	2
SMAD5	ENST00000545620					

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000671130	1	ENST00000509749, ENST00000508767	TGFBI
FFCP00000671129	1	ENST00000509749, ENST00000508767	TGFBI

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000423745	1	ENCP00000045720	CTC-321K16.4

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001290929 (promoter flanking region)	4	NHLF	DNase1, H3K36me3
		embryonic stem cell (H1ESC)	H3K27me3, DNase1
		Osteobl	H3K36me3, H3K27ac
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		muscle (HSMM)	H3K36me3
		breast (HMEC)	H3K36me3
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	DNase1, H3K4me1
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K9ac, H3K36me3, DNase1
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001290932 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	H3K27ac, DNase1
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K4me1, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H3K4me1, H3K27ac, H3K4me2, H3K36me3, DNase1
		liver (HepG2)	DNase1, H3K36me3, Cmyc, FOSL2, Jund, BHLHE40, H3K4me1, p300, H3K4me2, FOXA1, H3K9ac, H3K27ac
		lung (IMR90)	DNase1, H4K8ac, H3K18ac, H3K27ac, H3K36me3
		nervous (NH-A)	H3K9ac, H3K27ac, H3K36me3, DNase1
		skin (NHEK)	DNase1, H3K36me3, H3K4me1
		NHLF	DNase1, H3K4me1, H3K27ac, H3K36me3
		Osteobl	H3K36me3, H3K27ac
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K36me3
		cervix (HeLa-S3)	Cjun, Jund, H3K27ac, H3K4me1, Max, Cfos, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3

		endothelium (HUVEC)	Cjun, DNase1
		A549	H3K4me3, H3K4me1, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00001290934 (promoter flanking region)	5	embryonic stem cell (H1ESC)	DNase1, Rad21, CTCF
		HSMMtube	CTCF, H3K27ac, H3K36me3, DNase1
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K36me3, H3K4me1, CTCF, DNase1, H3K27ac
		muscle (HSMM)	H3K27ac, H3K36me3, DNase1, CTCF
		liver (HepG2)	FOSL2, Jund, FOXA2, DNase1, H3K4me1, p300, FOXA1, H3K27ac, CTCF, Cmyc, H3K36me3
		lung (IMR90)	DNase1, H3K4me1, H3K18ac, H3K27ac, H3K36me3, H3K4ac, CTCF
		nervous (NH-A)	DNase1, CTCF, H3K4me2, H3K27ac, H3K36me3, H3K9ac
		skin (NHEK)	CTCF, H3K36me3
		NHLF	DNase1, H3K4me1, CTCF, H3K27ac, H3K36me3
		Osteobl	H3K36me3, CTCF, H3K27ac
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K36me3, CTCF
		cervix (HeLa-S3)	DNase1, Cjun, Jund, CTCF, H3K36me3
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	Cjun, CTCF
		A549	H3K4me3, CTCF, DNase1, H3K27ac, H3K36me3
ENSR00001697850 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	CTCF, H3K27ac, DNase1
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K4me1, CTCF, DNase1, H3K27ac
		muscle (HSMM)	DNase1, CTCF, H3K27ac, H3K36me3
		liver (HepG2)	DNase1, H3K4me1, p300, FOXA1, H3K27ac, CTCF, Jund
		lung (IMR90)	CTCF, H3K4ac, H3K36me3, DNase1, H3K18ac, H3K27ac
		nervous (NH-A)	H3K9ac, CTCF, H3K4me2, H3K27ac, H3K36me3, DNase1
		skin (NHEK)	CTCF, H3K36me3
		NHLF	H3K27ac, CTCF, DNase1
		Osteobl	H3K36me3, CTCF, H3K27ac
		breast (HMEC)	H3K36me3, CTCF
		cervix (HeLa-S3)	Cjun, Jund, CTCF, DNase1
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	CTCF
		A549	H3K4me3, CTCF, DNase1, H3K27ac, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
TGFBI	upstream gene variant, downstream gene variant	802	ENST00000504185	?	?	6
TGFBI	upstream gene variant, downstream gene variant	530	ENST00000506699	?	?	5
TGFBI	upstream gene variant, downstream gene variant	531	ENST00000508076	?	ENSP00000423935	3
TGFBI	upstream gene variant, downstream gene variant	529	ENST00000442011	NM_000358.2	ENSP00000416330	5
TGFBI	upstream gene variant, downstream gene variant	199	ENST00000509485	?	ENSP00000426589	12
TGFBI	upstream gene variant, downstream gene variant	681	ENST00000507018	?	ENSP00000421540	5
TGFBI	upstream gene variant, downstream gene variant	974	ENST00000503087	?	ENSP00000427137	3
TGFBI	upstream gene variant, downstream gene variant	2260	ENST00000515433	?	?	11
TGFBI	upstream gene variant, downstream gene variant	118	ENST00000604555	?	ENSP00000474155	8
TGFBI	upstream gene variant, downstream gene variant	53	ENST00000509749	?	?	2
TGFBI	upstream gene variant, downstream gene variant	300	ENST00000514242	?	?	12
TGFBI	upstream gene variant, downstream gene variant	24	ENST00000508767	?	ENSP00000423871	5
TGFBI	upstream gene variant, downstream gene variant	210	ENST00000504411	?	?	13
TGFBI	upstream gene variant, downstream gene variant	528	ENST00000514554	?	ENSP00000421440	3
TGFBI	upstream gene variant, downstream gene variant	702	ENST00000513497	?	?	3

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
TGFBI	ENST00000442011	NM_000358.2	ENSP00000416330	L	ctC/ctG	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
TGFBI	ENST00000506699	?	?	20
TGFBI	ENST00000504185	?	?	5
TGFBI	ENST00000508076	?	ENSP00000423935	11
TGFBI	ENST00000442011	NM_000358.2	ENSP00000416330	20
TGFBI	ENST00000509485	?	ENSP00000426589	2
TGFBI	ENST00000507018	?	ENSP00000421540	20
TGFBI	ENST00000513497	?	?	11
TGFBI	ENST00000515433	?	?	2
TGFBI	ENST00000514554	?	ENSP00000421440	14
TGFBI	ENST00000509749	?	?	1
TGFBI	ENST00000514242	?	?	2
TGFBI	ENST00000508767	?	ENSP00000423871	2
TGFBI	ENST00000503087	?	ENSP00000427137	11
TGFBI	ENST00000604555	?	ENSP00000474155	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
TGFBI	ENST00000507018	?	ENSP00000421540	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
TGFBI	ENST00000506699	?	3
TGFBI	ENST00000515433	?	2
TGFBI	ENST00000504411	?	1