|  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Supplementary Table S1. Nucleotide sequence variants in the *EYS* gene in the Japanese population. | | | | | | | | | | | |
| No. | ID | Reference SNP | Nucleotide change | Protein change | MAF | | MAF Ratio (RP+KRP)/CT | Genome Variation Databases | | | |
| RP+KRP | CT | 1000Genome | ExAC | HGVD | iJGVD |
| 1 | Pr-1 | rs9453373 | c.-1237G>T |  | 0.0926 | - | - | 0.1915 | - | - | 0.0868 |
| 2 | Pr-2 | rs147501078 | c.-849\_-848insA |  | 0.2686 | - | - | 0.1116 | - | - | - |
| 3 | Pr-3 | rs9294647 | c.-804C>A |  | 0.3600 | - | - | 0.3361 | - | - | 0.4111 |
| 4 | Pr-4 | rs12660682 | c.-648G>T |  | 0.0544 | - | - | 0.0272 | - | - |  |
| 5 | Ex1-1 | rs1490127 | c.-500A>G |  | 0.0884 | 0.0809 | 1.09 | 0.1915 | - | - | 0.0867 |
| 6 | In3-1 | rs13217899 | c.-198+83T>A |  | - | - | - | 0.095 | - | - | 0.0583 |
| 7 | Ex4-1 | rs112609906 | c.334G>C | p.(Val112Leu) | 0.0053 | 0.0000 | - | 0.0012 | 0.0003543 | 0.0025 | - |
| 8 | Ex4-2 | rs12193967 | c.359C>T | p.(Thr120Met) | 0.3233 | 0.3100 | 1.04 | 0.1703 | 0.2026 | 0.2612 | 0.252 |
| 9 | Ex4-3 | rs780433094 | c.525\_527delGGA | p.(Glu176del) | 0.0053 | 0.0000 | - | - | 0.000115 | 0.0008 | - |
| 10 | Ex4-4 | rs772707303 | c.632G>A | p.(Cys211Tyr) | 0.0032 | 0.0000 | - | - | T：0.0002 | - | - |
| 11 | In4-1 | - | c.749-136T>A |  | 0.0015 | - | - | - | - | - | - |
| 12 | In4-2 | rs551877761 | c.749-132C>T |  | 0.0152 | - | - | 0.0004 | - | - | - |
| 13 | In5-1 | rs4710522 | c.862+87T>C |  | 0.0650 | - | - | 0.097 | - | - | 0.058 |
| 14 | In5-2 | rs4710521 | c.862+391A>T |  | 0.0660 | - | - | 0.097 | - | - | - |
| 15 | In5-3 | - | c.863-94A>G |  | 0.0015 | 0.0000 | - | - | - | - | - |
| 16 | In5-4 | rs68032536 | c.863-31\_863-30insTT/TTT |  |  |  | - | - | - | - | - |
| 17 | Ex6-1 | - | c.942delT | p.(Ala315Leufs\*24) | 0.0015 | 0.0000 | - | - | - | - | - |
| 18 | Ex6-2 | rs80095433 | c.1005G>T | p.(Glu335Asp) | 0.0076 | 0.0153 | 0.49 | 0.0006 | 0.00003 | 0.0058 | 0.0147 |
| 19 | Ex7-1 | rs974110 | c.1146T>C | p.(Asn382=) | 0.4635 | - | - | T:0.4940 | T：0.4835 | 0.4396 | 0.4457 |
| 20 | In7-1 | - | c.1184+124T>G |  | 0.0015 | - | - | - | - | - | - |
| 21 | In7-2 | rs373800075 | c.1184+261T>C |  | 0.0030 | - | - | 0.0004 | - | - | - |
| 22 | Ex8-1 | rs764163418 | c.1211dupA | p.(Asn404Lysfs\*3) | 0.0032 | 0.0000 | - | - | 0.000008 | - | - |
| 23 | In8-1 | rs1155668 | c.1299+357T>C |  | 0.4807 | 0.4516 | 1.06 | 0.4006 | - | - | 0.4354 |
| 24 | In8-2 | rs1155669 | c.1299+457G>A |  | 0.4739 | 0.4516 | 1.05 | 0.4008 | - | - | 0.4348 |
| 25 | In8-3 | - | c.1300-108T>C |  | 0.0000 | 0.0051 | 0.00 | - | - | - | - |
| 26 | In8-4 | rs1936439 | c.1300-3C>T |  | 0.4438 | 0.4444 | 1.00 | C:0.3488 | C：0.3254 | 0.433 | - |
| 27 | Ex9-1 | rs76754818 | c.1382G>A | p.(Cys461Tyr) | 0.0299 | 0.0152 | 1.97 | 0.002 | 0.0005 | 0.0314 | 0.036 |
| 28 | In9-1 | rs9453265 | c.1459+103C>T |  | 0.2505 | 0.2806 | 0.89 | 0.2394 | - | - | 0.2517 |
| 29 | In9-2 | rs201155768 | c.1459+229\_1459+230delTA |  | 0.0743 | 0.0707 | 1.05 | 0.0246 | - | - | - |
| 30 | In9-3 | rs781415405 | c.1460-5dupT |  | 0.0015 | 0.0000 | - | - | 0.0009 | - | - |
| 31 | Ex10-1 | - | c.1485\_1493delinsCGAAAAG | p.(Val496Glufs\*13) | 0.0015 | 0.0000 | - | - | - | - | - |
| 32 | In10-1 | rs190462222 | c.1600-126T>C |  | 0.0058 | 0.0100 | 0.58 | 0.0004 | - | - | - |
| 33 | In10-2 | rs77096978 | c.1600-82A>T |  | 0.0458 | 0.0500 | 0.92 | 0.0076 | - | - | 0.0634 |
| 34 | Ex11-1 | rs61753610 | c.1712A>G | p.(Gln571Arg) | 0.1151 | 0.1300 | 0.89 | 0.1242 | 0.1569 | 0.1112 | 0.1259 |
| 35 | Ex11-2 | rs527236072 | c.1750G>T | p.(Glu584\*) | 0.0021 | 0.0000 | - | - | - | - | - |
| 36 | In11-1 | rs76148513 | c.1766+61A>G |  | 0.0043 | 0.0050 | 0.85 | 0.0449 | 0.0618 | - | - |
| 37 | In11-2 | rs990273081 | c.1766+95G>A |  | 0.0021 | 0.0000 | - | - | - | - | - |
| 38 | In11-3 | rs188568438 | c.1767-87T>C |  | 0.0122 | 0.0147 | 0.83 | 0.0016 | - | - | 0.0159 |
| 39 | In11-4 | - | c.1767-49C>A |  | 0.0000 | 0.0074 | 0.00 | - | - | - | - |
| 40 | Ex12-1 | rs9345601 | c.1809T>C | p.(Val603=) | 0.0760 | 0.0882 | 0.86 | 0.4052 | 0.4189 | 0.0994 | 0.1055 |
| 41 | Ex12-2 | rs9342464 | c.1891G>A | p.(Gly631Ser) | 0.0760 | 0.0882 | 0.86 | 0.405 | 0.4202 | 0.1001 | 0.1054 |
| 42 | Ex12-3 | rs17411795 | c.1922A>T | p.(Glu641Val) | 0.0745 | 0.0882 | 0.84 | 0.1042 | 0.1615 | 0.0947 | 0.0994 |
| 43 | Ex12-4 | - | c.1929T>C | p.(Asp643=) | 0.0030 | 0.0000 | - | - | - | 0.0008 | - |
| 44 | Ex12-5 | rs144430026 | c.1985G>T | p.(Arg662Met) | 0.0395 | 0.1176 | 0.34 | 0.0108 | 0.002 | 0.033 | 0.0444 |
| 45 | In12-1 | rs374955689 | c.2023+15delT |  | 0.0836 | 0.0882 | 0.95 | - | 0.1934 | 0.054 | - |
| 46 | In12-2 | rs372205279 | c.2024-14delCinsTT |  | 0.0246 | - | - | - | - | 0.0395 | - |
| 47 | In13-1 | rs185460544 | c.2137+40A>C |  | 0.0015 | - | - | 0.0002 | 0.0003 | - | - |
| 48 | In13-2 | rs10455568 | c.2137+114C>T |  | 0.0231 | - | - | 0.0661 | - | 0.045 | 0.029 |
| 49 | In13-3 | rs10455567 | c.2137+239A>G |  | 0.0247 | - | - | 0.0669 | - | - | 0.0299 |
| 50 | In13-4 | rs117654837 | c.2137+320A>G |  | 0.0046 | - | - | 0.0044 | - | - | - |
| 51 | In13-5 | rs10455185 | c.2137+374T>A |  | 0.0231 | - | - | 0.0611 | - | - | 0.0296 |
| 52 | In13-6 | rs10455566 | c.2137+405G>C |  | 0.0231 | - | - | 0.0613 | - | - | 0.0295 |
| 53 | In13-7 | rs9453167 | c.2137+412T>A |  | 0.0463 | - | - | 0.2995 | - | - | 0.0611 |
| 54 | Ex14-1 | rs781359405 | c.2259C>T | p.(Leu753=) | 0.0046 | - | - | - | - | 0.0033 | - |
| 55 | In14-1 | rs9453115 | c.2260-76G>A |  | 0.3429 | 0.2647 | 1.30 | 0.3948 | - | 0.314 | 0.2761 |
| 56 | In15-1 | rs12196494 | c.2381+417T>C |  | 0.2197 | 0.1912 | 1.15 | 0.1709 | - | - | - |
| 57 | In15-2 | rs9445437 | c.2382-26C>G |  | 0.4499 | 0.4000 | 1.12 | 0.3534 | 0.3622 | - | 0.4563 |
| 58 | Ex16-1 | rs150194717 | c.2490T>C | p.(Pro830=) | 0.0021 | 0.0000 | - | 0.0004 | - | 0.0033 | - |
| 59 | Ex16-2 | rs74419361 | c.2528G>A | p.(Gly843Glu) | 0.0693 | 0.0190 | 3.64 | 0.0014 | - | 0.022 | 0.017 |
| 60 | Ex16-3 | rs9294631 | c.2555T>C | p.(Leu852Pro) | 0.4488 | 0.4000 | 1.12 | 0.3536 | 0.3893 | 0.4417 | 0.4483 |
| 61 | In16-1 | rs117137733 | c.2641+35A>G |  | 0.0512 | 0.0762 | 0.67 | 0.0182 | 0.0031 | 0.066 | 0.0623 |
| 62 | In16-2 | rs10046141 | c.2641+198A>C |  | 0.4455 | 0.3952 | 1.13 | 0.3556 | - | - | 0.4522 |
| 63 | In16-3 | rs183476685 | c.2641+205G>A |  | 0.0032 | 0.0048 | 0.67 | 0.0002 | - | - | - |
| 64 | In16-4 | rs67221279 | c.2641+254delT |  | 0.4048 | 0.4663 | 0.87 | 0.3361 | - | - | - |
| 65 | In17-1 | - | c.2739-42T>C |  | 0.0015 | - | - | - | - | - | - |
| 66 | In18-1 | rs59518422 | c.2846+53\_2846+54insTAAT |  | 0.3558 | - | - | 0.2867 | - | - | - |
| 67 | In18-2 | rs57706332 | c.2846+192A>G |  | 0.1603 | - | - | 0.1066 | - | - | 0.0612 |
| 68 | In18-3 | rs59191409 | c.2847-221C>T |  | 0.4894 | - | - | 0.2218 | - | - | 0.4308 |
| 69 | In18-4 | rs58449017 | c.2847-157A>G |  | 0.4894 | - | - | 0.2218 | - | - | 0.4313 |
| 70 | In18-5 | rs7743515 | c.2847-24C>T |  | 0.1049 | - | - | 0.1564 | 0.1707 | 0.1479 | 0.1615 |
| 71 | Ex19-1 | rs749081001 | c.2980C>G | p.(Pro994Ala) | 0.0030 | - | - | - | 0.00005 | 0.0004 | - |
| 72 | In20-1 | - | c.3164+296G>A |  | 0.0048 | - | - | - | - | - |  |
| 73 | In20-2 | rs188297913 | c.3164+360G>A |  | 0.0096 | - | - | 0.0004 | - | - | 0.0124 |
| 74 | In22-1 | rs9345532 | c.3443+160T>G |  | 0.3405 | - | - | 0.4201 | - | - | - |
| 75 | In22-2 | rs56215318 | c.3443+253delT |  | 0.1405 | - | - | 0.1459 | - | - | - |
| 76 | In22-3 | rs7760032 | c.3444-283A>G |  | 0.3591 | 0.4559 | 0.79 | A:0.4123 | - | - | 0.4529 |
| 77 | In22-4 | rs558307468 | c.3444-274C>A |  | 0.0030 | 0.0000 | - | 0.0002 | - | - | - |
| 78 | In22-5 | rs915649401 | c.3444-149A>G |  | 0.0091 | 0.0000 | - | - | - | - | - |
| 79 | In22-6 | rs548545956 | c.3444-141C>T |  | 0.0015 | 0.0074 | 0.21 | 0.0002 | - | - | - |
| 80 | In22-7 | rs9445051 | c.3444-5C>T |  | 0.2909 | 0.3284 | 0.89 | 0.1955 | 0.2032 | 0.3067 | 0.2959 |
| 81 | Ex23-1 | rs371491059 | c.3454G>A | p.(Gly1152Arg) | 0.0015 | 0.0000 | - | - | - | - | - |
| 82 | Ex23-2 | rs150951106 | c.3489T>A | p.(Asn1163Lys) | 0.0030 | 0.0000 | - | 0.0018 | 0.0004 | 0.0054 | - |
| 83 | In23-1 | rs62415853 | c.3568+99C>A |  | 0.2553 | 0.1194 | 2.14 | 0.099 | - | - | 0.1161 |
| 84 | In24-1 | - | c.3684+93G>T |  | 0.0024 | - | - | - | - | - | - |
| 85 | In24-2 | rs1009346790 | c.3684+101T>C |  | 0.0024 | - | - | - | - | - | - |
| 86 | In24-3 | - | c.3685-94T>C |  | 0.0030 | 0.0000 | - | - | - | - | - |
| 87 | Ex25-1 | rs17404123 | c.3787A>G | p.(Ile1263Val) | 0.2682 | 0.1223 | 2.19 | 0.0938 | 0.1012 | 0.1279 | 0.1174 |
| 88 | Ex25-2 | rs368856942 | c.3809T>G | p.(Val1270Gly) | 0.0045 | 0.0000 | - | 0.0004 | 0.00005 | 0.0045 | - |
| 89 | In25-1 | rs139235690 | c.3877+18\_3877+22delAGATA |  | 0.2652 | 0.1211 | 2.19 | 0.0911 | 0.1013 | - | - |
| 90 | In25-2 | rs553365985 | c.3877+93delT |  | 0.2470 | 0.2611 | 0.95 | 0.223 | - | - | - |
| 91 | In25-3 | rs16895524 | c.3877+112T>C |  | 0.2652 | 0.1250 | 2.12 | 0.0938 | - | - | 0.1176 |
| 92 | In25-4 | rs75420781 | c.3877+133T>A |  | 0.2652 | 0.1250 | 2.12 | 0.0938 | - | - | 0.1172 |
| 93 | In25-5 | rs12661208 | c.3877+282G>C |  | 0.2652 | 0.1169 | 2.27 | 0.0938 | - | - | 0.1172 |
| 94 | In25-6 | rs140222628 | c.3878-95G>A |  | 0.0161 | 0.0050 | 3.21 | 0.0048 | - | - | - |
| 95 | In25-7 | rs12663927 | c.3878-92C>G |  | 0.2291 | 0.1050 | 2.18 | 0.0938 | - | - | 0.1165 |
| 96 | In25-8 | rs74621474 | c.3878-31C>T |  | 0.0043 | 0.0000 | - | 0.0004 | - | - | - |
| 97 | Ex26-1 | rs12663916 | c.3906C>T | p.(His1302=) | 0.2308 | 0.1050 | 2.20 | 0.0938 | 0.1114 | 0.1267 | 0.1172 |
| 98 | Ex26-2 | rs12662610 | c.3936A>G | p.(Thr1312=) | 0.2308 | 0.1050 | 2.20 | 0.0966 | 0.1143 | 0.1277 | 0.1174 |
| 99 | Ex26-3 | rs12663622 | c.3973C>G | p.(Gln1325Glu) | 0.2308 | 0.1050 | 2.20 | 0.0938 | 0.1051 | 0.1261 | 0.1172 |
| 100 | Ex26-4 | rs12663619 | c.4026C>T | p.(Ser1342=) | 0.2308 | 0.1050 | 2.20 | 0.0938 | 0.1036 | 0.1274 | 0.1172 |
| 101 | Ex26-5 | rs17403955 | c.4081A>G | p.(Ile1361Val) | 0.2308 | 0.1050 | 2.20 | 0.0938 | 0.1034 | 0.1277 | 0.1175 |
| 102 | Ex26-6 | rs624851 | c.4256T>C | p.(Leu1419Ser) | 0.2468 | 0.2450 | 1.01 | 0.2185 | 0.253 | 0.2932 | 0.2606 |
| 103 | Ex26-7 | rs62415828 | c.4352T>C | p.(Ile1451Thr) | 0.2296 | 0.1150 | 2.00 | 0.094 | 0.1036 | 0.1277 | 0.1174 |
| 104 | Ex26-8 | rs527236075 | c.4387delA | p.(Arg1463Glyfs\*15) | 0.0011 | 0.0000 | - | - | - | - | - |
| 105 | Ex26-9 | rs527236073 | c.4395\_4402dupTCAAGAGG | p.(Asp1468Valfs\*13) | 0.0011 | 0.0000 | - | - | - | - | - |
| 106 | Ex26-10 | rs755759273 | c.4455A>G | p.(Arg1485=) | 0.0011 | 0.0000 | - | - | 0.00005 | - | - |
| 107 | Ex26-11 | rs62415827 | c.4543C>T | p.(Arg1515Trp) | 0.2313 | 0.1111 | 2.08 | 0.0938 | 0.1032 | 0.1268 | 0.1171 |
| 108 | Ex26-12 | rs62415826 | c.4549A>G | p.(Ser1517Gly) | 0.2313 | 0.1111 | 2.08 | 0.0938 | 0.1032 | 0.1265 | 0.1171 |
| 109 | Ex26-13 | rs62415825 | c.4593G>A | p.(Glu1531=) | 0.2313 | 0.1111 | 2.08 | 0.0938 | 0.1034 | 0.1224 | 0.1171 |
| 110 | Ex26-14 | rs77523865 | c.4897A>G | p.(Lys1633Glu) | 0.0032 | 0.0051 | 0.63 | 0.0002 | - | 0.0021 | - |
| 111 | Ex26-15 | - | c.4929A>G | p.(Ser1643=) | 0.0011 | 0.0000 | - | - | - | - | - |
| 112 | Ex26-16 | rs527236065 | c.4957dupA | p.(Ser1653Lysfs\*2) | 0.0970 | 0.0000 | - | - | - | 0.0021 | - |
| 113 | Ex26-17 | rs527236074 | c.5014C>T | p.(Gln1672\*) | 0.0011 | 0.0000 | - | - | - | - | - |
| 114 | Ex26-18 | rs527236071 | c.5202\_5203delGT | p.(Phe1735Glnfs\*6) | 0.0011 | 0.0000 | - | - | - | - | - |
| 115 | Ex26-19 | rs57312007 | c.5244A>C | p.(Leu1748Phe) | 0.0399 | 0.0550 | 0.72 | 0.0727 | 0.1029 | 0.0545 | 0.0685 |
| 116 | Ex26-20 | rs772953910 | c.5263G>A | p.(Asp1755Asn) | 0.0011 | 0.0000 | - | - | 0.00005 | - | - |
| 117 | Ex26-21 | rs16895517 | c.5617C>G | p.(Leu1873Val) | 0.2295 | 0.1150 | 2.00 | 0.0913 | 0.1097 | 0.1275 | 0.1172 |
| 118 | Ex27-1 | rs9353806 | c.5705A>T | p.(Asn1902Ile) | 0.3852 | - | - | 0.2913 | 0.2864 | 0.4399 | 0.4407 |
| 119 | In27-1 | rs3904083 | c.5835+155T>C |  | 0.3660 | - | - | 0.2863 | - | - | 0.4031 |
| 120 | In27-2 | - | c.5836-88\_5836-87delins　CATGTCTCTCCAAAGTGA |  | 0.0030 | 0.0000 | - | - | - | - | - |
| 121 | In28-1 | rs112848046 | c.5927+116T>C |  | 0.0195 | 0.0505 | 0.39 | 0.0296 | - | - | 0.0251 |
| 122 | In28-2 | rs1338831 | c.5927+392T>G |  | 0.4093 | 0.4899 | 0.84 | 0.2913 | - | - | 0.4329 |
| 123 | In28-3 | rs587278 | c.5928-35T>C |  | 0.3726 | - | - | 0.2919 | 0.2821 | 0.437 | 0.4439 |
| 124 | In29-1 | rs36133910 | c.6078+68A>G |  | 0.3726 | - | - | 0.2919 | - | - | 0.4435 |
| 125 | In29-2 | rs140892533 | c.6078+100G>A |  | 0.0096 | - | - | 0.0018 | - | - | - |
| 126 | In29-3 | rs36179168 | c.6078+335A>G |  | 0.3714 | - | - | 0.3946 | - | - | 0.4416 |
| 127 | In29-4 | rs36127403 | c.6078+451T>G |  | 0.3695 | - | - | 0.3944 | - | - | 0.4429 |
| 128 | In29-5 | rs115236489 | c.6079-85A>G |  | - | - | - | 0.0134 | - | - | 0.0111 |
| 129 | In29-6 | rs149568304 | c.6079-23C>A |  | - | - | - | 0.026 | 0.0143 | - | 0.03547 |
| 130 | In29-7 | rs35395170 | c.6079-4\_6079-3delTC |  |  |  | - | - | - | - | - |
| 131 | In30-1 | rs9362694 | c.6191+132G>A |  | 0.4757 | - | - | 0.2518 | - | - | 0.3547 |
| 132 | In30-2 | rs79945794 | c.6191+210A>T |  | 0.1466 | - | - | 0.0497 | - | - | 0.1358 |
| 133 | In31-1 | rs149522181 | c.6192-250\_6192-249delCA |  | 0.0845 | - | - | 0.0523 | - | - | - |
| 134 | In31-2 | rs990233632 | c.6192-145A>G |  | 0.0072 | - | - | - | - | - | - |
| 135 | In31-3 | rs190537633 | c.6424+37A>G |  | 0.0049 | - | - | 0.004 | 0.0007 | - | - |
| 136 | Ex32-1 | - | c.6474G>T | p.(Leu2158=) | 0.0011 | 0.0000 | - | - | - | 0.0008 | - |
| 137 | Ex32-2 | rs527236068 | c.6557G>A | p.(Gly2186Glu) | 0.0117 | 0.0000 | - | - | - | - | - |
| 138 | Ex32-3 | - | c.6563T>C | p.(Ile2188Thr) | 0.0064 | 0.0000 | - | - | - | - | - |
| 139 | In32-1 | - | c.6571+6T>A |  | 0.0011 | 0.0000 | - | - | - | - | - |
| 140 | In33-1 | - | c.6725+20G>A |  | 0.0034 | - | - | - | - | - | - |
| 141 | In34-1 | rs66502009 | c.6834+61T>G |  | 0.3883 | - | - | 0.2698 | - | - | 0.229 |
| 142 | In34-2 | rs76633698 | c.6834+268A>T |  | 0.3883 | - | - | 0.2919 | - | - | - |
| 143 | In34-3 | rs1482457 | c.6835-64C>T |  | 0.4304 | 0.3538 | 1.22 | 0.3484 | - | - | 0.343 |
| 144 | Ex35-1 | rs180912436 | c.6844G>A | p.(Glu2282Lys) | 0.0016 | 0.0000 | - | 0.0002 | - | - | - |
| 145 | Ex35-2 | rs4710457 | c.6977G>A | p.(Arg2326Gln) | 0.4290 | 0.3538 | 1.21 | 0.3496 | 0.3632 | 0.3489 | 0.3436 |
| 146 | Ex35-3 | - | c.7028\_7029delinsATCGT | p.(Leu2343delinsHisArg) | 0.0016 | 0.0000 | - | - | - | - | - |
| 147 | In35-1 | rs118052257 | c.7055+271C>T |  | 0.0379 | 0.0231 | 1.64 | 0.0038 | - | - | 0.025 |
| 148 | In35-2 | rs374537625 | c.7056-145A>G |  | 0.3349 | 0.2279 | 1.47 | - | - | - | - |
| 149 | In35-3 | rs139056492 | c.7056-15T>A |  | 0.0072 | 0.0217 | 0.33 | 0.0034 | 0.0007 | - | 0.0132 |
| 150 | Ex37-1 | - | c.7283C>A | p.(Ser2428\*) | 0.0011 | 0.0000 | - | - | - | - | - |
| 151 | Ex37-2 | rs145184183 | c.7394C>G | p.(Thr2465Ser) | 0.0332 | 0.0202 | 1.64 | 0.0006 | 0.00005 | 0.0296 | 0.029 |
| 152 | In37-1 | rs138375660 | c.7411+95\_7411+96insA |  | 0.0000 | 0.0051 | 0.00 | 0.0164 | - | - | - |
| 153 | In37-2 | - | c.7411+141T>A |  | 0.0011 | 0.0000 | - | - | - | - | - |
| 154 | In37-3 | rs973578989 | c.7411+142G>A |  | 0.0021 | 0.0051 | 0.42 | - | - | - | - |
| 155 | In37-4 | - | c.7411+154C>A |  | 0.0032 | 0.0000 | - | - | - | - | - |
| 156 | In37-5 | - | c.7411+332G>A |  | 0.0022 | 0.0000 | - | - | - | - | - |
| 157 | In37-6 | rs117410463 | c.7411+380C>A |  | 0.0535 | 0.0404 | 1.32 | 0.0052 | - | - | 0.0335 |
| 158 | In37-7 | rs2008452 | c.7411+411T>G |  | 0.2621 | 0.2172 | 1.21 | 0.1761 | - | - | 0.2131 |
| 159 | Ex39-1 | rs189406424 | c.7609G>A | p.(Ala2537Thr) | 0.0032 | 0.0101 | 0.32 | 0.0018 | 0.0001 | 0.005 | - |
| 160 | Ex39-2 | - | c.7616delG | p.(Gly2539Glufs\*14) | 0.0011 | 0.0000 | - | - | - | - | - |
| 161 | Ex39-3 | - | c.7665\_7666delCA | p.(Tyr2555\*) | 0.0011 | 0.0051 | 0.21 | - | - | - | - |
| 162 | Ex39-4 | rs66462731 | c.7666A>T | p.(Ser2556Cys) | 0.2719 | 0.2245 | 1.21 | 0.0899 | 0.0777 | 0.2347 | 0.2112 |
| 163 | Ex39-5 | - | c.7713T>G | p.(Asn2571Lys) | 0.0011 | 0.0000 | - | - | - | - | - |
| 164 | In39-1 | rs144282503 | c.7724-12C>T |  | 0.0021 | 0.0000 | - | 0.001 | 0.0004 | - | - |
| 165 | Ex40-1 | rs527236064 | c.7793G>A | p.(Gly2598Asp) | 0.0021 | 0.0000 | - | - | - | - | - |
| 166 | In40-1 | rs190814750 | c.7899-202G>A |  | 0.0021 | 0.0000 | - | 0.0006 | - | - | - |
| 167 | Ex41-1 | rs527236066 | c.7919G>A | p.(Trp2640\*) | 0.0064 | 0.0000 | - | - | - | - | - |
| 168 | Ex41-2 | rs527236076 | c.8012T>A | p.(Leu2671\*) | 0.0011 | 0.0000 | - | - | - | - | - |
| 169 | In41-1 | rs4710257 | c.8071+84T>G |  | 0.2249 | 0.2000 | 1.12 | 0.2883 | - | - | 0.1945 |
| 170 | In42-1 | rs182430037 | c.8134+12T>G |  | 0.0024 | - | - | 0.0002 | - | - | - |
| 171 | In42-2 | rs7764184 | c.8134+367T>A |  | 0.2561 | - | - | 0.3832 | - | - | 0.1902 |
| 172 | In43-1 | - | c.8297-95T>C |  | 0.0011 | 0.0000 | - | - | - | - | - |
| 173 | In43-2 | - | c.8297-88A>C |  | 0.0021 | 0.0000 | - | - | - | - | - |
| 174 | Ex44-1 | - | c.8331delC | p.(Val2778Phefs\*14) | 0.0011 | 0.0000 | - | - | - | - | - |
| 175 | Ex44-2 | rs527236070 | c.8439\_8442dupTGCA | p.(Glu2815Cysfs\*19) | 0.0011 | 0.0000 | - | - | - | - | - |
| 176 | Ex44-3 | - | c.8696C>G | p.(Ala2899Gly) | 0.0011 | 0.0000 | - | - | - | - | - |
| 177 | Ex44-4 | - | c.8759G>C | p.(Cys2920Ser) | 0.0011 | 0.0000 | - | - | - | - | - |
| 178 | Ex44-5 | rs527236067 | c.8868C>A | p.(Tyr2956\*) | 0.0480 | 0.0000 | - | - | - | 0.0029 | - |
| 179 | Ex44-6 | rs888739369 | c.9082G>T | p.(Asp3028Tyr) | 0.0011 | 0.0000 | - | - | - | - | - |
| 180 | Ex44-7 | - | c.9094A>G | p.(Ile3032Val) | 0.0011 | 0.0000 | - | - | - | - | - |
| 181 | Ex44-8 | rs779289332 | c.9164A>G | p.(Tyr3055Cys) | 0.0011 | 0.0000 | - | - | 0.00005 | - | - |
| 182 | Ex44-9 | rs183589498 | c.9272T>C | p.(Ile3091Thr) | 0.0021 | 0.0050 | 0.43 | 0.0004 | 0.0001 | 0.0021 | - |
| 183 | Ex44-10 | rs139944387 | c.9300A>G | p.(Leu3100=) | 0.0053 | 0.0100 | 0.53 | 0.0018 | 0.0001 | 0.0095 | 0.0168 |
| 184 | Ex44-11 | rs374161234 | c.9477T>G | p.(Asp3159Glu) | 0.0011 | 0.0000 | - | 0.0004 | 0.0002 | 0.0004 | - |
| 185 | Ex44-12 | - | c.\*183A>C |  | 0.0476 | 0.0000 | - | - | - | - | - |
| 186 | Ex44-13 | rs193035948 | c.\*529A>C |  | 0.0023 | 0.0000 | - | 0.0016 | - | - | - |
| An orange column indicates a deletion and insertion variant or nonsense variant. A yellow column indicates a putative pathogenic missense variant. MAF, minor allele frequency; HGVD, Human Genetic Variation Database; iJGVD, Integrative Japanese Genome Variation Database | | | | | | | | | | | |