|  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **PUBLICATION** | **Ethnicity** | **GRCh38 (hg38):** | **HGVS c.**  **NM\_014844.5** | **HGVS p.**  **NM\_014844.5** | **gnomAD v2.1.1 HET** | **gnomAD v3.1.2 HET** | **gnomAD HOM** | **gnomAD**  **All AF** | **SIFT** | **Polyphen-2** | **REVEL** | **Splice AI** | **ACMG/ACGS Class** |
| **Heimer *et al*** | Ashkenazi Jewish | Chr14:102414721C>T | c.566C>T | p.(Thr189Ile) | 0 | 0 | 0 | 0 | 0.00 | 0.697 | 0.288 | 0.00 | **VUS**  PM1(supporting), PM2, PM3 |
| **Neuser *et al*** | German | Chr14:102425055G>A | c.715G>A | p.(Gly239Arg) | 21 | 6 | 0 | 0.00007424 | 0.01 | 0.995 | 0.539 | 0.370 (acceptor gain) | **VUS**  PM1(supporting), PM2, PP3 |
| ***This paper*** | Palestinian | Chr14:102425085G>A | c.745G>A | p.(Gly249Arg) | 0 | 0 | 0 | 0 | 0.00 | 1.000 | 0.574 | 0.01 (acceptor gain) | **VUS**  PM1(supporting), PM2 (supporting), PP3 |
| **Guan *et al*** | Chinese | Chr14:102434546C>T | c.1729C>T | p.(His577Tyr) | 10 | 7 | 0 | 0.00004600 | 1.00 | 0.000 | 0.109 | 0.00 | **VUS**  PM2 (supporting), BP4 |
| **Covone *et al*** | Italian | Chr14:102434867C>G | c.2050C>G | p.(Leu684Val) | 11974 | 5078 | 404 | 0.04244 | 0.21 | 0.003 | 0.014 | 0.00 | **Benign**  BS1, BS2, BP4 |
| **Covone *et al*** | Italian | Chr14:102440565C>T | c.2708C>T | p.(Thr903Met) | 10 | 2 | 0 | 0.00003980 | 0.21 | 0.796 | 0.367 | 0.00 | **VUS**  PM2 |
| **Neuser *et al*** | Saudi Arabian | Chr14:102445870G>T | c.2998G>T | p.(Asp1000Tyr) | 0 | 0 | 0 | 0 | 0.00 | 1.000 | 0.801 | 0.00 | **VUS**  PM1(supporting), PM2 (supporting), PP3 |
| **Neuser *et al*** | Emirati | Chr14:102452405T>G | c.3418T>G | p.(Trp1140Gly) | 0 | 0 | 0 | 0 | 0.01 | 0.846 | 0.643 | 0.1 (acceptor loss) | **VUS**  PM1(supporting), PM2 (supporting), PP3 |
| **Neuser *et al*** | Hispanic | Chr14:102497644C>T | c.4006C>T | p.(Arg1336Trp) | 13 | 0 | 0 | 0.00004836 | 0.00 | 1.000 | 0.608 | 0.00 | **VUS**  PM1(supporting), PM2, PP3 |
| **Neuser *et al*** | German | Chr14:102497671G>C | c.4033G>C | p.(Ala1345Pro) | 224 | 2 | 0 | 0.0008361 | 0.01 | 1.000 | 0.512 | 0.01  (donor gain) | **VUS**  PM1(supporting), PP3 |
| **Guan *et al*** | Chinese | Chr14:102498210G>A | c.4189G>A | p.(Ala1397Thr) | 20 | 7 | 0 | 0.00007251 | 0.44 | 0.000 | 0.006 | 0.00 | **VUS**  PM2 (supporting), BP4 |

**Supplementary Table S1:** *TECPR2* missense variants published to date, including American College of Medical Genetics (ACMG)/ Association for Clinical Genomic Science (ACGS) classification of variant pathogenicity confirming all missense variants as variants of uncertain significance (VUS) or benign. Abbreviations: HGVS; Human Genome Variation Society, HET; heterozygous, HOM; homozygous, AF; allele frequency.