Locus 217 (rs6750059)

SNiPAcard

Block annotations

Block info

genomic range	chr2:109,594,969-109,613,765
block size	18,797 bp
variant count	12 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.183 [-1.417 – 2.379]	gene(s) hit or close-by	EDAR
phastCons	μ = 0.081 [0 – 0.895]	eQTL gene(s)	SULT1C3
GERP++	μ = -0.253 [-6.15 – 4.2]	potentially regulated gene(s)	EDAR, LIMS1
CADD score	μ = 5.145 [0.276 – 14.27]	disease gene(s)	EDAR

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
EDAR	HAIR MORPHOLOGY 1	OMIM	MIM:612630
EDAR	ECTODERMAL DYSPLASIA 10B, HYPOHIDROTIC/HAIR/TOOTH TYPE, [...]	OMIM	MIM:224900
EDAR	ECTODERMAL DYSPLASIA 10A, HYPOHIDROTIC/HAIR/NAIL TYPE, AUTOSOMAL [...]	OMIM	MIM:129490
EDAR	Autosomal dominant hypohidrotic ectodermal dysplasia	OrphaNet	OrphaNet:1810
EDAR	Autosomal recessive hypohidrotic ectodermal dysplasia	OrphaNet	OrphaNet:248

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SULT1C3	ENST00000329106	ILMN_1653463	b-cell	3.65×10-4 (p-value)	Fairfax et al.	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000258714	1	ENCP00000027465	EDAR
		ENCP00000027450	LIMS1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
EDAR	upstream gene variant	4040	ENST00000409271	?	ENSP00000386371	2
EDAR	upstream gene variant	3937	ENST00000376651	?	ENSP00000365839	2
EDAR	upstream gene variant	3937	ENST00000258443	NM_022336.3	ENSP00000258443	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
EDAR	ENST00000409271	?	ENSP00000386371	8
EDAR	ENST00000376651	?	ENSP00000365839	8
EDAR	ENST00000258443	NM_022336.3	ENSP00000258443	8