

## Block annotations

### Block info

genomic range	chr4:55,972,946-55,972,946 <i>e!</i>
block size	1 bp
variant count	1 variants

### Basic features

Conservation/deleteriousness		Linked genes	
phyloP	6.349	gene(s) hit or close-by	KDR <i>e!</i>
phastCons	1	eQTL gene(s)	-
GERP++	5.67	potentially regulated gene(s)	-
CADD score	25.4	disease gene(s)	KDR <i>e!</i>

## Trait annotations

### Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Clinvar: phenotype not specified	not provided	ClinVar	RCV000121295.1 <i>ClinVar</i>	1
Hemangioma, capillary infantile, susceptibility to	risk factor	ClinVar	RCV000013111.1 <i>ClinVar</i>	1
?	HGMD curated	HGMD	CM087498 <i>HGMD</i>	1
HEMANGIOMA, CAPILLARY INFANTILE, SUSCEPTIBILITY TO	OMIM curated	OMIM	MIM:191306 <i>OMIM</i>	1

### Disease gene annotation

gene	trait	source DB	source entry/link
KDR <i>e!</i>	HEMANGIOMA, CAPILLARY INFANTILE	OMIM	MIM:602089 <i>OMIM</i>
KDR <i>e!</i>	Familial capillary hemangioma	OrphaNet	OrphaNet:91415 <i>orphanet</i>

## Direct effect on transcript

### Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
KDR <i>e!</i>	missense variant	ENST00000263923 <i>e!</i>	NM_002253.2	ENSP00000263923	R/C	Cgt/Tgt	?	?	1

## Putative effect on transcript

### Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
KDR <i>e!</i>	ENST00000512566 <i>e!</i>	?	1

