# Locus 242 (rs9468569)

SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| genomic range | chr6:29,613,377-29,671,356 |
| block size | 57,980 bp |
| variant count | 27 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| phyloP | μ = -0.431 [-3.465 – 1.743] | gene(s) hit or close-by | MOG, ZDHHC20P1, ZFP57 |
| phastCons | μ = 0.044 [0 – 0.4] | eQTL gene(s) | – |
| GERP++ | μ = 0.146 [-1.75 – 3.54] | potentially regulated gene(s) | GABBR1, GABBR1, GABBR1, GABBR1, GABBR1, GABBR1, GABBR1, HCG4P5, HCG4P5, HCG4P5, HCG4P5, HCG4P5, HCG4P5, HCG4P5, HLA-A, HLA-A, HLA-A, HLA-A, HLA-A, HLA-A, HLA-A, HLA-A, HLA-F, HLA-F, HLA-F, HLA-F, HLA-F, HLA-F, HLA-F, HLA-H, HLA-H, HLA-H, HLA-H, HLA-H, HLA-H, HLA-H, HLA-H, MOG, MOG, MOG, MOG, MOG, MOG, MOG, MOG, PPP1R11, PPP1R11, PPP1R11, PPP1R11, PPP1R11, PPP1R11, PPP1R11, PPP1R11, ZNRD1-AS1 |
| CADD score | μ = 2.954 [0.116 – 10.4] | disease gene(s) | HLA-A, MOG, ZFP57, , , , , , , , |

## Trait annotations

### Disease gene annotation

| gene | trait | source DB | source entry/link | |
|---|---|---|---|---|
| HLA-A | Birdshot chorioretinopathy | OrphaNet | OrphaNet:179 | orphanet |
| MOG | Narcolepsy-cataplexy | OrphaNet | OrphaNet:2073 | orphanet |
| ZFP57 | Transient neonatal diabetes mellitus | OrphaNet | OrphaNet:99886 | orphanet |
| MOG | NARCOLEPSY 7 | OMIM | MIM:614250 | OMIM |
| | NARCOLEPSY 7 | OMIM | MIM:614250 | OMIM |
| | NARCOLEPSY 7 | OMIM | MIM:614250 | OMIM |
| | NARCOLEPSY 7 | OMIM | MIM:614250 | OMIM |
| ZFP57 | DIABETES MELLITUS, TRANSIENT NEONATAL, 1 | OMIM | MIM:601410 | OMIM |
| | NARCOLEPSY 7 | OMIM | MIM:614250 | OMIM |
| | NARCOLEPSY 7 | OMIM | MIM:614250 | OMIM |
| | NARCOLEPSY 7 | OMIM | MIM:614250 | OMIM |
| | NARCOLEPSY 7 | OMIM | MIM:614250 | OMIM |
| ZFP57 | Diabetes Mellitus, 6q24-Related Transient Neonatal | DECIPHER | MIM:601410 | OMIM |

## Putative effect on regulation

### FANTOM5 expressed promoter

| SNiPA promoter id | variant(s) | associated transcript(s) | gene |
|---|---|---|---|
| FFCP00000735239 | 1 | ENST00000376898, ENST00000494692, ENST00000376917, ENST00000490427, ENST00000376888, ENST00000396704, ENST00000416766, ENST00000431798, ENST00000483013, ENST00000376894, ENST00000396701 | MOG |

### ENCODE promoter-associated DHS

| SNiPA promoter id | variant(s) | associated gene(s) |
|---|---|---|
| ENCP00000047827 | | MOG |
| | | MOG |

| | | | |
|---|---|---|---|
| | | | MOG |
| | | | MOG |
| | | | MOG |
| | | | MOG |
| | | | MOG |

## ENCODE promoter-associated distal DHS (Enhancer)

| SNiPA enhancer id | variant(s) | associated SNiPA promoter id | associated gene(s) |
|---|---|---|---|
| ENCE00000444016 | 1 | ENCP00000047860 | HLA-A<br>HLA-A<br>HLA-A<br>HLA-A<br>HLA-A<br>HLA-A<br>HLA-A<br>HLA-A |
| | | ENCP00000047832 | HLA-F<br>HLA-F<br>HLA-F<br>HLA-F<br>HLA-F<br>HLA-F<br>HLA-F |
| | | ENCP00000047875 | PPP1R11<br>PPP1R11<br>PPP1R11<br>PPP1R11<br>PPP1R11<br>PPP1R11<br>PPP1R11<br>PPP1R11 |
| | | ENCP00000047871 | ZNRD1-AS1 |
| | | ENCP00000047861 | HCG4P5<br>HCG4P5<br>HCG4P5<br>HCG4P5<br>HCG4P5<br>HCG4P5<br>HCG4P5<br>HCG4P5 |
| | | ENCP00000047851 | HLA-H<br>HLA-H<br>HLA-H<br>HLA-H<br>HLA-H<br>HLA-H<br>HLA-H<br>HLA-H |
| | | ENCP00000047876 | PPP1R11<br>PPP1R11<br>PPP1R11<br>PPP1R11<br>PPP1R11<br>PPP1R11<br>PPP1R11<br>PPP1R11 |
| | | ENCP00000047824 | GABBR1<br>GABBR1<br>GABBR1<br>GABBR1<br>GABBR1<br>GABBR1<br>GABBR1 |

## Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|---|---|---|---|
| ENSR00000487538 | 2 | embryonic stem cell (H1ESC) | PolII, Rad21, CTCF, Yy1, H3K4me2, H3K4me3, DNase1 |

| | | | |
|---|---|---|---|
| (CTCF binding site) | | HSMMtube | H2AZ, H3K4me2, CTCF, DNase1 |
| | | blood (K562) | PolII, H3K4me2, CTCF, DNase1, H3K4me3, H2AZ, THAP1, H3K27me3, Yy1 |
| | | skin (NHDF-AD) | CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac |
| | | muscle (HSMM) | H2AZ, CTCF, H3K4me3, H3K4me2, DNase1 |
| | | liver (HepG2) | H3K4me3, ELF1, H2AZ, H3K4me2, CTCF, DNase1 |
| | | blood (GM12878) | H3K4me3, H3K27ac, H3K4me2, CTCF, PolII, H2AZ, DNase1, Yy1, ELF1 |
| | | lung (IMR90) | DNase1, H3K4me2, H3K4me3, CTCF |
| | | nervous (NH-A) | H3K9ac, CTCF, DNase1, H3K4me2 |
| | | skin (NHEK) | H3K4me2, CTCF, DNase1 |
| | | NHLF | DNase1, CTCF |
| | | Osteobl | CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ |
| | | blood (DND-41) | CTCF, H3K27ac, H3K4me3 |
| | | breast (HMEC) | CTCF, H3K4me2, H3K4me3, DNase1 |
| | | cervix (HeLa-S3) | H3K4me2, PolII, CTCF, DNase1 |
| | | monocytes (Monocytes-CD14+) | DNase1, CTCF, H3K4me2, H3K27ac, H3K9ac, H3K4me3 |
| | | endothelium (HUVEC) | H3K36me3, DNase1, CTCF, H3K4me2, PolII |
| | | A549 | DNase1, CTCF, H3K4me2, H3K4me3 |
| ENSR00001494451 e! (promoter flanking region) | 1 | embryonic stem cell (H1ESC) | DNase1, H3K4me1, H3K27me3, PolII, Rad21, CTCF, Yy1, H3K4me2, H3K4me3 |
| | | HSMMtube | DNase1, H2AZ, H3K4me2, CTCF |
| | | blood (K562) | Yy1, THAP1, H2AZ, PolII, H3K4me2, CTCF, DNase1, H3K4me3 |
| | | skin (NHDF-AD) | CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac |
| | | muscle (HSMM) | H2AZ, CTCF, H3K4me3, H3K4me2, DNase1 |
| | | liver (HepG2) | H3K4me3, ELF1, H2AZ, H3K4me2, CTCF, DNase1 |
| | | lung (IMR90) | DNase1, H3K4me2, H3K4me3, CTCF |
| | | blood (GM12878) | PolII, H2AZ, DNase1, Yy1, ELF1, H3K4me3, H3K27ac, H3K4me2, CTCF |
| | | nervous (NH-A) | DNase1, H3K4me2, CTCF, H3K9ac |
| | | skin (NHEK) | CTCF, DNase1, H3K4me2 |
| | | NHLF | DNase1, CTCF |
| | | Osteobl | CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ |
| | | blood (DND-41) | CTCF, H3K27ac, H3K4me3 |
| | | breast (HMEC) | DNase1, H3K4me3, H3K4me2, CTCF |
| | | cervix (HeLa-S3) | DNase1, H3K4me2, PolII, CTCF |
| | | monocytes (Monocytes-CD14+) | H3K27ac, H3K9ac, H3K4me3, H3K4me2, DNase1, CTCF |
| | | endothelium (HUVEC) | H3K36me3, H3K4me2, PolII, CTCF, DNase1 |
| | | A549 | DNase1, CTCF, H3K4me2, H3K4me3 |
| ENSR00000487541 e! (enhancer) | 1 | monocytes (Monocytes-CD14+) | H3K27me3 |
| | | embryonic stem cell (H1ESC) | H4K5ac, H3K27ac, DNase1 |
| | | endothelium (HUVEC) | H3K27me3 |
| | | HSMMtube | H3K27me3 |
| | | Osteobl | H3K27me3 |
| | | blood (K562) | H3K27me3 |
| ENSR00000487543 e! (TF binding site) | 5 | endothelium (HUVEC) | H3K27me3 |
| | | embryonic stem cell (H1ESC) | DNase1, H3K27ac, CTCF, PolII |
| | | blood (K562) | H3K27me3 |
| | | A549 | H3K27me3 |
| ENSR00000487548 e! (CTCF binding site) | 1 | embryonic stem cell (H1ESC) | Rad21, CTCF, DNase1 |
| | | HSMMtube | H3K27me3 |
| | | Osteobl | H3K27me3 |
| | | blood (K562) | H3K27me3, Rad21, CTCF |
| | | blood (DND-41) | H3K27me3 |
| | | cervix (HeLa-S3) | CTCF |
| | | endothelium (HUVEC) | H3K36me3, CTCF, H3K27me3 |
| | | skin (NHEK) | CTCF |

## Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| MOG e! | upstream gene variant, downstream gene variant | 1672 | ENST00000416766 e! | ? | ENSP00000409394 e! | 4 |
| MOG e! | upstream gene variant, downstream gene variant | 1672 | ENST00000376898 e! | NM_002433.4 | ENSP00000366095 e! | 4 |
| MOG e! | upstream gene variant, downstream gene variant | 1672 | ENST00000490427 e! | NM_001170418.1 | ENSP00000420350 e! | 4 |
| MOG e! | upstream gene variant, downstream gene variant | 1266 | ENST00000485885 e! | ? | ? | 12 |
| MOG e! | upstream gene variant, downstream gene variant | 1672 | ENST00000396704 e! | NM_001008228.2 | ENSP00000379932 e! | 4 |

| | | | | | | |
|---|---|---|---|---|---|---|
| MOG | upstream gene variant, downstream gene variant | 1443 | ENST00000376917 | NM_206809.3 | ENSP00000366115 | 4 |
| MOG | upstream gene variant, downstream gene variant | 259 | ENST00000376889 | ? | ENSP00000366086 | 6 |
| MOG | upstream gene variant, downstream gene variant | 1554 | ENST00000376894 | ? | ENSP00000366091 | 4 |
| MOG | upstream gene variant, downstream gene variant | 259 | ENST00000485211 | ? | ENSP00000418872 | 6 |
| MOG | upstream gene variant, downstream gene variant | 1338 | ENST00000476244 | ? | ? | 10 |
| MOG | upstream gene variant, downstream gene variant | 1672 | ENST00000483013 | ? | ENSP00000418090 | 4 |
| MOG | upstream gene variant, downstream gene variant | 1592 | ENST00000469353 | ? | ? | 2 |
| MOG | upstream gene variant, downstream gene variant | 921 | ENST00000376903 | ? | ENSP00000366101 | 9 |
| MOG | upstream gene variant, downstream gene variant | 1465 | ENST00000376888 | NM_206814.5 | ENSP00000366085 | 4 |
| MOG | upstream gene variant, downstream gene variant | 1672 | ENST00000376891 | NM_001008229.2 | ENSP00000366088 | 4 |
| MOG | upstream gene variant, downstream gene variant | 1672 | ENST00000431798 | NM_206812.3 | ENSP00000410866 | 4 |
| MOG | upstream gene variant, downstream gene variant | 598 | ENST00000469603 | ? | ? | 10 |
| MOG | upstream gene variant, downstream gene variant | 1672 | ENST00000396701 | NM_206810.3 | ENSP00000379929 | 4 |
| MOG | upstream gene variant, downstream gene variant | 1672 | ENST00000494692 | NM_206811.3 | ENSP00000417405 | 4 |
| ZDHHC20P1 | downstream gene variant | 4546 | ENST00000435787 | ? | ? | 2 |
| ZFP57 | downstream gene variant, upstream gene variant | 3541 | ENST00000488757 | NM_001109809.2 | ENSP00000418259 | 8 |
| ZFP57 | downstream gene variant, upstream gene variant | 3649 | ENST00000376881 | ? | ENSP00000366078 | 4 |
| ZFP57 | downstream gene variant, upstream gene variant | 288 | ENST00000376883 | ? | ENSP00000366080 | 5 |

## Putative effect on transcript

### Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| MOG | ENST00000396701 | NM_206810.3 | ENSP00000379929 | 12 |
| MOG | ENST00000490427 | NM_001170418.1 | ENSP00000420350 | 12 |
| MOG | ENST00000376888 | NM_206814.5 | ENSP00000366085 | 12 |
| MOG | ENST00000396704 | NM_001008228.2 | ENSP00000379932 | 12 |
| MOG | ENST00000494692 | NM_206811.3 | ENSP00000417405 | 12 |
| MOG | ENST00000483013 | ? | ENSP00000418090 | 12 |
| MOG | ENST00000431798 | NM_206812.3 | ENSP00000410866 | 12 |
| MOG | ENST00000376903 | ? | ENSP00000366101 | 4 |
| MOG | ENST00000485211 | ? | ENSP00000418872 | 10 |
| MOG | ENST00000376891 | NM_001008229.2 | ENSP00000366088 | 12 |
| MOG | ENST00000469353 | ? | ? | 2 |
| MOG | ENST00000469603 | ? | ? | 3 |
| MOG | ENST00000416766 | ? | ENSP00000409394 | 12 |
| MOG | ENST00000376898 | NM_002433.4 | ENSP00000366095 | 12 |
| MOG | ENST00000376894 | ? | ENSP00000366091 | 12 |
| MOG | ENST00000476244 | ? | ? | 3 |
| MOG | ENST00000376889 | ? | ENSP00000366086 | 10 |
| MOG | ENST00000376917 | NM_206809.3 | ENSP00000366115 | 12 |
| ZFP57 | ENST00000376883 | ? | ENSP00000366080 | 3 |

| | | | | |
|---|---|---|---|---|
| ZFP57 | ENST00000488757 | NM_001109809.2 | ENSP00000418259 | 3 |
| ZFP57 | ENST00000376881 | ? | ENSP00000366078 | 3 |