**Table** S1. *TCOF1*mutations and SNPs identified in TCS patients. [1-3]

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| **Location** | **cDNA** | **Amino acid change** | **Variant** | **Protein effect** | | **Clinical phenotype** | **Reference** |
| 5’-UTR | c.-89T>G |  | Polymorphism |  | | TCS | [4] |
| 5'-UTR | c.-59G>A |  | Unclassified variants |  | | TCS | [5] |
| 5’-UTR | c.-41G>T |  | Polymorphism |  | | TCS | [6] |
| 5’-UTR | c.-26T>A |  | Polymorphism |  | | TCS | [4] |
| exon 1 | c.1A>T | p.M1? | Mutation |  | | TCS | [5] |
| exon 1 | c.3G>A | p.M1? | Mutation |  | | TCS | [5,7] |
| exon 1 | c.40A>T | p.I14F | Mutation |  | | TCS | [8] |
| exon 1 | c.42C>G | p.I14M | Mutation |  | | TCS | [5] |
| exon 1 | c.50A>G | p.H17R | Mutation |  | | TCS | [5] |
| exon 1 | c.59delG |  | Mutation |  | | TCS | [8] |
| exon 1 | c.61G>C | p.A21P | Mutation |  | | TCS | [5] |
| exon 1 | c.77C>T | p.A26V | Mutation |  | | TCS | [9] |
| exon 1 | c.79G>A | p.A27T | Mutation |  | | TCS | [5] |
| exon 1 | c.87delA | p.V30\* | Mutation | STOP | | TCS | [5] |
| exon 1 | ND | p.Q36\* | Mutation | STOP | | TCS | [10] |
| intron 1 | c.108+62\_68del |  | Unclassified variants |  | | TCS | [5] |
| intron 1 | c.109-1delG |  | Mutation |  | | TCS | [10] |
| intron 1 | c.109-2A>T |  | Mutation |  | | TCS | [5] |
| intron 1 | c.109-28T>C |  | Unclassified variants |  | | TCS | [5] |
| exon 2 | c.122C>T | p.A41V | Polymorphism |  | | TCS | [11] |
| exon 2 | c.149A>G | p.Y50C | Mutation |  | | TCS | [12] |
| exon 2 | c.157T>C | p.W53R | Mutation |  | | TCS | [10,13] |
| exon 2 | c.159G>A | p.W53\* | Mutation | STOP | | TCS | [5] |
| exon 2 | c.163C>T | p.Q55\* | Mutation | STOP | | TCS | [5,9] |
| intron 2 | 1244G>T |  | Polymorphism |  | | TCS | [4] |
| exon 3 | c.165‑1G>A |  | Mutation | STOP | | TCS | [14] |
| exon 3 | c.218insAACC | p.A73fs | Mutation |  | | TCS | [15] |
| exon 3 | c.274 delG |  | Mutation |  | | TCS | [10] |
| exon 3 | ND | p.E93\* | Mutation |  | | TCS | [10] |
| exon 3 | c.303\_304delCA | p.A101Afsx73 | Mutation | STOP | | TCS | [6] |
| exon 3 | c.4369\_4373 | p.K1457-Efs\*12 | ND | affects NLS | | TCS | [16] |
| intron 3 | c.304+5G>C | splice | Mutation |  | | TCS | [5,8,10] |
| intron 3 | c.305-1 G>A |  | Mutation |  | | TCS | [12] |
| intron 3 | c.305-52A>G |  | Polymorphism |  | | TCS | [5] |
| exon 4 | c.343delG | p.121\* | Mutation | STOP | | TCS | [7] |
| exon 4 | c.357\_358delAA | p.S120fs | Mutation |  | | TCS | [5] |
| exon4/intron4 | c.376delAAGGTGAGTGGGACTGCC |  | ND | STOP | | TCS | [17] |
| intron 4 | c.379-2A>G |  | Mutation |  | | TCS | [7] |
| exon 5 | c.381\_382delAG | p.A127\* | Mutation | STOP | | TCS | [18] |
| exon 5 | c.389insA |  | Mutation |  | | TCS | [8] |
| exon 5 | c.390delGA | p.173\* | Mutation | STOP | | TCS | [7] |
| exon 5 | c.405\_406delTG | p.G136fs | Mutation |  | | TCS | [5,10,19] |
| exon 5 | c.408delG | p.214\* | Mutation |  | | TCS | [7] |
| exon 5 | c.422insA | p.H141Q | Mutation | STOP | | TCS | [20] |
| exon 5 | c.431delC | p.T144fs | Mutation |  | | TCS | [15] |
| exon 5 | c.475A>T | p.K159\* | Mutation | STOP | | TCS | [9] |
| exon 5 | c.484\_668ins |  | ND | STOP | | TCS | [21] |
| exon 5 | c.489delC | p.S164Qfs\*55 | Mutation |  | | TCS | [22] |
| exon 5 | c.497delATAC | p.N166I | Mutation | STOP | | TCS | [20] |
| exon 5 | c.503C>T | p.T168M | Polymorphism |  | | TCS | [7] |
| exon 5 | c.519delT | p.T173Tfsx46 | Mutation | STOP | | TCS | [6] |
| intron 5 | c.565+1G>A |  | Mutation |  | | TCS | [5] |
| intron 5 | c.565+18G>C |  | Polymorphism |  | | TCS | [7] |
| intron 5 | c.566-10C>A |  | Mutation |  | | TCS | [5] |
| exon 6 | c.574del(16bp) | p.209\* | Mutation | STOP | | TCS | [7] |
| exon 6 | c.579G>A | p.A193A | Unclassified variants |  | | TCS | [5] |
| exon 6 | c.599delG | p.S200Tfsx19 | Mutation | STOP | | TCS | [6] |
| exon 6 | c.618delC | p.S207fs | Mutation |  | | TCS | [5] |
| exon 6 | c.630 delAG |  | Mutation |  | | TCS | [10] |
| intron 6 | c.639+1G>A |  | Mutation |  | | TCS | [15] |
| intron 6 | c.639+32C>G |  | Polymorphism |  | | TCS | [6,23] |
| intron 6 | c.639+32C>T¹ |  | Polymorphism |  | | TCS | [7,23] |
| intron 6 | c.640-69T>C |  | Polymorphism |  | | TCS | [5] |
| intron 6 | c.14036G>A |  | Polymorphism |  | | TCS | [4] |
| exon 6A | c.648delC | p.S217Qfs\*2 | Mutation |  | | TCS | [22] |
| exon 6A | c.726delT | p.K244fs | Mutation |  | | TCS | [13] |
| exon 6A | c.827\_844del | p.G276\_E281del | Mutation |  | | TCS | [5] |
| exon 7 | IVS6-710 C-T |  | Mutation |  | | TCS | [24] |
| exon 7 | c.698delC |  | Mutation |  | | TCS | [8] |
| exon 7 | c.720-727delAGCACCCC |  | Mutation |  | | TCS | [12,24] |
| exon 7 | c.724insC |  | Mutation |  | | TCS | [10] |
| exon 7 | c.728insC | p.271\* | Mutation | STOP | | TCS | [7] |
| exon 7 | c.744insT |  | Mutation | STOP | | TCS | [25] |
| exon 7 | ND | p.Q252\* | Mutation |  | | TCS | [10] |
| exon 7 | c.768 G>A | p.G256G | Polymorphism |  | | TCS | [7] |
| exon 7 | c.786delA |  | Mutation |  | | TCS | [12] |
| exon 7 | c.786delAG |  | Mutation | STOP | | TCS | [25] |
| exon 7 | c.790\_791delAG | p.S264QfsX7 | Mutation |  | | TCS | [26] |
| exon 7 | c.797G>A | p.S266N | Mutation |  | | TCS | [7] |
| exon 7 | c.803A>G¹ | p.E268G | Polymorphism |  | | TCS | [23] |
| exon 7 | c.1016C>G | p.S339\* | Mutation | STOP | | TCS | [5] |
| exon 7 | c.1028G>A | p.S343N | Unclassified variants |  | | TCS | [5] |
| exon 7 | c.1347T>C | p.P439L | Polymorphism |  | | TCS | [6,20] |
| intron 7 | c.852+39A>G |  | Polymorphism |  | | TCS | [7] |
| intron 7 | c.1083+39G>A |  | Polymorphism |  | | TCS | [5] |
| exon 8 | c.864-865delAG |  | Mutation |  | | TCS | [8] |
| exon 8 | c.911C>T | p.S304L | Mutation |  | | TCS | [27] |
| exon 8 | c.998 C>T¹ | p.S333L | Polymorphism | affects ATM phoshorylation site | | TCS | [23] |
| exon 8 | c.1015 G>T |  | Mutation |  | | TCS | [12] |
| exon 8 | c.1086G>A | p.A362A | Unclassified variants |  | | TCS | [5] |
| exon 8 | c.1095\_1096delAG | p.G366fs | Mutation |  | | TCS | [8,15] |
| exon 8 | c.1096G>T | p.G366\* | Mutation | STOP | | TCS | [5] |
| exon 8 | c.1242C>T | p.S414S | Unclassified variants |  | | TCS | [5] |
| intron 8 | c.1047+60 G>C |  | Polymorphism |  | | TCS | [7] |
| intron 8 | c.1278+60G>C |  | Polymorphism |  | | TCS | [5] |
| exon 9 | c.1084G>A | p.A362T | Mutation |  | | TCS | [28] |
| exon 9 | c.1098delC |  | Mutation |  | | TCS | [12] |
| exon 9 | c.1099A>T | p.K367\* | Mutation | STOP | | TCS | [7] |
| exon 9 | c.1106insC |  | Mutation | affects ATM phoshorylation site | | TCS | [8] |
| exon 9 | c.1120delG |  | Mutation |  | | TCS | [10] |
| exon 9 | c.1142delC |  | Mutation |  | | TCS | [29] |
| exon 9 | c.1150G>A, 3987insG | p.A350A | Mutation |  | | TCS | [23] |
| exon 9 | c.1164insT |  | Mutation |  | | TCS | [23] |
| exon 9 | c.1215insA |  | Mutation |  | | TCS | [12] |
| exon 9 | c.1216insA |  | Mutation |  | | TCS | [10] |
| exon 9 | c.1230A>G | p.E410E | Polymorphism |  | | TCS | [7] |
| exon 9 | c.1247A >G | p.E419E | Polymorphism |  | | TCS | [28] |
| exon 9 | c.1281G>A | p.A427A | Polymorphism |  | | TCS | [5] |
| exon 9 | c.1298delC | p.A433fs | Mutation |  | | TCS | [15] |
| exon 9 | c.1303delC | p.Q435fs | Mutation |  | | TCS | [5] |
| exon 9 | c.1446dupA | p.D483fs | Mutation |  | | TCS | [5,12,19] |
| exon 9 | c.1473dupC | p.M492fs | Mutation |  | | TCS | [5] |
| exon 10 | c.1287insG |  | Mutation |  | | TCS | [23] |
| exon 10 | c.1316C>T | p.P439L | Polymorphism |  | | TCS | [10,20,23] |
| exon 10 | c.1327insA |  | Mutation |  | | TCS | [10] |
| exon 10 | c.1347T>C¹ | p.P449P | Mutation/Polymorphism |  | | TCS | [7,20,23,24,28,30] |
| exon 10 | c.1359G>A | p.G453G | Mutation |  | | TCS | [28] |
| exon 10 | c.1393 C>T | p.Q465\* | Mutation | affects NLS | | TCS | [29] |
| exon 10 | c.1406-1409delAGAG |  | Mutation |  | | TCS | [23,24] |
| exon 10 | c.1408-1409delAG | p.S470Q | Mutation | STOP | | TCS | [12,20,23,24] |
| exon 10 | c.1441insGG |  | Mutation |  | | TCS | [10] |
| exon 10 | c.1557dupA | p.G520fs | Mutation |  | | TCS | [5,31] |
| exon 10 | c.1578 T > C | p.P526P | Mutation/Polymorphism | Affects PolI interaction site | | TCS | [6,13,20] |
| exon 10 | c.1581delG | p.G587Gfsx69 | Mutation | STOP/Affects PolI interaction site | | TCS | [6] |
| exon 10 | c.1609C>T | p.Q537\* | Mutation | STOP/Affects PolI interaction site | | TCS | [15] |
| exon 10 | c.1639\_1640delAG | p.S547fs | Mutation | Affects PolI interaction site | | TCS | [5,6,12,20,32] |
| exon 10 | c.1702C>T | p.Q568\* | Mutation | STOP/Affects PolI interaction site | | TCS | [5] |
| intron 10 | c.1473+68C>T |  | Polymorphism |  | | TCS | [7] |
| exon 11 | c.1530G>T | p.G510G | Polymorphism | Affects PolI interaction site | | TCS | [7,23,28,30] |
| exon 11 | c.1552G>A | p.V518I | Polymorphism | Affects PolI interaction site | | TCS | [7,20,23] |
| exon 11 | c.[1552delG; 1565T< | p.518\* | Mutation | STOP/Affects PolI interaction site | | TCS | [7] |
| exon 11 | c.1611G>A | p.S537S | Mutation/Polymorphism | Affects PolI interaction site | | TCS | [7,20,23,24,28,30] |
| exon 11 | c.1729C>T | p.Q577\* | Mutation | STOP/Affects PolI interaction site | | TCS | [5] |
| exon 11 | c.1747C>T |  | Mutation | STOP/Affects PolI interaction site | | TCS | [33] |
| exon 11 | c.1761G>T | p.G587G | Polymorphism | Affects PolI interaction site | | TCS | [6,23] |
| exon 11 | c.1762C>G¹ | p.P588A | Mutation | Affects PolI interaction site | | TCS | [30] |
| exon 11 | c.1782\_1788del7 | p.Val595fs | Mutation | Affects PolI interaction site | | TCS | [5] |
| exon 11 | c.1813\_1814delAT | p.M605fs | Mutation | Affects PolI interaction site | | TCS | [5] |
| exon 11 | c.1837delG | p.E613fs | Mutation | Affects PolI interaction site | | TCS | [5] |
| exon 11 | c.1842A>G | p.S614S | Polymorphism | Affects PolI interaction site | | TCS | [6,20] |
| exon 11 | c.1854G>A | p.A618A | Unclassified variants | Affects PolI interaction site | | TCS | [5] |
| exon 11 | c.17681C>T | p.P526P | Polymorphism | Affects PolI interaction site | | TCS | [4] |
| exon 11 | c.17693G>A | p.G530G | Polymorphism | Affects PolI interaction site | | TCS | [4] |
| exon 12 | c.1687C>T | p.Q563\* | Mutation | STOP/Affects PolI interaction site | | TCS | [7] |
| exon 12 | c.1719delG | p.N574Tfs | Mutation | Affects PolI interaction site | | TCS | [29] |
| exon 12 | c.1742insC | p.620\* | Mutation | STOP/Affects PolI interaction site | | TCS | [7] |
| exon 12 | c.1762C>G | p.P588A | Polymorphism | Affects PolI interaction site | | TCS | [7,28] |
| exon 12 | c.1768insC | p.620\* | Mutation | STOP/Affects PolI interaction site | | TCS | [7] |
| exon 12 | c.1837G > C | p.A588P | Polymorphism | Affects PolI interaction site | | TCS | [6] |
| exon12 | c.1863A>G, 2355-2364delCAGGGCCAGA | p.E621E | Mutation | Affects PolI interaction site | | TCS | [23] |
| exon 12 | c.1866-1873delAGATAGTG |  | Mutation | Affects PolI interaction site | | TCS | [23] |
| exon 12 | c.1867-1868delGA | p.D623X | Mutation | STOP/Affects PolI interaction site | | TCS | [9,23] |
| exon 12 | c.1868delATAG | p.632\* | Mutation | STOP/Affects PolI interaction site | | TCS | [7] |
| exon 12 | c.1872-1875delTGAG |  | Mutation | Affects PolI interaction site | | TCS | [12] |
| exon 12 | c.1879 delGAGAA |  | Mutation | Affects PolI interaction site | | TCS | [10] |
| exon 12 | c.1907delT | p.L636fs | Mutation | Affects PolI interaction site | | TCS | [5] |
| exon 12 | c.1952\_1956delCTGCA | p.T651fs | Mutation | Affects PolI interaction site | | TCS | [5] |
| exon 12 | c.1953C>T | p.T651T | Unclassified variants | Affects PolI interaction site | | TCS | [5] |
| exon 12 | c.1973delC | p.P658Lfsx53 | Mutation | STOP/Affects PolI interaction site | | TCS | [6] |
| exon 12 | c.1993C>G | p.A665P | Polymorphism | Affects PolI interaction site | | TCS | [6] |
| exon 12 | c.1993delG | p.A665fs | Mutation | Affects PolI interaction site | | TCS | [5] |
| exon 12 | c.1999dupC | p.R667fs | Mutation | Affects PolI interaction site | | TCS | [5,7] |
| exon 12 | c.2065\_2075del11 | p.P689fs | Mutation | Affects PolI interaction site | | TCS | [5] |
| exon 12 | c.2098\_2099delGA | p.D700\* | Mutation | STOP/Affects PolI interaction site | | TCS | [5,23] |
| exon 12 | c.2099\_2102delATAG | p.D700fs | Mutation | Affects PolI interaction site | | TCS | [5,7] |
| exon 12 | c.2103\_2106delTGAG | p.S701fs | Mutation | Affects PolI interaction site | | TCS | [5,12] |
| exon 12 | c.18111A>G | p.S614S | Polymorphism | Affects PolI interaction site | | TCS | [4] |
| intron 12 | c.1911+1G>A |  | Mutation |  | | TCS | [7] |
| intron 12 | c.1911+36delC |  | Polymorphism |  | | TCS | [7] |
| intron 12 | c.2142+22C>T |  | Unclassified variants |  | | TCS | [5] |
| exon 13 | c.1611G>A |  | Mutation | Affects PolI interaction site | | TCS | [24] |
| exon 13 | c.18434G>C | p.A665P | Polymorphism | Affects PolI interaction site | | TCS | [4] |
| exon 13 | c.1915-1916delAA |  | Mutation | Affects PolI interaction site | | TCS | [8] |
| exon 13 | c.1926-1927insG |  | Mutation | Affects PolI interaction site | | TCS | [12] |
| exon 13 | c.1974G>C¹ | p.L658F | Polymorphism | Affects PolI interaction site | | TCS | [23] |
| exon 13 | c.1978delC |  | Mutation | STOP/Affects PolI interaction site | | TCS | [21] |
| exon 13 | c.2014insG |  | Mutation | Affects PolI interaction site | | TCS | [10] |
| exon 13 | c.2014C>T¹ | p.P672S | Polymorphism | Affects PolI interaction site | | TCS | [23] |
| exon 13 | c.2018-2025delCAGTCACC |  | Mutation | Affects PolI interaction site | | TCS | [23,24] |
| exon 13 | c.2019-2025delAGTCACC |  | Mutation | Affects PolI interaction site | | TCS | [8] |
| exon 13 | c.2026C>T | p.Q676\* | Mutation | STOP; affects ATM phosphorylation site, Affects PolI interaction site | | TCS | [8,12] |
| exon 13 | c.2055delAG |  | Mutation | STOP/Affects PolI interaction site | | TCS, AMD? | [34] |
| exon 13 | c.2059delAG |  | Mutation | STOP; Affects PolI interaction site | | TCS | [25] |
| exon 13 | c.2082-2085delTGAG |  | Mutation | Affects PolI interaction site | | TCS | [8] |
| exon 13 | c.2157dupG | p.K720fs | Mutation | Affects PolI interaction site | | TCS | [5,12] |
| exon 13 | c.2164delC | p.L722fs | Mutation | Affects PolI interaction site | | TCS | [5] |
| exon 13 | c.2167C>T | p.Q723\* | Mutation | STOP/Affects PolI interaction site | | TCS | [5] |
| exon 13 | c.2285\_2286delCT | p.S762\* | Mutation | STOP/Affects PolI interaction site | | TCS | [5] |
| exon 13 | c.2285\_2286delCT | p.S762fs | Mutation | STOP/Affects PolI interaction site | | TCS | [6] |
| exon 13 | c.2287G>T | p.E763\* | Mutation | STOP/Affects PolI interaction site | | TCS | [5] |
| intron 13 | c.2341-2A>G |  | Mutation |  | | TCS | [5] |
| exon 14 | c.2110delG |  | Mutation | Affects PolI interaction site | | TCS | [8] |
| exon 14 | c.2221C> T | p.Q741\* | Mutation | STOP/Affects PolI interaction site | | TCS | [28] |
| exon 14 | ND | p.K748K | Mutation | Affects PolI interaction site | | TCS | [19,23] |
| exon 14 | ND | p.K749K | Mutation | Affects PolI interaction site | | TCS | [10] |
| exon 14 | c.2205insTT; 2206delG | p.795\* | Mutation | STOP/Affects PolI interaction site | | TCS | [7] |
| exon 14 | c.2426delC | p.P809fs | Mutation | Affects PolI interaction site | | TCS | [5] |
| exon 14 | c.2473\_2476delTCCA | p.S825fs | Mutation | Affects PolI interaction site | | TCS | [5] |
| exon 14 | c.2478G>A |  | Mutation | Affects PolI interaction site | | TCS | [13,15,19,23] |
| intron 14 | c.2247+27G>A |  | Polymorphism |  | | TCS | [7] |
| intron 14 | c.2248-42insG |  | Polymorphism |  | | TCS | [7] |
| intron 14 | c.2478+5G>C |  | Mutation |  | | TCS | [5] |
| intron 14 | c.2478+5G>A |  | Mutation |  | | TCS | [22] |
| exon 15 | c.2272C>T | p.Q758\* | Mutation | STOP/Affects PolI interaction site | | TCS | [8] |
| exon 15 | c.2297delG |  | Mutation | Affects PolI interaction site | | TCS | [10] |
| exon 15 | c.2354-2363delCAGGGCCAGA |  | Mutation | Affects PolI interaction site | | TCS | [23] |
| exon 15 | c.2355-2356delAG |  | Mutation | Affects PolI interaction site | | TCS | [23] |
| exon 15 | c.2375-2376delGG |  | Mutation | Affects PolI interaction site | | TCS | [12] |
| exon 15 | c.238delG |  | Mutation | Affects PolI interaction site | | TCS | [10] |
| exon 15 | c.2394\_2395delAG | p.D799Qfs\* | Mutation | STOP | | TCS | [18] |
| exon 15 | c.2399delGTGA |  | Mutation | Affects PolI interaction site | | TCS | [10] |
| exon 15 | c.2399-2402delGTGA |  | Mutation | Affects PolI interaction site | | TCS | [8] |
| exon 15 | ND | p.E796\* | Mutation | STOP/Affects PolI interaction site | | TCS | [23] |
| exon 15 | c.2626\_2627delGA | p.D876Qfsx2 | Mutation | STOP/Affects PolI interaction site | | TCS | [6] |
| intron 15 | c.2428-20insCTCT |  | Polymorphism |  | | TCS | [20,23] |
| intron 15 | c.2428-20insCTCTC |  | Polymorphism |  | | TCS | [7,20] |
| intron 15 | c.2428-36A>C |  | Polymorphism |  | | TCS | [7] |
| intron 15 | c.2659-28delTCTC |  | Polymorphism |  | | TCS | [6] |
| exon 16 | c.2429C>T¹ | p.A810V | Mutation/Polymorphism | Affects PolI interaction site | | TCS | [7,10,20,23,28,30] |
| exon 16 | c.2442delG |  | Mutation | Affects PolI interaction site | | TCS | [10] |
| exon 16 | c.2452C>T | p.Q818\* | Mutation | STOP/Affects PolI interaction site | | TCS | [7] |
| exon 16 | c.2490delC | p.PR830–831PG² | Mutation | STOP/Affects PolI interaction site | | TCS | [20] |
| exon 16 | c.2526insA | p.854\* | Mutation | STOP/Affects PolI interaction site | | TCS | [7] |
| exon 16 | c.2526delAG | p.TG842–843TA² | Mutation | STOP/Affects PolI interaction site | | TCS | [20] |
| exon 16 | c.2527insA |  | Mutation | Affects PolI interaction site | | TCS | [23] |
| exon 16 | c.2529ins G |  | Mutation | STOP/Affects PolI interaction site | | TCS | [25] |
| exon 16 | c.2534C>T¹ | p.S845L | Polymorphism | Affects PolI interaction site | | TCS | [20,23] |
| exon 16 | c.2542C>T | p.G848\* | Mutation | STOP/Affects PolI interaction site | | TCS | [7] |
| exon 16 | c.2545delG | p.872\* | Mutation | STOP/Affects PolI interaction site | | TCS | [7] |
| exon 16 | c.2552delA; 2561delA | p.K851S | Mutation | STOP/Affects PolI interaction site | | TCS | [20] |
| exon 16 | c.2565delAG | p.SG855–856SE² | Mutation | STOP/Affects PolI interaction site | | TCS | [20] |
| exon 16 | c.2622insT |  | Mutation | Affects PolI interaction site | | TCS | [8] |
| exon 16 | c.2660C>T | p.A887V | Polymorphism | Affects PolI interaction site | | TCS | [6,20] |
| exon 16 | c.2683C>T | p.Q895\* | Mutation | STOP/Affects PolI interaction site | | TCS | [5,7] |
| exon 16 | c.2713G>T | p.E905\* | Mutation | STOP/Affects PolI interaction site | | TCS | [5] |
| exon 16 | c.2762C>T | p.P921L | Unclassified variants | Affects PolI interaction site | | TCS | [5] |
| exon 16 | c.2765C>T | p.S992L | Polymorphism | Affects PolI interaction site | | TCS | [6,20] |
| exon 16 | c.2831delA | p.E944Efsx6 | Mutation | Affects PolI interaction site | | TCS | [6] |
| intron 16 | c.2628+26A>G |  | Polymorphism |  | | TCS | [7] |
| intron 16 | c.2629-1G>A |  | Mutation |  | | TCS | [7] |
| intron 16 | c.2629-1G>C |  | Mutation |  | | TCS | [12] |
| intron 16 | c.2629-3A>G |  | Mutation |  | | TCS | [7] |
| intron 16 | c.2859+26A>G |  | Unclassified variants |  | | TCS | [5] |
| intron 16 | c.2859-30G>A |  | Polymorphism |  | | TCS | [6] |
| intron 16 | c.2860-27G>A |  | Polymorphism |  | | TCS | [5] |
| intron 16 | c.21761-21765delCTCTC |  | Polymorphism |  | | TCS | [4] |
| exon 16A | IVS16a +C/T |  | ND |  | | TCS | [30] |
| exon 16A | c.2919G>T | p.R973S | Unclassified variants |  | | TCS | [5] |
| intron 16A | exon 16A+4C>T |  | Polymorphism |  | | TCS | [7] |
| intron 16A | c.2859+3444C > T |  | Polymorphism |  | | TCS | [6] |
| exon 17 | c.2629G>A | p.V877M | Mutation |  | | TCS | [7] |
| exon 17 | c.2643C>T | p.P881P | Polymorphism |  | | TCS | [7] |
| exon 17 | c.2643C>T | p.P881P | Polymorphism |  | | TCS | [28] |
| exon 17 | c.2731C>T | p.R911\* | Mutation | STOP | | TCS | [25,33] |
| exon 17 | c.2740G>T | p.S914\* | Mutation | STOP | | TCS | [8] |
| exon 17 | c.2800C>T | p.Q934\* | Mutation | STOP; affects ATM phosphorylation site | | TCS | [7] |
| exon 17 | c.2802A>G | p.Q934Q | Polymorphism |  | | TCS | [28] |
| exon 17 | c.2924C>T | p.P975L | Polymorphism |  | | TCS | [6] |
| exon 17 | c.2962C>T | p.R988\* | Mutation | STOP | | TCS | [5] |
| exon 17 | c.2969C>A | p.S990\* | Mutation | STOP | | TCS | [5] |
| exon 17 | c.21786T>C | p.V887A | Polymorphism |  | | TCS | [4] |
| exon 17 | c.21968G>T | p.A948S | Polymorphism |  | | TCS | [4] |
| intron 17 | c.2815+1delG |  | Mutation |  | | TCS | [12] |
| intron 17 | c.3046+1G>A |  | Mutation |  | | TCS | [5] |
| intron 17 | c.3047-1G>A |  | Mutation |  | | TCS | [5] |
| intron 17 | c.3047-2A>T |  | Mutation |  | | TCS | [5] |
| intron 17 | c.3047-2A>G |  | Mutation |  | | TCS | [22] |
| exon 18 | c.2822-2823delGA |  | Mutation |  | | TCS | [23] |
| exon 18 | c.2846delC |  | Mutation |  | | TCS | [12] |
| exon 18 | c.2881delG |  | Mutation |  | | TCS | [12] |
| exon 18 | c.2952G>T | p.Q984H | Mutation | affects ATM phoshorylation site | | TCS | [25] |
| exon 18 | c.3053\_3054delGA | p.R1018fs | Mutation |  | | TCS | [15,23] |
| exon 18 | c.3112delG | p.A1038fs | Mutation |  | | TCS | [5,12] |
| exon 18 | c.3118\_3119dupG | p.A1040Gfx47 | Mutation |  | | TCS | [6] |
| exon 18 | c.3156C>T |  | Mutation |  | | TCS | [5] |
| exon 18 | c.3163C>T | p.Q1055\* | Mutation | STOP | | TCS | [5] |
| exon 18 | c.3183>4A |  | Mutation |  | | TCS | [5] |
| exon 19 | c.2998delC |  | Mutation |  | | TCS | [10] |
| exon 19 | c.3066+59C>T, 2355delCAGGGCCAGA |  | Mutation |  | | TCS | [23] |
| exon 19 | c.3156C>T | p.G1052G | ND | STOP | | TCS | [35] |
| intron 19 | c.3066+66C>T¹ |  | Polymorphism |  | | TCS | [7,23] |
| intron 19 | c.3067-72A>C |  | Polymorphism |  | | TCS | [7] |
| intron 19 | c.3197+66C>T |  | Polymorphism |  | | TCS | [6] |
| exon 20 | c.3084delA |  | Mutation |  | | TCS | [12] |
| exon 20 | c.3086-3092delCACTCCC |  | Mutation |  | | TCS | [23] |
| exon 20 | c.3100delA |  | Mutation |  | | TCS | [23] |
| exon 20 | ND | p.Q1041\* | Mutation | STOP | | TCS | [10] |
| exon 20 | c.3169C>T | p.Q1057\* | Mutation | STOP | | TCS | [7] |
| exon 20 | c.3279C>T¹ | p.H1093H | Polymorphism |  | | TCS | [23] |
| exon 20 | c.3314delG | p.G1105fs | Mutation |  | | TCS | [5] |
| exon 20 | c.3456\_63delTTCTTCAG | p.S1152Rfsx3 | Mutation | STOP | | TCS | [6] |
| intron 20 | c.3287-34G>A |  | Polymorphism |  | | TCS | [7] |
| intron 20 | c.3517-34G>A |  | Polymorphism |  | | TCS | [6] |
| exon 21 | c.2103\_2106delTGAG | p.S701fs | Mutation |  | | TCS | [12,15] |
| exon 21 | c.3276G>C | p.R1099P | Polymorphism |  | | TCS | [28] |
| exon 21 | c.3296C>G | p.P1099R | Polymorphism |  | | TCS | [7,23] |
| exon 21 | c.3311delC |  | Mutation | Affects TOPBP1 interaction site | | TCS | [23] |
| exon 21 | c.3527C>G | p.P1176R | Polymorphism |  | | TCS | [6,23] |
| exon 21 | c.3528C>T | p.P1176P | Unclassified variants |  | | TCS | [5] |
| intron 21 | IVS21-3C/T |  | ND |  | | TCS | [30] |
| intron 21 | c.3600+1delG |  | Mutation | Affects TOPBP1 interaction site | | TCS | [5] |
| intron 21 | c.3600+64dupT |  | Unclassified variants |  | | TCS | [5] |
| intron 21 | c.3370-3C>T |  | Polymorphism |  | | TCS | [6,7,12] |
| exon 22 | c.3376-3380delCTCTC |  | Mutation |  | | TCS | [8] |
| exon 22 | c.3389T>A | p.M1130K | Mutation |  | | TCS | [12] |
| exon 22 | c.3447T>C | p.D1149D | Polymorphism |  | | TCS | [28] |
| exon 22 | c.3498T>C | p.\*1168Q | Polymorphism |  | | TCS | [28] |
| exon 22 | c.3611C>G | p.S1204\* | Mutation | STOP | | TCS | [5] |
| exon 22 | c.3612A>C |  | Mutation | loss of exon 22 | | TCS | [11] |
| exon 22 | c.3613G>A |  | Mutation |  | | TCS | [5] |
| exon 22 | c.3700\_3704delACTCT | p.T1234Gfsx5 | Mutation |  | | TCS | [6] |
| exon 22 | c.3745\_3747delinsA | p.P1249fs | Mutation |  | | TCS | [5] |
| intron 22 | c.3550+1G-A |  | Mutation |  | | TCS | [10] |
| intron 22 | c.3781+2dupT |  | Mutation |  | | TCS | [5] |
| intron 22 | c.3781+8A>G |  | Unclassified variants |  | | TCS | [5] |
| exon 23 | c.3606delAG |  | Mutation |  | | TCS | [10] |
| exon 23 | c.3639delG |  | Mutation |  | | TCS | [12,24] |
| exon 23 | c.3700delG |  | Mutation |  | | TCS | [8] |
| exon 23 | c.3711-3712delAG |  | Mutation |  | | TCS | [23] |
| exon 23 | c.3779insC |  | Mutation |  | | TCS | [12] |
| exon 23 | c.3789G>A | p.K1263K | Mutation |  | | TCS | [28] |
| exon 23 | c.3802C>A¹ | p.R1268S | Polymorphism |  | | TCS | [23] |
| exon 23 | c.3819G>A¹ | p.S1273S | Polymorphism |  | | TCS | [23] |
| exon 23 | c.3823delC | p.R1275fs | Mutation |  | | TCS | [5] |
| exon 23 | c.3830delC |  | Mutation |  | | TCS | [12] |
| exon 23 | c.3853delC | p.Q1285fs | Mutation |  | | TCS | [15] |
| exon 23 | c.3853dupC | p.Q1285fs | Mutation |  | | TCS | [5,15] |
| exon 23 | c.3876 delCGGGGAAGGTGGGGAGGCCTCTGTT and insTTC |  | Mutation | STOP | | TCS | [25] |
| exon 23 | c.3899delT |  | Mutation |  | | TCS | [12] |
| exon 23 | c.3933insG |  | Mutation |  | | TCS | [12,24] |
| exon 23 | c.3938C>T¹ | p.A1313V | Mutation/Polymorphism |  | | TCS | [7,10,20,23,24] |
| exon 23 | c.3938T>C | p.V1313A | Polymorphism |  | | TCS | [28] |
| exon 23 | c.3942A>C | p.S1314S | Unclassified variants |  | | TCS | [5] |
| exon 23 | c.3975-3979delGAAAG |  | Mutation |  | | TCS | [12] |
| exon 23 | c.3983\_3986delAGAA | p.K1328fs | Mutation |  | | TCS | [5] |
| exon 23 | c.3987insG | p.1352\* | Mutation | STOP | | TCS | [7,12,23] |
| exon 23 | c.4019-4032delCAGAAGAGGAGCTT |  | Mutation |  | | TCS | [12] |
| exon 23 | c.4061G>C¹ | p.G1354A | Polymorphism |  | | TCS | [7,23] |
| exon 23 | c.4061delC | p.P1354fs | Mutation |  | | TCS | [12,15] |
| exon 23 | c.4108delAA |  | Mutation |  | | TCS | [10] |
| exon 23 | c.4111+5G>C |  | Mutation |  | | TCS | [29] |
| exon 23 | c.4127\_4128delCT | p.S1376fs | Mutation |  | | TCS | [5] |
| exon 23 | c.4131\_4132delTT | p.S1378fs | Mutation |  | | TCS | [5] |
| exon 23 | c.4177\_4178delGA | p.D1393fs | Mutation | Affects NLS | | TCS | [5] |
| exon 23 | c.4218dupG | p.S1407fs | Mutation |  | | TCS | [5,7,12,13,15,23] |
| exon 23 | c.4219delT | p.S1407fs | Mutation |  | | TCS | [5] |
| exon 23 | c.4231C>T | p.Q1411\* | Mutation | STOP; affects ATM phosphorylation site | | TCS | [3,5] |
| exon 23 | c.4295\_4296AT>GA | p.D1432G | Unclassified variants |  | | TCS | [5] |
| exon 23 | c.4321delG | p.E1441fs | Mutation | Affects NLS | | TCS | [5] |
| exon 23 | c.4339\_4340delAA | p.K1447fs | Mutation | Affects NLS | | TCS | [5,10] |
| exon 23 | c.4342A>T | p.R1448\* | Mutation | STOP; affects NLS | | TCS | [5] |
| intron 23 | c.4112-17T>A |  | Polymorphism |  | | TCS | [7] |
| exon 23B | c.4169C>T | p.A1390V | Polymorphism |  | | TCS | [6,23] |
| exon 23B | c.4292G>C | p.G1431A | Polymorphism |  | | TCS | [6,23] |
| exon 23B | c.4331C>T | p.Q1411\* | Mutation | STOP; affects ATM phosphorylation site | | TCS | [3,6] |
| exon 24 | IVS24+350C/G |  | Mutation |  | | TCS | [24] |
| exon 24 | c.4130delAAAAA |  | Mutation |  | | TCS | [10] |
| exon 24 | c.4130-4134delAAAAA |  | Mutation |  | | TCS | [12] |
| exon 24 | c.4131\_4135delAAAAG | p.K1380Efs\* | Mutation | STOP | | TCS | [18] |
| exon 24 | c.4131-4139delAAAAGAAAA | p.1378K-1380Kdel | Mutation |  | | TCS | [9] |
| exon 24 | c.4122delCA | p.1392\* | Mutation | STOP; affects NLS | | TCS | [7] |
| exon 24 | c.4124-4125delAA |  | Mutation |  | | TCS | [12] |
| exon 24 | c.4134delA |  | Mutation |  | | TCS | [12] |
| exon 24 | c.4135 del GAAAA |  | Mutation | STOP | | TCS | [8,10,12,23-25] |
| exon 24 | c.4135-4136insG |  | Mutation |  | | TCS | [12] |
| exon 24 | c.4138\_4142del | p.K1380 | Mutation | STOP | | TCS | [36] |
| exon 24 | c.4148insGAA | p.insK1382 | Polymorphism |  | | TCS | [7] |
| exon 24 | c.4156-4157delAA |  | Mutation |  | | TCS | [12] |
| exon 24 | c.4344dupA | p.R1448fs | Mutation | Affects NLS | | TCS | [15] |
| exon 24 | c.4355\_4356ins14 | p.1456Thrfs\*18 | Mutation | STOP | | TCS | [37] |
| exon 24 | c.4359\_4363delAAAAA | p.E1453Efsx16 | Mutation |  | | TCS | [6,10] |
| exon 24 | c.4361\_4365delAAAAA | p.K1454fs | Mutation | Affects NLS | | TCS | [10,12,15] |
| exon 24 | c.4362\_4365delAAAA | p.E1456fs | Mutation | Affects NLS | | TCS | [5] |
| exon 24 | c.4363\_4365delinsTAG | p.K1455\* | Mutation | STOP; affects NLS | | TCS | [5] |
| exon 24 | c.4365delA | p.E1456fs | Mutation | Affects NLS | | TCS | [5,12] |
| exon 24 | c.4366\_4369delGAAA | p.E1456fs | Mutation | Affects NLS | | TCS | [5] |
| exon 24 | c.4366\_4370delGAAAA | p.K1457fs | Mutation | Affects NLS | | TCS | [6,10,12,13,23] |
| exon 24 | c.4369\_4373delAAGAA | p.K1457fs | Mutation | Affects NLS | | TCS | [5,8,10,12,23,25,37] |
| exon 24 | c.4375\_4377delAAG | p.K1459del | Mutation | Affects NLS | | TCS | [15] |
| exon 24 | c.4377\_4379delGAA | p.K1460del | Unclassified variants | Affects NLS | | TCS | [5] |
| exon 24 | c.38922C>T | p.A1390V | Polymorphism |  | | TCS | [4] |
| intron 24 | IVS24+350C>G |  | Mutation |  | | TCS | [24] |
| intron 24 | IVS24+439C>A |  | Mutation |  | | TCS | [24] |
| intron 24 | c.4209+42C>A |  | Polymorphism |  | | TCS | [7] |
| intron 24 | c.4440+106G>T |  | Unclassified variants |  | | TCS | [5] |
| intron 24 | c.4440+108C>A |  | Polymorphism |  | | TCS | [5] |
| exon 25 | c.4224 G>A, 2822-2823delGA | p.E1508E | Mutation |  | | TCS | [23] |
| ND | c.2757\_2758delAG | p.G920fs | Mutation |  | | TCS | [13,20] |
| **Location** | **cDNA** | **Amino acid change** | **Variant** | **Protein effect** | **Clinical phenotype** | | **Reference** |
| 5’-UTR | c.-89T>G |  | Polymorphism |  | TCS | | [4] |
| 5'-UTR | c.-59G>A |  | Unclassified variants |  | TCS | | [5] |
| 5’-UTR | c.-41G>T |  | Polymorphism |  | TCS | | [6] |
| 5’-UTR | c.-26T>A |  | Polymorphism |  | TCS | | [4] |
| exon 1 | c.1A>T | p.M1? | Mutation |  | TCS | | [5] |
| exon 1 | c.3G>A | p.M1? | Mutation |  | TCS | | [5,7] |
| exon 1 | c.40A>T | p.I14F | Mutation |  | TCS | | [8] |
| exon 1 | c.42C>G | p.I14M | Mutation |  | TCS | | [5] |
| exon 1 | c.50A>G | p.H17R | Mutation |  | TCS | | [5] |
| exon 1 | c.59delG |  | Mutation |  | TCS | | [8] |
| exon 1 | c.61G>C | p.A21P | Mutation |  | TCS | | [5] |
| exon 1 | c.77C>T | p.A26V | Mutation |  | TCS | | [9] |
| exon 1 | c.79G>A | p.A27T | Mutation |  | TCS | | [5] |
| exon 1 | c.87delA | p.V30\* | Mutation | STOP | TCS | | [5] |
| exon 1 | ND | p.Q36\* | Mutation | STOP | TCS | | [10] |
| intron 1 | c.108+62\_68del |  | Unclassified variants |  | TCS | | [5] |
| intron 1 | c.109-1delG |  | Mutation |  | TCS | | [10] |
| intron 1 | c.109-2A>T |  | Mutation |  | TCS | | [5] |
| intron 1 | c.109-28T>C |  | Unclassified variants |  | TCS | | [5] |
| exon 2 | c.122C>T | p.A41V | Polymorphism |  | TCS | | [11] |
| exon 2 | c.149A>G | p.Y50C | Mutation |  | TCS | | [12] |
| exon 2 | c.157T>C | p.W53R | Mutation |  | TCS | | [10,13] |
| exon 2 | c.159G>A | p.W53\* | Mutation | STOP | TCS | | [5] |
| exon 2 | c.163C>T | p.Q55\* | Mutation | STOP | TCS | | [5,9] |
| intron 2 | 1244G>T |  | Polymorphism |  | TCS | | [4] |
| exon 3 | c.165‑1G>A |  | Mutation | STOP | TCS | | [14] |
| exon 3 | c.218insAACC | p.A73fs | Mutation |  | TCS | | [15] |
| exon 3 | c.274 delG |  | Mutation |  | TCS | | [10] |
| exon 3 | ND | p.E93\* | Mutation |  | TCS | | [10] |
| exon 3 | c.303\_304delCA | p.A101Afsx73 | Mutation | STOP | TCS | | [6] |
| exon 3 | c.4369\_4373 | p.K1457-Efs\*12 | ND | affects NLS | TCS | | [16] |
| intron 3 | c.304+5G>C | splice | Mutation |  | TCS | | [5,8,10] |
| intron 3 | c.305-1 G>A |  | Mutation |  | TCS | | [12] |
| intron 3 | c.305-52A>G |  | Polymorphism |  | TCS | | [5] |
| exon 4 | c.343delG | p.121\* | Mutation | STOP | TCS | | [7] |
| exon 4 | c.357\_358delAA | p.S120fs | Mutation |  | TCS | | [5] |
| exon4/intron4 | c.376delAAGGTGAGTGGGACTGCC |  | ND | STOP | TCS | | [17] |
| intron 4 | c.379-2A>G |  | Mutation |  | TCS | | [7] |
| exon 5 | c.381\_382delAG | p.A127\* | Mutation | STOP | TCS | | [18] |
| exon 5 | c.389insA |  | Mutation |  | TCS | | [8] |
| exon 5 | c.390delGA | p.173\* | Mutation | STOP | TCS | | [7] |
| exon 5 | c.405\_406delTG | p.G136fs | Mutation |  | TCS | | [5,10,19] |
| exon 5 | c.408delG | p.214\* | Mutation |  | TCS | | [7] |
| exon 5 | c.422insA | p.H141Q | Mutation | STOP | TCS | | [20] |
| exon 5 | c.431delC | p.T144fs | Mutation |  | TCS | | [15] |
| exon 5 | c.475A>T | p.K159\* | Mutation | STOP | TCS | | [9] |
| exon 5 | c.484\_668ins |  | ND | STOP | TCS | | [21] |
| exon 5 | c.489delC | p.S164Qfs\*55 | Mutation |  | TCS | | [22] |
| exon 5 | c.497delATAC | p.N166I | Mutation | STOP | TCS | | [20] |
| exon 5 | c.503C>T | p.T168M | Polymorphism |  | TCS | | [7] |
| exon 5 | c.519delT | p.T173Tfsx46 | Mutation | STOP | TCS | | [6] |
| intron 5 | c.565+1G>A |  | Mutation |  | TCS | | [5] |
| intron 5 | c.565+18G>C |  | Polymorphism |  | TCS | | [7] |
| intron 5 | c.566-10C>A |  | Mutation |  | TCS | | [5] |
| exon 6 | c.574del(16bp) | p.209\* | Mutation | STOP | TCS | | [7] |
| exon 6 | c.579G>A | p.A193A | Unclassified variants |  | TCS | | [5] |
| exon 6 | c.599delG | p.S200Tfsx19 | Mutation | STOP | TCS | | [6] |
| exon 6 | c.618delC | p.S207fs | Mutation |  | TCS | | [5] |
| exon 6 | c.630 delAG |  | Mutation |  | TCS | | [10] |
| intron 6 | c.639+1G>A |  | Mutation |  | TCS | | [15] |
| intron 6 | c.639+32C>G |  | Polymorphism |  | TCS | | [6,23] |
| intron 6 | c.639+32C>T¹ |  | Polymorphism |  | TCS | | [7,23] |
| intron 6 | c.640-69T>C |  | Polymorphism |  | TCS | | [5] |
| intron 6 | c.14036G>A |  | Polymorphism |  | TCS | | [4] |
| exon 6A | c.648delC | p.S217Qfs\*2 | Mutation |  | TCS | | [22] |
| exon 6A | c.726delT | p.K244fs | Mutation |  | TCS | | [13] |
| exon 6A | c.827\_844del | p.G276\_E281del | Mutation |  | TCS | | [5] |
| exon 7 | IVS6-710 C-T |  | Mutation |  | TCS | | [24] |
| exon 7 | c.698delC |  | Mutation |  | TCS | | [8] |
| exon 7 | c.720-727delAGCACCCC |  | Mutation |  | TCS | | [12,24] |
| exon 7 | c.724insC |  | Mutation |  | TCS | | [10] |
| exon 7 | c.728insC | p.271\* | Mutation | STOP | TCS | | [7] |
| exon 7 | c.744insT |  | Mutation | STOP | TCS | | [25] |
| exon 7 | ND | p.Q252\* | Mutation |  | TCS | | [10] |
| exon 7 | c.768 G>A | p.G256G | Polymorphism |  | TCS | | [7] |
| exon 7 | c.786delA |  | Mutation |  | TCS | | [12] |
| exon 7 | c.786delAG |  | Mutation | STOP | TCS | | [25] |
| exon 7 | c.790\_791delAG | p.S264QfsX7 | Mutation |  | TCS | | [26] |
| exon 7 | c.797G>A | p.S266N | Mutation |  | TCS | | [7] |
| exon 7 | c.803A>G¹ | p.E268G | Polymorphism |  | TCS | | [23] |
| exon 7 | c.1016C>G | p.S339\* | Mutation | STOP | TCS | | [5] |
| exon 7 | c.1028G>A | p.S343N | Unclassified variants |  | TCS | | [5] |
| exon 7 | c.1347T>C | p.P439L | Polymorphism |  | TCS | | [6,20] |
| intron 7 | c.852+39A>G |  | Polymorphism |  | TCS | | [7] |
| intron 7 | c.1083+39G>A |  | Polymorphism |  | TCS | | [5] |
| exon 8 | c.864-865delAG |  | Mutation |  | TCS | | [8] |
| exon 8 | c.911C>T | p.S304L | Mutation |  | TCS | | [27] |
| exon 8 | c.998 C>T¹ | p.S333L | Polymorphism | affects ATM phoshorylation site | TCS | | [23] |
| exon 8 | c.1015 G>T |  | Mutation |  | TCS | | [12] |
| exon 8 | c.1086G>A | p.A362A | Unclassified variants |  | TCS | | [5] |
| exon 8 | c.1095\_1096delAG | p.G366fs | Mutation |  | TCS | | [8,15] |
| exon 8 | c.1096G>T | p.G366\* | Mutation | STOP | TCS | | [5] |
| exon 8 | c.1242C>T | p.S414S | Unclassified variants |  | TCS | | [5] |
| intron 8 | c.1047+60 G>C |  | Polymorphism |  | TCS | | [7] |
| intron 8 | c.1278+60G>C |  | Polymorphism |  | TCS | | [5] |
| exon 9 | c.1084G>A | p.A362T | Mutation |  | TCS | | [28] |
| exon 9 | c.1098delC |  | Mutation |  | TCS | | [12] |
| exon 9 | c.1099A>T | p.K367\* | Mutation | STOP | TCS | | [7] |
| exon 9 | c.1106insC |  | Mutation | affects ATM phoshorylation site | TCS | | [8] |
| exon 9 | c.1120delG |  | Mutation |  | TCS | | [10] |
| exon 9 | c.1142delC |  | Mutation |  | TCS | | [29] |
| exon 9 | c.1150G>A, 3987insG | p.A350A | Mutation |  | TCS | | [23] |
| exon 9 | c.1164insT |  | Mutation |  | TCS | | [23] |
| exon 9 | c.1215insA |  | Mutation |  | TCS | | [12] |
| exon 9 | c.1216insA |  | Mutation |  | TCS | | [10] |
| exon 9 | c.1230A>G | p.E410E | Polymorphism |  | TCS | | [7] |
| exon 9 | c.1247A >G | p.E419E | Polymorphism |  | TCS | | [28] |
| exon 9 | c.1281G>A | p.A427A | Polymorphism |  | TCS | | [5] |
| exon 9 | c.1298delC | p.A433fs | Mutation |  | TCS | | [15] |
| exon 9 | c.1303delC | p.Q435fs | Mutation |  | TCS | | [5] |
| exon 9 | c.1446dupA | p.D483fs | Mutation |  | TCS | | [5,12,19] |
| exon 9 | c.1473dupC | p.M492fs | Mutation |  | TCS | | [5] |
| exon 10 | c.1287insG |  | Mutation |  | TCS | | [23] |
| exon 10 | c.1316C>T | p.P439L | Polymorphism |  | TCS | | [10,20,23] |
| exon 10 | c.1327insA |  | Mutation |  | TCS | | [10] |
| exon 10 | c.1347T>C¹ | p.P449P | Mutation/Polymorphism |  | TCS | | [7,20,23,24,28,30] |
| exon 10 | c.1359G>A | p.G453G | Mutation |  | TCS | | [28] |
| exon 10 | c.1393 C>T | p.Q465\* | Mutation | affects NLS | TCS | | [29] |
| exon 10 | c.1406-1409delAGAG |  | Mutation |  | TCS | | [23,24] |
| exon 10 | c.1408-1409delAG | p.S470Q | Mutation | STOP | TCS | | [12,20,23,24] |
| exon 10 | c.1441insGG |  | Mutation |  | TCS | | [10] |
| exon 10 | c.1557dupA | p.G520fs | Mutation |  | TCS | | [5,31] |
| exon 10 | c.1578 T > C | p.P526P | Mutation/Polymorphism | Affects PolI interaction site | TCS | | [6,13,20] |
| exon 10 | c.1581delG | p.G587Gfsx69 | Mutation | STOP/Affects PolI interaction site | TCS | | [6] |
| exon 10 | c.1609C>T | p.Q537\* | Mutation | STOP/Affects PolI interaction site | TCS | | [15] |
| exon 10 | c.1639\_1640delAG | p.S547fs | Mutation | Affects PolI interaction site | TCS | | [5,6,12,20,32] |
| exon 10 | c.1702C>T | p.Q568\* | Mutation | STOP/Affects PolI interaction site | TCS | | [5] |
| intron 10 | c.1473+68C>T |  | Polymorphism |  | TCS | | [7] |
| exon 11 | c.1530G>T | p.G510G | Polymorphism | Affects PolI interaction site | TCS | | [7,23,28,30] |
| exon 11 | c.1552G>A | p.V518I | Polymorphism | Affects PolI interaction site | TCS | | [7,20,23] |
| exon 11 | c.[1552delG; 1565T< | p.518\* | Mutation | STOP/Affects PolI interaction site | TCS | | [7] |
| exon 11 | c.1611G>A | p.S537S | Mutation/Polymorphism | Affects PolI interaction site | TCS | | [7,20,23,24,28,30] |
| exon 11 | c.1729C>T | p.Q577\* | Mutation | STOP/Affects PolI interaction site | TCS | | [5] |
| exon 11 | c.1747C>T |  | Mutation | STOP/Affects PolI interaction site | TCS | | [33] |
| exon 11 | c.1761G>T | p.G587G | Polymorphism | Affects PolI interaction site | TCS | | [6,23] |
| exon 11 | c.1762C>G¹ | p.P588A | Mutation | Affects PolI interaction site | TCS | | [30] |
| exon 11 | c.1782\_1788del7 | p.Val595fs | Mutation | Affects PolI interaction site | TCS | | [5] |
| exon 11 | c.1813\_1814delAT | p.M605fs | Mutation | Affects PolI interaction site | TCS | | [5] |
| exon 11 | c.1837delG | p.E613fs | Mutation | Affects PolI interaction site | TCS | | [5] |
| exon 11 | c.1842A>G | p.S614S | Polymorphism | Affects PolI interaction site | TCS | | [6,20] |
| exon 11 | c.1854G>A | p.A618A | Unclassified variants | Affects PolI interaction site | TCS | | [5] |
| exon 11 | c.17681C>T | p.P526P | Polymorphism | Affects PolI interaction site | TCS | | [4] |
| exon 11 | c.17693G>A | p.G530G | Polymorphism | Affects PolI interaction site | TCS | | [4] |
| exon 12 | c.1687C>T | p.Q563\* | Mutation | STOP/Affects PolI interaction site | TCS | | [7] |
| exon 12 | c.1719delG | p.N574Tfs | Mutation | Affects PolI interaction site | TCS | | [29] |
| exon 12 | c.1742insC | p.620\* | Mutation | STOP/Affects PolI interaction site | TCS | | [7] |
| exon 12 | c.1762C>G | p.P588A | Polymorphism | Affects PolI interaction site | TCS | | [7,28] |
| exon 12 | c.1768insC | p.620\* | Mutation | STOP/Affects PolI interaction site | TCS | | [7] |
| exon 12 | c.1837G > C | p.A588P | Polymorphism | Affects PolI interaction site | TCS | | [6] |
| exon12 | c.1863A>G, 2355-2364delCAGGGCCAGA | p.E621E | Mutation | Affects PolI interaction site | TCS | | [23] |
| exon 12 | c.1866-1873delAGATAGTG |  | Mutation | Affects PolI interaction site | TCS | | [23] |
| exon 12 | c.1867-1868delGA | p.D623X | Mutation | STOP/Affects PolI interaction site | TCS | | [9,23] |
| exon 12 | c.1868delATAG | p.632\* | Mutation | STOP/Affects PolI interaction site | TCS | | [7] |
| exon 12 | c.1872-1875delTGAG |  | Mutation | Affects PolI interaction site | TCS | | [12] |
| exon 12 | c.1879 delGAGAA |  | Mutation | Affects PolI interaction site | TCS | | [10] |
| exon 12 | c.1907delT | p.L636fs | Mutation | Affects PolI interaction site | TCS | | [5] |
| exon 12 | c.1952\_1956delCTGCA | p.T651fs | Mutation | Affects PolI interaction site | TCS | | [5] |
| exon 12 | c.1953C>T | p.T651T | Unclassified variants | Affects PolI interaction site | TCS | | [5] |
| exon 12 | c.1973delC | p.P658Lfsx53 | Mutation | STOP/Affects PolI interaction site | TCS | | [6] |
| exon 12 | c.1993C>G | p.A665P | Polymorphism | Affects PolI interaction site | TCS | | [6] |
| exon 12 | c.1993delG | p.A665fs | Mutation | Affects PolI interaction site | TCS | | [5] |
| exon 12 | c.1999dupC | p.R667fs | Mutation | Affects PolI interaction site | TCS | | [5,7] |
| exon 12 | c.2065\_2075del11 | p.P689fs | Mutation | Affects PolI interaction site | TCS | | [5] |
| exon 12 | c.2098\_2099delGA | p.D700\* | Mutation | STOP/Affects PolI interaction site | TCS | | [5,23] |
| exon 12 | c.2099\_2102delATAG | p.D700fs | Mutation | Affects PolI interaction site | TCS | | [5,7] |
| exon 12 | c.2103\_2106delTGAG | p.S701fs | Mutation | Affects PolI interaction site | TCS | | [5,12] |
| exon 12 | c.18111A>G | p.S614S | Polymorphism | Affects PolI interaction site | TCS | | [4] |
| intron 12 | c.1911+1G>A |  | Mutation |  | TCS | | [7] |
| intron 12 | c.1911+36delC |  | Polymorphism |  | TCS | | [7] |
| intron 12 | c.2142+22C>T |  | Unclassified variants |  | TCS | | [5] |
| exon 13 | c.1611G>A |  | Mutation | Affects PolI interaction site | TCS | | [24] |
| exon 13 | c.18434G>C | p.A665P | Polymorphism | Affects PolI interaction site | TCS | | [4] |
| exon 13 | c.1915-1916delAA |  | Mutation | Affects PolI interaction site | TCS | | [8] |
| exon 13 | c.1926-1927insG |  | Mutation | Affects PolI interaction site | TCS | | [12] |
| exon 13 | c.1974G>C¹ | p.L658F | Polymorphism | Affects PolI interaction site | TCS | | [23] |
| exon 13 | c.1978delC |  | Mutation | STOP/Affects PolI interaction site | TCS | | [21] |
| exon 13 | c.2014insG |  | Mutation | Affects PolI interaction site | TCS | | [10] |
| exon 13 | c.2014C>T¹ | p.P672S | Polymorphism | Affects PolI interaction site | TCS | | [23] |
| exon 13 | c.2018-2025delCAGTCACC |  | Mutation | Affects PolI interaction site | TCS | | [23,24] |
| exon 13 | c.2019-2025delAGTCACC |  | Mutation | Affects PolI interaction site | TCS | | [8] |
| exon 13 | c.2026C>T | p.Q676\* | Mutation | STOP; affects ATM phosphorylation site, Affects PolI interaction site | TCS | | [8,12] |
| exon 13 | c.2055delAG |  | Mutation | STOP/Affects PolI interaction site | TCS, AMD? | | [34] |
| exon 13 | c.2059delAG |  | Mutation | STOP; Affects PolI interaction site | TCS | | [25] |
| exon 13 | c.2082-2085delTGAG |  | Mutation | Affects PolI interaction site | TCS | | [8] |
| exon 13 | c.2157dupG | p.K720fs | Mutation | Affects PolI interaction site | TCS | | [5,12] |
| exon 13 | c.2164delC | p.L722fs | Mutation | Affects PolI interaction site | TCS | | [5] |
| exon 13 | c.2167C>T | p.Q723\* | Mutation | STOP/Affects PolI interaction site | TCS | | [5] |
| exon 13 | c.2285\_2286delCT | p.S762\* | Mutation | STOP/Affects PolI interaction site | TCS | | [5] |
| exon 13 | c.2285\_2286delCT | p.S762fs | Mutation | STOP/Affects PolI interaction site | TCS | | [6] |
| exon 13 | c.2287G>T | p.E763\* | Mutation | STOP/Affects PolI interaction site | TCS | | [5] |
| intron 13 | c.2341-2A>G |  | Mutation |  | TCS | | [5] |
| exon 14 | c.2110delG |  | Mutation | Affects PolI interaction site | TCS | | [8] |
| exon 14 | c.2221C> T | p.Q741\* | Mutation | STOP/Affects PolI interaction site | TCS | | [28] |
| exon 14 | ND | p.K748K | Mutation | Affects PolI interaction site | TCS | | [19,23] |
| exon 14 | ND | p.K749K | Mutation | Affects PolI interaction site | TCS | | [10] |
| exon 14 | c.2205insTT; 2206delG | p.795\* | Mutation | STOP/Affects PolI interaction site | TCS | | [7] |
| exon 14 | c.2426delC | p.P809fs | Mutation | Affects PolI interaction site | TCS | | [5] |
| exon 14 | c.2473\_2476delTCCA | p.S825fs | Mutation | Affects PolI interaction site | TCS | | [5] |
| exon 14 | c.2478G>A |  | Mutation | Affects PolI interaction site | TCS | | [13,15,19,23] |
| intron 14 | c.2247+27G>A |  | Polymorphism |  | TCS | | [7] |
| intron 14 | c.2248-42insG |  | Polymorphism |  | TCS | | [7] |
| intron 14 | c.2478+5G>C |  | Mutation |  | TCS | | [5] |
| intron 14 | c.2478+5G>A |  | Mutation |  | TCS | | [22] |
| exon 15 | c.2272C>T | p.Q758\* | Mutation | STOP/Affects PolI interaction site | TCS | | [8] |
| exon 15 | c.2297delG |  | Mutation | Affects PolI interaction site | TCS | | [10] |
| exon 15 | c.2354-2363delCAGGGCCAGA |  | Mutation | Affects PolI interaction site | TCS | | [23] |
| exon 15 | c.2355-2356delAG |  | Mutation | Affects PolI interaction site | TCS | | [23] |
| exon 15 | c.2375-2376delGG |  | Mutation | Affects PolI interaction site | TCS | | [12] |
| exon 15 | c.238delG |  | Mutation | Affects PolI interaction site | TCS | | [10] |
| exon 15 | c.2394\_2395delAG | p.D799Qfs\* | Mutation | STOP | TCS | | [18] |
| exon 15 | c.2399delGTGA |  | Mutation | Affects PolI interaction site | TCS | | [10] |
| exon 15 | c.2399-2402delGTGA |  | Mutation | Affects PolI interaction site | TCS | | [8] |
| exon 15 | ND | p.E796\* | Mutation | STOP/Affects PolI interaction site | TCS | | [23] |
| exon 15 | c.2626\_2627delGA | p.D876Qfsx2 | Mutation | STOP/Affects PolI interaction site | TCS | | [6] |
| intron 15 | c.2428-20insCTCT |  | Polymorphism |  | TCS | | [20,23] |
| intron 15 | c.2428-20insCTCTC |  | Polymorphism |  | TCS | | [7,20] |
| intron 15 | c.2428-36A>C |  | Polymorphism |  | TCS | | [7] |
| intron 15 | c.2659-28delTCTC |  | Polymorphism |  | TCS | | [6] |
| exon 16 | c.2429C>T¹ | p.A810V | Mutation/Polymorphism | Affects PolI interaction site | TCS | | [7,10,20,23,28,30] |
| exon 16 | c.2442delG |  | Mutation | Affects PolI interaction site | TCS | | [10] |
| exon 16 | c.2452C>T | p.Q818\* | Mutation | STOP/Affects PolI interaction site | TCS | | [7] |
| exon 16 | c.2490delC | p.PR830–831PG² | Mutation | STOP/Affects PolI interaction site | TCS | | [20] |
| exon 16 | c.2526insA | p.854\* | Mutation | STOP/Affects PolI interaction site | TCS | | [7] |
| exon 16 | c.2526delAG | p.TG842–843TA² | Mutation | STOP/Affects PolI interaction site | TCS | | [20] |
| exon 16 | c.2527insA |  | Mutation | Affects PolI interaction site | TCS | | [23] |
| exon 16 | c.2529ins G |  | Mutation | STOP/Affects PolI interaction site | TCS | | [25] |
| exon 16 | c.2534C>T¹ | p.S845L | Polymorphism | Affects PolI interaction site | TCS | | [20,23] |
| exon 16 | c.2542C>T | p.G848\* | Mutation | STOP/Affects PolI interaction site | TCS | | [7] |
| exon 16 | c.2545delG | p.872\* | Mutation | STOP/Affects PolI interaction site | TCS | | [7] |
| exon 16 | c.2552delA; 2561delA | p.K851S | Mutation | STOP/Affects PolI interaction site | TCS | | [20] |
| exon 16 | c.2565delAG | p.SG855–856SE² | Mutation | STOP/Affects PolI interaction site | TCS | | [20] |
| exon 16 | c.2622insT |  | Mutation | Affects PolI interaction site | TCS | | [8] |
| exon 16 | c.2660C>T | p.A887V | Polymorphism | Affects PolI interaction site | TCS | | [6,20] |
| exon 16 | c.2683C>T | p.Q895\* | Mutation | STOP/Affects PolI interaction site | TCS | | [5,7] |
| exon 16 | c.2713G>T | p.E905\* | Mutation | STOP/Affects PolI interaction site | TCS | | [5] |
| exon 16 | c.2762C>T | p.P921L | Unclassified variants | Affects PolI interaction site | TCS | | [5] |
| exon 16 | c.2765C>T | p.S992L | Polymorphism | Affects PolI interaction site | TCS | | [6,20] |
| exon 16 | c.2831delA | p.E944Efsx6 | Mutation | Affects PolI interaction site | TCS | | [6] |
| intron 16 | c.2628+26A>G |  | Polymorphism |  | TCS | | [7] |
| intron 16 | c.2629-1G>A |  | Mutation |  | TCS | | [7] |
| intron 16 | c.2629-1G>C |  | Mutation |  | TCS | | [12] |
| intron 16 | c.2629-3A>G |  | Mutation |  | TCS | | [7] |
| intron 16 | c.2859+26A>G |  | Unclassified variants |  | TCS | | [5] |
| intron 16 | c.2859-30G>A |  | Polymorphism |  | TCS | | [6] |
| intron 16 | c.2860-27G>A |  | Polymorphism |  | TCS | | [5] |
| intron 16 | c.21761-21765delCTCTC |  | Polymorphism |  | TCS | | [4] |
| exon 16A | IVS16a +C/T |  | ND |  | TCS | | [30] |
| exon 16A | c.2919G>T | p.R973S | Unclassified variants |  | TCS | | [5] |
| intron 16A | exon 16A+4C>T |  | Polymorphism |  | TCS | | [7] |
| intron 16A | c.2859+3444C > T |  | Polymorphism |  | TCS | | [6] |
| exon 17 | c.2629G>A | p.V877M | Mutation |  | TCS | | [7] |
| exon 17 | c.2643C>T | p.P881P | Polymorphism |  | TCS | | [7] |
| exon 17 | c.2643C>T | p.P881P | Polymorphism |  | TCS | | [28] |
| exon 17 | c.2731C>T | p.R911\* | Mutation | STOP | TCS | | [25,33] |
| exon 17 | c.2740G>T | p.S914\* | Mutation | STOP | TCS | | [8] |
| exon 17 | c.2800C>T | p.Q934\* | Mutation | STOP; affects ATM phosphorylation site | TCS | | [7] |
| exon 17 | c.2802A>G | p.Q934Q | Polymorphism |  | TCS | | [28] |
| exon 17 | c.2924C>T | p.P975L | Polymorphism |  | TCS | | [6] |
| exon 17 | c.2962C>T | p.R988\* | Mutation | STOP | TCS | | [5] |
| exon 17 | c.2969C>A | p.S990\* | Mutation | STOP | TCS | | [5] |
| exon 17 | c.21786T>C | p.V887A | Polymorphism |  | TCS | | [4] |
| exon 17 | c.21968G>T | p.A948S | Polymorphism |  | TCS | | [4] |
| intron 17 | c.2815+1delG |  | Mutation |  | TCS | | [12] |
| intron 17 | c.3046+1G>A |  | Mutation |  | TCS | | [5] |
| intron 17 | c.3047-1G>A |  | Mutation |  | TCS | | [5] |
| intron 17 | c.3047-2A>T |  | Mutation |  | TCS | | [5] |
| intron 17 | c.3047-2A>G |  | Mutation |  | TCS | | [22] |
| exon 18 | c.2822-2823delGA |  | Mutation |  | TCS | | [23] |
| exon 18 | c.2846delC |  | Mutation |  | TCS | | [12] |
| exon 18 | c.2881delG |  | Mutation |  | TCS | | [12] |
| exon 18 | c.2952G>T | p.Q984H | Mutation | affects ATM phoshorylation site | TCS | | [25] |
| exon 18 | c.3053\_3054delGA | p.R1018fs | Mutation |  | TCS | | [15,23] |
| exon 18 | c.3112delG | p.A1038fs | Mutation |  | TCS | | [5,12] |
| exon 18 | c.3118\_3119dupG | p.A1040Gfx47 | Mutation |  | TCS | | [6] |
| exon 18 | c.3156C>T |  | Mutation |  | TCS | | [5] |
| exon 18 | c.3163C>T | p.Q1055\* | Mutation | STOP | TCS | | [5] |
| exon 18 | c.3183>4A |  | Mutation |  | TCS | | [5] |
| exon 19 | c.2998delC |  | Mutation |  | TCS | | [10] |
| exon 19 | c.3066+59C>T, 2355delCAGGGCCAGA |  | Mutation |  | TCS | | [23] |
| exon 19 | c.3156C>T | p.G1052G | ND | STOP | TCS | | [35] |
| intron 19 | c.3066+66C>T¹ |  | Polymorphism |  | TCS | | [7,23] |
| intron 19 | c.3067-72A>C |  | Polymorphism |  | TCS | | [7] |
| intron 19 | c.3197+66C>T |  | Polymorphism |  | TCS | | [6] |
| exon 20 | c.3084delA |  | Mutation |  | TCS | | [12] |
| exon 20 | c.3086-3092delCACTCCC |  | Mutation |  | TCS | | [23] |
| exon 20 | c.3100delA |  | Mutation |  | TCS | | [23] |
| exon 20 | ND | p.Q1041\* | Mutation | STOP | TCS | | [10] |
| exon 20 | c.3169C>T | p.Q1057\* | Mutation | STOP | TCS | | [7] |
| exon 20 | c.3279C>T¹ | p.H1093H | Polymorphism |  | TCS | | [23] |
| exon 20 | c.3314delG | p.G1105fs | Mutation |  | TCS | | [5] |
| exon 20 | c.3456\_63delTTCTTCAG | p.S1152Rfsx3 | Mutation | STOP | TCS | | [6] |
| intron 20 | c.3287-34G>A |  | Polymorphism |  | TCS | | [7] |
| intron 20 | c.3517-34G>A |  | Polymorphism |  | TCS | | [6] |
| exon 21 | c.2103\_2106delTGAG | p.S701fs | Mutation |  | TCS | | [12,15] |
| exon 21 | c.3276G>C | p.R1099P | Polymorphism |  | TCS | | [28] |
| exon 21 | c.3296C>G | p.P1099R | Polymorphism |  | TCS | | [7,23] |
| exon 21 | c.3311delC |  | Mutation | Affects TOPBP1 interaction site | TCS | | [23] |
| exon 21 | c.3527C>G | p.P1176R | Polymorphism |  | TCS | | [6,23] |
| exon 21 | c.3528C>T | p.P1176P | Unclassified variants |  | TCS | | [5] |
| intron 21 | IVS21-3C/T |  | ND |  | TCS | | [30] |
| intron 21 | c.3600+1delG |  | Mutation | Affects TOPBP1 interaction site | TCS | | [5] |
| intron 21 | c.3600+64dupT |  | Unclassified variants |  | TCS | | [5] |
| intron 21 | c.3370-3C>T |  | Polymorphism |  | TCS | | [6,7,12] |
| exon 22 | c.3376-3380delCTCTC |  | Mutation |  | TCS | | [8] |
| exon 22 | c.3389T>A | p.M1130K | Mutation |  | TCS | | [12] |
| exon 22 | c.3447T>C | p.D1149D | Polymorphism |  | TCS | | [28] |
| exon 22 | c.3498T>C | p.\*1168Q | Polymorphism |  | TCS | | [28] |
| exon 22 | c.3611C>G | p.S1204\* | Mutation | STOP | TCS | | [5] |
| exon 22 | c.3612A>C |  | Mutation | loss of exon 22 | TCS | | [11] |
| exon 22 | c.3613G>A |  | Mutation |  | TCS | | [5] |
| exon 22 | c.3700\_3704delACTCT | p.T1234Gfsx5 | Mutation |  | TCS | | [6] |
| exon 22 | c.3745\_3747delinsA | p.P1249fs | Mutation |  | TCS | | [5] |
| intron 22 | c.3550+1G-A |  | Mutation |  | TCS | | [10] |
| intron 22 | c.3781+2dupT |  | Mutation |  | TCS | | [5] |
| intron 22 | c.3781+8A>G |  | Unclassified variants |  | TCS | | [5] |
| exon 23 | c.3606delAG |  | Mutation |  | TCS | | [10] |
| exon 23 | c.3639delG |  | Mutation |  | TCS | | [12,24] |
| exon 23 | c.3700delG |  | Mutation |  | TCS | | [8] |
| exon 23 | c.3711-3712delAG |  | Mutation |  | TCS | | [23] |
| exon 23 | c.3779insC |  | Mutation |  | TCS | | [12] |
| exon 23 | c.3789G>A | p.K1263K | Mutation |  | TCS | | [28] |
| exon 23 | c.3802C>A¹ | p.R1268S | Polymorphism |  | TCS | | [23] |
| exon 23 | c.3819G>A¹ | p.S1273S | Polymorphism |  | TCS | | [23] |
| exon 23 | c.3823delC | p.R1275fs | Mutation |  | TCS | | [5] |
| exon 23 | c.3830delC |  | Mutation |  | TCS | | [12] |
| exon 23 | c.3853delC | p.Q1285fs | Mutation |  | TCS | | [15] |
| exon 23 | c.3853dupC | p.Q1285fs | Mutation |  | TCS | | [5,15] |
| exon 23 | c.3876 delCGGGGAAGGTGGGGAGGCCTCTGTT and insTTC |  | Mutation | STOP | TCS | | [25] |
| exon 23 | c.3899delT |  | Mutation |  | TCS | | [12] |
| exon 23 | c.3933insG |  | Mutation |  | TCS | | [12,24] |
| exon 23 | c.3938C>T¹ | p.A1313V | Mutation/Polymorphism |  | TCS | | [7,10,20,23,24] |
| exon 23 | c.3938T>C | p.V1313A | Polymorphism |  | TCS | | [28] |
| exon 23 | c.3942A>C | p.S1314S | Unclassified variants |  | TCS | | [5] |
| exon 23 | c.3975-3979delGAAAG |  | Mutation |  | TCS | | [12] |
| exon 23 | c.3983\_3986delAGAA | p.K1328fs | Mutation |  | TCS | | [5] |
| exon 23 | c.3987insG | p.1352\* | Mutation | STOP | TCS | | [7,12,23] |
| exon 23 | c.4019-4032delCAGAAGAGGAGCTT |  | Mutation |  | TCS | | [12] |
| exon 23 | c.4061G>C¹ | p.G1354A | Polymorphism |  | TCS | | [7,23] |
| exon 23 | c.4061delC | p.P1354fs | Mutation |  | TCS | | [12,15] |
| exon 23 | c.4108delAA |  | Mutation |  | TCS | | [10] |
| exon 23 | c.4111+5G>C |  | Mutation |  | TCS | | [29] |
| exon 23 | c.4127\_4128delCT | p.S1376fs | Mutation |  | TCS | | [5] |
| exon 23 | c.4131\_4132delTT | p.S1378fs | Mutation |  | TCS | | [5] |
| exon 23 | c.4177\_4178delGA | p.D1393fs | Mutation | Affects NLS | TCS | | [5] |
| exon 23 | c.4218dupG | p.S1407fs | Mutation |  | TCS | | [5,7,12,13,15,23] |
| exon 23 | c.4219delT | p.S1407fs | Mutation |  | TCS | | [5] |
| exon 23 | c.4231C>T | p.Q1411\* | Mutation | STOP; affects ATM phosphorylation site | TCS | | [3,5] |
| exon 23 | c.4295\_4296AT>GA | p.D1432G | Unclassified variants |  | TCS | | [5] |
| exon 23 | c.4321delG | p.E1441fs | Mutation | Affects NLS | TCS | | [5] |
| exon 23 | c.4339\_4340delAA | p.K1447fs | Mutation | Affects NLS | TCS | | [5,10] |
| exon 23 | c.4342A>T | p.R1448\* | Mutation | STOP; affects NLS | TCS | | [5] |
| intron 23 | c.4112-17T>A |  | Polymorphism |  | TCS | | [7] |
| exon 23B | c.4169C>T | p.A1390V | Polymorphism |  | TCS | | [6,23] |
| exon 23B | c.4292G>C | p.G1431A | Polymorphism |  | TCS | | [6,23] |
| exon 23B | c.4331C>T | p.Q1411\* | Mutation | STOP; affects ATM phosphorylation site | TCS | | [3,6] |
| exon 24 | IVS24+350C/G |  | Mutation |  | TCS | | [24] |
| exon 24 | c.4130delAAAAA |  | Mutation |  | TCS | | [10] |
| exon 24 | c.4130-4134delAAAAA |  | Mutation |  | TCS | | [12] |
| exon 24 | c.4131\_4135delAAAAG | p.K1380Efs\* | Mutation | STOP | TCS | | [18] |
| exon 24 | c.4131-4139delAAAAGAAAA | p.1378K-1380Kdel | Mutation |  | TCS | | [9] |
| exon 24 | c.4122delCA | p.1392\* | Mutation | STOP; affects NLS | TCS | | [7] |
| exon 24 | c.4124-4125delAA |  | Mutation |  | TCS | | [12] |
| exon 24 | c.4134delA |  | Mutation |  | TCS | | [12] |
| exon 24 | c.4135 del GAAAA |  | Mutation | STOP | TCS | | [8,10,12,23-25] |
| exon 24 | c.4135-4136insG |  | Mutation |  | TCS | | [12] |
| exon 24 | c.4138\_4142del | p.K1380 | Mutation | STOP | TCS | | [36] |
| exon 24 | c.4148insGAA | p.insK1382 | Polymorphism |  | TCS | | [7] |
| exon 24 | c.4156-4157delAA |  | Mutation |  | TCS | | [12] |
| exon 24 | c.4344dupA | p.R1448fs | Mutation | Affects NLS | TCS | | [15] |
| exon 24 | c.4355\_4356ins14 | p.1456Thrfs\*18 | Mutation | STOP | TCS | | [37] |
| exon 24 | c.4359\_4363delAAAAA | p.E1453Efsx16 | Mutation |  | TCS | | [6,10] |
| exon 24 | c.4361\_4365delAAAAA | p.K1454fs | Mutation | Affects NLS | TCS | | [10,12,15] |
| exon 24 | c.4362\_4365delAAAA | p.E1456fs | Mutation | Affects NLS | TCS | | [5] |
| exon 24 | c.4363\_4365delinsTAG | p.K1455\* | Mutation | STOP; affects NLS | TCS | | [5] |
| exon 24 | c.4365delA | p.E1456fs | Mutation | Affects NLS | TCS | | [5,12] |
| exon 24 | c.4366\_4369delGAAA | p.E1456fs | Mutation | Affects NLS | TCS | | [5] |
| exon 24 | c.4366\_4370delGAAAA | p.K1457fs | Mutation | Affects NLS | TCS | | [6,10,12,13,23] |
| exon 24 | c.4369\_4373delAAGAA | p.K1457fs | Mutation | Affects NLS | TCS | | [5,8,10,12,23,25,37] |
| exon 24 | c.4375\_4377delAAG | p.K1459del | Mutation | Affects NLS | TCS | | [15] |
| exon 24 | c.4377\_4379delGAA | p.K1460del | Unclassified variants | Affects NLS | TCS | | [5] |
| exon 24 | c.38922C>T | p.A1390V | Polymorphism |  | TCS | | [4] |
| intron 24 | IVS24+350C>G |  | Mutation |  | TCS | | [24] |
| intron 24 | IVS24+439C>A |  | Mutation |  | TCS | | [24] |
| intron 24 | c.4209+42C>A |  | Polymorphism |  | TCS | | [7] |
| intron 24 | c.4440+106G>T |  | Unclassified variants |  | TCS | | [5] |
| intron 24 | c.4440+108C>A |  | Polymorphism |  | TCS | | [5] |
| exon 25 | c.4224 G>A, 2822-2823delGA | p.E1508E | Mutation |  | TCS | | [23] |
| ND | c.2757\_2758delAG | p.G920fs | Mutation |  | TCS | | [13,20] |

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