|  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Variant | Gene Role | Gene Locus | Strand/  Transcripts | Transcript Exons/  n aa | Disease association | Variant coordinates | Exon/  Intron | Mutation type | Frequency | Clinical effect |
| **NM\_007294.4(BRCA1):c.5444G>A (p.Trp1815Ter)** |  |  |  |  |  |  |  |  |  |  |
| **NM\_000059.4(BRCA2)**  **:c.1A>G (p.Met1Val)** |  |  |  |  |  |  |  |  |  |  |
| **NM\_000021.4(PSEN1):c.806G>A (p.Arg269His)** |  |  |  |  |  |  |  |  |  |  |
| **NM\_000249.4(MLH1):c.1763T>C (p.Leu588Pro)** |  |  |  |  |  |  |  |  |  |  |
| **NM\_000249.4(MLH1):c.381-1G>A** |  |  |  |  |  |  |  |  |  |  |
| NM\_024675.4(PALB2):c.1592del (p.Leu531fs) |  |  |  |  |  |  |  |  |  |  |
| **NM\_006015.6(ARID1A):c.5299\_5301delinsCTT (p.Glu1767Leu)** |  |  |  |  |  |  |  |  |  |  |