**Таблица 3. Обнаруженные патогенные мутации у пациентов с наследственными РМЖ и РЯ**

|  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Ген** |  | **Экзон** | **кДНК** | **Белок** | **Функция** | **dbSNP** | **Славяне** | **Татары** | **Башкиры** | **Другие** | **Всего** |
| *BRCA1* | 17:41209079T>TG | 19 | c.5266dupC | p.Q1756fs | frameshift ins | rs80357906 | 55 | 32 | 3 | 2 | 84 |
| *BRCA1* | 17:41258504A>C | 4 | c.181T>G | p.C61G | missense | rs28897672 | 4 | 7 |  |  | 9 |
| *BRCA1* | 17:41243512CT>C | 10 | c.4035delA | p.E1345fs | frameshift del | rs80357711 | 5 | 1 |  | 1 | 7 |
| *BRCA1* | 17:41215382G>A | 18 | c.5161C>T | p.Q1721X | stop gain | rs878854957 |  | 4 | 2 | 2 | 7 |
| *BRCA1* | 17:41209095G>A | 19 | c.5251C>T | p.R1751X | stop gain | rs80357123 | 7 |  |  |  | 7 |
| *BRCA1* | 17:41245586CT>C | 10 | c.1961delA | p.K654fs | frameshift del | rs80357522 | 6 |  |  |  | 6 |
| *BRCA2* | 13:32906576C>CAA | 10 | c.961\_962insAA | p.Q321fs | frameshift ins |  | 1 | 4 | 1 |  | 4 |
| *BRCA1* | 17:41226348C>T | 14 | c.4675G>A | p.E1559K | missense | rs80356988 | 4 |  |  |  | 4 |
| *BRCA2* | 13:32950929G>A | 21 | c.8754+1G>A | splicing | rs397508006 | 2 |  |  | 2 | 4 |
| *BRCA1* | 17:41243843GTTTAC>G | 10 | c.3700\_3704del | p.V1234fs | frameshift del | rs80357609 | 1 | 3 |  |  | 4 |
| *BRCA2* | 13:32911388ACT>A | 11 | c.2897\_2898del | p.T966fs | frameshift del | rs80359361 |  | 2 |  | 1 | 3 |
| *BRCA1* | 17:41276044ACT>A | 2 | c.68\_69del | p.E23fs | frameshift del | rs386833395 | 3 |  |  |  | 3 |
| *BRCA1* | 17:41223242G>C | 15 | c.4689C>G | p.Y1563X | stop gain | rs80357433 | 3 |  |  |  | 3 |
| *BRCA2* | 13:32912240G>GA | 11 | c.3749dupA | p.E1250fs | frameshift ins | rs397507683 |  | 3 |  |  | 3 |
| *BRCA2* | 13:32906915AAAAG>A | 10 | c.1301\_1304del | p.K434fs | frameshift del | rs80359277 | 2 |  |  |  | 2 |
| *BRCA1* | 17:41244404AC>A | 10 | c.3143delG | p.G1048fs | frameshift del | rs886040100 | 1 | 1 |  |  | 2 |
| *BRCA1* | 17:41243917ACT>A | 10 | c.3629\_3630del | p.E1210fs | frameshift del | rs80357589 |  |  |  | 2 | 2 |
| *BRCA1* | 17:41201137C>G | 21 | c.5406+1G>C | splicing |  |  | 2 |  |  | 2 |
| *BRCA1* | 17:41215969C>T | 16 | c.5075-1G>A | splicing | rs1800747 | 2 |  |  |  | 2 |
| *BRCA1* | 17:41234451G>A | 12 | c.4327C>T | p.R1443X | stop gain | rs41293455 | 2 |  |  |  | 2 |
| *BRCA2* | 13:32921034G>A | 13 | c.7007+1G>A | splicing | rs397507891 |  | 2 |  |  | 2 |
| *BRCA2* | 13:32954050G>A | 23 | c.9117G>A | p.P3039P | synonymous | rs28897756 | 1 | 1 |  |  | 2 |
| *BRCA2* | 13:32911297TAAAC>T | 11 | c.2806\_2809del | p.K936fs | frameshift del | rs80359351 | 2 |  |  | 1 | 2 |
| *BRCA2* | 13:32906625A>ATG | 10 | c.1010\_1011insTG | p.N337fs | frameshift ins |  |  |  |  | 1 | 1 |
| *BRCA1* | 17:41222939A>G | 15 | c.4986+6T>C | splicing | rs80358086 | 1 |  |  |  | 1 |
| *BRCA1* | 17:41276061A>G | 2 | c.53T>C | p.M18T | missense | rs80356929 | 1 |  |  |  | 1 |
| *BRCA2* | 13:32907434A>T | 10 | c.1819A>T | p.K607X | stop gain | rs80358471 | 1 |  |  |  | 1 |
| *BRCA1* | 17:41244839A>T | 10 | c.2709T>A | p.C903X | stop gain |  | 1 |  |  |  | 1 |
| *BRCA1* | 17:41197774A>T | 23 | c.5513T>A | p.V1838E | missense | rs80357107 | 1 |  |  |  | 1 |
| *BRCA2* | 13:32936733A>T | 17 | c.7879A>T | p.I2627F | missense | rs80359014 | 1 |  |  |  | 1 |
| *BRCA1* | 17:41244296AAGCAT>A | 10 | c.3247\_3251del | p.M1083fs | frameshift del |  | 1 |  |  |  | 1 |
| *BRCA1* | 17:41251880ACT>A | 7 | c.457\_458del | p.S153fs | frameshift del | rs397509185 |  |  |  | 1 | 1 |
| *BRCA1* | 17:41244056ACTAGTATCTTC>A | 10 | c.3481\_3491del | p.E1161fs | frameshift del | rs80357877 |  | 1 |  |  | 1 |
| *BRCA2* | 13:32911605AT>A | 11 | c.3114delT | p.Y1038fs | frameshift del |  |  |  |  | 1 | 1 |
| *BRCA2* | 13:32953959AT>A | 23 | c.9027delT | p.Y3009fs | frameshift del | rs80359742 |  |  |  | 1 | 1 |
| *BRCA1* | 17:41246073ATC>A | 10 | c.1473\_1474del | p.Q491fs | frameshift del |  | 1 |  |  |  | 1 |
| *BRCA1* | 17:41256249C>A | 6 | c.331G>T | p.E111X | stop gain |  | 1 |  |  |  | 1 |
| *BRCA1* | 17:41215890C>A | 17 | c.5152+1G>T | splicing | rs80358094 | 1 |  |  |  | 1 |
| *BRCA2* | 13:32914137C>A | 11 | c.5645C>A | p.S1882X | stop gain | rs80358785 | 1 |  |  |  | 1 |
| *BRCA2* | 13:32914800C>A | 11 | c.6308C>A | p.S2103X | stop gain | rs80358870 | 1 |  |  |  | 1 |
| *BRCA2* | 13:32954022C>CA | 23 | c.9090dupA | p.T3030fs | frameshift ins | rs397507419 |  | 1 |  |  | 1 |
| *BRCA2* | 13:32906565C>CA | 10 | c.951dupA | p.T317fs | frameshift ins | rs80359770 | 1 |  |  |  | 1 |
| *BRCA2* | 13:32954009C>CAGTT | 23 | c.9076\_9077insAGTT | p.Q3026fs | frameshift ins |  | 1 | 1 |  |  | 1 |
| *BRCA1* | 17:41276047C>CT | 2 | c.66dupA | p.E23fs | frameshift ins | rs80357783 | 1 |  |  |  | 1 |
| *BRCA2* | 13:32914647C>G | 11 | c.6155C>G | p.S2052X | stop gain | rs786202461 |  | 1 |  |  | 1 |
| *BRCA1* | 17:41258472C>T | 4 | c.212+1G>A | splicing | rs80358042 | 1 |  |  |  | 1 |
| *BRCA1* | 17:41226347C>T | 14 | c.4675+1G>A | splicing | rs80358044 |  | 1 |  |  | 1 |
| *BRCA1* | 17:41209068C>T | 19 | c.5277+1G>A | splicing | rs80358150 |  | 1 |  |  | 1 |
| *BRCA2* | 13:32915112CAA>C | 11 | c.6621\_6622del | p.T2207fs | frameshift del |  |  | 1 |  |  | 1 |
| *BRCA2* | 13:32890556CAG>C | 2 | r.spl |  | splicing | rs1064794852 |  | 1 |  |  | 1 |
| *BRCA2* | 13:32912899CAT>C | 11 | c.4408\_4409del | p.I1470fs | frameshift del | rs80359446 | 1 |  |  |  | 1 |
| *BRCA2* | 13:32907408CATCTT>C | 10 | c.1794\_1798del | p.T598fs | frameshift del | rs276174813 |  | 1 |  |  | 1 |
| *BRCA1* | 17:41246037CG>C | 10 | c.1510delC | p.R504fs | frameshift del | rs80357908 |  |  |  | 1 | 1 |
| *BRCA1* | 17:41245330CT>C | 10 | c.2217delA | p.K739fs | frameshift del |  | 1 |  |  |  | 1 |
| *BRCA2* | 13:32903604CTG>C | 8 | c.657\_658del | p.T219fs | frameshift del | rs80359604 |  |  |  | 1 | 1 |
| *BRCA2* | 13:32912171CTGAA>C | 11 | c.3680\_3683del | p.L1227fs | frameshift del | rs80359396 | 1 |  |  |  | 1 |
| *BRCA1* | 17:41246531CTT>C | 10 | c.1015\_1016del | p.K339fs | frameshift del |  | 1 | 1 |  |  | 1 |
| *BRCA2* | 13:32914766CTT>C | 11 | c.6275\_6276del | p.L2092fs | frameshift del | rs11571658 | 1 |  |  |  | 1 |
| *BRCA1* | 17:41245630G>A | 10 | c.1918C>T | p.Q640X | stop gain | rs886039981 |  | 1 |  |  | 1 |
| *BRCA1* | 17:41245210G>A | 10 | c.2338C>T | p.Q780X | stop gain | rs80356945 | 1 |  |  |  | 1 |
| *BRCA1* | 17:41244145G>A | 10 | c.3403C>T | p.Q1135X | stop gain | rs80357136 | 1 |  |  |  | 1 |
| *BRCA1* | 17:41243941G>A | 10 | c.3607C>T | p.R1203X | stop gain | rs62625308 | 1 |  |  |  | 1 |
| *BRCA1* | 17:41226471G>A | 14 | c.4552C>T | p.Q1518X | stop gain | rs80356881 | 1 |  |  |  | 1 |
| *BRCA2* | 13:32932019G>A | 16 | c.7758G>A | p.W2586X | stop gain | rs80359004 | 1 |  |  |  | 1 |
| *BRCA2* | 13:32907420G>GA | 10 | c.1806dupA | p.G602fs | frameshift ins | rs80359306 | 1 |  |  |  | 1 |
| *BRCA2* | 13:32911442G>GA | 11 | c.2951dupA | p.E984fs | frameshift ins | rs80359365 | 1 |  |  |  | 1 |
| *BRCA2* | 13:32918745G>GA | 12 | c.6893dupA | p.E2298fs | frameshift ins | rs886040933 |  |  |  | 1 | 1 |
| *BRCA2* | 13:32929255G>GAC | 14 | c.7265\_7266insAC | p.C2422\_V2423delinsX | stop gain |  |  | 1 |  |  | 1 |
| *BRCA2* | 13:32921023G>GT | 13 | c.6998dupT | p.V2333fs | frameshift ins | rs754611265 | 1 |  |  |  | 1 |
| *BRCA1* | 17:41226515GA>G | 14 | c.4507delT | p.S1503fs | frameshift del |  | 1 |  |  |  | 1 |
| *BRCA2* | 13:32954272GA>G | 24 | c.9247delA | p.K3083fs | frameshift del | rs397508041 | 1 |  |  |  | 1 |
| *BRCA2* | 13:32893347GAAACCATCTTATAATC>G | 3 | c.202\_217del | p.K68fs | frameshift del | rs1064792960 | 1 |  |  |  | 1 |
| *BRCA1* | 17:41242952GCTTTTTAC>G | 11 | c.4185+1\_4185+8delGTAAAAAG | splicing |  |  | 1 |  |  | 1 |
| *BRCA2* | 13:32936761GT>G | 17 | c.7908delT | p.C2636fs | frameshift del |  | 1 | 1 |  |  | 1 |
| *BRCA2* | 13:32953534GT>G | 22 | c.8836delT | p.L2946fs | frameshift del |  |  |  |  | 1 | 1 |
| *BRCA2* | 13:32914109GTAAT>G | 11 | c.5618\_5621del | p.V1873fs | frameshift del | rs80359526 |  |  |  | 1 | 1 |
| *BRCA1* | 17:41243584T>A | 10 | c.3964A>T | p.K1322X | stop gain | rs80357343 | 1 | 1 |  |  | 1 |
| *BRCA2* | 13:32914366T>A | 11 | c.5874T>A | p.C1958X | stop gain |  | 1 |  |  |  | 1 |
| *BRCA2* | 13:32913778T>G | 11 | c.5286T>G | p.Y1762X | stop gain |  | 1 |  |  |  | 1 |
| *BRCA2* | 13:32914069T>TA | 11 | c.5578dupA | p.I1859fs | frameshift ins | rs397507790 |  | 1 |  |  | 1 |
| *BRCA2* | 13:32914939T>TA | 11 | c.6448dupA | p.I2149fs | frameshift ins | rs1010863706 | 1 |  |  |  | 1 |
| *BRCA1* | 17:41243571T>TGCCTCATTTGTTTGGAAGAACCAATCAA | 10 | c.3976\_3977insTTGATTGGTTCTTCCAAACAAATGAGGC | p.H1326fs | frameshift ins |  |  |  |  | 1 | 1 |
| *BRCA1* | 17:41244016TA>T | 10 | c.3531delT | p.F1177fs | frameshift del | rs80357621 | 1 |  |  |  | 1 |
| *BRCA2* | 13:32912353TAATA>T | 11 | c.3862\_3865del | p.N1288fs | frameshift del | rs80359412 | 1 |  |  |  | 1 |
| *BRCA1* | 17:41243788TAGAC>T | 10 | c.3756\_3759del | p.L1252fs | frameshift del | rs80357868 | 1 |  |  |  | 1 |
| *BRCA1* | 17:41243775TC>T | 10 | c.3772delG | p.E1258fs | frameshift del |  | 1 |  |  |  | 1 |
| *BRCA1* | 17:41244341TTGAA>T | 10 | c.3203\_3206del | p.I1068fs | frameshift del | rs886040102 |  | 1 |  |  | 1 |