Locus 294 (rs2795502)

SNiPAcard

Block annotations

Block info

genomic range	chr10:43,337,842-43,423,626
block size	85,785 bp
variant count	54 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.516 [-3.961 – 2.125]	gene(s) hit or close-by	RP11-124O11.1
phastCons	μ = 0.078 [0 – 0.766]	eQTL gene(s)	ANKRA2
GERP++	μ = -0.233 [-6.18 – 1.58]	potentially regulated gene(s)	BMS1, CCNYL2, HNRNPF, RASGEF1A, RET, RET, RP11-168L22.2, RP11-517P14.2
CADD score	μ = 2.663 [0.012 – 11.37]	disease gene(s)	RET, BMS1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Progressive supranuclear palsy	<3.00×10-7	GWAS Catalog	21685912	1

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
RET	Familial medullary thyroid carcinoma	OrphaNet	OrphaNet:99361
RET	Bilateral renal agenesis	OrphaNet	OrphaNet:1848

Direct effect on regulation

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)

ANKRA2	ENST00000506130	ILMN_1687351	chr5	blood	3.54×10-7 (p-value)	Westra et al.	1
ANKRA2	ENST00000504641						
ANKRA2	ENST00000515804						
ANKRA2	ENST00000514916						
ANKRA2	ENST00000296785						

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000072560	1	ENCP00000008233	RASGEF1A
		ENCP00000008238	HNRNPF
		ENCP00000008224	BMS1
		ENCP00000008227	RET RET
		ENCP00000008239	HNRNPF
		ENCP00000008241	HNRNPF
		ENCP00000008235	RP11-168L22.2
		ENCP00000008242	RP11-517P14.2
		ENCP00000008212	CCNYL2
		ENCP00000008234	RASGEF1A

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000353984 (CTCF binding site)	2	endothelium (HUVEC)	H3K36me3, H3K27me3
		embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	H3K27me3
		liver (HepG2)	CTCF
		blood (K562)	CTCF, H3K27me3
ENSR00000353986 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, DNase1
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3, CTCF
		blood (DND-41)	H3K27me3
		breast (HMEC)	CTCF
		muscle (HSMM)	CTCF
		cervix (HeLa-S3)	CTCF, H3K27me3
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K36me3, CTCF, H3K27me3
		liver (HepG2)	CTCF, H3K27me3
		lung (IMR90)	H3K27me3, CTCF
		A549	CTCF, H3K27me3
		nervous (NH-A)	H3K27me3
		skin (NHEK)	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RP11-124O11.1	upstream gene variant	2463	ENST00000421320	?	?	1