# Locus 186 (rs34231037)

SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| genomic range | chr4:55,972,946-55,972,946 |
| block size | 1 bp |
| variant count | 1 variants |

### Basic features

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

## Trait annotations

### Variant annotation

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

### Disease gene annotation

| gene | trait | source DB | source entry/link |
|---|---|---|---|
| KDR | HEMANGIOMA, CAPILLARY INFANTILE | OMIM | MIM:602089 |
| KDR | Familial capillary hemangioma | OrphaNet | OrphaNet:91415 |

## 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 | missense variant | ENST00000263923 | 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 | ENST00000512566 | ? | 1 |