**Supplementary Table S1. Missense variants in cancer predisposition gene**

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Chr | Position | REF | ALT | | AC | GENE | c.HGVS | p.HGVS | M-LR Pred | M-LR Score | M-SVM Pred | M-SVM Score | Vest3 Score |
| 2 | 47641520 | T | C | | 1 | MSH2 | c.905T>C | p.Leu302Ser | D | 0.9417 | D | 1.0958 | 0.991 |
| 2 | 47643457 | G | A | | 1 | MSH2 | c.965G>A | p.Gly322Asp | T | 0.4851 | D | 0.0934 | 0.804 |
| 2 | 47707991 | A | G | | 1 | MSH2 | c.2615A>G | p.Lys872Arg | . | . | . | . | . |
| 2 | 48010488 | G | A | | 1 | MSH6 | c.116G>A | p.Gly39Glu | T | 0.0001 | T | -1.0974 | 0.144 |
| 2 | 48026308 | C | G | | 1 | MSH6 | c.1186C>G | p.Leu396Val | T | 0.4259 | T | -0.3029 | 0.276 |
| 2 | 48026630 | C | G | | 1 | MSH6 | c.1508C>G | p.Ser503Cys | D | 0.7495 | D | 0.6125 | 0.532 |
| 2 | 48027755 | T | C | | 1 | MSH6 | c.2633T>C | p.Val878Ala | T | 0.421 | T | -0.3685 | 0.279 |
| 2 | 48027910 | A | G | | 1 | MSH6 | c.2788A>G | p.Lys930Glu | D | 0.6125 | D | 0.0564 | 0.88 |
| 2 | 48028158 | AAAG | A | | 1 | MSH6 | c.3040\_3042delAAG | p.Lys1014del | . | . | . | . | . |
| 2 | 48030645 | C | T | | 1 | MSH6 | c.3259C>T | p.Pro1087Ser | D | 0.5847 | D | 0.0684 | 0.817 |
| 2 | 215593522 | T | C | | 1 | BARD1 | c.2212A>G | p.Ile738Val | T | 0.0176 | T | -1.0574 | 0.131 |
| 2 | 215595164 | G | A | | 1 | BARD1 | c.1972C>T | p.Arg658Cys | T | 0.077 | T | -0.8985 | 0.311 |
| 2 | 215610518 | C | T | | 1 | BARD1 | c.1738G>A | p.Glu580Lys | T | 0.1023 | T | -0.9597 | 0.055 |
| 2 | 215610571 | G | A | | 1 | BARD1 | c.1685C>T | p.Thr562Ile | T | 0.4491 | T | -0.5628 | 0.354 |
| 2 | 215617178 | C | G | | 1 | BARD1 | c.1670G>C | p.Cys557Ser | T | 0.2678 | T | -0.7955 | 0.375 |
| 2 | 215632255 | C | T | | 1 | BARD1 | c.1519G>A | p.Val507Met | T | 0 | T | -1.0971 | 0.018 |
| 2 | 215645464 | C | G | | 1 | BARD1 | c.1134G>C | p.Arg378Ser | T | 0 | T | -0.9215 | 0.294 |
| 2 | 215645502 |  |  | | 1 | BARD1 | c.1075\_1095del21 | p.Leu359\_Pro365del | . | . | . | . | . |
| 2 | 215674224 | G | A | | 1 | BARD1 | c.70C>T | p.Pro24Ser | T | 0 | T | -1.0427 | 0.109 |
| 3 | 37053550 | G | A | | 1 | MLH1 | c.637G>A | p.Val213Met | T | 0.241 | T | -0.2139 | 0.678 |
| 3 | 37053568 | A | G | | 1 | MLH1 | c.655A>G | p.Ile219Val | T | 0 | T | -1.0773 | 0.517 |
| 3 | 37089110 | T | C | | 1 | MLH1 | c.1832T>C | p.Ile611Thr | D | 0.8895 | D | 0.9765 | 0.893 |
| 3 | 37089130 | A | G | | 1 | MLH1 | c.1852A>G | p.Lys618Glu | D | 0.8885 | D | 0.8782 | 0.811 |
| 3 | 37089131 | A | C | | 1 | MLH1 | c.1853A>C | p.Lys618Thr | D | 0.8835 | D | 0.8682 | 0.927 |
| 5 | 131911535 | A | C | | 1 | RAD50 | c.280A>C | p.Ile94Leu | T | 0.0058 | T | -0.9428 | 0.223 |
| 5 | 131915673 | G | A | | 1 | RAD50 | c.671G>A | p.Arg224His | T | 0.0408 | T | -1.1491 | 0.5 |
| 5 | 131923710 | G | A | | 1 | RAD50 | c.980G>A | p.Arg327His | T | 0.0545 | T | -1.184 | 0.405 |
| 5 | 131927596 | A | G | | 2 | RAD50 | c.1663A>G | p.Ile555Val | T | 0.1918 | T | -0.845 | 0.257 |
| 7 | 6013049 | C | G | | 1 | PMS2 | c.2570G>C | p.Gly857Ala | T | 0.0003 | T | -1.1026 | 0.086 |
| 7 | 6017269 | G | A | | 1 | PMS2 | c.2395C>T | p.Arg799Trp | D | 0.8629 | D | 0.9478 | 0.95 |
| 7 | 6018236 | C | T | | 1 | PMS2 | c.2266G>A | p.Asp756Asn | T | 0.4086 | T | -0.4586 | 0.09 |
| 7 | 6018320 | T | C | | 1 | PMS2 | c.2182A>G | p.Thr728Ala | T | 0.2226 | T | -0.8766 | 0.091 |
| 7 | 6022617 | G | A | | 1 | PMS2 | c.2012C>T | p.Thr671Met | D | 0.5189 | T | -0.2292 | 0.268 |
| 7 | 6026530 | C | T | | 1 | PMS2 | c.1866G>A | p.Met622Ile | T | 0.0288 | T | -1.0187 | 0.24 |
| 7 | 6026607 | T | A | | 1 | PMS2 | c.1789A>T | p.Thr597Ser | T | 0.0256 | T | -1.0381 | 0.092 |
| 7 | 6026708 | C | A | | 1 | PMS2 | c.1688G>T | p.Arg563Leu | T | 0.0524 | T | -1.0586 | 0.686 |
| 7 | 6026775 | T | C | | 1 | PMS2 | c.1621A>G | p.Lys541Glu | T | 0 | T | -0.9487 | 0.353 |
| 7 | 6026865 | T | C | | 1 | PMS2 | c.1531A>G | p.Thr511Ala | T | 0.1508 | T | -0.8334 | 0.232 |
| 7 | 6026942 | G | T | | 1 | PMS2 | c.1454C>A | p.Thr485Lys | T | 0 | T | -1.0253 | 0.215 |
| 7 | 6026988 | G | A | | 1 | PMS2 | c.1408C>T | p.Pro470Ser | T | 0 | T | -1.0019 | 0.333 |
| 7 | 6027153 | C | T | | 1 | PMS2 | c.1243G>A | p.Val415Met | T | 0.3651 | T | -0.5348 | 0.429 |
| 7 | 6045627 | C | T | | 1 | PMS2 | c.59G>A | p.Arg20Gln | T | 0.0512 | T | -0.606 | 0.164 |
| 7 | 6045634 | T | C | | 1 | PMS2 | c.52A>G | p.Ile18Val | D | 0.7732 | D | 0.6865 | 0.727 |
| 8 | 90983460 | G | A | | 1 | NBN | c.643C>T | p.Arg215Trp | T | 0.4117 | D | 0.0544 | 0.796 |
| 8 | 90990479 | C | G | | 1 | NBN | c.553G>C | p.Glu185Gln | T | 0 | T | -0.9605 | 0.106 |
| 8 | 90993741 | T | C | | 1 | NBN | c.182A>G | p.Asp61Gly | T | 0.0388 | T | -1.0372 | 0.184 |
| 9 | 21970985 | C | G | 1 | | CDKN2A | c.373G>C | p.Asp125His | T | 0.2111 | T | -0.8577 | 0.698 |
| 9 | 21971008 | A | G | 1 | | CDKN2A | c.350T>C | p.Leu117Pro | D | 0.9112 | D | 1.0265 | 0.969 |
| 9 | 21971086 | A | T | 1 | | CDKN2A | c.272T>A | p.Leu91Gln | D | 0.9002 | D | 1.0025 | 0.919 |
| 11 | 94192599 | G | T | | 1 | MRE11A | c.1475C>A | p.Ala492Asp | T | 0.3439 | T | -0.4188 | 0.485 |
| 11 | 108098576 | C | G | | 1 | ATM | c.146C>G | p.Ser49Cys | T | 0.1138 | T | -0.9703 | 0.341 |
| 11 | 108106443 | T | A | | 1 | ATM | c.378T>A | p.Asp126Glu | T | 0.0001 | T | -1.0656 | 0.127 |
| 11 | 108119823 | T | C | | 4 | ATM | c.1229T>C | p.Val410Ala | T | 0.0108 | T | -1.0725 | 0.785 |
| 11 | 108124761 | T | C | | 1 | ATM | c.2119T>C | p.Ser707Pro | T | 0.1716 | T | -0.9444 | 0.311 |
| 11 | 108138003 | T | C | | 2 | ATM | c.2572T>C | p.Phe858Leu | T | 0.0049 | T | -0.9629 | 0.224 |
| 11 | 108139320 | C | T | | 1 | ATM | c.2822C>T | p.Ser941Phe | T | 0.2863 | T | -0.7625 | 0.41 |
| 11 | 108143456 | C | G | | 1 | ATM | c.3161C>G | p.Pro1054Arg | T | 0.3028 | T | -0.1752 | 0.443 |
| 11 | 108151744 | A | C | | 1 | ATM | c.3425A>C | p.Glu1142Ala | T | 0.2892 | T | -0.706 | 0.351 |
| 11 | 108160350 | C | T | | 1 | ATM | c.4258C>T | p.Leu1420Phe | T | 0.1289 | T | -0.8541 | 0.357 |
| 11 | 108170506 | A | C | | 1 | ATM | c.5071A>C | p.Ser1691Arg | T | 0.1507 | T | -1.0139 | 0.663 |
| 11 | 108175462 | G | A | | 2 | ATM | c.5557G>A | p.Asp1853Asn | T | 0.0019 | T | -1.0617 | 0.114 |
| 11 | 108183167 | A | G | | 1 | ATM | c.5948A>G | p.Asn1983Ser | T | 0 | T | -0.9322 | 0.063 |
| 11 | 108186556 | C | A | | 1 | ATM | c.6013C>A | p.Leu2005Ile | T | 0.0623 | T | -1.1735 | 0.668 |
| 11 | 108186610 | G | A | | 2 | ATM | c.6067G>A | p.Gly2023Arg | T | 0.0211 | T | -1.1196 | 0.889 |
| 11 | 108224555 | A | G | | 1 | ATM | c.8734A>G | p.Arg2912Gly | D | 0.7556 | D | 0.7055 | 0.998 |
| 13 | 32893271 | A | G | | 1 | BRCA2 | c.125A>G | p.Tyr42Cys | T | 0.0001 | T | -0.7612 | 0.341 |
| 13 | 32906480 | A | C | | 1 | BRCA2 | c.865A>C | p.Asn289His | T | 0.0002 | T | -0.8732 | 0.516 |
| 13 | 32906480 | A | G | | 1 | BRCA2 | c.865A>G | p.Asn289Asp | T | 0.0025 | T | -0.872 | 0.518 |
| 13 | 32906729 | A | C | | 1 | BRCA2 | c.1114A>C | p.Asn372His | T | 0 | T | -0.9508 | 0.193 |
| 13 | 32907407 | A | G | | 1 | BRCA2 | c.1792A>G | p.Thr598Ala | T | 0.0014 | T | -0.9029 | 0.807 |
| 13 | 32910456 | C | G | | 1 | BRCA2 | c.1964C>G | p.Pro655Arg | T | 0.0026 | T | -0.8896 | 0.595 |
| 13 | 32911172 | G | A | | 1 | BRCA2 | c.2680G>A | p.Val894Ile | T | 0.0803 | T | -1.017 | 0.326 |
| 13 | 32911295 | G | A | | 1 | BRCA2 | c.2803G>A | p.Asp935Asn | T | 0.1074 | T | -1.0439 | 0.399 |
| 13 | 32911295 | G | C | | 1 | BRCA2 | c.2803G>C | p.Asp935His | T | 0.1083 | T | -1.0309 | 0.699 |
| 13 | 32911463 | A | G | | 1 | BRCA2 | c.2971A>G | p.Asn991Asp | T | 0 | T | -1.0039 | 0.154 |
| 13 | 32912750 | G | T | | 1 | BRCA2 | c.4258G>T | p.Asp1420Tyr | T | 0.0025 | T | -0.9105 | 0.625 |
| 13 | 32914236 | C | T | | 1 | BRCA2 | c.5744C>T | p.Thr1915Met | T | 0.0454 | T | -1.0364 | 0.309 |
| 13 | 32929080 | G | A | | 1 | BRCA2 | c.7090G>A | p.Glu2364Lys | T | 0.2798 | T | -0.8698 | 0.739 |
| 13 | 32929387 | T | C | | 1 | BRCA2 | c.7397T>C | p.Val2466Ala | T | 0 | T | -0.9296 | 0.128 |
| 13 | 32937488 | G | T | | 1 | BRCA2 | c.8149G>T | p.Ala2717Ser | D | 0.5275 | T | -0.3119 | 0.654 |
| 13 | 32937521 | G | A | | 1 | BRCA2 | c.8182G>A | p.Val2728Ile | T | 0.1014 | T | -0.7942 | 0.811 |
| 13 | 32945172 | A | C | | 1 | BRCA2 | c.8567A>C | p.Glu2856Ala | T | 0.2119 | T | -0.5935 | 0.639 |
| 13 | 32953550 | G | A | | 1 | BRCA2 | c.8851G>A | p.Ala2951Thr | T | 0.2378 | T | -0.2327 | 0.727 |
| 13 | 32972884 | A | G | | 1 | BRCA2 | c.10234A>G | p.Ile3412Val | T | 0 | T | -0.932 | 0.065 |
| 14 | 45605457 | T | G | | 1 | FANCM | c.223T>G | p.Phe75Val | T | 0.0865 | T | -1.072 | 0.747 |
| 14 | 45605463 | A | G | | 1 | FANCM | c.229A>G | p.Thr77Ala | T | 0.0021 | T | -0.9904 | 0.04 |
| 14 | 45606287 | C | T | | 1 | FANCM | c.524C>T | p.Ser175Phe | T | 0 | T | -0.9171 | 0.064 |
| 14 | 45606290 | C | T | | 1 | FANCM | c.527C>T | p.Thr176Ile | T | 0.0261 | T | -1.0502 | 0.174 |
| 14 | 45606387 | A | G | | 1 | FANCM | c.624A>G | p.Ile208Met | T | 0.0321 | T | -1.0719 | 0.237 |
| 14 | 45636328 | A | G | | 1 | FANCM | c.1964A>G | p.Asn655Ser | T | 0.0075 | T | -0.9727 | 0.016 |
| 14 | 45644589 | G | T | | 1 | FANCM | c.2632G>T | p.Val878Leu | T | 0 | T | -0.9027 | 0.021 |
| 14 | 45644816 | A | C | | 1 | FANCM | c.2859A>C | p.Lys953Asn | T | 0.0821 | T | -1.0365 | 0.145 |
| 14 | 45645715 | A | G | | 1 | FANCM | c.3758A>G | p.Asn1253Ser | T | 0.0104 | T | -1.0365 | 0.048 |
| 14 | 45650900 | A | G | | 2 | FANCM | c.4378A>G | p.Ile1460Val | T | 0.0004 | T | -1.0163 | 0.06 |
| 14 | 45658024 | C | T | | 1 | FANCM | c.4799C>T | p.Thr1600Ile | T | 0.1628 | T | -0.8012 | 0.188 |
| 14 | 45665468 | C | G | | 2 | FANCM | c.5434C>G | p.Pro1812Ala | T | 0.002 | T | -1.0509 | 0.216 |
| 14 | 45665613 | G | A | | 2 | FANCM | c.5579G>A | p.Arg1860His | T | 0.1884 | T | -0.739 | 0.731 |
| 14 | 45665661 | A | G | | 1 | FANCM | c.5627A>G | p.Asn1876Ser | T | 0.0106 | T | -1.0727 | 0.061 |
| 14 | 45665717 | T | C | | 2 | FANCM | c.5683T>C | p.Cys1895Arg | T | 0.3818 | T | -0.2914 | 0.984 |
| 16 | 23634293 | C | T | | 1 | PALB2 | c.2993G>A | p.Gly998Glu | T | 0.099 | T | -0.9106 | 0.375 |
| 16 | 23635370 | C | T | | 1 | PALB2 | c.2794G>A | p.Val932Met | T | 0.0981 | T | -0.8493 | 0.525 |
| 16 | 23641461 | C | G | | 1 | PALB2 | c.2014G>C | p.Glu672Gln | T | 0.0136 | T | -1.0459 | 0.058 |
| 16 | 23646191 | T | C | | 1 | PALB2 | c.1676A>G | p.Gln559Arg | T | 0 | T | -0.9795 | 0.01 |
| 16 | 23646678 | T | A | | 1 | PALB2 | c.1189A>T | p.Thr397Ser | T | 0.1609 | T | -0.6232 | 0.657 |
| 16 | 23646866 | T | C | | 1 | PALB2 | c.1001A>G | p.Tyr334Cys | T | 0.0297 | T | -1.0008 | 0.151 |
| 16 | 23649399 | G | A | | 1 | PALB2 | c.100C>T | p.Arg34Cys | T | 0.1516 | T | -0.8749 | 0.605 |
| 16 | 68772239 | C | A | | 1 | CDH1 | c.88C>A | p.Pro30Thr | D | 0.5465 | T | -0.3193 | 0.857 |
| 16 | 68867325 | G | C | | 1 | CDH1 | c.2572G>C | p.Asp858His | D | 0.759 | D | 0.7647 | 0.871 |
| 17 | 7572980 | T | G | | 1 | TP53 | c.1129A>C | p.Thr377Pro | D | 0.8633 | D | 0.4181 | 0.21 |
| 17 | 7577577 | T | C | | 15 | TP53 | c.704A>G | p.Asn235Ser | D | 0.9811 | D | 1.0418 | 0.291 |
| 17 | 7579472 | G | C | | 2 | TP53 | c.215C>G | p.Pro72Arg | T | 0 | T | -0.9287 | 0.267 |
| 17 | 33433487 | C | T | | 1 | RAD51D | c.494G>A | p.Arg165Gln | T | 0.0001 | T | -1.0318 | 0.025 |
| 17 | 41222975 | C | T | | 1 | BRCA1 | c.4956G>A | p.Met1652Ile | T | 0.1432 | T | -0.8538 | 0.681 |
| 17 | 41223094 | T | C | | 1 | BRCA1 | c.4837A>G | p.Ser1613Gly | T | 0 | T | -1.0796 | 0.454 |
| 17 | 41226488 | C | A | | 1 | BRCA1 | c.4535G>T | p.Ser1512Ile | D | 0.6215 | T | -0.12 | 0.759 |
| 17 | 41243509 | T | C | | 1 | BRCA1 | c.4039A>G | p.Arg1347Gly | T | 0.4499 | T | -0.3762 | 0.711 |
| 17 | 41244000 | T | C | | 1 | BRCA1 | c.3548A>G | p.Lys1183Arg | T | 0 | T | -1.0241 | 0.258 |
| 17 | 41244429 | C | T | | 1 | BRCA1 | c.3119G>A | p.Ser1040Asn | T | 0.3572 | T | -0.545 | 0.469 |
| 17 | 41244435 | T | C | | 1 | BRCA1 | c.3113A>G | p.Glu1038Gly | T | 0 | T | -1.094 | 0.252 |
| 17 | 41244936 | G | A | | 1 | BRCA1 | c.2612C>T | p.Pro871Leu | T | 0 | T | -0.9367 | 0.255 |
| 17 | 41245471 | C | T | | 1 | BRCA1 | c.2077G>A | p.Asp693Asn | T | 0.0226 | T | -0.9508 | 0.265 |
| 17 | 41246481 | T | C | | 2 | BRCA1 | c.1067A>G | p.Gln356Arg | T | 0.0737 | T | -0.8452 | 0.648 |
| 17 | 56772522 | G | A | | 2 | RAD51C | c.376G>A | p.Ala126Thr | T | 0.1836 | T | -0.7852 | 0.303 |
| 17 | 56787304 | G | A | | 2 | RAD51C | c.790G>A | p.Gly264Ser | T | 0.2265 | T | -0.861 | 0.677 |
| 17 | 56798128 | A | G | | 2 | RAD51C | c.859A>G | p.Thr287Ala | T | 0.1243 | T | -0.9304 | 0.442 |
| 17 | 59763347 | A | G | | 1 | BRIP1 | c.2755T>C | p.Ser919Pro | T | 0 | T | -0.9367 | 0.026 |
| 17 | 59885956 | G | A | | 1 | BRIP1 | c.790C>T | p.Arg264Trp | D | 0.5349 | T | -0.0027 | 0.576 |
| 17 | 59924512 | C | T | | 1 | BRIP1 | c.577G>A | p.Val193Ile | T | 0.1941 | T | -0.9417 | 0.033 |
| 22 | 29090054 | G | A | | 2 | CHEK2 | c.1427C>T | p.Thr476Met | T | 0.4373 | D | 0.0662 | 0.833 |
| 22 | 29121019 | G | A | | 1 | CHEK2 | c.538C>T | p.Arg180Cys | D | 0.6639 | D | 0.4216 | 0.9 |

a The reference sequence for the position is Hg19; VUS: Variant of Uncertain Significance; Pred – Prediction; C: Deleterious; T-Tolerated; M-SVM: MetaSVM; M-LR: MetaLR; AC: Allele Count.

**Supplementary Table S2. Predicted deleterious missense variants in cancer predisposition gene**

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Chr | Positiona | REF | ALT | GENE | c.HGVS | p.HGVS | M-LR Pred | M-LR Score | M-LR Rank | M-SVM Pred | M-SVM Score | M-SVM Rank | Vest3 Score | ClinVar |
| 11 | 108224555 | A | G | ATM | c.8734A>G | p.Arg2912Gly | D | 0.7556 | 0.92296 | D | 0.7055 | 0.93747 | 0.998 | VUS |
| 16 | 68867325 | G | C | CDH1 | c.2572G>C | p.Asp858His | D | 0.759 | 0.92407 | D | 0.7647 | 0.94428 | 0.871 | VUS |
| 9 | 21971008 | A | G | CDKN2A | c.350T>C | p.Leu117Pro | D | 0.97284 | 0.9112 | D | 0.97835 | 1.0265 | 0.969 | VUS |
| 9 | 21971086 | A | T | CDKN2A | c.272T>A | p.Leu91Gln | D | 0.96944 | 0.9002 | D | 0.97447 | 1.0025 | 0.919 | VUS |
| 22 | 29121019 | G | A | CHEK2 | c.538C>T | p.Arg180Cys | D | 0.6639 | 0.89171 | D | 0.4216 | 0.90145 | 0.9 | VUS |
| 3 | 37089131 | A | C | MLH1 | c.1853A>C | p.Lys618Thr | D | 0.8835 | 0.9643 | D | 0.8682 | 0.95622 | 0.927 | Benign |
| 3 | 37089110 | T | C | MLH1 | c.1832T>C | p.Ile611Thr | D | 0.8895 | 0.96614 | D | 0.9765 | 0.97053 | 0.893 | VUS |
| 3 | 37089130 | A | G | MLH1 | c.1852A>G | p.Lys618Glu | D | 0.8885 | 0.96583 | D | 0.8782 | 0.9574 | 0.811 | VUS |
| 2 | 47641520 | T | C | MSH2 | c.905T>C | p.Leu302Ser | D | 0.9417 | 0.98239 | D | 1.0958 | 0.99521 | 0.991 | VUS |
| 2 | 48027910 | A | G | MSH6 | c.2788A>G | p.Lys930Glu | D | 0.6125 | 0.87229 | D | 0.0564 | 0.84408 | 0.88 | VUS |
| 2 | 48030645 | C | T | MSH6 | c.3259C>T | p.Pro1087Ser | D | 0.5847 | 0.86079 | D | 0.0684 | 0.84621 | 0.817 | VUS |
| 2 | 48026630 | C | G | MSH6 | c.1508C>G | p.Ser503Cys | D | 0.7495 | 0.92097 | D | 0.6125 | 0.92642 | 0.532 | Benign |
| 7 | 6017269 | G | A | PMS2 | c.2395C>T | p.Arg799Trp | D | 0.8629 | 0.95787 | D | 0.9478 | 0.96647 | 0.95 | VUS |
| 7 | 6045634 | T | C | PMS2 | c.52A>G | p.Ile18Val | D | 0.7732 | 0.9287 | D | 0.6865 | 0.93526 | 0.727 | VUS |

a The reference sequence for the position is Hg19; VUS: Variant of Uncertain Significance; Pred – Prediction; C: Deleterious; T-Tolerated; M-SVM: MetaSVM; M-LR: MetaLR; Rank: Rank Score