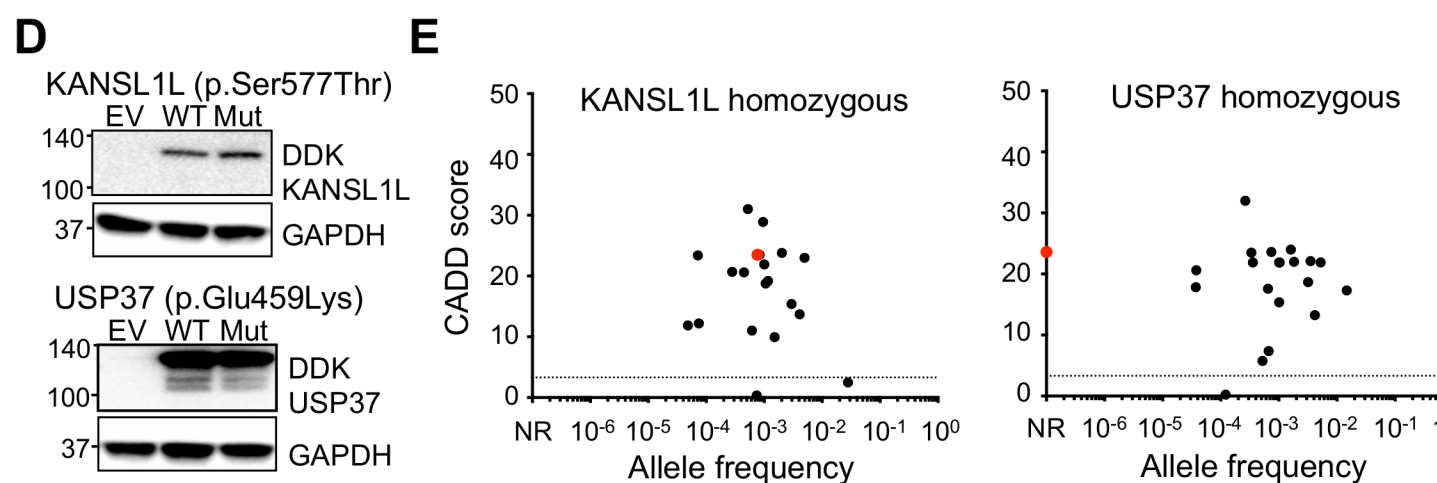


**C Rare homozygous variants within the linked region**

| Chr | Gene    | Position  | Variant | Consequence    | Global frequency | Homozygotes reported in databases (n)* | CADD   | MSC   |
|-----|---------|-----------|---------|----------------|------------------|--|--------|-------|
| 2   | CD28    | 204571471 | G/A     | G/R and Splice | Not reported     | Not reported                           | 13.790 | 9.194 |
| 2   | KANSL1L | 210940301 | C/G     | S/T            | 0.0009167        | 4                                      | 23.500 | 3.313 |
| 2   | USP37   | 219360580 | C/T     | E/K            | Not reported     | Not reported                           | 23.600 | 3.313 |

\*gnomAD, Bravo TOPMed, GME Variome and ATAVDB



**F**

|                         | KANSL1L |   |   |   |   |   |   |   |   |   | USP37 |   |   |   |   |   |   |   |   |   |   |   |
|-------------------------|---------|---|---|---|---|---|---|---|---|---|-------|---|---|---|---|---|---|---|---|---|---|---|
| Homo sapiens            | K       | L | Y | R | L | S | P | T | F | Y | W     | E | P | V | S | G | E | E | N | S | P | D |
| Pan troglodytes         | K       | L | Y | R | L | S | P | T | F | Y | W     | E | P | V | S | G | E | E | N | S | P | D |
| Gorilla gorilla         | K       | L | Y | R | L | S | P | T | F | Y | W     | E | P | V | S | G | E | E | N | S | P | D |
| Nomascus leucogenys     | K       | L | Y | R | L | S | P | T | F | Y | W     | E | P | V | L | G | E | E | N | S | P | D |
| Macaca Mulatta          | K       | L | Y | R | L | S | P | T | F | Y | W     | E | P | V | L | G | E | E | N | S | P | D |
| Rattus norvegicus       | K       | L | Y | R | L | S | P | T | F | Y | W     | E | P | V | P | G | E | E | N | S | P | D |
| Mus musculus            | K       | L | Y | R | L | S | P | T | F | Y | W     | E | P | V | L | G | E | E | N | L | P | D |
| Canis familiaris        | K       | L | Y | R | L | S | P | T | F | Y | W     | E | P | V | P | G | E | E | N | S | P | D |
| Felis catus             | K       | L | Y | R | L | S | P | T | F | Y | W     | E | P | V | P | G | E | E | T | S | S | D |
| Bos taurus              | K       | L | Y | R | L | S | P | T | F | Y | W     | E | P | V | P | G | E | E | N | S | P | D |
| Monodelphis domestica   | K       | L | Y | R | L | S | P | V | C | Y | L     | E | P | I | S | G | E | E | N | S | P | D |
| Gallus gallus           | K       | L | Y | R | M | H | G | A | C | D | W     | E | P | V | P | N | D | D | S | S | P | D |
| Danio rerio             | R       | L | F | L | M | D | R | L | S | S | G     | D | E | L | S | S | G | D | D | V | P | D |
| Drosophila melanogaster | K       | L | F | Q | T | T | N | M | H | T | I     |   |   |   |   |   |   |   |   |   |   |   |

