**Table I.** Distribution of mutations in the studied group

|  |  |  |  |
| --- | --- | --- | --- |
| **High penetrance breast genes** | | | |
| **Gene** | **Cases** | **Pathogenic Mutation** | **VUS mutation** |
| BRCA1 | 25 | c.3607C>T (7), c.5266dupC (4), c.181T>G (2), c.4035delA, c.3726 C>T, c.2241dupC, c.135-2A>G, c.1789G>A, c.737delT, c.3700\_3704delGTAAA, c.843\_846delCTCA, c.4035delA (3), c5329\_5330insC | - |
| BRCA2 | 11 | c.9371A>T (3), c.8755-1G>A (3), c.1528G>T, c.8695 C>T | c.3547G>C, c.3562A>G (2) |
| TP53 | 4 | c.469G>T (2) | c.847C>T, c.480G>A |
| STK 11 | 3 | - | c.1225C>T (3) |
| CDH1 | 1 | - | c.1840A>G |
| **Moderate penetrance breast genes** | | | |
| **Gene** | **Cases** | **Pathogenic Mutation** | **VUS mutation** |
| PALB2 | 6 | c.93dupA (2), c.3549C>G, c.509\_510delGA, c.79G>T | c.2461A>T |
| CHEK2 | 10 | c.470T>C (5), c.1232G>A (2), 1283C>T | c.1521T>C, c.1313A>T |
| ATM | 7 | c.7630-2A>C, c.2250G>A, c.1564\_1565delGA | c.9077T>G, c8734A>G, c.2735G>A, c.4768C>T |
| **Additional genes** | | | |
| **Gene** | **Cases** | **Pathogenic Mutation** | **VUS mutation** |
| BARD 1 | 3 | - | c.1333G>A, c.2282G>A, c.2640del15 |
| PMS2 | 3 | - | c.620G>A, c.2012C>T, c.852A>G |
| BLM | 4 | - | c.3014T>C, c.3879A>G, c.4076+4T>G, c.1642C>T |
| MLH1 | 1 | c.2041G>A | - |
| MSH2 | 1 | - | c.1597C>G |
| MSH6 | 2 | c.2136delG | c.1068T>G |
| NBN | 1 | - | c.511A>G |
| RAD 50 | 4 | c.2165dupA | c.785T>G, c900G>A, c.1663A>G |
| Rad51C | 2 | - | c.790G>A (2) |
| MUTYH | 2 | c.1187G>A | c.158-3C>T |
| MEN1 | 1 | - | c.777G>A |
| NBN | 1 | c657\_661delACAAA | - |
| MRE11A | 1 | - | c.1091G>A |