Locus 234 (rs260692)

SNiPAcard

Block annotations

Block info

genomic range	chr2:109,574,150-109,581,319
block size	7,170 bp
variant count	14 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.658 [-2.622 – 1.528]	gene(s) hit or close-by	EDAR
phastCons	μ = 0.068 [0 – 0.9]	eQTL gene(s)	–
GERP++	μ = -0.616 [-5.82 – 4.76]	potentially regulated gene(s)	–
CADD score	μ = 3.340 [0.472 – 9.224]	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

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000596156 (enhancer)	1	HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		cervix (HeLa-S3)	DNase1
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K27me3
		lung (IMR90)	H3K27me3
		skin (NHEK)	H3K4me1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
EDAR	ENST00000409271	?	ENSP00000386371	14
EDAR	ENST00000376651	?	ENSP00000365839	14
EDAR	ENST00000258443	NM_022336.3	ENSP00000258443	14