|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| Supplementary Table S3. Patients carrying possible pathogenic variants or rare variants with an MAF of <0.01 in the *EYS* gene. | | | | | | | |
| No. | Patient ID | Inheritance pattern | Allele 1 | Allele 2 | Possible damaging variant or rare variant | CNVs | Reference |
| 1 | RP003 | ar | JV1 |  |  | ND | 8 |
| 2 | RP004 | s | JV1 | JV1 |  |  | 8 |
| 3 | RP008 | s | JV1 | JV3 |  |  | 8 |
| 4 | RP010 | ar |  |  | c.3489T>A (p.(Asn1163Lys)) c.3685-94T>C c.6078+100G>A |  | 8 |
| 5 | RP012 | s | JV1 | JV1 | c.1929T>C (p.(Asp643=)) c.6192-145A>G |  | 8 |
| 6 | RP013 | s |  |  | c.8297-88A>C |  |  |
| 7 | RP014 | s |  |  | c.3809T>G (p.(Val1270Gly)) c.3444-149A>G |  | 8 |
| 8 | RP016 | ar | JV1 | c.1211dupA (p.(Asn404Lysfs\*3)) |  |  | 8 |
| 9 | RP018 | s |  |  | c.3684+101T>C |  |  |
| 10 | RP019 | s |  |  | c.4929A>G |  |  |
| 11 | RP020 | s | JV4 | JV4 |  |  |  |
| 12 | RP021 | ar | JV3 |  |  | ND | 8 |
| 13 | RP026 | ar | JV2 |  |  | ND | 8 |
| 14 | RP028 | s | JV1 | JV3 |  |  | 8 |
| 15 | RP029 | s | JV2 | c.7665\_7666delCA (p.(Tyr2555\*)) |  |  | 8 |
| 16 | RP030 | s |  |  | c.9094A>G (p.(Ile3032Val)) |  |  |
| 17 | RP034 | ad |  |  | c.3878-31C>T c.5927+116T>C |  |  |
| 18 | RP036 | ar |  |  | c.1766+61A>G |  |  |
| 19 | RP037 | ad | JV1 | JV1 |  |  | 8 |
| 20 | RP038 | s | JV1 | JV1 |  |  | 8 |
| 21 | RP041 | s |  |  | c.3878-95G>A |  |  |
| 22 | RP043 | ar | JV1 | JV3 |  |  | 8 |
| 23 | RP044 | ar | JV1 | JV2 JV3 |  |  | 8 |
| 24 | RP045 | ar | JV2 |  |  | ND | 8 |
| 25 | RP048 | ar |  |  | c.3878-31C>T |  |  |
| 26 | RP049 | ar | JV2 JV3 | JV2 JV3 |  |  | 8 |
| 27 | RP050 | s | JV3 | c.7283C>A (p.(Ser2428\*)) |  |  | 8 |
| 28 | RP051 | ar |  |  | c.3164+360G>A |  |  |
| 29 | RP054 | s | c.1485\_1493delGGTTATT GAinsCGAAAAG (p.(Val496Glufs\*1)) |  | c.7028\_7029delTGinsATCGT (p.(Leu2343delinsHisArg)) c.2259C>T (p.(Leu753=)) |  | 8 |
| 30 | RP055 | s | JV1 | JV3 | c.632G>A (p.(Cys211Tyr)) |  | 8 |
| 31 | RP057 | s | JV1 | JV1 |  |  | 8 |
| 32 | RP061 | s | JV1 | JV2 |  |  | 8 |
| 33 | RP062 | ar | JV1 | JV3 |  |  | 8 |
| 34 | RP063 | ar | JV1 | JV1 |  |  | 8 |
| 35 | RP066 | ar | JV3 |  |  | ND | 8 |
| 36 | RP068 | s | JV1 | JV4 |  |  | 8 |
| 37 | RP106 | s | JV1 | JV1 |  |  |  |
| 38 | RP109 | ad |  |  | c.2980C>G (p.(Pro994Ala)) c.863-94A>G c.3444-274C>A |  |  |
| 39 | RP111 | ar |  |  | c.5836-88\_5836-87delinsCA TGTCTCTCCAAAGTGA |  |  |
| 40 | RP112 | s |  |  | c.9272T>C (p.(Ile3091Thr)) |  |  |
| 41 | RP113 | ar |  |  | c.1600-126T>C |  |  |
| 42 | RP115 | ar |  |  | c.3878-31C>T |  |  |
| 43 | RP119 | s |  |  | c.\*529A>C |  |  |
| 44 | RP122 | ar | JV1 | JV3 |  |  |  |
| 45 | RP125 | s | JV3 |  |  | ND |  |
| 46 | RP127 | ar | JV3 |  |  | ND |  |
| 47 | RP131 | ar | JV1 | JV1 |  |  |  |
| 48 | RP134 | ar | JV2 | JV3 |  |  |  |
| 49 | RP135 | s |  |  | c.7899-202G>A |  |  |
| 50 | RP136 | s |  |  | c.3878-31C>T |  |  |
| 51 | RP137 | s | JV3 |  |  | ND |  |
| 52 | RP140 | s | JV2 | JV2 |  |  |  |
| 53 | RP141 | ar | JV3 |  |  | ND |  |
| 54 | RP143 | ar |  |  | c.1766+61A>G c.3878-95G>A |  |  |
| 55 | RP145 | s | JV2 |  | c.334G>C (p.(Glu335Asp)) |  |  |
| 56 | RP146 | s | JV4 |  |  | ND |  |
| 57 | RP150 | ar | JV1 |  |  | ND |  |
| 58 | RP151 | ad | JV1 | JV3 | c.749-132C>T |  |  |
| 59 | RP153 | ad |  |  | c.749-136T>A c.1184+261T>C c.1600-126T>C |  |  |
| 60 | RP156 | s | JV1 | JV1 |  |  |  |
| 61 | RP159 | xl |  |  | c.7899-202G>A |  |  |
| 62 | RP161 | ar | JV1 |  |  | ND |  |
| 63 | RP162 | s | JV2 |  |  | ND |  |
| 64 | RP164 | ar | JV2 | JV2 |  |  |  |
| 65 | RP165 | s | JV1 | c.1211dupA  (p.(Asn404Lysfs\*3)) |  |  |  |
| 66 | RP166 | uncertain | JV3 |  | c.3164+296G>A c.3164+360G>A | ND |  |
| 67 | RP167 | s | JV1 | JV3 | c.7411+141T>A |  |  |
| 68 | RP169 | s |  |  | c.8759G>C (p.(Cys2920Ser)) c.3164+296G>A c.3164+360G>A |  |  |
| 69 | RP171 | s | JV3 | JV5 |  |  |  |
| 70 | RP172 | s | JV1 | JV1 |  |  |  |
| 71 | RP174 | ad | JV1 | JV2 | c.334G>C (p.(Glu335Asp)) |  |  |
| 72 | RP176 | s | JV1 | JV1 |  |  |  |
| 73 | RP177 | s | JV2 | Exon 1 del |  | Exon 1 del |  |
| 74 | RP178 | s | JV1 |  | c.9082G>T (p.(Asp3028Tyr)) | ND |  |
| 75 | RP180 | s |  |  | c.2259C>T (p.(Leu753=)) |  |  |
| 76 | RP182 | ar |  |  | c.4897A>G (p.(Lys1633Glu)) c.2641+205G>A c.6078+100G>A c.7609G>A (p.(Ala2537Thr)) |  |  |
| 77 | RP184 | ar | JV1 | c.7919G>A (p.(Trp2640\*)) |  |  |  |
| 78 | RP185 | ar | JV1 | JV3 |  |  |  |
| 79 | RP187 | s |  |  | c.2259C>T (p.(Leu753=)) |  |  |
| 80 | RP188 | ar | JV1 | JV4 |  |  |  |
| 81 | RP191 | s |  |  | c.6844G>A (p.(Glu2282Lys)) |  |  |
| 82 | RP193 | s | JV3 | JV3 |  |  |  |
| 83 | RP195 | s | JV1 | JV2 |  |  |  |
| 84 | RP196 | s | JV3 | c.942delT (p.(Ala315Leufs\*24)) |  |  |  |
| 85 | RP198 | s | c.7616delG (p.(Gly2539Glufs\*14)) |  | c.1184+124T>G | ND |  |
| 86 | RP199 | s |  |  | c.1766+95G>A |  |  |
| 87 | RP200 | s |  |  | c.1766+95G>A |  |  |
| 88 | RP203 | s | JV1 |  |  | ND |  |
| 89 | RP206 | s | JV1 | JV3 |  |  |  |
| 90 | RP208 | s |  |  | c.7724-12C>T |  |  |
| 91 | RP210 | s | JV2 | JV4 |  |  |  |
| 92 | RP211 | s | JV1 | JV3 |  |  |  |
| 93 | RP213 | ar | JV3 | JV3 | c.632G>A (p.(Cys211Tyr)) |  |  |
| 94 | RP214 | s |  |  | c.2980C>G (p.(Pro994Ala)) c.3444-274C>A |  |  |
| 95 | RP216 | s | JV2 |  |  | ND |  |
| 96 | RP219 | s | JV3 | JV3 |  |  |  |
| 97 | RP222 | s |  |  | c.9300A>G (p.(Leu3100=)) |  |  |
| 98 | RP227 | s |  |  | c.3454G>A (p.(Gly1152Arg)) c.7411+142G>A c.9300A>G (p.(Leu3100=)) |  |  |
| 99 | RP232 | ar | JV3 | c.525\_527delGGA (p.(Glu176del)) |  |  |  |
| 100 | RP233 | ad | JV3 | JV5 | c.5263G>A (p.(Asp1755Asn)) |  |  |
| 101 | RP237 | ar | JV1 | JV1 |  |  |  |
| 102 | RP239 | s | JV1 | Exon 31 del |  | Exon 31 del |  |
| 103 | RP241 | s | JV1 | Exon 1-Intron 1 del | c.6424+37A>G | Exon 1-Intron 1 del |  |
| 104 | RP242 | ar |  |  | c.7713T>G (p.(Asn2571Lys)) |  |  |
| 105 | RP243 | s |  |  | c.7793G>A (p.(Gly2598Asp)) c.5927+116T>C |  |  |
| 106 | RP244 | ar | JV2 | JV3 |  |  |  |
| 107 | RP246 | ar |  |  | c.1766+61A>G |  |  |
| 108 | RP247 | s |  |  | c.9477T>G (p.(Asp3159Glu)) |  |  |
| 109 | RP248 | s |  |  | c.1460-5dupT c.2739-42T>C |  |  |
| 110 | RP249 | s | JV2 JV3 | JV3 |  |  |  |
| 111 | RP250 | s | JV2 | Exon 33 del | c.334G>C (p.(Glu335Asp)) | Exon 33 del |  |
| 112 | RP252 | s |  |  | c.3489T>A (p.(Asn1163Lys)) c.3685-94T>C |  |  |
| 113 | RP259 | ar |  |  | c.749-132C>T |  |  |
| 114 | RP262 | s | JV1 | JV3 |  |  |  |
| 115 | RP263 | ar | JV1 |  |  | ND |  |
| 116 | RP264 | s | JV3 |  |  |  |  |
| 117 | RP265 | ar |  |  | c.6192-145A>G |  |  |
| 118 | RP272 | ar | JV1 | JV4 |  |  |  |
| 119 | RP274 | s |  |  | c.9300A>G (p.(Leu3100=)) |  |  |
| 120 | RP279 | s | JV1 | JV2 |  |  |  |
| 121 | RP280 | s | JV2 |  |  |  |  |
| 122 | RP281 | s |  |  | c.2137+320A>G |  |  |
| 123 | RP283 | s |  |  | c.2490T>C (p.(Pro830=)) |  |  |
| 124 | RP285 | s | c.1211dupA (p.(Asn404Lysfs\*3)) |  | c.334G>C (p.(Glu335Asp)) c.5836-88\_5836-87delinsCA TGTCTCTCCAAAGTGA |  |  |
| 125 | RP288 | ad | JV1 | c.525\_527delGGA (p.(Glu176del)) | c.3684+93G>T |  |  |
| 126 | RP290 | s | JV3 |  |  |  |  |
| 127 | RP291 | ar | JV1 |  |  |  |  |
| 128 | RP294 | s ( LCA) |  |  | c.4897A>G (p.(Lys1633Glu)) c.2641+205G>A c.7609G>A (p.(Ala2537Thr)) c.6078+100G>A |  |  |
| 129 | KRP005 | s | JV1 | c.8331delC (p.(Val2778Phefs\*14)) | c.3809T>G (p.(Val1270Gly)) |  |  |
| 130 | KRP006 | ar | JV1 |  | c.6474G>T (p.(Leu2158=)) |  |  |
| 131 | KRP007 | s | JV1 |  | c.7793G>A (p.(Gly2598Asp)) | ND | 10 |
| 132 | KRP008 | s | JV2 | c.525\_527delGGA (p.(Glu176del)) |  |  |  |
| 133 | KRP009 | ar | JV3 |  |  | ND |  |
| 134 | KRP012 | s | JV1 |  |  | ND |  |
| 135 | KRP014 | s | JV1 | JV1 |  |  | 10 |
| 136 | KRP019 | ar | JV1 | JV3 |  |  |  |
| 137 | KRP021 | ar | JV1 | JV1 |  |  | 10 |
| 138 | KRP022 | s | c.7919G>A (p.(Trp2640\*)) | c.7919G>A (p.(Trp2640\*)) | c.334G>C (p.(Val112Leu)) c.3444-149A>G c.7411+154C>A |  | 10 |
| 139 | KRP025 | ar |  |  | c.7411+142G>A c.9300A>G (p.(Leu3100=)) |  |  |
| 140 | KRP031 | ar | c.7919G>A (p.(Trp2640\*)) | c.7919G>A (p.(Trp2640\*)) | c.334G>C (p.(Val112Leu)) c.3444-149A>G c.7411+154C>A |  | 10 |
| 141 | KRP033 | ar | JV1 | c.1750G>T (p.(Glu584\*)) | c.9272T>C (p.(Ile3091Thr)) |  | 10 |
| 142 | KRP034 | s |  |  | c.4897A>G (p.(Lys1633Glu)) c.7609G>A (p.(Ala2537Thr)) c.2641+205G>A c.3164+360G>A c.6078+100G>A |  |  |
| 143 | KRP036 | ar | JV4 | JV4 |  |  |  |
| 144 | KRP037 | ar | JV1 | JV1 |  |  | 10 |
| 145 | KRP038 | ar | JV1 | JV2 | c.4455A>G (p.(Arg1485=)) |  |  |
| 146 | KRP042 | s | JV3 |  |  | ND |  |
| 147 | KRP044 | ar | JV1 | c.525\_527delGGA (p.(Glu176del)) |  |  |  |
| 148 | KRP048 | s | JV2 |  |  | ND |  |
| 149 | KRP049 | s | JV1 |  | c.6571+6T>A | ND |  |
| 150 | KRP050 | ar | JV3 | c.1750G>T (p.(Glu584\*)) |  |  |  |
| 151 | KRP054 | s | JV4 | Exon 6-8 del |  | Exon 6-8 del |  |
| 152 | KRP055 | s | JV1 | JV2 |  |  | 10 |
| 153 | KRP056 | s | JV1 | JV3 |  |  |  |
| 154 | KRP057 | ar | JV2 |  |  | ND |  |
| 155 | KRP058 | ar | c.5202\_5203delGT (p.(Phe1735Glnfs\*6)) | c.8439\_8442dupTGCA (p.(Glu2815Cysfs\*19)) |  |  | 10 |
| 156 | KRP064 | s | JV2 |  |  | ND |  |
| 157 | KRP065 | s |  |  | c.1766+61A>G |  |  |
| 158 | KRP066 | s | JV2 | JV3 |  |  |  |
| 159 | KRP072 | s | JV1 |  | c.2137+320A>G | ND |  |
| 160 | KRP074 | s | JV3 | JV3 | c.632G>A (p.(Cys211Tyr)) c.9164A>G (p.(Tyr3055Cys)) |  |  |
| 161 | KRP077 | ar | JV1 | Exon 33 del |  | Exon 33 del |  |
| 162 | KRP078 | ar | JV2 |  | c.334G>C (p.(Glu335Asp)) c.8134+12T>G | ND |  |
| 163 | KRP079 | s | JV3 |  |  | ND |  |
| 164 | KRP080 | s | JV2 | JV5 |  |  |  |
| 165 | KRP082 | s | JV3 |  |  | ND |  |
| 166 | KRP083 | s | JV3 |  |  | ND |  |
| 167 | KRP084 | s | JV2 |  |  | ND |  |
| 168 | KRP085 | s | JV1 |  | c.8696C>G (p.(Ala2899Gly)) c.6725+20G>A | ND |  |
| 169 | KRP088 | ar | JV1 |  |  | ND |  |
| 170 | KRP090 | s | JV2 | c.4395\_4402dupTCAAGAGG (p.(Asp1468Valfs\*13)) |  |  | 10 |
| 171 | KRP093 | s |  |  | c.2490T>C (p.(Pro830=)) |  |  |
| 172 | KRP099 | s |  |  | c.334G>C (p.(Val112Leu)) |  |  |
| 173 | KRP101 | s |  |  | c.9300A>G (p.(Leu3100=)) |  |  |
| 174 | KRP102 | ar | JV2 | JV3 |  |  |  |
| 175 | KRP109 | s | JV2 | JV3 |  |  |  |
| 176 | KRP112 | s | JV3 |  |  | ND |  |
| 177 | KRP115 | s |  |  | c.1600-126T>C |  |  |
| 178 | KRP116 | s | JV1 | JV2 |  |  | 10 |
| 179 | KRP121 | ar | JV1 | JV1 |  |  | 10 |
| 180 | KRP128 | ar | JV3 | JV4 |  |  |  |
| 181 | KRP133 | s | JV1 |  | c.1929T>C (p.(Asp643=)) | ND |  |
| 182 | KRP134 | ar | JV3 |  |  | ND |  |
| 183 | KRP135 | s | JV2 | c.5014C>T (p.(Gln1672\*)) |  |  | 10 |
| 184 | KRP139 | s |  |  | c.8297-88A>C |  |  |
| 185 | KRP140 | s | c.525\_527delGGA (p.(Glu176del)) |  |  |  |  |
| 186 | KRP141 | s | JV1 |  |  | ND |  |
| 187 | KRP143 | s | JV1 | JV3 |  |  |  |
| 188 | KRP144 | s | JV3 |  |  | ND |  |
| 189 | KRP150 | s | JV1 | JV2 |  |  | 10 |
| 190 | KRP152 | s | JV1 | JV2 |  |  | 10 |
| 191 | KRP153 | s |  |  | c.1600-126T>C |  |  |
| 192 | KRP155 | s | c.4387delA (p.(Arg1463Glyfs\*15)) |  |  |  | 10 |
| 193 | KRP157 | s | JV1 | JV5 |  |  |  |
| 194 | KRP161 | ar | JV2 | JV3 |  |  |  |
| 195 | KRP162 | s | JV3 |  |  | ND |  |
| 196 | KRP168 | s | JV3 |  | c.3809T>G (p.(Val1270Gly)) | ND |  |
| 197 | KRP169 | ar | JV5 |  |  | ND |  |
| 198 | KRP170 | s | JV2 | c.8012T>A (p.(Leu2671\*)) | c.2137+40A>C |  | 10 |
| 199 | KRP171 | s |  |  | c.7724-12C>T |  |  |
| 200 | KRP178 | s | JV1 |  |  | ND |  |
| 201 | KRP180 | s | JV1 |  |  | ND |  |
| 202 | KRP181 | s |  |  | c.8297-95T>C |  |  |
| 203 | KRP183 | s | JV1 | JV2 |  |  | 10 |
| 204 | KRP185 | s | JV3 | JV3 |  |  |  |
| 205 | KRP186 | s | JV3 |  | c.6424+37A>G | ND |  |
| 206 | KRP188 | ar | JV3 | c.7919G>A (p.(Trp2640\*)) | c.334G>C (p.(Val112Leu)) c.3444-149A>G c.7411+154C>A |  |  |
| 207 | KRP190 | s | JV1 | Exon 39-42 dup |  | Exon 39-42 dup |  |
| 208 | KRP199 | s |  |  | c.1600-126T>C |  |  |
| 209 | KRP202 | s | JV2 |  |  | ND |  |
| 210 | KRP204 | s | JV3 | JV5 |  |  |  |
| An inheritance pattern was inferred based on the pedigree structure of each family. ar, autosomal recessive; ad, autosomal dominant; s, simplex; xl, X-linked; LCA, Leber's congenital amaurosis; JV1, c.4957dupA (p.(Ser1653Lysfs\*2)); JV2, c.8868C>A (p.(Tyr2956\*)); JV3, c.2528G>A (p.(Gly843Glu)); JV4, c.6557G>A (p.(Gly2186Glu)); JV5, c.6563T>C (p.(Ile2188Thr)); ND, not detected; del, deletion; dup, duplication | | | | | | | |