**Table S2. 849 unique nonsense mutations from LOVD, HGMD, ClinVar and cohort of internal patients.**

| **EXON** | **CDS** | **PROTEIN** | **hg19** | **Reference** | **Disease** | **Patients**  **(from LOVD)** |
| --- | --- | --- | --- | --- | --- | --- |
| 1 | c.8G>A | p.(Trp3\*) | g.33229422C>T | [1] | BMD | 1 |
| 1 | c.9G>A | p.[Trp3\*, Leu2\_Met124del, Leu2\_Met128del] | g.33229421C>T | [2] | BMD | 27 |
| 1 | c.11G>A | p.(Trp4\*) | g.33229419C>T | [3, 4] | BMD | 6 |
| 1 | c.12G>A | p.(Trp4\*) | g.33229418C>T | [3] | DMD | 1 |
| 2 | c.49C>T | p.(Gln17\*) | g.33038300G>A | [5] | BMD | 1 |
| 2 | c.67A>T | p.(Lys23\*) | g.33038282T>A | [1] | BMD | 1 |
| 2 | c.72G>A | p.(Trp24\*) | g.33038277C>T | [6] | BMD | 1 |
| 2 | c.82C>T | p.(Gln28\*) | g.33038267G>A | [2] | ND | 2 |
| 3 | c.100A>T | p.(Lys34\*) | g.32867931T>A | [7] | DMD/BMD | 1 |
| 3 | c.103C>T | p.(Gln35\*) | g.32867928G>A | [8] | BMD | 2 |
| 3 | c.133C>T | p.(Gln45\*) | g.32867898G>A | [4, 9] | DMD | 9 |
| 3 | c.163G>T | p.[Glu55\*, Phe32Metfs\*13] | g.32867868C>A | [10] | BMD | 1 |
| 3 | c.178C>T | p.(Gln60\*) | g.32867853G>A | [11] | DMD | 3 |
| 4 | c.193G>T | p.(Glu65\*) | g.32862971C>A | [12] | DMD | 1 |
| 4 | c.196A>T | p.(Lys66\*) | g.32862968T>A | [13] | DMD | 1 |
| 4 | c.199G>T | p.(Gly67\*) | g.32862965C>A | [14] | DMD | 4 |
| 4 | c.253C>T | p.(Gln85\*) | g.32862911G>A | [11] | DMD | 4 |
| 5 | c.272T>A | p.(Leu91\*) | g.32841497A>T | [15] | DMD | 1 |
| 5 | c.283G>T | p.(Gly95\*) | g.32841486C>A | [16] | DMD/BMD | 1 |
| 5 | c.313A>T | p.(Lys105\*) | g.32841456T>A | [17] | DMD/BMD | 3 |
| 5 | c.336G>A | p.(Trp112\*) | g.32841433C>T | [3] | BMD | 3 |
| 5 | c.353G>A | p.(Trp118\*) | g.32841416C>T | [4] | DMD | 1 |
| 5 | c.354G>A | p.(Trp118\*) | g.32841415C>T | [18] | DMD | 2 |
| 5 | c.355C>T | p.(Gln119\*) | g.32841414G>A | [19] | DMD/BMD | 7 |
| 6 | c.361A>T | p.(Lys121\*) | g.32834754T>A | [20] | DMD/BMD | 1 |
| 6 | c.394C>T | p.(Gln132\*) | g.32834721G>A | [21] | DMD | 2 |
| 6 | c.397C>T | p.(Gln133\*) | g.32834718G>A | [1] | DMD | 1 |
| 6 | c.409G>T | p.(Glu137\*) | g.32834706C>A | [22] | DMD | 2 |
| 6 | c.428G>A | p.(Trp143\*) | g.32834687C>T | [13] | DMD | 1 |
| 6 | c.429G>A | p.(Trp143\*) | g.32834686C>T | [19] | DMD | 1 |
| 6 | c.433C>T | p.(Arg145\*) | g.32834682G>A | [19, 23] | DMD | 38 |
| 6 | c.436C>T | p.(Gln146\*) | g.32834679G>A | [24] | DMD | 5 |
| 6 | c.440C>A | p.(Ser147\*) | g.32834675G>T | [25] | DMD | 1 |
| 6 | c.440C>G | p.(Ser147\*) | g.32834675G>C | [19, 26] | DMD | 4 |
| 6 | c.453T>A | p.(Tyr151\*) | g.32834662A>T | [27] | DMD | 1 |
| 6 | c.457C>T | p.(Gln153\*) | g.32834658G>A | [19] | DMD | 2 |
| 6 | c.488G>A | p.(Trp163\*) | g.32834627C>T | [28] | DMD | 3 |
| 6 | c.489G>A | p.(Trp163\*) | g.32834626C>T | [1] | DMD/BMD | 1 |
| 7 | c.549G>A | p.(Trp183\*) | g.32827710C>T | [19] | DMD/BMD | 1 |
| 7 | c.564C>A | p.(Cys188\*) | g.32827695G>T | [29] | DMD | 1 |
| 7 | c.565C>T | p.(Gln189\*) | g.32827694G>A | [30] | DMD | 4 |
| 7 | c.568C>T | p.(Gln190\*) | g.32827691G>A | [19] | DMD | 10 |
| 7 | c.572C>G | p.(Ser191\*) | g.32827687G>C | [28] | DMD | 4 |
| 7 | c.580C>T | p.(Gln194\*) | g.32827679G>A | [19, 21] | DMD | 5 |
| 7 | c.583C>T | p.(Arg195\*) | g.32827676G>A | [10] | DMD | 37 |
| 7 | c.589G>T | p.(Glu197\*) | g.32827670C>A | [31] | ND | 1 |
| 7 | c.615T>A | p.(Tyr205\*) | g.32827644A>T | [25] | DMD | 3 |
| 7 | c.616C>T | p.(Gln206\*) | g.32827643G>A | [32] | DMD | 1 |
| 7 | c.620T>G | p.(Leu207\*) | g.32827639A>C | [20] | DMD | 3 |
| 7 | c.646G>T | p.(Glu216\*) | g.32827613C>A | [1] | DMD | 1 |
| 8 | c.673A>T | p.(Lys225\*) | g.32717387T>A | [12] | DMD | 1 |
| 8 | c.686T>G | p.(Leu229\*) | g.32717374A>C | [16] | DMD | 1 |
| 8 | c.686T>A | p.(Leu229\*) | g.32717374A>T | [19] | OTHER | 2 |
| 8 | c.693C>A | p.(Tyr231\*) | g.32717367G>T | [4] | DMD | 1 |
| 8 | c.701C>A | p.(Ser234\*) | g.32717359G>T | [33] | DMD | 6 |
| 8 | c.701C>G | p.(Ser234\*) | g.32717359G>C | [13] | DMD | 1 |
| 8 | c.709C>T | p.(Gln237\*) | g.32717351G>A | [34] | DMD | 4 |
| 8 | c.721C>T | p.(Gln241\*) | g.32717339G>A | [10] | DMD | 2 |
| 8 | c.724C>T | p.(Gln242\*) | g.32717336G>A | [35] | DMD | 9 |
| 8 | c.745C>T | p.(Gln249\*) | g.32717315G>A | [36] | DMD | 3 |
| 8 | c.748G>T | p.(Glu250\*) | g.32717312C>A | [11] | DMD | 2 |
| 8 | c.754G>T | p.(Glu252\*) | g.32717306C>A | [34] | DMD | 3 |
| 8 | c.775A>T | p.(Lys259\*) | g.32717285T>A | [20] | DMD | 1 |
| 8 | c.799C>T | p.(Gln267\*) | g.32717261G>A | [19] | DMD | 4 |
| 8 | c.811C>T | p.(Gln271\*) | g.32717249G>A | [20] | DMD | 3 |
| 8 | c.826C>T | p.(Gln276\*) | g.32717234G>A | [3] | DMD | 2 |
| 8 | c.829C>T | p.(Gln277\*) | g.32717231G>A | [37][3] | DMD | 9 |
| 9 | c.853G>T | p.(Gly285\*) | g.32716094C>A | [13, 38] | DMD | 4 |
| 9 | c.858T>G | p.(Tyr286\*) | g.32716089A>C | [1] | DMD | 1 |
| 9 | c.883C>T | p.[Arg295\*,Ile278\_Gln320del] | g.32716064G>A | [38] | DMD/BMD | 9 |
| 9 | c.897T>G | p.[Tyr299\*; Ile278\_Gln320del] | g.32716050A>C | [1] | DMD | 1 |
| 9 | c.903C>A | p.(Tyr301\*) | g.32716044G>T | [39] | DMD | 1 |
| 9 | c.903C>G | p.(Tyr301\*) | g.32716044G>C | [19] | DMD | 2 |
| 9 | c.907C>T | p.(Gln303\*) | g.32716040G>A | [28] | ND | 2 |
| 9 | c.956C>G | p.(Ser319\*) | g.32715991G>C | [39] | DMD | 1 |
| 10 | c.965T>A | p.(Leu322\*) | g.32663265A>T | [25] | DMD | 1 |
| 10 | c.986C>G | p.(Ser329\*) | g.32663244G>C | [1] | ND | 1 |
| 10 | c.998C>A | p.(Ser333\*) | g.32663232G>T | [25] | DMD | 5 |
| 10 | c.1006G>T | p.(Glu336\*) | g.32663224C>A | [40] | DMD | 2 |
| 10 | c.1012G>T | p.(Glu338\*) | g.32663218C>A | [19] | DMD | 1 |
| 10 | c.1033C>T | p.(Gln345\*) | g.32663197G>A | [20] | DMD | 3 |
| 10 | c.1045G>T | p.(Glu349\*) | g.32663185C>A | [1] | DMD | 1 |
| 10 | c.1048G>T | p.(Glu350\*) | g.32663182C>A | [1] | DMD/BMD | 2 |
| 10 | c.1055T>G | p.(Leu352\*) | g.32663175A>C | [41] | DMD | 2 |
| 10 | c.1061G>A | p.(Trp354\*) | g.32663169C>T | [13, 19] | DMD/BMD | 4 |
| 10 | c.1062G>A | p.(Trp354\*) | g.32663168C>T | [10] | DMD | 5 |
| 10 | c.1075G>T | p.(Glu359\*) | g.32663155C>A | [16] | ND | 1 |
| 10 | c.1087C>T | p.(Gln363\*) | g.32663143G>A | [18] | DMD | 1 |
| 10 | c.1093C>T | p.(Gln365\*) | g.32663137G>A | [22] | DMD | 4 |
| 10 | c.1099G>T | p.(Glu367\*) | g.32663131C>A | [21] | DMD | 1 |
| 10 | c.1117G>T | p.(Glu373\*) | g.32663113C>A | [19] | DMD | 3 |
| 10 | c.1132C>T | p.(Gln378\*) | g.32663098G>A | [28] | DMD | 2 |
| 11 | c.1177C>T | p.(Gln393\*) | g.32662403G>A | [21] | DMD | 2 |
| 11 | c.1201C>T | p.(Gln401\*) | g.32662379G>A | [7] | DMD/BMD | 2 |
| 11 | c.1207G>T | p.(Gly403\*) | g.32662373C>A | [37] | ND | 1 |
| 11 | c.1238C>A | p.(Ser413\*) | g.32662342G>T | [1] | DMD | 1 |
| 11 | c.1238C>G | p.(Ser413\*) | g.32662342G>C | [42] | DMD | 1 |
| 11 | c.1249G>T | p.(Glu417\*) | g.32662331C>A | [23] | DMD | 1 |
| 11 | c.1255G>T | p.(Glu419\*) | g.32662325C>A | [32] | DMD | 2 |
| 11 | c.1261C>T | p.(Gln421\*) | g.32662319G>A | [26] | DMD | 4 |
| 11 | c.1264G>T | p.(Glu422\*) | g.32662316C>A | [1] | DMD | 1 |
| 11 | c.1267C>T | p.(Gln423\*) | g.32662313G>A | [19] | DMD | 6 |
| 11 | c.1286C>A | p.(Ser429\*) | g.32662294G>T | [19] | DMD/BMD | 2 |
| 11 | c.1286C>G | p.(Ser429\*) | g.32662294G>C | [1] | DMD/BMD | 1 |
| 11 | c.1288A>T | p.(Arg430\*) | g.32662292T>A | [16] | ND | 1 |
| 11 | c.1292G>A | p.(Trp431\*) | g.32662288C>T | [25] | DMD | 3 |
| 11 | c.1293G>A | p.(Trp431\*) | g.32662287C>T | [1] | DMD | 2 |
| 11 | c.1324C>T | p.(Gln442\*) | g.32662256G>A | [10] | DMD | 1 |
| 12 | c.1334T>G | p.(Leu445\*) | g.32632568A>C | [1] | DMD | 1 |
| 12 | c.1339A>T | p.(Arg447\*) | g.32632563T>A | [13] | DMD | 2 |
| 12 | c.1357C>T | p.(Gln453\*) | g.32632545G>A | [13, 37] | DMD | 4 |
| 12 | c.1363C>T | p.(Gln455\*) | g.32632539G>A | [3, 21] | DMD | 3 |
| 12 | c.1366A>T | p.(Lys456\*) | g.32632536T>A | [1] | DMD | 1 |
| 12 | c.1372A>T | p.(Lys458\*) | g.32632530T>A | [13] | DMD | 1 |
| 12 | c.1388G>A | p.(Trp463\*) | g.32632514C>T | [13, 19] | DMD | 4 |
| 12 | c.1389G>A | p.(Trp463\*) | g.32632513C>T | [1] | DMD | 1 |
| 12 | c.1417A>T | p.(Lys473\*) | g.32632485T>A | [16] | DMD/BMD | 1 |
| 12 | c.1426G>T | p.(Glu476\*) | g.32632476C>A | [23] | DMD | 2 |
| 12 | c.1429G>T | p.(Glu477\*) | g.32632473C>A | [19] | DMD/BMD | 3 |
| 12 | c.1438G>T | p.(Gly480\*) | g.32632464C>A | [4] | DMD/BMD | 2 |
| 12 | c.1465C>T | p.(Gln489\*) | g.32632437G>A | [10] | DMD/BMD | 4 |
| 12 | c.1471C>T | p.(Gln491\*) | g.32632431G>A | [1] | ND | 1 |
| 12 | c.1474C>T | p.(Gln492\*) | g.32632428G>A | [18, 26] | DMD | 4 |
| 13 | c.1489C>T | p.(Gln497\*) | g.32613987G>A | [43] | DMD | 4 |
| 13 | c.1504C>T | p.(Gln502\*) | g.32613972G>A | [13] | DMD | 1 |
| 13 | c.1507G>T | p.(Glu503\*) | g.32613969C>A | [1] | OTHER | 1 |
| 13 | c.1510C>T | p.(Gln504\*) | g.32613966G>A | [4, 44] | DMD | 4 |
| 13 | c.1555G>T | p.(Glu519\*) | g.32613921C>A | [45] | DMD | 1 |
| 13 | c.1594C>T | p.(Gln532\*) | g.32613882G>A | [30] | DMD | 6 |
| 14 | c.1609G>T | p.(Gly537\*) | g.32591957C>A | [28] | ND | 1 |
| 14 | c.1615C>T | p.(Arg539\*) | g.32591951G>A | [20, 40] | DMD | 21 |
| 14 | c.1619G>A | p.(Trp540\*) | g.32591947C>T | [46] | DMD | 3 |
| 14 | c.1620G>A | p.(Trp540\*) | g.32591946C>T | [1] | DMD | 1 |
| 14 | c.1633A>T | p.(Arg545\*) | g.32591933T>A | [12] | DMD | 1 |
| 14 | c.1637G>A | p.(Trp546\*) | g.32591929C>T | [37] | DMD | 3 |
| 14 | c.1638G>A | p.(Trp546\*) | g.32591928C>T | [3, 23, 43] | DMD | 4 |
| 14 | c.1642G>T | p.(Glu548\*) | g.32591924C>A | [1] | DMD | 1 |
| 14 | c.1652G>A | p.(Trp551\*) | g.32591914C>T | [13] | DMD | 4 |
| 14 | c.1653G>A | p.(Trp551\*) | g.32591913C>T | [1] | DMD | 1 |
| 14 | c.1663C>T | p.(Gln555\*) | g.32591903G>A | [46] | DMD | 15 |
| 14 | c.1682G>A | p.(Trp561\*) | g.32591884C>T | [30] | DMD | 1 |
| 14 | c.1683G>A | p.(Trp561\*) | g.32591883C>T | [25] | DMD | 3 |
| 14 | c.1684C>T | p.(Gln562\*) | g.32591882G>A | [18] | DMD | 3 |
| 14 | c.1699G>T | p.(Glu567\*) | g.32591867C>A | [28] | ND | 1 |
| 14 | c.1702C>T | p.(Gln568\*) | g.32591864G>A | [34] | DMD | 9 |
| 15 | c.1707C>A | p.(Cys569\*) | g.32591752G>T | [13] | DMD/BMD | 2 |
| 15 | c.1721G>A | p.(Trp574\*) | g.32591738C>T | [47] | DMD | 2 |
| 15 | c.1727C>A | p.(Ser576\*) | g.32591732G>T | [19] | DMD/BMD | 2 |
| 15 | c.1729G>T | p.(Glu577\*) | g.32591730C>A | [7] | DMD/BMD | 2 |
| 15 | c.1732A>T | p.(Lys578\*) | g.32591727T>A | [13] | DMD | 2 |
| 15 | c.1777C>T | p.(Gln593\*) | g.32591682G>A | [3] | DMD | 2 |
| 15 | c.1783G>T | p.(Glu595\*) | g.32591676C>A | [19] | DMD | 3 |
| 15 | c.1793C>G | p.(Ser598\*) | g.32591666G>C | [18, 19] | DMD | 3 |
| 16 | c.1817T>G | p.(Leu606\*) | g.32583994A>C | [1] | DMD | 2 |
| 16 | c.1831G>T | p.(Glu611\*) | g.32583980C>A | [7] | DMD/BMD | 1 |
| 16 | c.1837A>T | p.(Lys613\*) | g.32583974T>A | [21] | DMD | 1 |
| 16 | c.1843C>T | p.(Gln615\*) | g.32583968G>A | [3, 23] | DMD | 3 |
| 16 | c.1865C>G | p.(Ser622\*) | g.32583946G>C | [17] | DMD | 2 |
| 16 | c.1865C>A | p.(Ser622\*) | g.32583946G>T | [16] | ND | 1 |
| 16 | c.1873C>T | p.(Gln625\*) | g.32583938G>A | [48] | DMD | 1 |
| 16 | c.1886C>G | p.(Ser629\*) | g.32583925G>C | [49] | DMD | 3 |
| 16 | c.1886C>A | p.(Ser629\*) | g.32583925G>T | [16] | ND | 1 |
| 16 | c.1900A>T | p.(Lys634\*) | g.32583911T>A | [1] | DMD/BMD | 1 |
| 16 | c.1904C>G | p.(Ser635\*) | g.32583907G>C | [20] | DMD | 2 |
| 16 | c.1912C>T | p.(Gln638\*) | g.32583899G>A | [7] | DMD/BMD | 1 |
| 16 | c.1928G>A | p.(Trp643\*) | g.32583883C>T | [50] | DMD | 2 |
| 16 | c.1952G>A | p.(Trp651\*) | g.32583859C>T | [11] | DMD | 2 |
| 16 | c.1961T>A | p.(Leu654\*) | g.32583850A>T | [19] | DMD | 2 |
| 16 | c.1961T>G | p.(Leu654\*) | g.32583850A>C | [1] | DMD/BMD | 2 |
| 16 | c.1966C>T | p.(Gln656\*) | g.32583845G>A | [19] | DMD | 3 |
| 16 | c.1975G>T | p.(Glu659\*) | g.32583836C>A | [1] | DMD | 1 |
| 16 | c.1990C>T | p.(Gln664\*) | g.32583821G>A | [46] | DMD | 7 |
| 17 | c.1997C>A | p.(Ser666\*) | g.32563447G>T | [3] | DMD | 1 |
| 17 | c.1999C>T | p.(Gln667\*) | g.32563445G>A | [1] | DMD | 1 |
| 17 | c.2017C>T | p.(Gln673\*) | g.32563427G>A | [13] | DMD | 3 |
| 17 | c.2032C>T | p.(Gln678\*) | g.32563412G>A | [4, 13] | DMD | 5 |
| 17 | c.2047G>T | p.(Glu683\*) | g.32563397C>A | [18] | DMD | 3 |
| 17 | c.2074G>T | p.(Glu692\*) | g.32563370C>A | [21] | ND | 1 |
| 17 | c.2077C>T | p.(Gln693\*) | g.32563367G>A | [19] | DMD | 5 |
| 17 | c.2089A>T | p.(Lys697\*) | g.32563355T>A | [18] | DMD | 1 |
| 17 | c.2098C>T | p.(Gln700\*) | g.32563346G>A | [51] | DMD | 1 |
| 17 | c.2101G>T | p.(Glu701\*) | g.32563343C>A | [16] | OTHER | 1 |
| 17 | c.2104G>T | p.(Glu702\*) | g.32563340C>A | [19] | OTHER | 3 |
| 17 | c.2125C>T | p.(Gln709\*) | g.32563319G>A | [52] | DMD | 4 |
| 17 | c.2128A>T | p.(Lys710\*) | g.32563316T>A | [30] | DMD | 1 |
| 17 | c.2137C>T | p.(Gln713\*) | g.32563307G>A | [53] | DMD | 4 |
| 18 | c.2201G>A | p.(Trp734\*) | g.32536216C>T | Internal patient | DMD | 1 |
| 18 | c.2202G>A | p.(Trp734\*) | g.32536215C>T | [16] | ND | 1 |
| 18 | c.2213C>G | p.(Ser738\*) | g.32536204G>C | [7] | DMD/BMD | 3 |
| 18 | c.2215G>T | p.(Glu739\*) | g.32536202C>A | [1] | DMD | 1 |
| 18 | c.2227C>T | p.(Gln743\*) | g.32536190G>A | [4] | DMD | 6 |
| 18 | c.2236G>T | p.(Glu746\*) | g.32536181C>A | [18] | DMD | 3 |
| 18 | c.2257G>T | p.(Glu753\*) | g.32536160C>A | [23] | DMD | 2 |
| 18 | c.2270C>G | p.(Ser757\*) | g.32536147G>C | [13, 48, 50] | DMD | 4 |
| 18 | c.2276T>A | p.(Leu759\*) | g.32536141A>T | [16] | DMD | 1 |
| 18 | c.2276T>G | p.(Leu759\*) | g.32536141A>C | [10] | DMD | 1 |
| 18 | c.2281G>T | p.(Glu761\*) | g.32536136C>A | [10] | DMD | 1 |
| 19 | c.2299G>T | p.(Glu767\*) | g.32519953C>A | [30] | DMD | 2 |
| 19 | c.2302C>T | p.(Arg768\*) | g.32519950G>A | [3] | DMD | 33 |
| 19 | c.2308A>T | p.(Lys770\*) | g.32519944T>A | [11, 28] | DMD | 4 |
| 19 | c.2314G>T | p.[Glu772\*, Ala765Argfs\*15] | g.32519938C>A | [1] | DMD | 2 |
| 19 | c.2317A>T | p.(Lys773\*) | g.32519935T>A | [19] | DMD | 1 |
| 19 | c.2332C>T | p.(Gln778\*) | g.32519920G>A | [37] | DMD | 2 |
| 19 | c.2353C>T | p.(Gln785\*) | g.32519899G>A | [37] | DMD | 4 |
| 19 | c.2365G>T | p.(Glu789\*) | g.32519887C>A | [18] | DMD | 3 |
| 19 | c.2368C>T | p.(Gln790\*) | g.32519884G>A | [34] | DMD | 3 |
| 19 | c.2380G>T | p.(Glu794\*) | g.32519872C>A | [52] | DMD | 1 |
| 20 | c.2392G>T | p.(Glu798\*) | g.32509624C>A | [16] | ND | 1 |
| 20 | c.2404A>T | p.(Lys802\*) | g.32509612T>A | [19] | DMD | 1 |
| 20 | c.2407C>T | p.(Gln803\*) | g.32509609G>A | [50] | DMD | 3 |
| 20 | c.2414C>G | p.(Ser805\*) | g.32509602G>C | [52] | ND | 1 |
| 20 | c.2416G>T | p.(Glu806\*) | g.32509600C>A | [19] | DMD | 1 |
| 20 | c.2419C>T | p.(Gln807\*) | g.32509597G>A | [18] | DMD/BMD | 5 |
| 20 | c.2435G>A | p.(Trp812\*) | g.32509581C>T | [13, 19] | DMD | 4 |
| 20 | c.2436G>A | p.(Trp812\*) | g.32509580C>T | [1] | DMD/BMD | 2 |
| 20 | c.2440G>T | p.(Glu814\*) | g.32509576C>A | [19] | OTHER | 2 |
| 20 | c.2448C>A | p.(Cys816\*) | g.32509568G>T | [13] | DMD | 2 |
| 20 | c.2449C>T | p.(Gln817\*) | g.32509567G>A | [18] | DMD | 2 |
| 20 | c.2474G>A | p.(Trp825\*) | g.32509542C>T | [54] | DMD | 1 |
| 20 | c.2475G>A | p.(Trp825\*) | g.32509541C>T | [55] | DMD | 1 |
| 20 | c.2479G>T | p.(Glu827\*) | g.32509537C>A | [19] | DMD | 3 |
| 20 | c.2484T>A | p.(Tyr828\*) | g.32509532A>T | [23] | DMD | 1 |
| 20 | c.2484T>G | p.(Tyr828\*) | g.32509532A>C | [1] | ND | 1 |
| 20 | c.2485C>T | p.(Gln829\*) | g.32509531G>A | [19] | DMD | 1 |
| 20 | c.2512C>T | p.(Gln838\*) | g.32509504G>A | [55] | DMD | 1 |
| 20 | c.2518C>T | p.(Gln840\*) | g.32509498G>A | [4] | DMD | 2 |
| 20 | c.2521C>T | p.(Gln841\*) | g.32509495G>A | [25] | DMD | 4 |
| 20 | c.2525T>A | p.(Leu842\*) | g.32509491A>T | [1] | DMD | 1 |
| 20 | c.2527G>T | p.(Glu843\*) | g.32509489C>A | [40] | DMD/BMD | 4 |
| 20 | c.2530C>T | p.(Gln844\*) | g.32509486G>A | [56] | DMD | 2 |
| 20 | c.2548G>T | p.(Glu850\*) | g.32509468C>A | [33] | OTHER | 1 |
| 20 | c.2555G>A | p.(Trp852\*) | g.32509461C>T | [16] | ND | 1 |
| 20 | c.2558T>A | p.(Leu853\*) | g.32509458A>T | [39] | DMD | 1 |
| 20 | c.2560A>T | p.(Lys854\*) | g.32509456T>A | [1] | DMD | 1 |
| 20 | c.2566C>T | p.(Gln856\*) | g.32509450G>A | [1] | DMD | 2 |
| 20 | c.2582C>G | p.(Ser861\*) | g.32509434G>C | [19] | DMD | 1 |
| 20 | c.2584G>T | p.(Glu862\*) | g.32509432C>A | [13] | DMD | 1 |
| 20 | c.2599A>T | p.(Lys867\*) | g.32509417T>A | [1] | DMD | 1 |
| 20 | c.2605C>T | p.(Gln869\*) | g.32509411G>A | [15] | DMD | 5 |
| 20 | c.2609T>G | p.(Leu870\*) | g.32509407A>C | [20] | DMD/BMD | 1 |
| 20 | c.2611A>T | p.(Lys871\*) | g.32509405T>A | [19] | DMD | 2 |
| 21 | c.2626G>T | p.(Glu876\*) | g.32503213C>A | [1] | ND | 1 |
| 21 | c.2642C>G | p.(Ser881\*) | g.32503197G>C | [1] | ND | 1 |
| 21 | c.2650C>T | p.(Gln884\*) | g.32503189G>A | [25] | DMD | 3 |
| 21 | c.2656C>T | p.(Gln886\*) | g.32503183G>A | [33] | OTHER | 1 |
| 21 | c.2665C>T | p.(Arg889\*) | g.32503174G>A | [28, 37, 53] | DMD | 5 |
| 21 | c.2669T>G | p.(Leu890\*) | g.32503170A>C | [15] | DMD | 2 |
| 21 | c.2669T>A | p.(Leu890\*) | g.32503170A>T | [16] | DMD/BMD | 1 |
| 21 | c.2671A>T | p.(Lys891\*) | g.32503168T>A | [12] | DMD | 1 |
| 21 | c.2677C>T | p.(Gln893\*) | g.32503162G>A | [18] | DMD | 2 |
| 21 | c.2701G>T | p.(Gly901\*) | g.32503138C>A | [25] | DMD | 1 |
| 21 | c.2704C>T | p.(Gln902\*) | g.32503135G>A | [13] | BMD | 2 |
| 21 | c.2707G>T | p.(Gly903\*) | g.32503132C>A | [28] | ND | 1 |
| 21 | c.2755A>T | p.(Lys919\*) | g.32503084T>A | [57] | DMD | 2 |
| 21 | c.2758C>T | p.(Gln920\*) | g.32503081G>A | [19] | DMD | 7 |
| 21 | c.2776C>T | p.(Gln926\*) | g.32503063G>A | [45] | DMD | 3 |
| 21 | c.2788A>T | p.(Lys930\*) | g.32503051T>A | [34] | DMD | 3 |
| 21 | c.2791G>T | p.(Glu931\*) | g.32503048C>A | [58] | DMD | 2 |
| 21 | c.2797C>T | p.(Gln933\*) | g.32503042G>A | [10] | DMD | 6 |
| 22 | c.2816T>A | p.(Leu939\*) | g.32490414A>T | [53] | DMD | 3 |
| 22 | c.2832T>A | p.(Tyr944\*) | g.32490398A>T | [3] | DMD | 1 |
| 22 | c.2832T>G | p.(Tyr944\*) | g.32490398A>C | [59] | ND | 1 |
| 22 | c.2833C>T | p.(Gln945\*) | g.32490397G>A | [14] | DMD | 4 |
| 22 | c.2836G>T | p.(Glu946\*) | g.32490394C>A | [23] | DMD | 2 |
| 22 | c.2861G>A | p.(Trp954\*) | g.32490369C>T | [16] | DMD | 1 |
| 22 | c.2862G>A | p.(Trp954\*) | g.32490368C>T | [19] | DMD | 1 |
| 22 | c.2866C>T | p.(Gln956\*) | g.32490364G>A | [49] | DMD/BMD | 5 |
| 22 | c.2869C>T | p.(Gln957\*) | g.32490361G>A | [3, 28] | DMD | 5 |
| 22 | c.2873C>G | p.(Ser958\*) | g.32490357G>C | [28] | ND | 1 |
| 22 | c.2916T>A | p.(Tyr972\*) | g.32490314A>T | [1] | DMD | 1 |
| 22 | c.2926G>T | p.(Glu976\*) | g.32490304C>A | [60] | ND | 1 |
| 22 | c.2929C>T | p.(Gln977\*) | g.32490301G>A | [47] | DMD | 3 |
| 22 | c.2941G>T | p.(Glu981\*) | g.32490289C>A | [1] | DMD | 2 |
| 22 | c.2947C>T | p.(Gln983\*) | g.32490283G>A | [54] | DMD | 1 |
| 23 | c.2954T>A | p.(Leu985\*) | g.32486823A>T | [34] | DMD | 2 |
| 23 | c.2956C>T | p.(Gln986\*) | g.32486821G>A | [54] | DMD | 3 |
| 23 | c.2968C>T | p.(Gln990\*) | g.32486809G>A | [30] | DMD | 5 |
| 23 | c.2971G>T | p.(Glu991\*) | g.32486806C>A | [21, 42] | OTHER | 3 |
| 23 | c.2974C>T | p.(Gln992\*) | g.32486803G>A | [28] | DMD | 3 |
| 23 | c.2977C>T | p.(Gln993\*) | g.32486800G>A | [1] | DMD/BMD | 1 |
| 23 | c.2991C>G | p.(Tyr997\*) | g.32486786G>C | [19] | DMD | 7 |
| 23 | c.2994T>A | p.(Tyr998\*) | g.32486783A>T | [1] | DMD/BMD | 1 |
| 23 | c.3013G>T | p.(Glu1005\*) | g.32486764C>A | [13] | DMD | 1 |
| 23 | c.3022A>T | p.(Lys1008\*) | g.32486755T>A | [1] | DMD/BMD | 1 |
| 23 | c.3037G>T | p.(Glu1013\*) | g.32486740C>A | [61] | DMD | 1 |
| 23 | c.3049A>T | p.(Lys1017\*) | g.32486728T>A | [1] | DMD | 1 |
| 23 | c.3054T>G | p.(Tyr1018\*) | g.32486723A>C | [19] | DMD/BMD | 1 |
| 23 | c.3055C>T | p.(Gln1019\*) | g.32486722G>A | [62] | DMD | 2 |
| 23 | c.3059C>G | p.(Ser1020\*) | g.32486718G>C | [1] | DMD/BMD | 2 |
| 23 | c.3061G>T | p.(Glu1021\*) | g.32486716C>A | [39] | DMD | 1 |
| 23 | c.3067G>T | p.(Glu1023\*) | g.32486710C>A | [1] | DMD | 2 |
| 23 | c.3076G>T | p.(Glu1026\*) | g.32486701C>A | [63] | DMD/BMD | 3 |
| 23 | c.3079G>T | p.(Gly1027\*) | g.32486698C>A | [13] | DMD | 1 |
| 23 | c.3086G>A | p.(Trp1029\*) | g.32486691C>T | [28] | DMD | 2 |
| 23 | c.3087G>A | p.(Trp1029\*) | g.32486690C>T | [3, 19] | DMD | 5 |
| 23 | c.3103C>T | p.(Gln1035\*) | g.32486674G>A | [44] | DMD | 2 |
| 23 | c.3121C>T | p.(Gln1041\*) | g.32486656G>A | [11, 19] | DMD | 7 |
| 23 | c.3124A>T | p.(Lys1042\*) | g.32486653T>A | [1] | DMD/BMD | 1 |
| 23 | c.3136C>T | p.(Gln1046\*) | g.32486641G>A | [19] | DMD | 4 |
| 23 | c.3151C>T | p.(Arg1051\*) | g.32486626G>A | [3, 47] | DMD | 23 |
| 23 | c.3154A>T | p.(Lys1052\*) | g.32486623T>A | [61] | DMD | 1 |
| 24 | c.3172C>T | p.(Gln1058\*) | g.32482807G>A | [44] | DMD/BMD | 3 |
| 24 | c.3184A>T | p.(Lys1062\*) | g.32482795T>A | [1] | DMD | 1 |
| 24 | c.3188G>A | p.(Trp1063\*) | g.32482791C>T | [11] | DMD | 3 |
| 24 | c.3196G>T | p.(Glu1066\*) | g.32482783C>A | [12] | DMD | 1 |
| 24 | c.3217G>T | p.(Glu1073\*) | g.32482762C>A | [19] | DMD | 2 |
| 24 | c.3220G>T | p.(Glu1074\*) | g.32482759C>A | [30] | DMD | 3 |
| 24 | c.3224G>A | p.(Trp1075\*) | g.32482755C>T | [19] | DMD | 1 |
| 24 | c.3242C>A | p.(Ser1081\*) | g.32482737G>T | [33] | ND | 2 |
| 24 | c.3244G>T | p.(Glu1082\*) | g.32482735C>A | [45] | DMD | 3 |
| 24 | c.3253A>T | p.(Lys1085\*) | g.32482726T>A | [23] | DMD | 1 |
| 24 | c.3256A>T | p.(Lys1086\*) | g.32482723T>A | [64] | DMD | 3 |
| 24 | c.3259C>T | p.(Gln1087\*) | g.32482720G>A | [17] | DMD | 8 |
| 24 | c.3268C>T | p.(Gln1090\*) | g.32482711G>A | [28] | DMD | 2 |
| 24 | c.3274A>T | p.(Arg1092\*) | g.32482705T>A | [12] | DMD | 1 |
| 25 | c.3281T>A | p.[Leu1094\*; Leu1093\_Gln1144del] | g.32481707A>T | [19] | DMD | 1 |
| 25 | c.3281T>G | p.(Leu1094\*) | g.32481707A>C | [19] | DMD | 1 |
| 25 | c.3295C>T | p.[Gln1099\*,Leu1093\_Gln1144del] | g.32481693G>A | [38] | DMD/BMD | 7 |
| 25 | c.3304C>T | p.[Gln1102\*,Leu1093\_Gln1144del] | g.32481684G>A | [13, 38] | DMD/BMD | 6 |
| 25 | c.3328G>T | p.[Glu1110\*, Leu1093\_Gln1144del] | g.32481660C>A | [10] | BMD | 1 |
| 25 | c.3337C>T | p.[Gln1113\*,Leu1093\_Gln1144del] | g.32481651G>A | [13, 38] | DMD/BMD | 8 |
| 25 | c.3340A>T | p.(Lys1114\*) | g.32481648T>A | [19] | BMD | 3 |
| 25 | c.3346A>T | p.(Lys1116\*) | g.32481642T>A | [7] | ND | 2 |
| 25 | c.3352G>T | p.(Glu1118\*) | g.32481636C>A | [27] | BMD | 2 |
| 25 | c.3358G>T | p.(Glu1120\*) | g.32481630C>A | [56] | BMD | 1 |
| 25 | c.3376A>T | p.(Arg1126\*) | g.32481612T>A | [65] | DMD | 1 |
| 25 | c.3397G>T | p.(Glu1133\*) | g.32481591C>A | [66] | DMD | 4 |
| 25 | c.3409C>T | p.(Gln1137\*) | g.32481579G>A | [22] | BMD | 3 |
| 25 | c.3413G>A | p.(Trp1138\*) | g.32481575C>T | [34] | BMD | 4 |
| 25 | c.3414G>A | p.(Trp1138\*) | g.32481574C>T | [23] | DMD | 3 |
| 25 | c.3426C>A | p.(Cys1142\*) | g.32481562G>T | [3] | DMD | 2 |
| 25 | c.3427C>T | p.(Gln1143\*) | g.32481561G>A | [39] | DMD | 3 |
| 25 | c.3430C>T | p.[Gln1144\*,Leu1093\_Gln1144del] | g.32481558G>A | [13, 38] | DMD/BMD | 6 |
| 26 | c.3445A>T | p.(Lys1149\*) | g.32472937T>A | [20] | ND | 2 |
| 26 | c.3448G>T | p.(Glu1150\*) | g.32472934C>A | [13] | DMD | 1 |
| 26 | c.3460G>T | p.(Gly1154\*) | g.32472922C>A | [29] | DMD | 1 |
| 26 | c.3469G>T | p.(Glu1157\*) | g.32472913C>A | [1] | DMD | 1 |
| 26 | c.3472A>T | p.(Lys1158\*) | g.32472910T>A | [18] | DMD | 1 |
| 26 | c.3487C>T | p.(Gln1163\*) | g.32472895G>A | [43] | DMD | 3 |
| 26 | c.3500C>G | p.(Ser1167\*) | g.32472882G>C | [19] | DMD | 5 |
| 26 | c.3502G>T | p.(Glu1168\*) | g.32472880C>A | [1] | ND | 1 |
| 26 | c.3511G>T | p.(Glu1171\*) | g.32472871C>A | [23] | DMD | 3 |
| 26 | c.3515G>A | p.[Trp1172\*, Val1145\_Lys1201del] | g.32472867C>T | [15] | BMD | 1 |
| 26 | c.3516G>A | p.(Trp1172\*) | g.32472866C>T | [47] | OTHER | 1 |
| 26 | c.3523C>T | p.(Gln1175\*) | g.32472859G>A | [34] | DMD | 1 |
| 26 | c.3532G>T | p.(Glu1178\*) | g.32472850C>A | [16] | DMD/BMD | 1 |
| 26 | c.3535G>T | p.(Glu1179\*) | g.32472847C>A | [1] | ND | 3 |
| 26 | c.3544G>T | p.(Glu1182\*) | g.32472838C>A | [19] | DMD | 1 |
| 26 | c.3556G>T | p.(Glu1186\*) | g.32472826C>A | [27] | ND | 1 |
| 26 | c.3562A>T | p.(Lys1188\*) | g.32472820T>A | [67] | DMD | 2 |
| 26 | c.3578T>A | p.(Leu1193\*) | g.32472804A>T | [4] | DMD | 2 |
| 26 | c.3578T>G | p.(Leu1193\*) | g.32472804A>C | [23] | DMD | 1 |
| 26 | c.3580C>T | p.(Gln1194\*) | g.32472802G>A | [18] | DMD | 15 |
| 26 | c.3595G>T | p.(Glu1199\*) | g.32472787C>A | [1] | ND | 1 |
| 27 | c.3622C>T | p.(Gln1208\*) | g.32466737G>A | [3, 19] | DMD | 4 |
| 27 | c.3625C>T | p.(Gln1209\*) | g.32466734G>A | [52] | DMD | 2 |
| 27 | c.3631G>T | p.[Glu1211\*; Arg1202\_1262del; Arg1202\_1357del] | g.32466728C>A | [18] | BMD | 1 |
| 27 | c.3679C>T | p.(Gln1227\*) | g.32466680G>A | [30] | DMD | 1 |
| 27 | c.3700G>T | p.(Glu1234\*) | g.32466659C>A | [19] | BMD | 1 |
| 27 | c.3709A>T | p.(Lys1237\*) | g.32466650T>A | [68] | DMD | 2 |
| 27 | c.3715G>T | p.(Glu1239\*) | g.32466644C>A | [23] | DMD | 1 |
| 27 | c.3742C>T | p.(Gln1248\*) | g.32466617G>A | [19] | OTHER | 2 |
| 27 | c.3746G>A | p.(Trp1249\*) | g.32466613C>T | [1] | DMD | 1 |
| 27 | c.3747G>A | p.(Trp1249\*) | g.32466612C>T | [69] | DMD | 1 |
| 27 | c.3769A>T | p.(Lys1257\*) | g.32466590T>A | [3] | DMD | 1 |
| 27 | c.3774C>A | p.(Cys1258\*) | g.32466585G>T | [7] | ND | 1 |
| 27 | c.3784G>T | p.(Glu1262\*) | g.32466575C>A | [69] | DMD | 1 |
| 28 | c.3795G>A | p.(Trp1265\*) | g.32459423C>T | [18] | DMD/BMD | 2 |
| 28 | c.3801T>A | p.(Cys1267\*) | g.32459417A>T | [70] | OTHER | 1 |
| 28 | c.3804G>A | p.(Trp1268\*) | g.32459414C>T | [19] | DMD | 1 |
| 28 | c.3818C>A | p.(Ser1273\*) | g.32459400G>T | [20] | DMD | 1 |
| 28 | c.3822C>G | p.[Tyr1274\*, Glu1263\_Asp1307del, Glu1263\_Glu1357del] | g.32459396G>C | [21, 33] | DMD | 5 |
| 28 | c.3822C>A | p.(Tyr1274\*) | g.32459396G>T | [19] | DMD/BMD | 5 |
| 28 | c.3838A>T | p.(Lys1280\*) | g.32459380T>A | [16] | DMD/BMD | 1 |
| 28 | c.3843G>A | p.(Trp1281\*) | g.32459375C>T | [69] | BMD | 2 |
| 28 | c.3850G>T | p.[Glu1284\*, Glu1263\_Asp1307del, Glu1263\_Glu1357del] | g.32459368C>A | [4] | BMD | 2 |
| 28 | c.3862A>T | p.(Lys1288\*) | g.32459356T>A | [4] | DMD | 2 |
| 28 | c.3868A>T | p.(Lys1290\*) | g.32459350T>A | [19] | DMD | 1 |
| 28 | c.3892G>T | p.(Gly1298\*) | g.32459326C>A | [1] | OTHER | 1 |
| 29 | c.3923C>G | p.(Ser1308\*) | g.32456506G>C | [1] | ND | 1 |
| 29 | c.3935T>A | p.[Leu1312\*, Glu1263\_Glu1357del, Ser1308\_Glu1357del] | g.32456494A>T | [1] | BMD | 1 |
| 29 | c.3940C>T | p.[Arg1314\*, Ser1308\_Glu1357del, Glu1263\_Glu1357del] | g.32456489G>A | [23] | BMD | 31 |
| 29 | c.3947C>A | p.(Ser1316\*) | g.32456482G>T | [1] | DMD | 1 |
| 29 | c.3964C>T | p.(Gln1322\*) | g.32456465G>A | [40] | ND | 1 |
| 29 | c.3982C>T | p.[Gln1328\*, Glu1263\_Asp1307del,Glu1263\_Glu1357del] | g.32456447G>A | [19, 23] | DMD/BMD | 10 |
| 29 | c.4000G>T | p.[Gly1334\*, Glu1263\_Glu1357del, Ser1308\_Glu1357del] | g.32456429C>A | [1] | BMD | 1 |
| 29 | c.4012G>T | p.(Glu1338\*) | g.32456417C>A | [19] | BMD | 1 |
| 29 | c.4027G>T | p.(Glu1343\*) | g.32456402C>A | [1] | ND | 3 |
| 29 | c.4033G>T | p.(Glu1345\*) | g.32456396C>A | [1] | DMD | 2 |
| 29 | c.4057G>T | p.(Glu1353\*) | g.32456372C>A | [48] | DMD | 3 |
| 29 | c.4066G>T | p.(Glu1356\*) | g.32456363C>A | [13] | DMD | 2 |
| 30 | c.4084C>T | p.(Gln1362\*) | g.32430018G>A | [8] | DMD | 2 |
| 30 | c.4087A>T | p.(Lys1363\*) | g.32430015T>A | [12] | DMD | 1 |
| 30 | c.4099C>T | p.(Gln1367\*) | g.32430003G>A | [10] | DMD | 3 |
| 30 | c.4108C>T | p.(Gln1370\*) | g.32429994G>A | [25] | DMD | 7 |
| 30 | c.4117C>T | p.(Gln1373\*) | g.32429985G>A | [3, 21, 22, 71] | DMD | 7 |
| 30 | c.4120G>T | p.(Glu1374\*) | g.32429982C>A | [72] | DMD | 2 |
| 30 | c.4126G>T | p.(Glu1376\*) | g.32429976C>A | [13] | DMD | 2 |
| 30 | c.4142T>G | p.(Leu1381\*) | g.32429960A>C | [3] | DMD | 1 |
| 30 | c.4147C>T | p.(Gln1383\*) | g.32429955G>A | [73] | DMD | 2 |
| 30 | c.4150G>T | p.(Glu1384\*) | g.32429952C>A | [19, 56] | DMD | 2 |
| 30 | c.4174C>T | p.(Gln1392\*) | g.32429928G>A | [21, 23, 53] | DMD | 5 |
| 30 | c.4213C>T | p.(Gln1405\*) | g.32429889G>A | [11] | DMD/BMD | 2 |
| 30 | c.4222C>T | p.(Gln1408\*) | g.32429880G>A | [16] | ND | 1 |
| 30 | c.4231C>T | p.(Gln1411\*) | g.32429871G>A | [53, 65] | DMD | 2 |
| 31 | c.4240C>T | p.(Gln1414\*) | g.32408292G>A | [19] | BMD | 1 |
| 31 | c.4250T>A | p.[Leu1417\*; Lys1412\_Gln1448del] | g.32408282A>T | [74] | BMD | 4 |
| 31 | c.4285A>T | p.(Lys1429\*) | g.32408247T>A | [19] | BMD | 1 |
| 31 | c.4294C>T | p.[Gln1432\*,Ile1413\_Lys1449del] | g.32408238G>A | [38] | BMD | 3 |
| 31 | c.4303G>T | p.[Glu1435\*, Lys1412\_Gln1448del] | g.32408229C>A | [18] | DMD/BMD | 1 |
| 31 | c.4312C>T | p.(Gln1438\*) | g.32408220G>A | [16] | ND | 1 |
| 31 | c.4315A>T | p.(Arg1439\*) | g.32408217T>A | [16] | DMD/BMD | 1 |
| 32 | c.4352T>A | p.(Leu1451\*) | g.32407784A>T | [19] | DMD | 1 |
| 32 | c.4354C>T | p.(Gln1452\*) | g.32407782G>A | [1] | DMD | 1 |
| 32 | c.4375C>T | p.(Arg1459\*) | g.32407761G>A | [3, 13, 21] | DMD | 14 |
| 32 | c.4384C>T | p.(Gln1462\*) | g.32407752G>A | [10] | DMD | 3 |
| 32 | c.4405C>T | p.(Gln1469\*) | g.32407731G>A | [30] | DMD | 7 |
| 32 | c.4414C>T | p.(Gln1472\*) | g.32407722G>A | [11] | DMD | 7 |
| 32 | c.4480G>T | p.(Glu1494\*) | g.32407656C>A | [11] | DMD | 1 |
| 32 | c.4483C>T | p.(Gln1495\*) | g.32407653G>A | [3, 19, 37] | DMD | 5 |
| 32 | c.4486G>T | p.(Glu1496\*) | g.32407650C>A | [26, 75] | DMD | 2 |
| 32 | c.4495C>T | p.(Gln1499\*) | g.32407641G>A | [13] | DMD | 3 |
| 32 | c.4499C>A | p.(Ser1500\*) | g.32407637G>T | [21] | DMD | 1 |
| 32 | c.4501C>T | p.(Gln1501\*) | g.32407635G>A | [13] | DMD | 2 |
| 33 | c.4527T>A | p.(Tyr1509\*) | g.32404574A>T | [1] | DMD | 1 |
| 33 | c.4527T>G | p.(Tyr1509\*) | g.32404574A>C | [18] | DMD | 4 |
| 33 | c.4540G>T | p.(Glu1514\*) | g.32404561C>A | [13] | DMD | 1 |
| 33 | c.4558G>T | p.(Glu1520\*) | g.32404543C>A | [1] | DMD | 1 |
| 33 | c.4559G>T | p.(Glu1520\*) | g.32404542C>A | [76] | DMD | 1 |
| 33 | c.4570A>T | p.(Lys1524\*) | g.32404531T>A | [27] | DMD/BMD | 1 |
| 33 | c.4576G>T | p.(Gly1526\*) | g.32404525C>A | [1] | BMD | 1 |
| 33 | c.4582C>T | p.(Gln1528\*) | g.32404519G>A | [3, 23] | DMD | 2 |
| 33 | c.4591C>T | p.(Gln1531\*) | g.32404510G>A | [15] | OTHER | 3 |
| 33 | c.4600C>T | p.(Gln1534\*) | g.32404501G>A | [54] | DMD | 7 |
| 33 | c.4606G>T | p.(Glu1536\*) | g.32404495C>A | [37] | DMD | 2 |
| 33 | c.4618G>T | p.(Glu1540\*) | g.32404483C>A | [19] | DMD | 4 |
| 33 | c.4660G>T | p.(Glu1554\*) | g.32404441C>A | [3] | DMD | 1 |
| 33 | c.4666G>T | p.(Gly1556\*) | g.32404435C>A | [18] | DMD | 3 |
| 34 | c.4687A>T | p.(Lys1563\*) | g.32398785T>A | [18] | DMD | 1 |
| 34 | c.4690C>T | p.(Gln1564\*) | g.32398782G>A | [52] | DMD | 1 |
| 34 | c.4693C>T | p.(Gln1565\*) | g.32398779G>A | [30] | DMD | 4 |
| 34 | c.4697T>A | p.(Leu1566\*) | g.32398775A>T | [19] | DMD | 1 |
| 34 | c.4702A>T | p.(Lys1568\*) | g.32398770T>A | [1] | DMD | 1 |
| 34 | c.4711A>T | p.(Lys1571\*) | g.32398761T>A | [25] | DMD | 4 |
| 34 | c.4729C>T | p.(Arg1577\*) | g.32398743G>A | [10] | DMD | 31 |
| 34 | c.4732A>T | p.(Lys1578\*) | g.32398740T>A | [23] | DMD | 2 |
| 34 | c.4735G>T | p.(Glu1579\*) | g.32398737C>A | [1] | DMD/BMD | 1 |
| 34 | c.4757G>A | p.(Trp1586\*) | g.32398715C>T | [18] | DMD | 1 |
| 34 | c.4758G>A | p.(Trp1586\*) | g.32398714C>T | [54] | DMD | 1 |
| 34 | c.4777G>T | p.(Glu1593\*) | g.32398695C>A | [39] | DMD | 1 |
| 34 | c.4781T>A | p.(Leu1574\*) | g.32398691A>T | [29] | DMD | 1 |
| 34 | c.4793C>A | p.(Ser1598\*) | g.32398679G>T | [19] | DMD | 1 |
| 34 | c.4838G>A | p.(Trp1613\*) | g.32398634C>T | [4, 56] | DMD | 3 |
| 34 | c.4843A>T | p.(Lys1615\*) | g.32398629T>A | [1] | DMD/BMD | 1 |
| 35 | c.4852C>T | p.(Gln1618\*) | g.32383310G>A | [1] | DMD | 3 |
| 35 | c.4858G>T | p.(Glu1620\*) | g.32383304C>A | [7] | DMD/BMD | 1 |
| 35 | c.4864G>T | p.(Glu1622\*) | g.32383298C>A | [19] | DMD | 1 |
| 35 | c.4870C>T | p.(Gln1624\*) | g.32383292G>A | [30] | DMD | 8 |
| 35 | c.4915A>T | p.(Lys1639\*) | g.32383247T>A | [69] | DMD | 2 |
| 35 | c.4933A>T | p.(Lys1645\*) | g.32383229T>A | [55] | DMD | 1 |
| 35 | c.4948G>T | p.(Glu1650\*) | g.32383214C>A | [1] | ND | 1 |
| 35 | c.4954A>T | p.(Lys1652\*) | g.32383208T>A | [63] | DMD | 2 |
| 35 | c.4979G>A | p.(Trp1660\*) | g.32383183C>T | [52] | BMD | 1 |
| 35 | c.4980G>A | p.(Trp1660\*) | g.32383182C>T | [1] | BMD | 1 |
| 35 | c.4996C>T | p.(Arg1666\*) | g.32383166G>A | [30] | DMD | 30 |
| 35 | c.5002G>T | p.(Glu1668\*) | g.32383160C>A | [21] | BMD | 1 |
| 35 | c.5009G>A | p.(Trp1670\*) | g.32383153C>T | [34] | DMD | 1 |
| 35 | c.5021T>A | p.(Leu1674\*) | g.32383141A>T | [1] | DMD/BMD | 1 |
| 35 | c.5024T>A | p.(Leu1675\*) | g.32383138A>T | [16] | ND | 1 |
| 36 | c.5031C>A | p.(Tyr1677\*) | g.32382822G>T | [10] | DMD | 1 |
| 36 | c.5032C>T | p.(Gln1678\*) | g.32382821G>A | [30] | DMD | 1 |
| 36 | c.5044G>T | p.(Glu1682\*) | g.32382809C>A | [13, 18, 67] | DMD | 3 |
| 36 | c.5082G>A | p.(Trp1694\*) | g.32382771C>T | [13] | DMD | 2 |
| 36 | c.5089C>T | p.(Gln1697\*) | g.32382764G>A | [1] | DMD | 2 |
| 36 | c.5110G>T | p.(Glu1704\*) | g.32382743C>A | [16] | ND | 1 |
| 36 | c.5119A>T | p.(Lys1707\*) | g.32382734T>A | [3] | DMD | 1 |
| 36 | c.5131C>T | p.(Gln1711\*) | g.32382722G>A | [37] | DMD | 5 |
| 36 | c.5134C>T | p.(Gln1712\*) | g.32382719G>A | [19] | DMD | 4 |
| 36 | c.5140G>T | p.(Glu1714\*) | g.32382713C>A | [19] | DMD | 3 |
| 37 | c.5159T>A | p.(Leu1720\*) | g.32381071A>T | [66] | DMD | 1 |
| 37 | c.5167G>T | p.(Glu1723\*) | g.32381063C>A | [21, 69] | DMD | 3 |
| 37 | c.5188A>T | p.(Lys1730\*) | g.32381042T>A | [1] | DMD | 1 |
| 37 | c.5209C>T | p.(Gln1737\*) | g.32381021G>A | [22, 71] | DMD | 4 |
| 37 | c.5255T>G | p.(Leu1752\*) | g.32380975A>C | [19] | DMD | 2 |
| 37 | c.5260G>T | p.(Glu1754\*) | g.32380970C>A | [77] | BMD | 1 |
| 37 | c.5266C>T | p.(Gln1756\*) | g.32380964G>A | [34] | DMD | 3 |
| 37 | c.5287C>T | p.[Arg1763\*, Arg1719\_Lys1775del] | g.32380943G>A | [25] | BMD | 22 |
| 37 | c.5308A>T | p.(Arg1770\*) | g.32380922T>A | [1] | DMD | 1 |
| 37 | c.5314A>T | p.(Lys1772\*) | g.32380916T>A | [39] | DMD | 1 |
| 38 | c.5341A>T | p.(Lys1781\*) | g.32366630T>A | [66] | DMD/BMD | 3 |
| 38 | c.5344G>T | p.(Glu1782\*) | g.32366627C>A | [19] | DMD | 2 |
| 38 | c.5350G>T | p.(Glu1784\*) | g.32366621C>A | [18] | DMD | 2 |
| 38 | c.5353C>T | p.(Gln1785\*) | g.32366618G>A | [23] | DMD/BMD | 2 |
| 38 | c.5363C>G | p.(Ser1788\*) | g.32366608G>C | [16] | DMD/BMD | 1 |
| 38 | c.5371C>T | p.[Gln1791\*,Ala1776\_Met1816del] | g.32366600G>A | [13, 38] | DMD/BMD | 11 |
| 38 | c.5398G>T | p.(Glu1800\*) | g.32366573C>A | [19] | BMD | 3 |
| 38 | c.5404C>T | p.(Gln1802\*) | g.32366567G>A | [78] | BMD | 5 |
| 38 | c.5407C>T | p.[Gln1803\*, Ala1776\_Met1816del] | g.32366564G>A | [13, 38] | BMD | 5 |
| 39 | c.5452G>T | p.(Glu1818\*) | g.32364194C>A | [1] | BMD | 1 |
| 39 | c.5461G>T | p.(Glu1821\*) | g.32364185C>A | [12] | DMD | 1 |
| 39 | c.5476G>T | p.(Glu1826\*) | g.32364170C>A | [22, 54] | DMD | 3 |
| 39 | c.5480T>A | p.[Leu1827\*, Ala1776\_Lys1862del, Asn1817\_Lys1862del] | g.32364166A>T | [1] | BMD | 2 |
| 39 | c.5485C>T | p.(Gln1829\*) | g.32364161G>A | [49] | DMD | 1 |
| 39 | c.5488A>T | p.(Arg1830\*) | g.32364158T>A | [9] | DMD/BMD | 5 |
| 39 | c.5503C>T | p.(Gln1835\*) | g.32364143G>A | [13] | DMD | 1 |
| 39 | c.5506C>T | p.(Gln1836\*) | g.32364140G>A | [16] | ND | 1 |
| 39 | c.5521G>T | p.(Glu1841\*) | g.32364125C>A | [37] | ND | 1 |
| 39 | c.5530C>T | p.(Arg1844\*) | g.32364116G>A | [3, 13, 19, 30] | DMD | 28 |
| 39 | c.5533G>T | p.(Glu1845\*) | g.32364113C>A | [79] | OTHER | 1 |
| 39 | c.5542A>T | p.(Lys1848\*) | g.32364104T>A | [30] | DMD | 1 |
| 39 | c.5551C>T | p.[Gln1851\*; Asn1817\_Lys1862del] | g.32364095G>A | [58] | DMD | 7 |
| 39 | c.5554C>T | p.(Gln1852\*) | g.32364092G>A | [15, 69] | OTHER | 4 |
| 39 | c.5561T>G | p.(Leu1854\*) | g.32364085A>C | [1] | ND | 1 |
| 39 | c.5563C>T | p.(Gln1855\*) | g.32364083G>A | [54] | DMD | 7 |
| 40 | c.5599C>T | p.(Gln1867\*) | g.32361391G>A | [13] | DMD | 1 |
| 40 | c.5602A>T | p.(Arg1868\*) | g.32361388T>A | [13] | DMD | 1 |
| 40 | c.5608A>T | p.(Lys1870\*) | g.32361382T>A | [19] | DMD | 3 |
| 40 | c.5611A>T | p.(Lys1871\*) | g.32361379T>A | [4, 20] | DMD | 4 |
| 40 | c.5632C>T | p.(Gln1878\*) | g.32361358G>A | [21, 53] | OTHER | 4 |
| 40 | c.5637G>A | p.(Trp1879\*) | g.32361353C>T | [25] | DMD | 3 |
| 40 | c.5640T>A | p.(Tyr1880\*) | g.32361350A>T | [1] | DMD/BMD | 2 |
| 40 | c.5641C>T | p.(Gln1881\*) | g.32361349G>A | [30] | DMD | 2 |
| 40 | c.5646C>A | p.(Tyr1882\*) | g.32361344G>T | [19] | DMD | 3 |
| 40 | c.5646C>G | p.(Tyr1882\*) | g.32361344G>C | [56] | DMD | 1 |
| 40 | c.5647A>T | p.(Lys1883\*) | g.32361343T>A | [16] | ND | 1 |
| 40 | c.5653C>T | p.(Gln1885\*) | g.32361337G>A | [10] | DMD | 3 |
| 40 | c.5671A>T | p.(Lys1891\*) | g.32361319T>A | [1] | DMD/BMD | 2 |
| 40 | c.5699T>G | p.(Leu1900\*) | g.32361291A>C | [1] | DMD/BMD | 2 |
| 40 | c.5713G>T | p.(Glu1905\*) | g.32361277C>A | [16] | ND | 1 |
| 40 | c.5725G>T | p.(Glu1909\*) | g.32361265C>A | [19] | BMD | 1 |
| 41 | c.5740G>T | p.(Glu1914\*) | g.32360399C>A | [1] | ND | 1 |
| 41 | c.5752G>T | p.(Glu1918\*) | g.32360387C>A | [19] | DMD/BMD | 3 |
| 41 | c.5758C>T | p.(Gln1920\*) | g.32360381G>A | [19] | DMD | 5 |
| 41 | c.5761A>T | p.(Lys1921\*) | g.32360378T>A | [69] | DMD | 1 |
| 41 | c.5764A>T | p.(Lys1922\*) | g.32360375T>A | [7] | DMD/BMD | 1 |
| 41 | c.5770G>T | p.(Glu1924\*) | g.32360369C>A | [13] | DMD | 2 |
| 41 | c.5773G>T | p.(Glu1925\*) | g.32360366C>A | [54] | DMD/BMD | 6 |
| 41 | c.5800G>T | p.(Glu1934\*) | g.32360339C>A | [30] | DMD | 2 |
| 41 | c.5807T>A | p.(Leu1936\*) | g.32360332A>T | [1] | DMD/BMD | 1 |
| 41 | c.5835G>T | p.(Glu1946\*) | g.32360304C>A | [1] | BMD | 1 |
| 41 | c.5845C>T | p.(Gln1949\*) | g.32360294G>A | [1] | ND | 1 |
| 41 | c.5851C>T | p.(Gln1951\*) | g.32360288G>A | [3, 20] | DMD | 6 |
| 41 | c.5867G>A | p.(Trp1956\*) | g.32360272C>T | [19] | DMD | 1 |
| 41 | c.5868G>A | p.(Trp1956\*) | g.32360271C>T | [21] | ND | 1 |
| 41 | c.5872G>T | p.(Glu1958\*) | g.32360267C>A | [16] | ND | 1 |
| 41 | c.5878G>T | p.(Glu1960\*) | g.32360261C>A | [37] | DMD | 3 |
| 41 | c.5893C>T | p.(Gln1965\*) | g.32360246G>A | [79] | OTHER | 2 |
| 41 | c.5899C>T | p.[Arg1967\*,Glu1914\_Ile1974del] | g.32360240G>A | [38] | DMD | 35 |
| 41 | c.5902A>T | p.(Arg1968\*) | g.32360237T>A | [19] | DMD | 1 |
| 41 | c.5917C>T | p.(Gln1973\*) | g.32360222G>A | [19, 46] | DMD | 2 |
| 42 | c.5938G>T | p.(Glu1980\*) | g.32328378C>A | [37] | DMD/BMD | 2 |
| 42 | c.5985T>G | p.[Tyr1995\*, His1975\_Tyr1995del] | g.32328331A>C | [13, 18] | DMD | 3 |
| 42 | c.6000T>A | p.(Tyr2000\*) | g.32328316A>T | [16] | DMD/BMD | 1 |
| 42 | c.6007G>T | p.(Glu2003\*) | g.32328309C>A | [21] | ND | 1 |
| 42 | c.6023C>G | p.(Ser2008\*) | g.32328293G>C | [23] | DMD | 1 |
| 42 | c.6023C>A | p.(Ser2008\*) | g.32328293G>T | [52] | DMD | 1 |
| 42 | c.6035T>G | p.(Leu2012\*) | g.32328281A>C | [7] | DMD/BMD | 1 |
| 42 | c.6037G>T | p.(Glu2013\*) | g.32328279C>A | [19] | DMD | 1 |
| 42 | c.6072T>A | p.(Cys2024\*) | g.32328244A>T | [54] | DMD | 1 |
| 42 | c.6085G>T | p.(Glu2029\*) | g.32328231C>A | [69] | DMD | 1 |
| 42 | c.6103G>T | p.(Glu2035\*) | g.32328213C>A | [19] | DMD | 3 |
| 42 | c.6106G>T | p.(Glu2036\*) | g.32328210C>A | [23] | DMD | 1 |
| 43 | c.6136C>T | p.(Gln2046\*) | g.32305800G>A | [13] | DMD | 1 |
| 43 | c.6139C>T | p.(Gln2047\*) | g.32305797G>A | [1] | ND | 1 |
| 43 | c.6208G>T | p.(Glu2070\*) | g.32305728C>A | [15] | DMD | 2 |
| 43 | c.6223C>T | p.(Gln2075\*) | g.32305713G>A | [28] | DMD | 2 |
| 43 | c.6226G>T | p.(Glu2076\*) | g.32305710C>A | [1] | DMD/BMD | 1 |
| 43 | c.6238C>T | p.(Gln2080\*) | g.32305698G>A | [25] | DMD | 2 |
| 43 | c.6250C>T | p.(Gln2084\*) | g.32305686G>A | [37] | ND | 1 |
| 43 | c.6255G>A | p.(Trp2085\*) | g.32305681C>T | [19] | DMD | 5 |
| 43 | c.6256G>T | p.(Glu2086\*) | g.32305680C>A | [3] | DMD | 1 |
| 43 | c.6259A>T | p.(Lys2087\*) | g.32305677T>A | [80] | DMD | 1 |
| 43 | c.6276C>G | p.(Tyr2092\*) | g.32305660G>C | [39] | DMD | 1 |
| 43 | c.6277A>T | p.(Lys2093\*) | g.32305659T>A | [71] | ND | 2 |
| 43 | c.6283C>T | p.(Arg2095\*) | g.32305653G>A | [10] | DMD | 27 |
| 43 | c.6286C>T | p.(Gln2096\*) | g.32305650G>A | [4] | DMD | 1 |
| 44 | c.6292C>T | p.(Arg2098\*) | g.32235179G>A | [10] | DMD | 27 |
| 44 | c.6310G>T | p.(Glu2104\*) | g.32235161C>A | [3] | DMD | 2 |
| 44 | c.6317G>A | p.(Trp2106\*) | g.32235154C>T | [16] | ND | 1 |
| 44 | c.6318G>A | p.(Trp2106\*) | g.32235153C>T | [9, 19] | DMD/BMD | 3 |
| 44 | c.6340A>T | p.(Lys2114\*) | g.32235131T>A | [1] | DMD/BMD | 1 |
| 44 | c.6352C>T | p.(Gln2118\*) | g.32235119G>A | [19] | DMD | 4 |
| 44 | c.6356G>A | p.(Trp2119\*) | g.32235115C>T | [13] | DMD | 1 |
| 44 | c.6364G>T | p.(Glu2122\*) | g.32235107C>A | [25] | DMD | 1 |
| 44 | c.6370G>T | p.(Glu2124\*) | g.32235101C>A | [3] | DMD | 1 |
| 44 | c.6373C>T | p.(Gln2125\*) | g.32235098G>A | [3] | DMD | 4 |
| 44 | c.6382A>T | p.(Arg2128\*) | g.32235089T>A | [1] | DMD | 1 |
| 44 | c.6391C>T | p.(Gln2131\*) | g.32235080G>A | [19] | DMD | 2 |
| 44 | c.6407G>A | p.(Trp2136\*) | g.32235064C>T | [1] | ND | 1 |
| 44 | c.6408G>A | p.(Trp2136\*) | g.32235063C>T | [7] | DMD/BMD | 1 |
| 44 | c.6423C>A | p.(Tyr2141\*) | g.32235048G>T | [27] | DMD | 5 |
| 44 | c.6423C>G | p.(Tyr2141\*) | g.32235048G>C | [23] | DMD | 2 |
| 44 | c.6424A>T | p.(Lys2142\*) | g.32235047T>A | [16] | DMD | 1 |
| 44 | c.6428G>A | p.(Trp2143\*) | g.32235043C>T | [1] | ND | 1 |
| 44 | c.6429G>A | p.(Trp2143\*) | g.32235042C>T | [28] | DMD | 1 |
| 44 | c.6432T>A | p.(Tyr2144\*) | g.32235039A>T | [18] | DMD | 2 |
| 44 | c.6436A>T | p.(Lys2146\*) | g.32235035T>A | [12] | DMD | 3 |
| 44 | c.6439G>T | p.(Gln2147\*) | g.31986631C>A | [81] | ND | 1 |
| 45 | c.6445C>T | p.(Gln2149\*) | g.31986625G>A | [19] | DMD | 1 |
| 45 | c.6460C>T | p.(Gln2154\*) | g.31986610G>A | [12] | DMD | 4 |
| 45 | c.6499G>T | p.(Glu2167\*) | g.31986571C>A | [1] | DMD | 1 |
| 45 | c.6502G>T | p.(Glu2168\*) | g.31986568C>A | [1] | ND | 1 |
| 45 | c.6544C>T | p.(Gln2182\*) | g.31986526G>A | [19, 55] | DMD | 6 |
| 45 | c.6550A>T | p.(Lys2184\*) | g.31986520T>A | [44, 59] | ND | 1 |
| 45 | c.6576G>A | p.(Trp2192\*) | g.31986494C>T | [54] | DMD | 1 |
| 45 | c.6577C>T | p.(Gln2193\*) | g.31986493G>A | [1] | DMD | 1 |
| 45 | c.6592C>T | p.(Gln2198\*) | g.31986478G>A | [19] | DMD/BMD | 2 |
| 45 | c.6607A>T | p.(Lys2203\*) | g.31986463T>A | [23] | DMD | 1 |
| 46 | c.6641C>A | p.(Ser2214\*) | g.31950318G>T | [19] | DMD | 1 |
| 46 | c.6649C>T | p.(Gln2217\*) | g.31950310G>A | [19] | DMD | 2 |
| 46 | c.6659T>G | p.(Leu2220\*) | g.31950300A>C | [13] | DMD | 1 |
| 46 | c.6674T>A | p.(Leu2225\*) | g.31950285A>T | [54] | DMD | 1 |
| 46 | c.6674T>G | p.(Leu2225\*) | g.31950285A>C | [28] | DMD | 1 |
| 46 | c.6677G>A | p.(Trp2226\*) | g.31950282C>T | [1] | ND | 2 |
| 46 | c.6678G>A | p.(Trp2226\*) | g.31950281C>T | [52] | DMD | 3 |
| 46 | c.6721G>T | p.(Gly2241\*) | g.31950238C>A | [19] | DMD/BMD | 1 |
| 46 | c.6730C>T | p.(Gln2244\*) | g.31950229G>A | [15] | DMD | 1 |
| 46 | c.6733C>T | p.(Gln2245\*) | g.31950226G>A | [1] | DMD | 1 |
| 46 | c.6742G>T | p.(Glu2248\*) | g.31950217C>A | [25] | DMD | 1 |
| 46 | c.6754C>T | p.(Gln2252\*) | g.31950205G>A | [23] | DMD | 1 |
| 47 | c.6790C>T | p.(Gln2264\*) | g.31947835G>A | [11] | DMD | 2 |
| 47 | c.6805C>T | p.(Gln2269\*) | g.31947820G>A | [18] | DMD | 2 |
| 47 | c.6805C>A | p.(Gln2269\*) | g.31947820G>T | [18] | DMD | 1 |
| 47 | c.6809T>G | p.(Leu2270\*) | g.31947816A>C | [82] | DMD | 1 |
| 47 | c.6856G>T | p.(Glu2286\*) | g.31947769C>A | [19] | DMD | 2 |
| 47 | c.6862C>T | p.(Gln2288\*) | g.31947763G>A | [18] | DMD | 1 |
| 47 | c.6868A>T | p.(Lys2290\*) | g.31947757T>A | [19] | DMD | 1 |
| 47 | c.6880A>T | p.(Lys2294\*) | g.31947745T>A | [39] | DMD | 1 |
| 47 | c.6889C>T | p.(Gln2297\*) | g.31947736G>A | [1] | DMD | 1 |
| 47 | c.6905G>A | p.(Trp2302\*) | g.31947720C>T | [10] | DMD | 6 |
| 47 | c.6906G>A | p.(Trp2302\*) | g.31947719C>T | [19] | DMD/BMD | 2 |
| 48 | c.6926T>A | p.(Leu2309\*) | g.31893477A>T | [1] | DMD | 1 |
| 48 | c.6943G>T | p.(Glu2315\*) | g.31893460C>A | [71] | DMD/BMD | 7 |
| 48 | c.6949G>T | p.(Glu2317\*) | g.31893454C>A | [13] | DMD | 1 |
| 48 | c.6955C>T | p.(Gln2319\*) | g.31893448G>A | [83] | DMD | 1 |
| 48 | c.6973C>T | p.(Gln2325\*) | g.31893430G>A | [13, 50] | DMD | 3 |
| 48 | c.6975C>T | p.(Gln2325\*) | g.31893428G>A | [13] | DMD | 1 |
| 48 | c.6979G>T | p.(Glu2327\*) | g.31893424C>A | [1] | ND | 1 |
| 48 | c.6982A>T | p.(Lys2328\*) | g.31893421T>A | [16] | DMD/BMD | 1 |
| 48 | c.7006C>T | p.(Gln2336\*) | g.31893397G>A | [33] | DMD/BMD | 3 |
| 48 | c.7010T>G | p.(Leu2337\*) | g.31893393A>C | [1] | DMD | 1 |
| 48 | c.7029G>A | p.(Trp2343\*) | g.31893374C>T | [44] | DMD | 2 |
| 48 | c.7031T>A | p.(Leu2344\*) | g.31893372A>T | [36] | ND | 1 |
| 48 | c.7054G>T | p.(Glu2352\*) | g.31893349C>A | [12] | DMD | 1 |
| 48 | c.7075C>T | p.(Gln2359\*) | g.31893328G>A | [19] | DMD | 1 |
| 49 | c.7105G>T | p.[Glu2369\*, Glu2367\_Lys2400del] | g.31854930C>A | [25] | BMD | 5 |
| 49 | c.7126C>T | p.(Gln2376\*) | g.31854909G>A | [13] | DMD | 1 |
| 49 | c.7159C>T | p.[Gln2387\*, Glu2367\_Lys2400del] | g.31854876G>A | [84] | ND | 2 |
| 49 | c.7170C>G | p.(Tyr2390\*) | g.31854865G>C | [16] | ND | 1 |
| 49 | c.7189C>T | p.(Gln2397\*) | g.31854846G>A | [16] | DMD/BMD | 1 |
| 50 | c.7204A>T | p.(Lys2402\*) | g.31838197T>A | [1] | ND | 1 |
| 50 | c.7229G>A | p.(Trp2410\*) | g.31838172C>T | [1] | DMD/BMD | 1 |
| 50 | c.7255G>T | p.(Glu2419\*) | g.31838146C>A | [18] | DMD | 2 |
| 50 | c.7288G>T | p.(Gly2430\*) | g.31838113C>A | [3] | DMD | 1 |
| 51 | c.7318C>T | p.(Gln2440\*) | g.31792301G>A | [85] | OTHER | 1 |
| 51 | c.7339C>T | p.(Gln2447\*) | g.31792280G>A | [12] | DMD | 1 |
| 51 | c.7402G>T | p.(Glu2468\*) | g.31792217C>A | [42] | DMD | 1 |
| 51 | c.7436G>A | p.(Trp2479\*) | g.31792183C>T | [1] | DMD | 1 |
| 51 | c.7471C>T | p.(Glu2491\*) | g.31792148G>A | [1] | DMD | 1 |
| 51 | c.7510G>T | p.(Glu2504\*) | g.31792109C>A | [49] | DMD | 1 |
| 51 | c.7537C>T | p.(Gln2513\*) | g.31792082G>A | [86] | ND | 1 |
| 51 | c.7561G>T | p.(Glu2521\*) | g.31747847C>A | [37] | ND | 1 |
| 52 | c.7564C>T | p.(Gln2522\*) | g.31747844G>A | [87] | DMD | 2 |
| 52 | c.7576C>T | p.(Gln2526\*) | g.31747832G>A | [19] | DMD | 2 |
| 52 | c.7582G>T | p.(Glu2528\*) | g.31747826C>A | [28] | DMD | 1 |
| 52 | c.7585G>T | p.(Glu2529\*) | g.31747823C>A | [20] | DMD | 2 |
| 52 | c.7657C>T | p.(Arg2553\*) | g.31747751G>A | [10] | DMD | 33 |
| 53 | c.7672C>T | p.(Gln2558\*) | g.31697692G>A | [30] | DMD | 2 |
| 53 | c.7682G>A | p.(Trp2561\*) | g.31697682C>T | [19] | DMD | 1 |
| 53 | c.7683G>A | p.(Trp2561\*) | g.31697681C>T | [1] | DMD/BMD | 1 |
| 53 | c.7693C>T | p.(Gln2565\*) | g.31697671G>A | [19] | DMD | 2 |
| 53 | c.7696G>T | p.(Glu2566\*) | g.31697668C>A | [1] | DMD | 1 |
| 53 | c.7717C>T | p.(Gln2573\*) | g.31697647G>A | [7] | DMD/BMD | 1 |
| 53 | c.7720C>T | p.(Gln2574\*) | g.31697644G>A | [19] | DMD | 1 |
| 53 | c.7736T>A | p.(Leu2579\*) | g.31697628A>T | [1] | DMD | 4 |
| 53 | c.7736T>G | p.(Leu2579\*) | g.31697628A>C | [1] | DMD | 1 |
| 53 | c.7750C>T | p.(Gln2584\*) | g.31697614G>A | [13] | DMD | 1 |
| 53 | c.7755G>A | p.(Trp2585\*) | g.31697609C>T | [27] | DMD | 1 |
| 53 | c.7771G>T | p.(Glu2591\*) | g.31697593C>A | [1] | DMD/BMD | 1 |
| 53 | c.7792C>T | p.(Gln2598\*) | g.31697572G>A | [21] | ND | 2 |
| 53 | c.7798A>T | p.(Arg2600\*) | g.31697566T>A | [1] | DMD/BMD | 1 |
| 53 | c.7814C>G | p.(Ser2605\*) | g.31697550G>C | [19] | DMD | 2 |
| 53 | c.7817G>A | p.(Trp2606\*) | g.31697547C>T | [18] | DMD | 1 |
| 53 | c.7818G>A | p.(Trp2606\*) | g.31697546C>T | [1] | DMD/BMD | 1 |
| 53 | c.7822G>T | p.(Glu2608\*) | g.31697542C>A | [19] | DMD | 2 |
| 53 | c.7855A>T | p.(Lys2619\*) | g.31697509T>A | [18] | DMD | 1 |
| 53 | c.7864G>T | p.(Glu2622\*) | g.31697500C>A | [7] | DMD/BMD | 1 |
| 54 | c.7873C>T | p.(Gln2625\*) | g.31676261G>A | [13] | DMD | 1 |
| 54 | c.7894C>T | p.(Gln2632\*) | g.31676240G>A | [13] | DMD | 3 |
| 54 | c.7898G>A | p.(Trp2633\*) | g.31676236C>T | [80] | DMD | 1 |
| 54 | c.7899G>A | p.(Trp2633\*) | g.31676235C>T | [43] | ND | 1 |
| 54 | c.7969A>T | p.(Arg2657\*) | g.31676165T>A | [23] | DMD | 1 |
| 54 | c.8009G>A | p.(Trp2670\*) | g.31676125C>T | [63] | DMD | 4 |
| 54 | c.8010G>A | p.(Trp2670\*) | g.31676124C>T | [13] | DMD | 3 |
| 55 | c.8038C>T | p.(Arg2680\*) | g.31645969G>A | [3, 53, 71] | DMD | 18 |
| 55 | c.8053G>T | p.(Glu2685\*) | g.31645954C>A | [13] | DMD | 2 |
| 55 | c.8069T>G | p.(Leu2690\*) | g.31645938A>C | [69] | DMD | 5 |
| 55 | c.8074C>T | p.(Gln2692\*) | g.31645933G>A | [28] | DMD | 2 |
| 55 | c.8077C>T | p.(Gln2693\*) | g.31645930G>A | [1] | DMD | 1 |
| 55 | c.8098A>T | p.(Lys2700\*) | g.31645909T>A | [45] | DMD | 1 |
| 55 | c.8146C>T | p.(Gln2716\*) | g.31645861G>A | [19] | DMD | 1 |
| 55 | c.8161A>T | p.(Lys2721\*) | g.31645846T>A | [19] | DMD | 2 |
| 55 | c.8176G>T | p.(Glu2726\*) | g.31645831C>A | [39] | DMD | 1 |
| 55 | c.8194A>T | p.(Lys2732\*) | g.31645813T>A | [3] | DMD | 2 |
| 55 | c.8197G>T | p.(Gln2733\*) | g.31645810C>A | [44] | ND | 1 |
| 55 | c.8209C>T | p.(Gln2737\*) | g.31645798G>A | [40] | ND | 1 |
| 55 | c.8213G>A | p.(Trp2738\*) | g.31645794C>T | [37] | DMD | 1 |
| 55 | c.8214G>A | p.(Trp2738\*) | g.31645793C>T | [4] | DMD | 5 |
| 55 | c.8215C>T | p.(Gln2739\*) | g.31645792G>A | [3] | DMD/BMD | 2 |
| 56 | c.8269G>T | p.(Glu2757\*) | g.31525519C>A | [55] | DMD | 1 |
| 56 | c.8299G>T | p.(Glu2767\*) | g.31525489C>A | [13] | DMD | 2 |
| 56 | c.8326C>T | p.(Gln2776\*) | g.31525462G>A | [1] | DMD | 1 |
| 56 | c.8353A>T | p.(Lys2785\*) | g.31525435T>A | [19] | BMD | 1 |
| 56 | c.8357G>A | p.(Trp2786\*) | g.31525431C>T | [45] | DMD | 2 |
| 56 | c.8358G>A | p.(Trp2786\*) | g.31525430C>T | [1] | DMD/BMD | 1 |
| 56 | c.8371A>T | p.(Lys2791\*) | g.31525417T>A | [34] | DMD | 2 |
| 56 | c.8374A>T | p.(Lys2792\*) | g.31525414T>A | [10] | DMD | 1 |
| 57 | c.8416C>T | p.(Gln2806\*) | g.31515036G>A | [13, 28] | DMD | 4 |
| 57 | c.8420G>A | p.(Trp2807\*) | g.31515032C>T | [25] | DMD | 4 |
| 57 | c.8422A>T | p.(Lys2808\*) | g.31515030T>A | [52] | DMD | 1 |
| 57 | c.8443C>T | p.(Gln2815\*) | g.31515009G>A | [3, 55] | DMD | 5 |
| 57 | c.8459G>A | p.(Trp2820\*) | g.31514993C>T | [13] | DMD | 1 |
| 57 | c.8460G>A | p.(Trp2820\*) | g.31514992C>T | [19] | DMD | 3 |
| 57 | c.8464C>T | p.(Gln2822\*) | g.31514988G>A | [37] | DMD | 3 |
| 57 | c.8479G>T | p.(Glu2827\*) | g.31514973C>A | [13] | DMD | 1 |
| 57 | c.8483T>G | p.(Leu2828\*) | g.31514969A>C | [37] | ND | 1 |
| 57 | c.8491C>T | p.(Gln2831\*) | g.31514961G>A | [19] | DMD | 1 |
| 57 | c.8527A>T | p.(Lys2843\*) | g.31514925T>A | [19] | DMD | 1 |
| 58 | c.8560G>T | p.(Glu2854\*) | g.31497208C>A | [13] | DMD | 1 |
| 58 | c.8575G>T | p.(Glu2859\*) | g.31497193C>A | [21] | DMD | 2 |
| 58 | c.8608C>T | p.(Arg2870\*) | g.31497160G>A | [10] | DMD | 45 |
| 58 | c.8626C>T | p.(Gln2876\*) | g.31497142G>A | [13] | DMD | 1 |
| 58 | c.8647A>T | p.(Lys2883\*) | g.31497121T>A | [16] | ND | 1 |
| 58 | c.8655C>A | p.(Tyr2885\*) | g.31497113G>T | [1] | OTHER | 1 |
| 58 | c.8656C>T | p.(Gln2886\*) | g.31497112G>A | [88] | DMD | 1 |
| 59 | c.8680G>T | p.(Glu2894\*) | g.31496480C>A | [19] | DMD/BMD | 1 |
| 59 | c.8686A>T | p.(Arg2896\*) | g.31496474T>A | [12] | DMD/BMD | 1 |
| 59 | c.8692C>T | p.(Gln2898\*) | g.31496468G>A | [13, 61] | DMD | 3 |
| 59 | c.8713C>T | p.(Arg2905\*) | g.31496447G>A | [19, 55] | DMD | 34 |
| 59 | c.8728G>T | p.(Glu2910\*) | g.31496432C>A | [25] | DMD | 1 |
| 59 | c.8745G>A | p.(Trp2915\*) | g.31496415C>T | [27] | OTHER | 2 |
| 59 | c.8746G>T | p.(Glu2916\*) | g.31496414C>A | [18] | DMD | 1 |
| 59 | c.8774G>A | p.(Trp2925\*) | g.31496386C>T | [28] | ND | 2 |
| 59 | c.8775G>A | p.(Trp2925\*) | g.31496385C>T | [19] | DMD | 1 |
| 59 | c.8776C>T | p.(Gln2926\*) | g.31496384G>A | [89] | DMD | 1 |
| 59 | c.8791G>T | p.(Glu2931\*) | g.31496369C>A | [1] | DMD | 1 |
| 59 | c.8800G>T | p.(Glu2934\*) | g.31496360C>A | [90] | ND | 1 |
| 59 | c.8812G>T | p.(Glu2938\*) | g.31496348C>A | [3] | DMD | 2 |
| 59 | c.8854C>T | p.(Gln2952\*) | g.31496306G>A | [10] | DMD | 1 |
| 59 | c.8872G>T | p.(Gly2958\*) | g.31496288C>A | [19] | DMD | 1 |
| 59 | c.8879G>A | p.(Trp2960\*) | g.31496281C>T | [1] | DMD | 2 |
| 59 | c.8880G>A | p.(Trp2960\*) | g.31496280C>T | [52] | ND | 1 |
| 59 | c.8914C>T | p.[Gln2972\*, Val2955\_Lys2979del] | g.31496246G>A | [28] | DMD | 4 |
| 59 | c.8929A>T | p.(Lys2977\*) | g.31496231T>A | [1] | OTHER | 1 |
| 60 | c.8944C>T | p.(Arg2982\*) | g.31462738G>A | [30] | DMD | 25 |
| 60 | c.8947G>T | p.(Gly2983\*) | g.31462735C>A | [10] | DMD | 1 |
| 60 | c.9001C>T | p.(Gln3001\*) | g.31462681G>A | [46] | DMD | 3 |
| 60 | c.9029C>G | p.(Ser3010\*) | g.31462653G>C | [3] | DMD | 2 |
| 60 | c.9036T>A | p.(Tyr3012\*) | g.31462646A>T | [13] | DMD | 1 |
| 60 | c.9071G>A | p.(Trp3024\*) | g.31462611C>T | [1] | DMD | 1 |
| 60 | c.9072G>A | p.(Trp3024\*) | g.31462610C>T | [21, 53, 55] | DMD | 3 |
| 60 | c.9082C>T | p.(Gln3028\*) | g.31462600G>A | [19] | DMD | 2 |
| 61 | c.9100C>T | p.(Arg3034\*) | g.31366736G>A | [19, 20, 37, 71] | DMD | 14 |
| 61 | c.9109C>T | p.(Gln3037\*) | g.31366727G>A | [28] | DMD | 1 |
| 61 | c.9148C>T | p.(Gln3050\*) | g.31366688G>A | [39] | DMD | 3 |
| 62 | c.9182G>A | p.(Trp3061\*) | g.31341757C>T | [10] | DMD | 3 |
| 62 | c.9183G>A | p.(Trp3061\*) | g.31341756C>T | [13] [54] | DMD | 3 |
| 62 | c.9197C>A | p.(Ser3066\*) | g.31341742G>T | [11] | DMD | 2 |
| 62 | c.9216C>A | p.(Tyr3072\*) | g.31341723G>T | [91] | DMD | 1 |
| 62 | c.9216C>G | p.(Tyr3072\*) | g.31341723G>C | [18] | DMD | 2 |
| 63 | c.9249G>A | p.(Trp3083\*) | g.31279109C>T | [20] | DMD | 2 |
| 63 | c.9276C>A | p.(Tyr3092\*) | g.31279082G>T | [37] | ND | 1 |
| 63 | c.9284T>G | p.(Leu3095\*) | g.31279074A>C | [21] | OTHER | 2 |
| 64 | c.9337C>T | p.(Arg3113\*) | g.31241188G>A | [19] | DMD | 27 |
| 64 | c.9346C>T | p.(Gln3116\*) | g.31241179G>A | [30] | DMD | 6 |
| 64 | c.9360C>A | p.(Cys3120\*) | g.31241165G>T | [27] | DMD | 1 |
| 65 | c.9380C>G | p.(Ser3127\*) | g.31227798G>C | [30] | DMD | 9 |
| 65 | c.9398T>A | p.(Leu3133\*) | g.31227780A>T | [1] | DMD/BMD | 1 |
| 65 | c.9403C>T | p.(Gln3135\*) | g.31227775G>A | [25] | DMD | 4 |
| 65 | c.9427C>T | p.(Gln3143\*) | g.31227751G>A | [30] | DMD | 4 |
| 65 | c.9445C>T | p.(Gln3149\*) | g.31227733G>A | [39] | DMD | 5 |
| 65 | c.9459T>A | p.(Cys3153\*) | g.31227719A>T | [1] | ND | 1 |
| 65 | c.9461T>A | p.(Leu3154\*) | g.31227717A>T | [28] | DMD | 2 |
| 65 | c.9474T>G | p.(Tyr3158\*) | g.31227704A>C | [33] | ND | 1 |
| 65 | c.9522C>A | p.(Cys3174\*) | g.31227656G>T | [21] | DMD | 4 |
| 65 | c.9542G>A | p.(Trp3181\*) | g.31227636C>T | [13] | DMD | 2 |
| 66 | c.9568C>T | p.[Arg3190\*, Gly3189Profs\*13] | g.31224780G>A | [49] | DMD | 47 |
| 66 | c.9621T>A | p.(Cys3207\*) | g.31224727A>T | [16] | ND | 1 |
| 66 | c.9640A>T | p.(Lys3214\*) | g.31224708T>A | [1] | DMD | 1 |
| 67 | c.9651C>A | p.(Tyr3217\*) | g.31222234G>T | [1] | OTHER | 1 |
| 67 | c.9661C>T | p.(Gln3231\*) | g.31222224G>A | [37] | ND | 1 |
| 67 | c.9691C>T | p.(Gln3231\*) | g.31222194G>A | [25, 91] | DMD | 2 |
| 67 | c.9739C>T | p.(Gln3247\*) | g.31222146G>A | [15] | DMD | 3 |
| 67 | c.9748G>T | p.(Glu3250\*) | g.31222137C>A | [12] | DMD | 1 |
| 68 | c.9829G>T | p.(Glu3277\*) | g.31201000C>A | [52] | ND | 2 |
| 68 | c.9851G>A | p.(Trp3284\*) | g.31200978C>T | [37] | DMD | 4 |
| 68 | c.9862G>T | p.(Glu3288\*) | g.31200967C>A | [28] | DMD/BMD | 1 |
| 68 | c.9882G>A | p.(Trp3294\*) | g.31200947C>T | [77] | DMD/BMD | 2 |
| 68 | c.9898A>T | p.(Arg3300\*) | g.31200931T>A | [20] | DMD | 1 |
| 68 | c.9913G>T | p.(Glu3305\*) | g.31200916C>A | [18] | DMD | 1 |
| 68 | c.9928C>T | p.(Gln3310\*) | g.31200901G>A | [13, 19, 46] | DMD | 5 |
| 68 | c.9948C>A | p.(Cys3316\*) | g.31200881G>T | [13] | DMD | 1 |
| 68 | c.9952G>T | p.(Glu3318\*) | g.31200877C>A | [19] | DMD | 1 |
| 69 | c.9978C>G | p.(Tyr3326\*) | g.31198595G>C | [19] | DMD | 1 |
| 69 | c.10002T>G | p.(Tyr3334\*) | g.31198571A>C | [19] | DMD/BMD | 1 |
| 69 | c.10012C>T | p.(Gln3338\*) | g.31198561G>A | [19] | DMD | 1 |
| 69 | c.10033C>T | p.(Arg3345\*) | g.31198540G>A | [13, 20, 38, 53] | DMD | 19 |
| 69 | c.10062T>A | p.(Tyr3354\*) | g.31198511A>T | [16] | ND | 1 |
| 69 | c.10072G>T | p.(Glu3358\*) | g.31198501C>A | [12] | DMD | 1 |
| 69 | c.10077T>A | p.(Tyr3359\*) | g.31198496A>T | [28] | DMD | 1 |
| 70 | c.10094C>G | p.(Ser3365\*) | g.31196915G>C | [1] | DMD/BMD | 1 |
| 70 | c.10096G>T | p.(Gly3366\*) | g.31196913C>A | [13] | DMD | 1 |
| 70 | c.10108C>T | p.(Arg3370\*) | g.31196901G>A | [10] | DMD | 44 |
| 70 | c.10135A>T | p.(Lys3379\*) | g.31196874T>A | [25, 91] | DMD | 2 |
| 70 | c.10141C>T | p.(Arg3381\*) | g.31196868G>A | [10] | DMD | 55 |
| 70 | c.10147A>T | p.(Lys3383\*) | g.31196862T>A | [1] | DMD | 1 |
| 70 | c.10171C>T | p.(Arg3391\*) | g.31196838G>A | [92] | DMD | 46 |
| 70 | c.10192C>T | p.(Gln3398\*) | g.31196817G>A | [1] | DMD/BMD | 1 |
| 70 | c.10202T>G | p.(Leu3401\*) | g.31196807A>C | [33] | DMD | 2 |
| 70 | c.10219G>T | p.(Glu3407\*) | g.31196790C>A | [71] | ND | 2 |
| 71 | c.10247G>A | p.(Trp3416\*) | g.31196064C>T | [93] | OTHER | 1 |
| 72 | c.10279C>T | p.[Gln3427\*, Pro3422\_Arg3443del] | g.31191705G>A | [25] | BMD | 7 |
| 72 | c.10320T>A | p.(Tyr3440\*) | g.31191664A>T | [13] | DMD/BMD | 3 |
| 73 | c.10362T>A | p.(Tyr3454\*) | g.31190497A>T | [1] | BMD | 1 |
| 73 | c.10387G>T | p.(Glu3463\*) | g.31190472C>A | [1] | ND | 1 |
| 74 | c.10412T>G | p.(Leu3471\*) | g.31187701A>C | [74] | DMD | 1 |
| 74 | c.10412T>A | p.(Leu3471\*) | g.31187701A>T | [74] | BMD | 2 |
| 74 | c.10429C>T | p.(Gln3477\*) | g.31187684G>A | [1] | BMD | 1 |
| 74 | c.10477C>T | p.(Gln3493\*) | g.31187636G>A | [84] | DMD/BMD | 5 |
| 74 | c.10493T>A | p.(Leu3498\*) | g.31187620A>T | [1] | DMD | 1 |
| 74 | c.10495G>T | p.(Glu3499\*) | g.31187618C>A | [13] | DMD | 2 |
| 74 | c.10504G>T | p.(Glu3502\*) | g.31187609C>A | [34] | DMD | 1 |
| 74 | c.10519G>T | p.(Glu3507\*) | g.31187594C>A | [16] | ND | 1 |
| 74 | c.10543G>T | p.[Glu3515\*, Ile3465\_Arg3518delinsMet] | g.31187570C>A | [15] | BMD | 1 |
| 74 | c.10546G>T | p.(Glu3516\*) | g.31187567C>A | [42, 53] | DMD/BMD | 3 |
| 75 | c.10572T>A | p.(Tyr3524\*) | g.31165617A>T | [1] | DMD | 1 |
| 75 | c.10588C>T | p.(Gln3530\*) | g.31165601G>A | [1] | DMD | 3 |
| 75 | c.10651C>T | p.(Gln3551\*) | g.31165538G>A | [13] | DMD | 3 |
| 75 | c.10759G>T | p.(Glu3587\*) | g.31165430C>A | [1] | BMD | 1 |
| 75 | c.10765C>T | p.(Gln3589\*) | g.31165424G>A | [1] | ND | 1 |
| 75 | c.10769T>A | p.(Leu3590\*) | g.31165420A>T | [1] | OTHER | 1 |
| 75 | c.10783C>T | p.(Gln3595\*) | g.31165406G>A | [71] | DMD/BMD | 3 |
| 75 | c.10792G>T | p.(Glu3598\*) | g.31165397C>A | [13] | BMD | 1 |
| 76 | c.10801C>T | p.(Gln3601\*) | g.31164528G>A | [19] | DMD | 3 |
| 76 | c.10855C>T | p.(Gln3619\*) | g.31164474G>A | [94] | DMD/BMD | 1 |
| 76 | c.10873C>T | p.(Gln3625\*) | g.31164456G>A | [18] | BMD | 1 |
| 76 | c.10888C>T | p.(Arg3630\*) | g.31164441G>A | [19] | BMD | 1 |
| 76 | c.10903C>T | p.(Gln3635\*) | g.31164426G>A | [30] | DMD | 3 |
| 76 | c.10910C>A | p.(Ser3637\*) | g.31164419G>T | [25, 91] | BMD | 4 |

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