# Locus 162 (rs3740909)

SNiPAcard

## Block annotations

### Block info

| | |
|---|---|
| genomic range | chr11:125,882,173-125,901,248 |
| block size | 19,076 bp |
| variant count | 43 variants |

### Basic features

| Conservation/deleteriousness | | Linked genes | |
|---|---|---|---|
| phyloP | μ = -0.312 [-4.048 – 2.849] | gene(s) hit or close-by | CDON |
| phastCons | μ = 0.064 [0 – 1] | eQTL gene(s) | – |
| GERP++ | μ = -0.344 [-8.68 – 4.03] | potentially regulated gene(s) | – |
| CADD score | μ = 4.002 [0.107 – 22.5] | disease gene(s) | CDON |

## Trait annotations

### Disease gene annotation

| gene | trait | source DB | source entry/link |
|---|---|---|---|
| CDON | HOLOPROSENCEPHALY 11 | OMIM | MIM:614226 |
| CDON | Holoprosencephaly 11 | DECIPHER | MIM:614226 |
| CDON | Semilobar holoprosencephaly | OrphaNet | OrphaNet:220386 |
| CDON | Alobar holoprosencephaly | OrphaNet | OrphaNet:93925 |
| CDON | Microform holoprosencephaly | OrphaNet | OrphaNet:280200 |
| CDON | Midline interhemispheric variant of holoprosencephaly | OrphaNet | OrphaNet:93926 |
| CDON | Septopreoptic holoprosencephaly | OrphaNet | OrphaNet:280195 |
| CDON | Lobar holoprosencephaly | OrphaNet | OrphaNet:93924 |

## 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) |
|---|---|---|---|---|---|---|---|---|---|
| CDON | missense variant | ENST00000392693 | NM_001243597.1 | ENSP00000376458 | K/E | Aaa/Gaa | ? | ? | 1 |
| CDON | missense variant | ENST00000534661 | ? | ENSP00000436755 | K/E | Aaa/Gaa | ? | ? | 1 |
| CDON | missense variant | ENST00000263577 | NM_016952.4 | ENSP00000263577 | K/E | Aaa/Gaa | ? | ? | 1 |
| CDON | missense variant | ENST00000531586 | ? | ENSP00000434212 | K/E | Aaa/Gaa | ? | ? | 1 |

## Putative effect on regulation

### Regulatory feature cluster

| element id | variant(s) | tissue/cell | factors |
|---|---|---|---|
| ENSR00001608006 (enhancer) | 3 | liver (HepG2) | H3K4me1 |
| | | Osteobl | H3K27me3 |
| | | blood (DND-41) | H3K27me3 |

### Variation proximal to gene

| gene | variant type | min(distance) | transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|---|---|
| CDON | downstream gene variant, upstream gene variant | 339 | ENST00000525625 | ? | ? | 18 |

| | | | | | | |
|---|---|---|---|---|---|---|
| CDON | upstream gene variant | 244 | ENST00000531830 | ? | ENSP00000432571 | 11 |
| CDON | downstream gene variant | 56 | ENST00000527967 | ? | ENSP00000436940 | 17 |
| CDON | downstream gene variant | 1168 | ENST00000534818 | ? | ENSP00000437176 | 16 |
| CDON | downstream gene variant | 45 | ENST00000531586 | ? | ENSP00000434212 | 16 |
| CDON | upstream gene variant | 289 | ENST00000534661 | ? | ENSP00000436755 | 3 |

## Putative effect on transcript

### Intron variant

| gene | affected transcript | RefSeq id | protein | variant(s) |
|---|---|---|---|---|
| CDON | ENST00000531586 | ? | ENSP00000434212 | 16 |
| CDON | ENST00000525625 | ? | ? | 22 |
| CDON | ENST00000531830 | ? | ENSP00000432571 | 22 |
| CDON | ENST00000534661 | ? | ENSP00000436755 | 31 |
| CDON | ENST00000263577 | NM_016952.4 | ENSP00000263577 | 42 |
| CDON | ENST00000527967 | ? | ENSP00000436940 | 16 |
| CDON | ENST00000392693 | NM_001243597.1 | ENSP00000376458 | 42 |
| CDON | ENST00000534818 | ? | ENSP00000437176 | 11 |

### Non-coding exon variant

| gene | affected transcript | RefSeq id | variant(s) |
|---|---|---|---|
| CDON | ENST00000525625 | ? | 4 |