# Locus 363 (rs858267)

SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| genomic range | chr7:23,206,329-23,321,521 |
| block size | 115,193 bp |
| variant count | 33 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| phyloP | μ = -0.086 [-3.04 – 1.42] | gene(s) hit or close-by | GPNMB, KLHL7 |
| phastCons | μ = 0.123 [0 – 0.996] | eQTL gene(s) | AC005082.12 |
| GERP++ | μ = -0.219 [-6.54 – 3.09] | potentially regulated gene(s) | GPNMB, TOMM7 |
| CADD score | μ = 5.205 [0.224 – 17.15] | disease gene(s) | KLHL7 |

## Trait annotations

### Disease gene annotation

| gene | trait | source DB | source entry/link |
|---|---|---|---|
| KLHL7 | RETINITIS PIGMENTOSA 42 | OMIM | MIM:612943 |
| KLHL7 | Retinitis pigmentosa | OrphaNet | OrphaNet:791 |

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| AC005082.12 | ? | ENSG00000226816 | blood | $3.64\times10^{-11}$ (p-value) | GTEx Portal V6 | 29 |
| AC005082.12 | ? | ENSG00000226816 | subcutaneous adipocytes | $5.45\times10^{-8}$ (p-value) | GTEx Portal V6 | 8 |

## Putative effect on regulation

### FANTOM5 expressed promoter

| SNiPA promoter id | variant(s) | associated transcript(s) | gene |
|---|---|---|---|
| FFCP00000781966 | 1 | ENST00000381990, ENST00000258733, ENST00000465673 | GPNMB |
| FFCP00000781967 | 1 | ENST00000381990, ENST00000258733, ENST00000465673 | GPNMB |

### ENCODE promoter-associated distal DHS (Enhancer)

| SNiPA enhancer id | variant(s) | associated SNiPA promoter id | associated gene(s) |
|---|---|---|---|
| ENCE00000476393 | 1 | ENCP00000051286 | TOMM7 |

### Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|---|---|---|---|
| ENSR00000065469 (promoter flanking region) | 2 | embryonic stem cell (H1ESC) | DNase1, H3K27me3, H3K4me2, USF1 |
| | | HSMMtube | DNase1, H3K27me3 |
| | | blood (K562) | H3K27me3 |
| | | skin (NHDF-AD) | H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac |
| | | muscle (HSMM) | DNase1 |
| | | liver (HepG2) | H3K4me2, H3K27me3 |
| | | lung (IMR90) | H3K27me3 |

| | | | |
|---|---|---|---|
| | | blood (GM12878) | H3K4me2, H3K27me3, PU1, H2AZ |
| | | nervous (NH-A) | DNase1, H3K4me2, H3K27me3 |
| | | skin (NHEK) | DNase1, H3K4me1, H3K4me2, H3K4me3, H3K36me3 |
| | | NHLF | H3K4me3, H3K27me3, DNase1 |
| | | Osteobl | H3K4me2, H2AZ, H3K27ac, H3K4me3 |
| | | blood (DND-41) | H3K27me3 |
| | | breast (HMEC) | H3K4me2, H3K4me3, DNase1 |
| | | monocytes (Monocytes-CD14+) | DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3 |
| | | endothelium (HUVEC) | H3K27me3 |
| | | A549 | H3K27me3 |
| ENSR00001558007 (promoter flanking region) | 1 | HSMMtube | DNase1, H3K27me3 |
| | | Osteobl | H3K4me2 |
| | | blood (DND-41) | H3K27me3 |
| | | blood (K562) | H3K27me3 |
| | | skin (NHDF-AD) | H3K36me3 |
| | | muscle (HSMM) | DNase1 |
| | | monocytes (Monocytes-CD14+) | H3K27me3 |
| | | endothelium (HUVEC) | H3K27me3 |
| | | lung (IMR90) | H3K27me3 |
| | | A549 | H3K27me3 |
| | | nervous (NH-A) | H3K27me3 |
| | | skin (NHEK) | H3K36me3 |
| ENSR00000065473 (CTCF binding site) | 1 | embryonic stem cell (H1ESC) | Rad21, CTCF, DNase1 |
| | | HSMMtube | CTCF |
| | | blood (K562) | DNase1, Rad21, CTCF |
| | | skin (NHDF-AD) | CTCF |
| | | muscle (HSMM) | CTCF |
| | | liver (HepG2) | CTCF, Rad21 |
| | | lung (IMR90) | CTCF |
| | | blood (GM12878) | Rad21, CTCF |
| | | nervous (NH-A) | CTCF |
| | | skin (NHEK) | H3K36me3, CTCF |
| | | NHLF | CTCF |
| | | Osteobl | CTCF |
| | | blood (DND-41) | CTCF |
| | | breast (HMEC) | CTCF |
| | | cervix (HeLa-S3) | CTCF |
| | | monocytes (Monocytes-CD14+) | CTCF |
| | | endothelium (HUVEC) | H3K36me3, CTCF |
| | | A549 | CTCF |

## Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| GPNMB | downstream gene variant | 1404 | ENST00000409458 | ? | ENSP00000386476 | 3 |
| GPNMB | downstream gene variant | 786 | ENST00000459927 | ? | ? | 3 |
| GPNMB | downstream gene variant | 889 | ENST00000474157 | ? | ? | 2 |
| GPNMB | downstream gene variant | 926 | ENST00000492858 | ? | ? | 3 |
| GPNMB | upstream gene variant, downstream gene variant | 1782 | ENST00000492512 | ? | ? | 4 |
| GPNMB | downstream gene variant | 43 | ENST00000487890 | ? | ? | 4 |
| GPNMB | downstream gene variant | 778 | ENST00000465673 | ? | ? | 3 |
| GPNMB | upstream gene variant, downstream gene variant | 1455 | ENST00000478451 | ? | ? | 5 |
| GPNMB | downstream gene variant | 1077 | ENST00000381990 | NM_001005340.1 | ENSP00000371420 | 2 |
| GPNMB | upstream gene variant, downstream gene variant | 25 | ENST00000468723 | ? | ? | 6 |
| GPNMB | upstream gene variant, downstream gene variant | 223 | ENST00000479625 | ? | ? | 6 |
| GPNMB | downstream gene variant | 1076 | ENST00000258733 | NM_002510.2 | ENSP00000258733 | 2 |
| GPNMB | upstream gene variant, downstream gene variant | 650 | ENST00000470994 | ? | ? | 6 |
| GPNMB | upstream gene variant, downstream gene variant | 323 | ENST00000463011 | ? | ? | 6 |

| | | | | | | |
|---|---|---|---|---|---|---|
| GPNMB | upstream gene variant, downstream gene variant | 323 | ENST00000463011 | ? | ? | 6 |
| KLHL7 | downstream gene variant | 625 | ENST00000469576 | ? | ? | 10 |
| KLHL7 | upstream gene variant, downstream gene variant | 885 | ENST00000469845 | ? | ? | 11 |

## Putative effect on transcript

### Synonymous coding variant

| gene | affected transcript | RefSeq id | protein | AA's | exchanged codons | variant(s) |
|---|---|---|---|---|---|---|
| GPNMB | ENST00000409458 | ? | ENSP00000386476 | D | gaT/gaC | 1 |
| GPNMB | ENST00000258733 | NM_002510.2 | ENSP00000258733 | D | gaT/gaC | 1 |
| GPNMB | ENST00000381990 | NM_001005340.1 | ENSP00000371420 | D | gaT/gaC | 1 |

### Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| GPNMB | ENST00000409458 | ? | ENSP00000386476 | 4 |
| GPNMB | ENST00000487890 | ? | ? | 3 |
| GPNMB | ENST00000258733 | NM_002510.2 | ENSP00000258733 | 14 |
| GPNMB | ENST00000479625 | ? | ? | 3 |
| GPNMB | ENST00000492858 | ? | ? | 3 |
| GPNMB | ENST00000492512 | ? | ? | 1 |
| GPNMB | ENST00000381990 | NM_001005340.1 | ENSP00000371420 | 14 |
| GPNMB | ENST00000459927 | ? | ? | 2 |
| GPNMB | ENST00000470994 | ? | ? | 3 |
| GPNMB | ENST00000465673 | ? | ? | 5 |
| KLHL7 | ENST00000521082 | ? | ENSP00000430351 | 12 |
| KLHL7 | ENST00000469576 | ? | ? | 2 |
| KLHL7 | ENST00000339077 | NM_001031710.2 | ENSP00000343273 | 12 |
| KLHL7 | ENST00000409689 | NM_018846.4 | ENSP00000386263 | 12 |

### 3'-UTR variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| GPNMB | ENST00000409458 | ? | ENSP00000386476 | 1 |
| GPNMB | ENST00000258733 | NM_002510.2 | ENSP00000258733 | 2 |
| GPNMB | ENST00000381990 | NM_001005340.1 | ENSP00000371420 | 2 |
| KLHL7 | ENST00000521082 | ? | ENSP00000430351 | 1 |
| KLHL7 | ENST00000339077 | NM_001031710.2 | ENSP00000343273 | 1 |
| KLHL7 | ENST00000409689 | NM_018846.4 | ENSP00000386263 | 1 |

### Non-coding exon variant

| gene | affected transcript | RefSeq id | variant(s) |
|---|---|---|---|
| GPNMB | ENST00000478451 | ? | 1 |
| GPNMB | ENST00000492858 | ? | 1 |
| GPNMB | ENST00000465673 | ? | 1 |