## Supplemental Figure S1

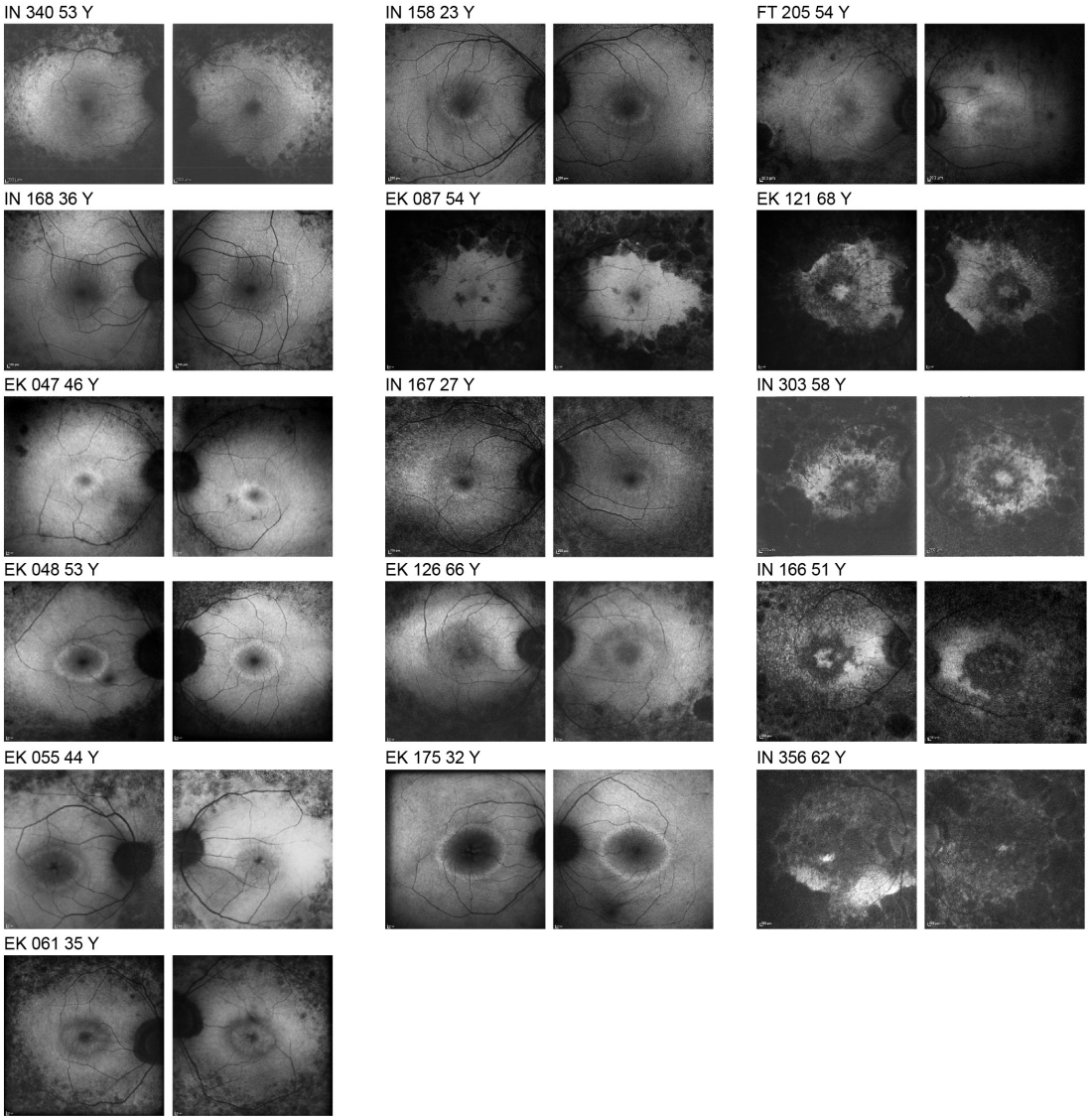


**Figure S1.** FAF patterns of right and left eyes of *ADGVR1* patients. ID and age of each patient is stated above the two images of each patient (in each column, the right eye is shown on the left and left eye is shown on the right). First two columns show hyperautofluorescent rings with preserved central retina while the third column shows advanced disease - hyperautofluorescent patch and atrophy. Note older ages of patients in the last column. Note also good interocular symmetry.

## Supplemental Table S1

Pathogenic variants of studied patients.

| Treatrush ID | Reference ID | Reference | CENTRE | GENE | Variant 1 | Variant2 |
|---|---|---|---|---|---|---|
| TR_EK-009 | DE03U06091 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Cys536ter) | p.(Glu767Serfs*21) |
| TR_EK-010 | DE03U07100 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Arg626ter) | p.(Gly516Arg) |
| TR_EK-012 | DE03U13121 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Arg3538ter) | p.(Glu767Serfs*21) |
| TR_EK-013 | DE03U13121 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Arg3538ter) | p.(Glu767Serfs*21) |
| TR_EK-014 | DE03U21141 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Glu767Serfs*21) | p.(Ile3103Val) |

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|---|---|---|---|---|---|---|
| TR_EK-016 | DE03U25161 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Val218Glu) | p.(Cys419Phe) |
| TR_EK-029 | DE03U26290 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Glu4458Aspfs*3) | p.(Cys4140Phe) |
| TR_EK-032 | DE03U27321+ | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(=,Tyr318Cysfs*17) | p.(Ile5166Val) |
| TR_EK-035 | DE03U27321+ | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(=,Tyr318Cysfs*17) | p.(Ile5166Val) |
| TR_EK-036 | DE03U28360 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Leu2380Profs*37) | p.(Gly3647Ser) |
| TR_EK-038 | DE03U29381 | Bonnet et al, 2016 | GERMANY | *USH2A* | c.5776+1G>A | p.(Ala3944Asp) |
| TR_EK-039 | DE03U30390 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Trp2945ter) | c.4627+25435_4987+660del |
| TR_EK-040 | DE03U31401 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Glu767Serfs*21) | c.(3157+1_3158-1)_(4627+1_4628-1)del |
| TR_EK-046 | DE03U33461 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Trp3955ter) | p.(Ser259Phefs*63) |
| TR_EK-049 | DE03U34490 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Glu767Serfs*21) | c.11549-1G>A |
| TR_EK-051 | DE03U35510 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Gln4711ter) | c.5776+2T>C |
| TR_EK-053 | DE03U36530 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Glu767Serfs*21) | p.(Trp2075Cys) |
| TR_EK-054 | DE03U37541 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Glu767Serfs*21) | p.(Trp3521Arg) |
| TR_EK-056 | DE03U38561 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Glu767Serfs*21) | p.(Cys2309Phe) |
| TR_EK-058 | DE03U02581 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Glu767Serfs*21) | p.(Glu767Serfs*21) |
| TR_EK-066 | DE03U39661 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Cys870ter) | p.(Arg4192Cys) |
| TR_EK-067 | DE03U40671 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Glu767Serfs*21) | c.7595-2144A>G |
| TR_EK-069 | DE03U41701+ | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Glu767Serfs*21) | c.(11548+1_11549-1)_(11711+1_11712-1)del |
| TR_EK-070 | DE03U41701+ | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Glu767Serfs*21) | c.(11548+1_11549-1)_(11711+1_11712-1)del |
| TR_EK-071 | DE03U42710 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Trp3955ter) | p.(=,Tyr318Cysfs*17) |
| TR_EK-072 | N/A | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Trp3955ter) | p.(Glu4445_Ser4449delinsAspLeu) |
| TR_EK-073 | DE03U43730 | Bonnet et al, 2016 | GERMANY | *USH2A* | c.11549-1G>A | c.(2993+1_2994-1)_(13811+1_13812-1)dup |
| TR_EK-075 | DE03U44750 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Trp3702ter) | p.(Trp3702ter) |
| TR_EK-077 | DE03U45770 | Bonnet et al, 2016 | GERMANY | *USH2A* | c.5777-2A>G | p.(Trp3521Arg) |
| TR_EK-080 | DE03U03800 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Glu767Serfs*21) | p.(Glu767Serfs*21) |
| TR_EK-081 | DE03U46811 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Trp3955ter) | p.(Asn346His) |
| TR_EK-082 | DE03U47821 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Cys870ter) | p.(Thr2919Pro) |
| TR_EK-084 | DE03U48840 | Bonnet et al, 2016 | GERMANY | *USH2A* | c.14791+2T>C | c.9259-2402_9371+1537del |
| TR_EK-085 | DE03U49851 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Trp3702ter) | p.(Trp3702ter) |
| TR_EK-088 | N/A | Glocke et al, 2014 | GERMANY | *USH2A* | p.(Val4367Argfs*22) | c.8682-1665_8845+140del |
| TR_EK-093 | DE03U04931 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Glu767Serfs*21) | p.(Glu767Serfs*21) |
| TR_EK-095 | DE03U50950 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Asn2651Glnfs*10) | p.(=,Tyr318Cysfs*17) |

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|---|---|---|---|---|---|---|
| TR_EK-096 | DE03U51960 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Glu767Serfs*21) | p.(Glu4458Aspfs*3) |
| TR_EK-097 | N/A | | GERMANY | *USH2A* | c.5776+1G>A | c.5776+1G>A |
| TR_EK-098 | DE03U52980 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Trp3955ter) | p.(Gln4676Profs*7) |
| TR_EK-099 | DE03U53990 | Bonnet et al, 2016 | GERMANY | *USH2A* | c.785-6636_1840+208del | p.(Trp3521Arg) |
| TR_EK-100 | DE03U081001 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Cys520Arg) | p.(Cys520Arg) |
| TR_EK-101 | DE03U091011 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Glu767Serfs*21) | p.(Trp3521Arg) |
| TR_EK-102 | DE03U101020 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Trp2945ter) | p.(Cys2128Tyr) |
| TR_EK-104 | DE03U111041 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Gln4711ter) | p.(Trp3521Arg) |
| TR_EK-114 | DE03U121140 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Asn4079Trpfs*19) | p.(Asn4079Trpfs*19) |
| TR_EK-120 | DE03U141201 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Arg4935ter) | p.(Gly3142ter) |
| TR_EK-124 | DE03U011240 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Glu767Serfs*21) | p.(Glu767Serfs*21) |
| TR_EK-125 | DE03U151250 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Tyr1103ter) | p.(His340Asp) |
| TR_EK-127 | N/A | | GERMANY | *USH2A* | p.(Glu767Serfs*21) | p.(Glu767Serfs*21) |
| TR_EK-128 | DE03U161282 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Gln4235ter) | p.(Glu767Serfs*21) |
| TR_EK-129 | DE03U171292 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Trp3955ter) | c.(11548+1_11549-1)_(11711+1_11712-1)del |
| TR_EK-130 | DE03U181302 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Cys3090ter) | p.(Trp3955ter) |
| TR_EK-131 | DE03U191312 | Bonnet et al, 2016 | GERMANY | *USH2A* | c.6805+2T>C | p.(Arg334Trp) |
| TR_EK-134 | DE03U201342 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Trp2841ter) | c.9258+1G>A |
| TR_EK-148 | DE03U221482 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Trp3955ter) | c.1144-2A>T |
| TR_EK-149 | DE03U231492 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Glu767Serfs*21) | p.(Val218Glu) |
| TR_EK-150 | DE03U241502 | Bonnet et al, 2016 | GERMANY | *USH2A* | p.(Trp3955ter) | p.(Val218Glu) |
| TR_EK-160 | N/A | | GERMANY | *USH2A* | p.(His308Serfs*16) | p.(Glu767Serfs*21) |
| TR_EK-179 | N/A | Neuhaus et al 2017 | GERMANY | *USH2A* | p.(Trp3955ter) | p.(Leu2215Serfs*16) |
| TR_EK-259 | N/A | | GERMANY | *USH2A* | p.(Trp2841ter) | p.(Gly3142ter) |
| TR_EK-278 | N/A | | GERMANY | *USH2A* | c.7301-1G>A | p.(Glu2238Ala) |
| TR_EK-279 | N/A | | GERMANY | *USH2A* | c.(11711+1_11712-1)_(12066+1_12067-1)del | c.(11711+1_11712-1)_(12066+1_12067-1)del |
| TR_FT-115 | IT02U111320 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Trp1084ter) | p.(Glu767Serfs*21) |
| TR_FT-184 | IT02U2420650 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Phe4993Profs*7) | p.(Phe4993Profs*7) |
| TR_FT-187 | IT02U1916760 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Tyr4329ter) | p.(Gly516Val) |
| TR_FT-189 | IT02U1414820 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Trp211ter) | p.(Gly4340Arg) |
| TR_FT-191 | IT02U1314641 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Glu767Serfs*21) | p.(Pro309Leu) |
| TR_FT-197 | IT02U0610141 | Bonnet et al, 2016 | ITALY | *USH2A* | c.1841-2A>G | p.(Leu3606Pro) |
| TR_FT-199 | IT02U2521551 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Cys1228ter) | p.(Thr3571Met) |
| TR_FT-200 | IT02U337021 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Phe1297Serfs*17) | p.(Thr3571Met) |
| TR_FT-203 | IT02U304071 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Trp3955ter) | p.(Glu4963Glyfs*38) |
| TR_FT-211 | IT02U2219552 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Tyr2566Leufs*11) | p.(Arg3119Cys) |
| TR_FT-212 | IT02U0710160 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Gln1573ter) | p.(Thr3571Met) |

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|---|---|---|---|---|---|---|
| TR_FT-213 | IT02U0410110 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Arg737ter) | p.(Ile285Thr) |
| TR_FT-214 | IT02U0310050 | Bonnet et al, 2016 | ITALY | *USH2A* | c.14791+4A>G | p.(Thr3571Met) |
| TR_FT-216 | IT02U293311 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Arg2509Glyfs*19) | p.(Arg1777Trp) |
| TR_FT-217 | IT02U272481 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Trp3955ter) | c.(3316+1_3317-1)_(4627+1_4628-1)del |
| TR_FT-219 | IT02U348231 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Phe1297Serfs*17) | p.(Thr3571Met) |
| TR_FT-220 | IT02U262321 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Ser2639Pro) | p.(Ser2639Pro) |
| TR_FT-223 | IT02U2319930 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Glu1492ter) | p.(Val2700Glu) |
| TR_FT-294 | IT02U1816640 + | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Ser2969ter) | p.(Ser2969ter) |
| TR_FT-295 | IT02U1816640 + | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Ser2969ter) | p.(Ser2969ter) |
| TR_FT-301 | IT02U2017672 + | Bonnet et al, 2016 | ITALY | *USH2A* | c.7452-1G>A | c.7452-1G>A |
| TR_FT-345 | IT02U1012180 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Thr3571Met) | p.(Thr352Ile) |
| TR_FT-418 | IT02U0110011 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Cys953ter) | p.(Trp3955ter) |
| TR_FT-419 | IT02U1615200 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Trp3955ter) | p.(Glu767Serfs*21) |
| TR_FT-426 | IT02U0810602 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Phe4993Profs*7) | p.(Phe4993Profs*7) |
| TR_FT-427 | IT02U0210041 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Asn330Lysfs*8) | p.(Thr3571Met) |
| TR_FT-428 | IT02U1816640 + | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Ser2969ter) | p.(Ser2969ter) |
| TR_FT-429 | IT02U1214361 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Tyr3715ter) | p.(Thr3571Met) |
| TR_FT-435 | IT02U2118741 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Glu2242ter) | p.(Arg303His) |
| TR_FT-436 | IT02U0510130 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Ser1350Leufs*16) | p.(Gly268Arg) |
| TR_FT-438 | IT02U31410 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Thr3571Met) | p.(Thr3571Met) |
| TR_FT-439 | IT02U091070 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Glu767Serfs*21) | p.(Pro2241Thr) |
| TR_FT-442 | IT02U282841 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Thr3571Met) | p.(Thr3571Met) |
| TR_FT-443 | IT02U1515091 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Phe4993Profs*7) | p.(Tyr90Cys) |
| TR_FT-445 | IT02U1715361 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Gln4750ter) | p.(Met3271Cysfs*30) |
| TR_FT-449 | IT02U2017672 + | Bonnet et al, 2016 | ITALY | *USH2A* | c.7452-1G>A | c.7452-1G>A |
| TR_FT-451 | IT02U32601 | Bonnet et al, 2016 | ITALY | *USH2A* | p.(Phe1297Serfs*17) | c.(4251+1_4252-1)_(4396+1_4397-1)del |
| TR_IN-024 | U6 | Bonnet et al, 2011 | FRANCE | *USH2A* | p.(Glu3562ter) | p.(Glu767Serfs*21) |
| TR_IN-025 | U21 | Bonnet et al, 2011 | FRANCE | *USH2A* | c.10586-1G>C | p.(Tyr1730Trpfs*6) |
| TR_IN-026 | U24 | Bonnet et al, 2011 | FRANCE | *USH2A* | p.(Ser1307ter) | p.(Cys536Arg) |
| TR_IN-027 | U30 | Bonnet et al, 2011 | FRANCE | *USH2A* | p.(Glu767Serfs*21) | p.(Arg303His) |
| TR_IN-028 | FR02U06392+ | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Glu767Serfs*21) | p.(Tyr4128Hisfs*24) |
| TR_IN-159 | FR02U113302 | Bonnet et al, 2011 | FRANCE | *USH2A* | p.(Tyr4031ter) | p.(Phe4697Leufs*2) |
| TR_IN-161 | FR02U06392+ | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Glu767Serfs*21) | p.(Tyr4128Hisfs*24) |
| TR_IN-162 | FR02U102062 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(His308Serfs*16) | c.(1644+1_1645-1)_(4627+1_4628-1)del |

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|---|---|---|---|---|---|---|
| TR_IN-163 | FR02U091752 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Arg4115Cys)/p.(Thr4425Met) | p.(Thr4439Ile) |
| TR_IN-164 | FR02U08872 | Bonnet et al, 2016 | FRANCE | *USH2A* | c.12067-2A>G | c.12067-2A>G |
| TR_IN-176 | FR02U4660482 | Bonnet et al, 2016 | FRANCE | *USH2A* | c.7595-2144A>G | c.7595-3Cys>G |
| TR_IN-177 | FR02U4256782 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Arg34ter) | p.(Arg3719His) |
| TR_IN-180 | FR02U4559922 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Glu767Serfs*21) | p.(Val218Glu) |
| TR_IN-185 | FR02U2225512 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Glu287ter) | p.(Asn346His) |
| TR_IN-186 | FR02U146642 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Gln919ter) | p.(Glu767Serfs*21) |
| TR_IN-209 | FR02U123922 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Arg4935ter) | p.(His308Serfs*16) |
| TR_IN-238 | FR02U07472+ | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Trp3955ter) | p.(Thr1443Pro) |
| TR_IN-239 | FR02U07472+ | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Trp3955ter) | p.(Thr1443Pro) |
| TR_IN-243 | FR02U1610392+ | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Glu284Aspfs*38) | c.7595-2144A>G |
| TR_IN-258 | FR02U159252 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(=,Tyr318Cysfs*17) | p.(Arg998Lys) |
| TR_IN-262 | FR02U2529982 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Pro5127Argfs*8) | p.(Pro5127Argfs*8) |
| TR_IN-264 | FR02U1610392+ | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Glu284Aspfs*38) | c.7595-2144A>G |
| TR_IN-268 | FR02U2119962 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Leu1317ter) | p.(Glu767Serfs*21) |
| TR_IN-273 | FR02U135262 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Glu767Serfs*21) | p.(Thr4809Ile) |
| TR_IN-274 | FR02U3953202 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Val218Glu) | p.(Gly660arg) |
| TR_IN-275 | FR02U2651202 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(His308Serfs*16) | p.(Asn346His) |
| TR_IN-276 | FR02U2934662+ | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Gly3264Valfs*37) | p.(Arg5143His) |
| TR_IN-281 | FR02U2934662+ | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Gly3264Valfs*37) | p.(Arg5143His) |
| TR_IN-282 | FR02U1815092 | Bonnet et al, 2016 | FRANCE | *USH2A* | c.12067-2A>G | p.(Pro4735Arg) |
| TR_IN-285 | FR02U3342772 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Gly1841Glu) | p.(Gly268Arg) |
| TR_IN-296 | FR02U3137132 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Glu2288ter) | p.(Thr4999Ile) |
| TR_IN-306 | FR02U75U1540 | Bonnet et al, 2016 | FRANCE | *USH2A* | c.(651+1_652-1)_(784+1_785-1)del | p.(Ser3005Thr) |
| TR_IN-316 | FR02U92U741 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Pro3116Hisfs*13) | p.(Thr4337Met) |
| TR_IN-317 | FR02U05242 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Ile1557Leufs*18) | c.3317-1G>A |
| TR_IN-320 | FR02U2730432 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Ser1247Lysfs*4) | c.9371+1G>T |
| TR_IN-321 | FR02U2327632 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Glu767Serfs*21) | c.3317-2A>G |
| TR_IN-322 | FR02U3750672 | Bonnet et al, 2016 | FRANCE | *USH2A* | c.13811+2T>G | p.(Pro3272Leu) |
| TR_IN-324 | FR02U2830812 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Gly4095Ser) | p.(Cys759Phe) |
| TR_IN-326 | FR02U76U1651 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Gln3959Asnfs*53) | p.(Ala3660fsVal*14) |
| TR_IN-327 | FR02U77U1680 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Lys811ter) | c.7595-2144A>G |
| TR_IN-328 | FR02U85U2531 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Leu4840Pro) | p.(Leu4840Pro) |

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|---|---|---|---|---|---|---|
| TR_IN-330 | FR02U82U2210 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Asn4079Trpfs*19) | p.(Glu511Lys) |
| TR_IN-331 | FR02U72S57220 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Trp3955ter) | p.(Pro1843Leu) |
| TR_IN-332 | FR02U3646012 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Cys759Phe) | p.(Leu4282Pro) |
| TR_IN-336 | FR02U0163292 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Glu767Serfs*21) | p.(Glu767Serfs*21) |
| TR_IN-344 | FR02U5163062 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Glu767Serfs*21) | p.(Thr4337Met) |
| TR_IN-348 | FR02U3443182 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Glu767Serfs*21) | p.(Ala1711Serfs*6) |
| TR_IN-349 | FR02U71S27881 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Glu767Serfs*21) | c.(6163+1_6164-1)_(6325+1_6326-1)del |
| TR_IN-350 | FR02U5061812 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Thr4337Met) | p.(Cys717Gly) |
| TR_IN-351 | FR02U91U720 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Gln4635ter) | p.(Ile4059Asnfs*40) |
| TR_IN-357 | FR02U3241292 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Trp3955ter) | p.(Arg2509Glyfs*19) |
| TR_IN-359 | FR02U081112 | Bonnet et al, 2016 | FRANCE | *USH2A* | c.785-6636_1840+208del | c.785-6636_1840+208del |
| TR_IN-365 | FR02U93U920 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Val2714ter) | c.7595-2144A>G |
| TR_IN-366 | FR02U81U2181 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Pro3116Hisfs*13) | p.(Pro3116Hisfs*13) |
| TR_IN-367 | FR02U94U991 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Glu767Serfs*21) | p.(Ser3856Valfs*28) |
| TR_IN-368 | FR01U03A1430 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Leu4282Pro) | p.(Leu4282Pro) |
| TR_IN-372 | FR02U83U2220 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Glu767Serfs*21) | p.(Leu2319Argfs*7) |
| TR_IN-373 | FR02U3036172 | Bonnet et al, 2016 | FRANCE | *USH2A* | c.11548+2T>G | p.(Glu767Serfs*21) |
| TR_IN-377 | FR02U1915842 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Glu767Serfs*21) | p.(Val218Glu) |
| TR_IN-401 | FR02U2429002 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Glu767Serfs*21) | p.(Gly4095Ser) |
| TR_IN-402 | FR02U3545182 | Bonnet et al, 2016 | FRANCE | *USH2A* | p.(Arg1653ter) | p.(Cys5122arg) |
| TR_UE-136 | SLO01U0411442 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Trp3955ter) |
| TR_UE-140 | SLO01U1411072 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | c.4627+25435_4987+660del | c.4627+25435_4987+660del |
| TR_UE-145 | SLO01U2811582 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | c.(784+1_785-1)_(848+1_849-1)del | p.(Gly4857Ala) |
| TR_UE-152 | SLO01U1110012 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Cys870ter) | p.(Trp3955ter) |
| TR_UE-171 | SLO01U1211052 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Gly1645ter) |
| TR_UE-172 | SLO01U2111392 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Gly1645ter) |
| TR_UE-173 | SLO01U1311062 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Phe78Val) |
| TR_UE-174 | SLO01U15111 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Cys870ter) | p.(Cys870ter) |

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| TR_UE-225 | SLO01U10STG02062 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Trp3955ter) |
| TR_UE-227 | SLO01U1611132 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Thr352Ile) |
| TR_UE-228 | SLO01U0111142 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Trp3955ter) |
| TR_UE-233 | SLO01U1711192 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Thr3676Ile) |
| TR_UE-234 | SLO01U1811212 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Gly1645ter) |
| TR_UE-235 | N/A | | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Cys870ter) |
| TR_UE-289 | SLO01U2211422 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Arg303His) |
| TR_UE-292 | SLO01U2011362 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Arg2914ter) |
| TR_UE-381 | SLO01U0211322 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Trp3955ter) |
| TR_UE-382 | SLO01U0311332 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Trp3955ter) |
| TR_UE-384 | SLO01U1911352 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Gly4763arg) |
| TR_UE-385 | N/A | | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Trp3955ter) |
| TR_UE-386 | SLO01U2311432 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Gln3292ter) |
| TR_UE-387 | SLO01U2411452 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Arg303His) |
| TR_UE-388 | N/A | | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Phe78Val) |
| TR_UE-389 | SLO01U2511542 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Cys870ter) | p.(Thr4315Pro) |
| TR_UE-391 | SLO01U2611562 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Cys870ter) |
| TR_UE-392 | SLO01U2711572 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Arg1549ter) |
| TR_UE-395 | SLO01U2912142 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Arg626ter) |
| TR_UE-396 | SLO01U3012252 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Arg626ter) |
| TR_UE-397 | SLO01U3112272 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Cys870ter) | p.(Cys870ter) |
| TR_UE-399 | N/A | | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Arg626ter) |
| TR_UE-403 | SLO01U05123 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Trp3955ter) |

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| | 62 | | | | | |
| TR_UE-404 | SLO01U06123 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Trp3955ter) |
| | 72 | | | | | |
| TR_UE-405 | N/A | | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Trp3955ter) |
| TR_UE-406 | N/A | | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Trp3955ter) |
| TR_UE-407 | N/A | | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Trp3955ter) |
| TR_UE-408 | SLO01U35126 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Cys870ter) | p.(Cys870ter) |
| | 72 | | | | | |
| TR_UE-409 | SLO01U32125 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Arg626ter) | p.(Phe78Val) |
| | 12 | | | | | |
| TR_UE-411 | SLO01U07125 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Trp3955ter) |
| | 62 | | | | | |
| TR_UE-412 | SLO01U33125 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Pro1243Leufs*6) |
| | 82 | | | | | |
| TR_UE-413 | SLO01U36127 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Cys870ter) | p.(Arg303His) |
| | 22 | | | | | |
| TR_UE-414 | N/A | | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Pro1243Leufs*6) |
| TR_UE-415 | SLO01U34126 | Bonnet et al, 2016 | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Pro2241His) |
| | 12 | | | | | |
| TR_UE-416 | N/A | | SLOVENIA | *USH2A* | p.(Trp3955ter) | p.(Trp3955ter) |
| TR_EK-047 | DE03G03480+ | Bonnet et al, 2016 | GERMANY | *ADGRV 1* | p.(Gln753Leufs*8) | p.(Lys1786Ilefs*8) |
| TR_EK-048 | DE03G03480+ | Bonnet et al, 2016 | GERMANY | *ADGRV 1* | p.(Gln753Leufs*8) | p.(Lys1786Ilefs*8) |
| TR_EK-055 | DE03G04551 | Bonnet et al, 2016 | GERMANY | *ADGRV 1* | p.(Tyr536ter) | p.(Tyr536ter) |
| TR_EK-061 | DE03G05611 | Bonnet et al, 2016 | GERMANY | *ADGRV 1* | p.(Phe5328Serfs*41) | p.(Glu2669Lysfs*4) |
| TR_EK-087 | DE03G06870 | Bonnet et al, 2016 | GERMANY | *ADGRV 1* | p.(Trp3486ter) | p.(Glu6295Alafs*31) |
| TR_EK-121 | DE03G011211 | Bonnet et al, 2016 | GERMANY | *ADGRV 1* | p.(Arg3293ter) | p.(Ser1358Pro) |
| | + | | | | | |
| TR_EK-126 | DE03G011211 | Bonnet et al, 2016 | GERMANY | *ADGRV 1* | p.(Arg3293ter) | p.(Ser1358Pro) |
| | + | | | | | |
| TR_EK-175 | DE03G021752 | Bonnet et al, 2016 | GERMANY | *ADGRV 1* | p.(Gly2045ter) | c.10054-1G>T |
| TR_EK-260 | DE03G072602 | Bonnet et al, 2016 | GERMANY | *ADGRV 1* | p.(Arg920ter) | c.(4752+1_4753-1)_(16196+1_16197-1)del |
| TR_FT-205 | IT02G012020 | Bonnet et al, 2016 | ITALY | *ADGRV 1* | p.(Leu568Cysfs*8) | p.(Cys5970Ser) |
| TR_IN-158 | P0473 | Bonnet et al, 2011 | FRANCE | *ADGRV 1* | p.(Pro522Serfs*8) | p.(Met5890Valfs*10) |

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| TR_IN-166 | FR02G028272 | Bonnet et al, 2016 | FRANCE | *ADGRV1* | p.(Arg493ter) | c.1509+3A>G |
| TR_IN-167 | FR02G0315122 | Bonnet et al, 2016 | FRANCE | *ADGRV1* | p.(Gln5626ter) | p.(Gln5626ter) |
| TR_IN-168 | FR02G0746112 | Bonnet et al, 2016 | FRANCE | *ADGRV1* | p.(Ser4048ter) | p.(Asp3992Asn) |
| TR_IN-280 | FR02G0534762 | Bonnet et al, 2016 | FRANCE | *ADGRV1* | p.(Gln3490ter) | p.(Gln3490ter) |
| TR_IN-303 | FR02G0636522 | Bonnet et al, 2016 | FRANCE | *ADGRV1* | p.(Ala2032Argfs*27) | c.9184+3A>G |
| TR_IN-340 | FR02G0850662 | Bonnet et al, 2016 | FRANCE | *ADGRV1* | p.(Ser4441Leufs*9) | p.(Val5647Glyfs*7) |
| TR_IN-356 | FR02G011822 | Bonnet et al, 2016 | FRANCE | *ADGRV1* | p.(Gly2329Argfs*17) | c.9906+1G>A |

The data of majority of the patients has been previously reported, as a part of genetic report on Treatrush study - see references. + symbol denotes siblings. Phenotypes of TR_EK-047 and TR_EK-048 have been described previously (Ebermann et al, 2009).