Locus 414 (rs2506028)

SNiPAcard

Block annotations

Block info

genomic range	chr10:43,435,628-43,447,847
block size	12,220 bp
variant count	12 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.714 [-2.896 – 0.183]	gene(s) hit or close-by	–
phastCons	μ = 0.058 [0 – 0.377]	eQTL gene(s)	RET
GERP++	μ = -2.080 [-8.59 – 0.481]	potentially regulated gene(s)	AL022344.5, CCNYL2, RET, RET, RP11-124O11.1
CADD score	μ = 3.098 [0.535 – 14.03]	disease gene(s)	RET

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
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
RET	RENAL AGENESIS	DECIPHER	MIM:191830
RET	Multiple endocrine neoplasia IIB	DECIPHER	MIM:162300
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

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RET	?	ENSG00000165731	unexposed skin	1.48×10-7 (p-value)	GTEx Portal V6	12

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)

ENCE00000072563	1	ENCP00000008221	AL022344.5
		ENCP00000008225	RP11-124O11.1
		ENCP00000008213	CCNYL2
		ENCP00000008212	CCNYL2
		ENCP00000008227	RET RET

Locus 414 (sentinel SNP rs2506028 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.