# SNiPAcard

## Block annotations

### Block info

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|---|---|
| **genomic range** | chr19:55,562,797-55,566,258 |
| **block size** | 3,462 bp |
| **variant count** | 22 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| **phyloP** | μ = -0.703 [-2.297 – 0.425] | **gene(s) hit or close-by** | CTC-550B14.6, CTC-550B14.7, RDH13 |
| **phastCons** | μ = 0.016 [0 – 0.209] | **eQTL gene(s)** | GP6 |
| **GERP++** | μ = 0.034 [-0.765 – 0.547] | **potentially regulated gene(s)** | AC009892.10, AC009892.10, AC009892.5, AC011515.2, AC011515.2, CTC-550B14.1, FCAR, FCAR, FCAR, FCAR, FCAR, FCAR, FCAR, FCAR, FCAR, FCAR, KIR2DL1, KIR2DL1, KIR2DL1

## Direct effect on regulation

### *cis*-eQTL

| gene | transcript | probe | tissue | min(statistic) (type) | source | variant(s) |
|---|---|---|---|---|---|---|
| GP6 | ? | ENSG00000088053 | sun exposed skin | $1.22\times10^{-7}$ (p-value) | GTEx Portal V6 | 21 |
| GP6 | ? | ENSG00000088053 | unexposed skin | $1.93\times10^{-9}$ (p-value) | GTEx Portal V6 | 21 |

## Putative effect on regulation

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

| SNiPA enhancer id | variant(s) | associated SNiPA promoter id | associated gene(s) |
|---|---|---|---|
| ENCE00000233760 | 1 | ENCP00000024802 | LILRB1<br>LILRB1<br>LILRB1<br>LILRB1<br>LILRB1 |
| | | ENCP00000024807 | LILRB4<br>LILRB4<br>LILRB4<br>LILRB4<br>LILRB4 |
| | | ENCP00000024823 | KIR2DS4<br>KIR2DS4<br>KIR2DS4<br>KIR2DS4<br>KIR2DS4<br>KIR2DS4<br>KIR2DS4<br>KIR2DS4<br>KIR2DS4<br>KIR2DS4<br>KIR2DS4<br>KIR2DS4<br>KIR2DS4<br>KIR2DS4<br>KIR2DS4<br>KIR2DS4<br>KIR2DS4<br>KIR2DS4<br>KIR2DS4<br>KIR2DS4<br>KIR2DS4<br>KIR2DS4 |
| | | ENCP00000024813 | LILRP1<br>LILRP1<br>LILRP1<br>LILRP1<br>LILRP1<br>LILRP1<br>LILRP1<br>LILRP1<br>LILRP1<br>LILRP1<br>LILRP1<br>LILRP1<br>LILRP1<br>LILRP1<br>LILRP1<br>LILRP1<br>LILRP1<br>LILRP1<br>LILRP1<br>LILRP1<br>LILRP1<br>LILRP1 |

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| ENCP00000024799 | LILRA1<br>LILRA1<br>LILRA1<br>LILRA1 |
| ENCP00000024812 | AC011515.2<br>AC011515.2 |
| ENCP00000024817 | KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3 |
| ENCP00000024811 | LILRB4<br>LILRB4<br>LILRB4<br>LILRB4<br>LILRB4 |
| ENCP00000024829 | NLRP2<br>NLRP2<br>NLRP2<br>NLRP2<br>NLRP2<br>NLRP2<br>NLRP2<br>NLRP2<br>NLRP2 |
| ENCP00000024822 | KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1 |
| ENCP00000024818 | KIR2DP1<br>KIR2DP1<br>KIR2DP1<br>KIR2DP1<br>KIR2DP1<br>KIR2DP1<br>KIR2DP1<br>KIR2DP1 |

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| ENCP00000024816 | KIR3DL3 |
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| ENCP00000024803 | LILRB1 |
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| ENCP00000024808 | VN1R105P |
| ENCP00000024798 | LILRA2 |
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| ENCP00000024806 | AC009892.10 |
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| ENCP00000024824 | KIR3DL2 |
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| ENCP00000024797 | LILRA2 |
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| ENCP00000024800 | AC009892.5 |
| ENCP00000024821 | KIR2DL4 |
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| ENCP00000024825 | FCAR |
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| ENCP00000024810 | LILRB4 |
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| ENCP00000024819 | KIR2DL1 |
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| ENCE00000233761 | 1 | ENCP00000024802 | LILRB1 |
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| | | ENCP00000024807 | LILRB4 |
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| ENCP00000024799 | LILRA1<br>LILRA1<br>LILRA1<br>LILRA1 |
| ENCP00000024812 | AC011515.2<br>AC011515.2 |
| ENCP00000024817 | KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3<br>KIR2DL3 |
| ENCP00000024811 | LILRB4<br>LILRB4<br>LILRB4<br>LILRB4<br>LILRB4 |
| ENCP00000024822 | KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1<br>KIR3DL1 |
| ENCP00000024818 | KIR2DP1<br>KIR2DP1<br>KIR2DP1<br>KIR2DP1<br>KIR2DP1<br>KIR2DP1<br>KIR2DP1<br>KIR2DP1<br>KIR2DP1<br>KIR2DP1<br>KIR2DP1<br>KIR2DP1<br>KIR2DP1<br>KIR2DP1<br>KIR2DP1<br>KIR2DP1<br>KIR2DP1<br>KIR2DP1 |

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| ENCP00000024803 | LILRB1<br>LILRB1<br>LILRB1<br>LILRB1<br>LILRB1 |
| ENCP00000024827 | NLRP2<br>NLRP2<br>NLRP2<br>NLRP2<br>NLRP2<br>CTC-550B14.1<br>NLRP2<br>NLRP2<br>NLRP2<br>NLRP2<br>NLRP2 |
| ENCP00000024808 | VN1R105P |
| ENCP00000024798 | LILRA2<br>LILRA2<br>LILRA2<br>LILRA2 |
| ENCP00000024806 | AC009892.10<br>AC009892.10 |
| ENCP00000024797 | LILRA2<br>LILRA2<br>LILRA2<br>LILRA2 |
| ENCP00000024824 | KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2<br>KIR3DL2 |
| ENCP00000024800 | AC009892.5 |
| ENCP00000024821 | KIR2DL4<br>KIR2DL4<br>KIR2DL4<br>KIR2DL4<br>KIR2DL4<br>KIR2DL4<br>KIR2DL4<br>KIR2DL4<br>KIR2DL4<br>KIR2DL4<br>KIR2DL4 |

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| ENCP00000024810 | LILRB4 |
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| ENCP00000024819 | KIR2DL1 |
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## Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| CTC-550B14.6 | upstream gene variant | 56 | ENST00000585492 | ? | ? | 9 |
| CTC-550B14.7 | downstream gene variant | 3343 | ENST00000593060 | ? | ? | 3 |
| RDH13 | downstream gene variant | 1763 | ENST00000586945 | ? | ENSP00000466708 | 21 |
| RDH13 | downstream gene variant | 4363 | ENST00000587026 | ? | ENSP00000467754 | 8 |

## Putative effect on transcript

### Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| CTC-550B14.6 | ENST00000585492 | ? | ? | 13 |
| RDH13 | ENST00000589305 | ? | ? | 22 |
| RDH13 | ENST00000591960 | ? | ENSP00000468422 | 22 |
| RDH13 | ENST00000291892 | ? | ENSP00000291892 | 22 |
| RDH13 | ENST00000396247 | NM_138412.3 | ENSP00000379547 | 22 |
| RDH13 | ENST00000587721 | ? | ENSP00000467208 | 22 |
| RDH13 | ENST00000592573 | ? | ENSP00000465538 | 22 |
| RDH13 | ENST00000589605 | ? | ? | 22 |
| RDH13 | ENST00000415061 | NM_001145971.1 | ENSP00000391121 | 22 |
| RDH13 | ENST00000610356 | ? | ENSP00000477732 | 22 |

### Non-coding exon variant

| gene | affected transcript | RefSeq id | variant(s) |
|---|---|---|---|
| CTC-550B14.6 | ENST00000585492 | ? | 1 |