Locus 207 (rs2505535)

SNiPAcard

Block annotations

Block info

genomic range	chr10:43,552,931-43,605,902
block size	52,972 bp
variant count	42 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.800 [-2.85 – 0.801]	gene(s) hit or close-by	RET
phastCons	μ = 0.048 [0 – 0.998]	eQTL gene(s)	CSGALNACT2, RET, RP11-351D16.3, ZNF33B
GERP++	μ = -0.954 [-6.48 – 2.97]	potentially regulated gene(s)	BMS1, HNRNPF, RASGEF1A, RET, RET, RP11-168L22.2, RP11-517P14.2, ZNF239
CADD score	μ = 3.994 [0.038 – 19.65]	disease gene(s)	RET, BMS1

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Clinvar: phenotype not specified	benign	ClinVar	RCV000153833.3	1
Clinvar: phenotype not specified	benign	ClinVar	RCV000127776.1	1
Hirschsprung disease 1	risk factor	ClinVar	RCV000014980.1	1
Hirschsprung disease 1	risk factor	ClinVar	RCV000014967.1	1
?	HGMD curated	HGMD	CR110781	1
?	HGMD curated	HGMD	CR051713	1
?	HGMD curated	HGMD	CM020759	1
HIRSCHSPRUNG DISEASE, SUSCEPTIBILITY TO, 1	OMIM curated	OMIM	MIM:164761	2

Disease gene annotation

gene	trait	source DB	source entry/link
RET	RENAL AGENESIS	DECIPHER	MIM:191830
RET	Multiple endocrine neoplasia IIB	DECIPHER	MIM:162300
BMS1	APLASIA CUTIS CONGENITA, NONSYNDROMIC	OMIM	MIM:107600
RET	PHEOCHROMOCYTOMAPHEOCHROMOCYTOMA, SUSCEPTIBILITY TO	OMIM	MIM:171300
RET	CENTRAL HYPOVENTILATION SYNDROME, CONGENITAL	OMIM	MIM:209880
RET	MULTIPLE ENDOCRINE NEOPLASIA, TYPE IIA	OMIM	MIM:171400
RET	THYROID CARCINOMA, FAMILIAL MEDULLARY	OMIM	MIM:155240
RET	MULTIPLE ENDOCRINE NEOPLASIA, TYPE IIB	OMIM	MIM:162300
RET	COLORECTAL CANCER	OMIM	MIM:114500
RET	HIRSCHSPRUNG DISEASE, SUSCEPTIBILITY TO, 1	OMIM	MIM:142623
RET	THYROID CARCINOMA, PAPILLARY	OMIM	MIM:188550
BMS1	Circumscribed cutaneous aplasia of the vertex	OrphaNet	OrphaNet:1114
RET	Unilateral renal dysplasia	OrphaNet	OrphaNet:93172
RET	Bilateral renal dysplasia	OrphaNet	OrphaNet:93173
RET	Haddad syndrome	OrphaNet	OrphaNet:99803
RET	Hirschsprung disease	OrphaNet	OrphaNet:388
RET	Multiple endocrine neoplasia type 2A	OrphaNet	OrphaNet:247698
RET	Papillary or follicular thyroid carcinoma	OrphaNet	OrphaNet:146

RET	Multiple endocrine neoplasia type 2B	OrphaNet	OrphaNet:247709	orphanet
RET	Familial medullary thyroid carcinoma	OrphaNet	OrphaNet:99361	orphanet
RET	Bilateral renal agenesis	OrphaNet	OrphaNet:1848	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CSGALNACT2	?	ENSG00000169826	pancreas	9.10×10^{-8} (p-value)	GTEx Portal V6	34
CSGALNACT2	?	ENSG00000169826	lung	1.31×10^{-8} (p-value)	GTEx Portal V6	38
RET	?	ENSG00000165731	lung	4.00×10^{-8} (p-value)	GTEx Portal V6	39
CSGALNACT2	?	ENSG00000169826	tibial nerve	3.21×10^{-23} (p-value)	GTEx Portal V6	42
RET	?	ENSG00000165731	tibial nerve	8.42×10^{-9} (p-value)	GTEx Portal V6	40
RP11-351D16.3	?	ENSG00000273008	tibial nerve	2.66×10^{-6} (p-value)	GTEx Portal V6	5
CSGALNACT2	?	ENSG00000169826	breast	1.76×10^{-7} (p-value)	GTEx Portal V6	26
RP11-351D16.3	?	ENSG00000273008	tibial artery	1.56×10^{-6} (p-value)	GTEx Portal V6	18
CSGALNACT2	?	ENSG00000169826	tibial artery	5.33×10^{-6} (p-value)	GTEx Portal V6	12
CSGALNACT2	?	ENSG00000169826	pituitary	3.52×10^{-8} (p-value)	GTEx Portal V6	35
CSGALNACT2	?	ENSG00000169826	thyroid	7.04×10^{-11} (p-value)	GTEx Portal V6	41
CSGALNACT2	?	ENSG00000169826	testis	3.34×10^{-7} (p-value)	GTEx Portal V6	30
CSGALNACT2	?	ENSG00000169826	ovary	4.00×10^{-9} (p-value)	GTEx Portal V6	38
CSGALNACT2	?	ENSG00000169826	left ventricle	1.61×10^{-7} (p-value)	GTEx Portal V6	39
RET	?	ENSG00000165731	subcutaneous adipocytes	2.81×10^{-7} (p-value)	GTEx Portal V6	19
ZNF33B	?	ENSG00000196693	subcutaneous adipocytes	1.49×10^{-5} (p-value)	GTEx Portal V6	1
RP11-351D16.3	?	ENSG00000273008	subcutaneous adipocytes	8.39×10^{-6} (p-value)	GTEx Portal V6	9
CSGALNACT2	?	ENSG00000169826	subcutaneous adipocytes	1.48×10^{-6} (p-value)	GTEx Portal V6	22
CSGALNACT2	?	ENSG00000169826	muscularis mucosae	1.73×10^{-6} (p-value)	GTEx Portal V6	26
CSGALNACT2	?	ENSG00000169826	adrenal gland	3.70×10^{-6} (p-value)	GTEx Portal V6	14
CSGALNACT2	?	ENSG00000169826	atrial appendage	7.94×10^{-6} (p-value)	GTEx Portal V6	1
?	?	ILMN_1889106	blood	1.29×10^{-4} (p-value)	Westra et al.	2
RP11-351D16.3	?	ENSG00000273008	sun exposed skin	1.65×10^{-5} (p-value)	GTEx Portal V6	2
CSGALNACT2	?	ENSG00000169826	anterior cingulate cortex	5.75×10^{-7} (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000008227		RET

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000072662	1	ENCP00000008233	RASGEF1A
		ENCP00000008238	HNRNPF
		ENCP00000008242	RP11-517P14.2
		ENCP00000008224	BMS1
		ENCP00000008227	RET RET
		ENCP00000008239	HNRNPF
ENCE00000072664	1	ENCP00000008233	RASGEF1A
		ENCP00000008238	HNRNPF
		ENCP00000008224	BMS1
		ENCP00000008227	RET RET
		ENCP00000008239	HNRNPF
		ENCP00000008241	HNRNPF
		ENCP00000008242	RP11-517P14.2
		ENCP00000008234	RASGEF1A
ENCE00000072665	1	ENCP00000008241	HNRNPF
		ENCP00000008233	RASGEF1A
		ENCP00000008238	HNRNPF
		ENCP00000008242	RP11-517P14.2
		ENCP00000008224	BMS1
		ENCP00000008227	RET RET
		ENCP00000008239	HNRNPF
ENCE00000072668	1	ENCP00000008233	RASGEF1A
		ENCP00000008238	HNRNPF
		ENCP00000008224	BMS1
		ENCP00000008227	RET RET
		ENCP00000008239	HNRNPF
		ENCP00000008241	HNRNPF
		ENCP00000008235	RP11-168L22.2
		ENCP00000008242	RP11-517P14.2
		ENCP00000008234	RASGEF1A
ENCE00000072933	1	ENCP00000008246	ZNF239

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001422878 (CTCF binding site)	1	cervix (HeLa-S3)	H3K27me3
		embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3, Rad21
ENSR00000115942 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, DNase1
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K36me3, H3K27me3
		liver (HepG2)	CTCF, H3K27me3
		lung (IMR90)	H3K27me3
		A549	H3K27me3

		nervous (NH-A)	H3K27me3
		skin (NHEK)	CTCF
ENSR00000354005 (TF binding site)	1	embryonic stem cell (H1ESC)	DNase1, H3K4me3, H3K4me2, H3K27me3, Rad21
		endothelium (HUVEC)	H3K27me3, DNase1
		lung (IMR90)	H3K27me3
		A549	H3K27me3, H3K4me3
		blood (K562)	H3K27me3, DNase1
		breast (HMEC)	H3K4me2
ENSR00000354008 (promoter flanking region)	3	embryonic stem cell (H1ESC)	USF1
		HSMMtube	DNase1, H3K27ac
		blood (K562)	H3K27me3, USF1
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	H3K36me3, DNase1
		cervix (HeLa-S3)	DNase1
		liver (HepG2)	USF1, H3K27me3
		blood (GM12878)	USF1
		skin (NHEK)	CTCF, H3K4me1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RET	upstream gene variant	487	ENST00000355710	NM_020975.4	ENSP00000347942	3
RET	upstream gene variant	15	ENST00000498820	?	ENSP00000419080	4
RET	upstream gene variant	10	ENST00000615310	?	ENSP00000480088	4
RET	upstream gene variant	20	ENST00000340058	NM_020630.4	ENSP00000344798	4
RET	upstream gene variant, downstream gene variant	1235	ENST00000479913	?	?	10

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
RET	ENST00000340058	NM_020630.4	ENSP00000344798	A	gcA/gcG	1
RET	ENST00000615310	?	ENSP00000480088	A	gcA/gcG	1
RET	ENST00000355710	NM_020975.4	ENSP00000347942	A	gcA/gcG	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
RET	ENST00000340058	NM_020630.4	ENSP00000344798	33
RET	ENST00000615310	?	ENSP00000480088	33
RET	ENST00000355710	NM_020975.4	ENSP00000347942	33
RET	ENST00000498820	?	ENSP00000419080	33
RET	ENST00000479913	?	?	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
RET	ENST00000355710	NM_020975.4	ENSP00000347942	1