Table S1

Variation identified at coding exons and exon-intron (+/-10 base pairs) boundaries in the proband\*.

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| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| No. | Gene | Transcript | Nucleotide change | Amino acid change | Gene locus | Heterozygosity | Rs No. | Functional change | Mutational type |
| 1 | *ATM* | NM\_000051 | c.3285-10delT | - | Intron22 | Het | rs1799757 | Splice | Benign |
| 2 | *ATM* | NM\_000051 | c.5557G>A | p.Asp1853Asn | CDS36 | Het | rs1801516 | Missense | Benign |
| 3 | *BARD1* | NM\_000465 | c.1519G>A | p.Val507Met | CDS6 | Het | rs2070094 | Missense | Benign |
| 4 | *BARD1* | NM\_000465 | c.1134G>C | p.Arg378Ser | CDS4 | Het | rs2229571 | Missense | Benign |
| 5 | *BARD1* | NM\_000465 | c.70C>T | p.Pro24Ser | CDS1 | Het | rs1048108 | Missense | Benign |
| 6 | *BRCA1* | NM\_007294 | c.4837A>G | p.Ser1613Gly | CDS14 | Het | rs1799966 | Missense | Benign |
| 7 | *BRCA1* | NM\_007294 | c.3548A>G | p.Lys1183Arg | CDS9 | Het | rs16942 | Missense | Benign |
| 8 | *BRCA1* | NM\_007294 | c.3113A>G | p.Glu1038Gly | CDS9 | Het | rs16941 | Missense | Benign |
| 9 | *BRCA1* | NM\_007294 | c.2612C>T | p.Pro871Leu | CDS9 | Het | rs799917 | Missense | Benign |
| 10 | *BRCA2* | NM\_000059 | c.1114A>C | p.Asn372His | CDS9 | Het | rs144848 | Missense | Benign |
| 11 | *BRCA2* | NM\_000059 | c.10234A>G | p.Ile3412Val | CDS26 | Het | rs1801426 | Missense | Benign |
| 12 | *BRIP1* | NM\_032043 | c.2755T>C | p.Ser919Pro | CDS18 | Hom | rs4986764 | Missense | Benign |
| 13 | *CDH1* | NM\_004360 | c.48+6C>T | - | Intron1 | Hom | rs3743674 | Splice | Benign |
| 14 | *MSH2* | NM\_000251 | c.211+9C>G | - | Intron1 | Hom | rs2303426 | Splice | Benign |
| 15 | *MSH2* | NM\_000251 | c.2006-6T>C | - | Intron12 | Hom | rs2303428 | Splice | Benign |
| 16 | *PMS2* | NM\_000535 | c.2006+6G>A | - | Intron11 | Het | rs111905775 | Splice | Benign |
| 17 | *PMS2* | NM\_000535 | c.1454C>A | p.Thr485Lys | CDS11 | Het | rs1805323 | Missense | Benign |
| 18 | *PMS2* | NM\_000535 | c.1408C>T | p.Pro470Ser | CDS11 | Het | rs1805321 | Missense | Benign |
| 19 | *PMS2* | NM\_000535 | c.706-4delT | - | Intron6 | Hom | rs549498051 | Splice | Benign |
| 20 | *STK11* | NM\_000455 | c.920+7G>C | - | Intron7 | Het | rs2075607 | Splice | Benign |
| 21 | *TP53* | NM\_000546 | c.215C>G | p.Pro72Arg | CDS3 | Het | rs1042522 | Missense | Benign |
| 22 | *BRCA1* | NM\_007294 | EX8 | - | EX8 | Het | - | Deletion | Pathogenic |

\*The proband aged at 58 received the 21-gene panel germline sequencing for hereditary breast or ovarian cancer syndrome at January 7th, 2018 after being diagnosed as ovarian cancer. The result identified a positive *BRCA1* heterozygous mutation (NM\_007294) which had been reported for its pathogenicity. The germline mutation was located in the region of EX8 by autosomal recessive inheritance resulting in exon deletion and associated amino acid change. It was studied and proved in a research by Engert S et al. (Engert S, Wappenschmidt B, Betz B, Kast K, Kutsche M, Hellebrand H, Goecke TO, Kiechle M, Niederacher D, Schmutzler RK, Meindl A. MLPA screening in the BRCA1 gene from 1,506 German hereditary breast cancer cases: novel deletions, frequent involvement of exon 17, and occurrence in single early-onset cases. Hum Mutat. 2008;29(7):948-958.) The analysis in the *ATM*, *BARD1*, *BRCA1*, *BRCA2*, *BRIP1*, *CDH1*, *MSH2*, *PMS2*, *STK11* and *TP53* revealed several benign variants. The analysis in the *CHEK2,* *PALB2*, *EPCAM*, *PTEN*, *MLH1*, *MRE11A*, *MSH6*, *MUTYH*, *NBN*, *NF1*, *PMS1*, *RAD50*, *RAD51C*, *RAD51D* and *SMARCA4* revealed no variants.