Table EV2. Variant Detail in the infertility Family

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| Gene | *LRRC23* | *SCN5A* | *NOX1* | *PRRG3* |
| Genome position | 12-7016610 | 3-38647444 | X-100117436 | X-150869452 |
| (Chr-Position) |
| Transcript position | NM\_001135217 | NM\_000335 | NM\_001271815 | NM\_024082 |
| c.621+1G>A | c.1336G>A | c.505T>C | c.643C>G |
| Mutation Type | Splice donor | Nonsynonymous | Nonsynonymous | Nonsynonymous |
| Mutation | c.621+1G>A | p.E446K | p.Y169H | p.P215A |
| ClinVar Allele ID | **.** | 78852 | **.** | **.** |
| dbSNP | **.** | rs199473339 | **.** | **.** |
| gnomAD MAF | 0.00003233 | 0.00003233 | **.** | **.** |
| Bravo MAF | **.** | 0.000047783 | **.** | **.** |
| MetaSVM | T | D | D | D |
| SIFT | **.** | D | **.** | D |
| PolyPhen | **.** | D | D | D |
| CADD | 26.3 | 31 | 24.4 | 24.8 |
| GTEx TPM in testis\* | 58.01 | 0.75 | 0.44 | 0.36 |
| GTEx Pext in testis# | 0.6 | 0.79 | 1 | 0.86 |
| Cosegregation  | Yes | No | No | No |
| \*GTEx TPM: The Genotype-Tissue Expression (GTEx) project V7, Transcripts Per Million. |
| #GTEx Pext: GTEx V7, Proportion EXpression across Transcripts. |