**Table S2. Genotype-phenotype correlation and allelic hierarchy model classification in USH2A-related disease.**

| **Study** | **Diagnosis** | **Allele 1** | **Allele 2** | **Allele 1** | **Allele 2** | **Total of cases** | **Model Y/N** |
| --- | --- | --- | --- | --- | --- | --- | --- |
| Carss et al 2017 | Classic USH2 | c.3831\_3834delACTAinsG, p.Leu1278del | c.920\_923dupGCCA, p.His308GlnfsTer16 | Unknown | non-specific | 1 | Y |
| Carss et al 2017 | Classic USH2 | c.2299delG, p.Glu767SerfsTer21 | c.2299delG, p.Glu767SerfsTer21 | non-specific | non-specific | 1 | Y |
| Carss et al 2017 | Classic USH2 | c.12819T>A, p.Tyr4273Ter | c.1055C>T, p.Thr352Ile | Unknown | non-specific | 1 | Y |
| Carss et al 2017 | Classic USH2 | c.12954C>A, p.Tyr4318Ter | c.5603T>G, p.Phe1868Cys | non-specific | Unknown | 1 | Y |
| Carss et al 2017 | Classic USH2 | c.2299delG, p.Glu767SerfsTer21 | c.2081G>A, p.Cys694Tyr | non-specific | Unknown | 1 | Y |
| Carss et al 2017 | Classic USH2 | c.14426C>T, p.Thr4809Ile | c.2299delG, p.Glu767SerfsTer21 | non-specific | non-specific | 1 | Y |
| Carss et al 2017 | Classic USH2 | c.7595-3C>G, p.? | c.2299delG, p.Glu767SerfsTer21 | non-specific | non-specific | 1 | Y |
| Carss et al 2017 | Classic USH2 | c.10561T>C, p.Trp3521Arg | c.7595-2144A>G, p.? | non-specific | non-specific | 1 | Y |
| Carss et al 2017 | Classic USH2 | c.12309delC, p.Phe4103LeufsTer11 | c.12309delC, p.Phe4103LeufsTer11 | Unknown | Unknown | 1 | Y |
| Carss et al 2017 | Classic USH2 | c.2299delG, p.Glu767SerfsTer21 | Deletion 1:216240159-222780953 | non-specific | Unknown | 1 | Y |
| Carss et al 2017 | Classic USH2 | c.2299delG, p.Glu767SerfsTer21 | c.820C>G, p.Arg274Gly | non-specific | non-specific | 1 | Y |
| Carss et al 2017 | Classic USH2 | c.3395G>A,  p.Gly1132Asp | c.2994A>T, p.Arg998Ser;  c.895delC, p.Gln299AsnfsTer37 | non-specific | Unknown | 1 | Y |
| Carss et al 2017 | Classic USH2 | c.11694delC, p.Asn3899ThrfsTer34 | c.3158-6A>G, p.? | Unknown | Unknown | 1 | Y |
| Carss et al 2017 | Classic USH2 | c.9860\_9873delATGATGGCCATGGC,  p.His3287ProfsTer54 | c.9860\_9873delATGATGGCCATGGC,  p.His3287ProfsTer54 | Unknown | Unknown | 1 | Y |
| Carss et al 2017 | nsRP | c.6050-1G>A, p.? | c.2299delG, p.Glu767SerfsTer21 | Unknown | non-specific | 1 | Y |
| Carss et al 2017 | nsRP | c.7334C>T, p.Ser2445Phe | c.3902G>T, p.Gly1301Val | Unknown | nsRP-enriched | 1 | Y |
| Carss et al 2017 | nsRP | c.9882C>G, p.Cys3294Trp | c.653T>A, p.Val218Glu | nsRP-enriched | non-specific | 1 | Y |
| Carss et al 2017 | nsRP | c.10342G>A, p.Glu3448Lys | c.6670G>T, p.Gly2224Cys | nsRP-enriched | unknown | 1 | Y |
| Carss et al 2017 | nsRP | c.10073G>A, p.Cys3358Tyr | c.920\_923dupGCCA, p.His308GlnfsTer16 | nsRP-enriched | non-specific | 1 | Y |
| Carss et al 2017 | nsRP | c.10073G>A, p.Cys3358Tyr | c.2299delG, p.Glu767SerfsTer21 | nsRP-enriched | non-specific | 1 | Y |
| Carss et al 2017 | nsRP | c.13576C>T, p.Arg4526Ter | c.4222C>T, p.Gln1408Ter | unknown | unknown | 1 | Y |
| Carss et al 2017 | nsRP | c.13335\_13347delGAACATGGACTCTinsCTTG,  p.Glu4445\_Ser4449delinsAspLeu | c.11864G>A, p.Trp3955Ter | nsRP-enriched | non-specific | 1 | Y |
| Carss et al 2017 | nsRP | c.12575G>A, p.Arg4192His | c.10073G>A, p.Cys3358Tyr | nsRP-enriched | nsRP-enriched | 1 | Y |
| Carss et al 2017 | nsRP | c.5012G>A, p.Gly1671Asp | c.5012G>A, p.Gly1671Asp | unknown | unknown | 1 | Y |
| Carss et al 2017 | nsRP | c.6446C>A, p.Pro2149Gln | c.2276G>T, p.Cys759Phe | unknown | nsRP-enriched | 1 | Y |
| Carss et al 2017 | nsRP | c.4510dupA, p.Arg1504LysfsTer26 | c.2276G>T, p.Cys759Phe | unknown | nsRP-enriched | 1 | Y |
| Carss et al 2017 | nsRP | c.7358T>A, p.Val2453Asp | c.4027A>C, p.Asn1343His | unknown | unknown | 1 | Y |
| Carss et al 2017 | nsRP | c.10073G>A, p.Cys3358Tyr | c.2276G>T, p.Cys759Phe | nsRP-enriched | nsRP-enriched | 1 | Y |
| Carss et al 2017 | nsRP | c.13335\_13347delGAACATGGACTCTinsCTTG,  p.Glu4445\_Ser4449delinsAspLeu | Deletion 1:216259365-216318209 | nsRP-enriched | unknown | 1 | Y |
| Carss et al 2017 | nsRP | c.10073G>A, p.Cys3358Tyr | c.2276G>T, p.Cys759Phe | nsRP-enriched | nsRP-enriched | 1 | Y |
| Carss et al 2017 | nsRP | c.12145G>A, p.Ala4049Thr | c.9785G>T, p.Gly3262Val | unknown | unknown | 1 | Y |
| Carss et al 2017 | nsRP | c.5012G>A, p.Gly1671Asp | c.5012G>A, p.Gly1671Asp | unknown | unknown | 1 | Y |
| Carss et al 2017 | nsRP | c.2276G>T, p.Cys759Phe | c.2276G>T, p.Cys759Phe | nsRP-enriched | nsRP-enriched | 1 | Y |
| Carss et al 2017 | nsRP | c.13126T>G, p.Trp4376Gly | c.12874A>G, p.Asn4292Asp | nsRP-enriched | nsRP-enriched | 1 | Y |
| Carss et al 2017 | nsRP | c.2802T>G, p.Cys934Trp | Deletion 1:216009683-216011948 | non-specific | unknown | 1 | Y |
| Carss et al 2017 | nsRP | c.13396C>T, p.Pro4466Ser ;  c.13396C>T, p.Pro4466Ser | Deletion 1:215836170-215851932 | Unknown | unknown | 1 | Y |
| Carss et al 2017 | nsRP | c.13331C>T, p.Pro4444Leu | c.8223+1G>C, NA | nsRP-enriched | Unknown | 1 | Y |
| Carss et al 2017 | nsRP | c.2299delG, p.Glu767SerfsTer21 | c.2276G>T, p.Cys759Phe | non-specific | nsRP-enriched | 1 | Y |
| Carss et al 2017 | nsRP | c.9571-2A>G, p.? | c.2299delG, p.Glu767SerfsTer21 | nsRP-enriched | non-specific | 1 | Y |
| Carss et al 2017 | nsRP | c.13331C>T, p.Pro4444Leu | c.13126T>G, p.Trp4376Gly | nsRP-enriched | nsRP-enriched | 1 | Y |
| Carss et al 2017 | nsRP | c.11507C>T, p.Pro3836Leu | c.9571-2A>G, p.? | unknown | nsRP-enriched | 1 | Y |
| Carss et al 2017 | Classic USH2 | c.2299delG, p.Glu767SerfsTer21 | c.2276G>T, p.Cys759Phe | non-specific | nsRP-enriched | 1 | N |
| Carss et al 2017 | nsRP | c.13274C>T, p.Thr4425Met | c.8981G>A, p.Trp2994Ter | non-specific | non-specific | 1 | N |
| Carss et al 2017 | nsRP | c.13316C>T, p.Thr4439Ile | c.12574C>T, p.Arg4192Cys | non-specific | non-specific | 1 | N |
| Carss et al 2017 | nsRP | c.11156G>A, p.Arg3719His | c.920\_923dupGCCA, p.His308GlnfsTer16 | non-specific | non-specific | 1 | N |
| Carss et al 2017 | nsRP | c.12505A>G, p.Thr4169Ala | c.12505A>G, p.Thr4169Ala | non-specific | non-specific | 1 | N |
| Carss et al 2017 | nsRP | c.926C>T, p.Pro309Leu | c.100C>T, p.Arg34Ter | non-specific | non-specific | 1 | N |
| Carss et al 2017 | nsRP | c.15433G>A, p.Val5145Ile | c.2522C>A, p.Ser841Tyr | non-specific | non-specific | 1 | N |
| Carss et al 2017 | nsRP | c.13274C>T, p.Thr4425Met | c.11875\_11876delCA, p.Gln3959AsnfsTer53 | non-specific | non-specific | 1 | N |
| Pierrache et al 2016 | Classic USH2 | c.187C>T, p.(Arg63\*) | c.949C>A, p.[(Tyr318CysfsX17)] | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.486-14G>A, p.? | c.12729G>A, p.(Trp4243\*) | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.653T>A, p.(Val218Glu) | c.6967C>T \*, p.(Arg2323\*) | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.785-?\_+5572+?dup deletion exon 5-27 | c.1256G>T, p.(Cys419Phe) | unknown | non-specific | 2 | Y |
| Pierrache et al 2016 | nsRP | c.917\_918insGCTGp.(Ser307Leufs\*17) | c.11007C>A, p.(Ser3669Arg) | unknown | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.920\_923dup, p.(His308fs) | c.7950dup, p.(Asn2651Glnfs\*10) | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.949C>A, p.[(Tyr318CysfsX17)] | c.4773del, p.(Val1592\*) | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.949C>A, p.[(Tyr318CysfsX17)] | c.11676del, p.Lys3892Asnfs\*41 | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.949C>A, p.[(=,Tyr318CysfsX17)] | c.11864G>A, p.(Trp3955\*) | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.1036A>C, p.(Asn346His) | c.4810G>A, p.(Asp1604Asn) | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.1036A>C, p.(Asn346His) | c.7301-?\_10939+?39-55indel | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.1036A>C, p.(Asn346His) | c.10525A>T \*\*, p.(Lys3509\*) | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | nsRP | c.1036A>C, p.(Asn346His) | c.14545T>C, p.(Trp4849Arg) | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.1039G>C, p.(Asp347His) | c.1039G>C, p.(Asp347His) | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.1227G>A, p.(Trp409\*) | c.1227G>A, p.(Trp409\*) | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.1227G>A, p.(Trp409\*) | c.1256G>T, p.(Cys419Phe) | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.1227G>A, p.(Trp409\*) | c.11875C>T, p.(Gln3959\*) | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | nsRP | c.1227G>A, p.(Trp409\*) | c.12575G>A, p.(Arg4192His) | non-specific | nsRP-enriched | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.1227G>A, p.(Trp409\*) | c.13262T>C, p.(Leu4421Pro) | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.1256G>T, p.(Cys419Phe) | c.1606T>C, p.(Cys536Arg) | non-specific | non-specific | 5 | Y |
| Pierrache et al 2016 | Classic USH2 | c.1256G>T, p.(Cys419Phe) | c.4056G>A, p.(Trp1352\*) | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | nsRP | c.1256G>T, p.(Cys419Phe) | c.4106C>T, p.(Ser1369Leu) | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.1256G>T, p.(Cys419Phe) | c.7121-8313\_11048-962delins12 | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.1256G>T, p.(Cys419Phe) | c.8079G>A, p.Trp2693X | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.1256G>T, p.(Cys419Phe) | c.10387+2T>C | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.1256G>T, p.(Cys419Phe) | c.10525A>T, p.(Lys3509\*) | non-specific | non-specific | 4 | Y |
| Pierrache et al 2016 | nsRP | c.1256G>T, p.(Cys419Phe) | c.10901A>C, p.(His3634Pro) | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.1256G>T, p.(Cys419Phe) | c.11864G>A, p.(Trp3955\*) | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.1256G>T, p.(Cys419Phe) | c.14545T>C, p.(Trp4849Arg) | non-specific | unknown | 2 | Y |
| Pierrache et al 2016 | Classic USH2 | c.1328+1G>A, p.? | c.2140C>T, p.(Gln714\*) | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.1606T>C, p.(Cys536Arg) | c.1606T>C, p.(Cys536Arg) | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.1606T>C, p.(Cys536Arg) | c.6658-2A>G, p.? | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.1606T>C, p.(Cys536Arg) | c.10525A>T, p.(Lys3509\*) | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.1606T>C, p.(Cys536Arg) | c.14131C>T, p.(Gln4711\*) | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.1808G>A, p.(Gly603Glu) | c.7501C>T, p.(Gln2501\*) | unknown | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.1876C>T, p.(Arg626\*) | c.1876C>T, p.(Arg626\*) | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | nsRP | c.1965T>G, p.(Cys655Thr) | c.6926G>T, p.(Cys2309Phe) | unknown | non-specific | 1 | Y |
| Pierrache et al 2016 | nsRP | c.1978G>A, p.(Gly660Arg) | c.9258G>T, p.Gln3086His | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.2014C>T, p.(Gln672\*) | c.2014C>T, p.(Gln672\*) | unknown | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.2242C>T, p.(Gln748\*) | c.4405C>T, p.(Gln1469\*) | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | nsRP | c.2276G>T, p.(Cys759Phe) | c.486-14G>A, p.? | nsRP-enriched | non-specific | 4 | Y |
| Pierrache et al 2016 | nsRP | c.2276G>T, p.(Cys759Phe) | c.949C>A, p.[(Tyr318CysfsX17)] | nsRP-enriched | non-specific | 1 | Y |
| Pierrache et al 2016 | nsRP | c.2276G>T, p.(Cys759Phe) | c.1256G>T, p.(Cys419Phe) | nsRP-enriched | non-specific | 1 | Y |
| Pierrache et al 2016 | nsRP | c.2276G>T, p.(Cys759Phe) | c.2276G>T, p.(Cys759Phe) | nsRP-enriched | nsRP-enriched | 2 | Y |
| Pierrache et al 2016 | nsRP | c.2276G>T, p.(Cys759Phe) | c.2299del, p.(Glu767Serfs\*21) | nsRP-enriched | non-specific | 6 | Y |
| Pierrache et al 2016 | nsRP | c.2276G>T, p.(Cys759Phe) | c.5516T>A, p.(Val1839Glu) | nsRP-enriched | unknown | 1 | Y |
| Pierrache et al 2016 | nsRP | c.2276G>T, p.(Cys759Phe) | c.5576T>G, p.(Phe1859Cys) | nsRP-enriched | unknown | 1 | Y |
| Pierrache et al 2016 | nsRP | c.2276G>T, p.(Cys759Phe) | c.7121-?\_11047+?, p.? | nsRP-enriched | unknown | 1 | Y |
| Pierrache et al 2016 | nsRP | c.2276G>T, p.(Cys759Phe) | c.7950dup, p.(Asn2651Glnfs\*10) | nsRP-enriched | non-specific | 1 | Y |
| Pierrache et al 2016 | nsRP | c.2276G>T, p.(Cys759Phe) | c.8723\_8724del, p.(Val2908Glyfs\*29) | nsRP-enriched | unknown | 1 | Y |
| Pierrache et al 2016 | nsRP | c.2276G>T, p.(Cys759Phe) | c.10073G>A, p.(Cys3358Tyr) | nsRP-enriched | nsRP-enriched | 1 | Y |
| Pierrache et al 2016 | nsRP | c.2276G>T, p.(Cys759Phe) | c.10525A>T, p.(Lys3509\*) | nsRP-enriched | non-specific | 1 | Y |
| Pierrache et al 2016 | nsRP | c.2276G>T, p.(Cys759Phe) | c.10689T>A, p.(Tyr3563\*) | nsRP-enriched | unknown | 1 | Y |
| Pierrache et al 2016 | nsRP | c.2276G>T, p.(Cys759Phe) | c.14174G>A \*, p.(Trp4725\*) | nsRP-enriched | unknown | 1 | Y |
| Pierrache et al 2016 | nsRP | c.2276G>T, p.(Cys759Phe) | c.14803C>T, p.(Arg4935\*) | nsRP-enriched | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.2299del, p.(Glu767Serfs\*21) | c.920\_923dup, p.(His308fs) | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.2299del, p.(Glu767Serfs\*21) | c.1036A>C, p.(Asn346His) | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.2299del, p.(Glu767Serfs\*21) | c.1039G>C, p.(Asp347His) | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.2299del, p.(Glu767Serfs\*21) | c.1227G>A, p.(Trp409\*) | non-specific | non-specific | 2 | Y |
| Pierrache et al 2016 | Classic USH2 | c.2299del, p.(Glu767Serfs\*21) | c.1256G>T, p.(Cys419Phe) | non-specific | non-specific | 5 | Y |
| Pierrache et al 2016 | Classic USH2 | c.2299del, p.(Glu767Serfs\*21) | c.1606T>C, p.(Cys536Arg) | non-specific | non-specific | 4 | Y |
| Pierrache et al 2016 | Classic USH2 | c.2299del, p.(Glu767Serfs\*21) | c.2168-1G>C, p.(Leu724Valfs\*31) | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.2299del, p.(Glu767Serfs\*21) | c.2299del, p.(Glu767Serfs\*21) | non-specific | non-specific | 15 | Y |
| Pierrache et al 2016 | Classic USH2 | c.2299del, p.(Glu767Serfs\*21) | c.4732C>T, p.(Arg1578Cys) | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.2299del, p.(Glu767Serfs\*21) | c.4957C>T, p.(Arg1653\*) | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.2299del, p.(Glu767Serfs\*21) | c.5516T>A, p.(Val1839Glu) | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.2299del, p.(Glu767Serfs\*21) | c.6722C>T, p.(Pro2241Leu) | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.2299del, p.(Glu767Serfs\*21) | c.7121-8313\_11048-962delins12 | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.2299del, p.(Glu767Serfs\*21) | c.7187G>A, p.(Trp2396\*) | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.2299del, p.(Glu767Serfs\*21) | c.9815C>T, p.(Pro3272Leu) | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.2299del, p.(Glu767Serfs\*21) | c.10525A>T, p.(Lys3509\*) | non-specific | non-specific | 2 | Y |
| Pierrache et al 2016 | Classic USH2 | c.2299del, p.(Glu767Serfs\*21) | c.13316C>T, p.(Thr4439lle) | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.2299del, p.(Glu767Serfs\*21) | c.13508\_13523delinsAGp.(Val4503Glufs\*54) | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.2299del, p.(Glu767Serfs\*21) | c.15053-2A>T \*, p.? | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.2299del, p.(Glu767Serfs\*21) | deletion exon 12-13 | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.2391\_2392del, p.(Cys797X) | c.7595-2144A>G, p.(Lys2532Thrfs\*56) | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.2555-1G>C, p.? | c.2710\_2720dup, p.(Leu908Profs\*63) | unknown | unknown | 1 | Y |
| Pierrache et al 2016 | nsRP | c.3045C>G, p.(His1015Gln) | c.12394del, p.(Leu4132Trpfs\*35) | unknown | unknown | 1 | Y |
| Pierrache et al 2016 | nsRP | c.3368A>G, p.(Tyr1123Cys) | c.7054C>T, p.(Pro2352Ser) | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | nsRP | c.4362\_4367delinsACTC | c.13274C>T, p.(Thre4425Met) | unknown | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.4645C>T, p.(Arg1549\*) | c.4645C>T, p.(Arg1549\*) | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.6544\_6548dup, p.(His2183fs) | c.14289del, p.(Ile4764Serfs\*42) | unknown | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.6722C>T, p.(Pro2241Leu) | c.13316C>T, p.(Thr4439lle) | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.6722C>T, p.(Pro2241Leu) | c.7121-?\_11074+?p? | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.7121-8313\_11048-962delins12 | c.7121-8313\_11048-962delins12 | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.7121-8313\_11048-962delins12 | c.8954delG, p.(Gly2985Alafs\*3) | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | nsRP | c.7853G>A, p.(Trp2618\*) | c.9258G>T, p.? [p.Gln3086His] | unknown | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.7931G>A, p.(Trp2644\*) | c.11819A>C, p.(Tyr3940Ser) | unknown | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.8079G>A, p.(Trp2876\*) | c.12806C>A, p.(Pro4269His) | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.8628G>A, p.(Trp2876\*) | c.14977\_14978del, p.(Phe4993Profs\*7) | unknown | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.8723\_8724del, p.(Val2908Glyfs\*29) | c.10525A>T, p.(Lys3509\*) | unknown | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.9372-?\_9570+? | c.9372-?\_9570+? | unknown | unknown | 1 | Y |
| Pierrache et al 2016 | nsRP | c.9433C>T, p.(Leu3145Phe) | c.9815C>T, p.(Pro3272Leu) | unknown | non-specific | 1 | Y |
| Pierrache et al 2016 | nsRP | c.10073G>A, p.(Cys3358Tyr) | c.13274C>T, c.(Thr4425Met) | nsRP-enriched | non-specific | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.10561T>C, p.(Trp3521Arg) | c.14802C>G, p.(Tyr4934\*) | non-specific | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.12992A>G, p.(Tyr4331Cys) | c.12992A>G, p.(Tyr4331Cys) | unknown | unknown | 1 | Y |
| Pierrache et al 2016 | Classic USH2 | c.14803C>T, p.(Arg4935\*) | c.15089C>A, p.(Ser5030\*) | non-specific | non-specific | 1 | Y |
| Pierrache et al 2016 | nsRP | c.1256G>T, p.(Cys419Phe) | c.13274C>T, c.(Thr4425Met) | non-specific | non-specific | 1 | N |
| Pierrache et al 2016 | Classic USH2 | c.2276G>T, p.(Cys759Phe) | c.1206G>T, p.(Lys402Asn) | nsRP-enriched | unknown | 1 | N |
| Pierrache et al 2016 | Classic USH2 | c.2276G>T, p.(Cys759Phe) | c.1256G>T, p.(Cys419Phe) | nsRP-enriched | non-specific | 2 | N |
| Pierrache et al 2016 | Classic USH2 | c.2276G>T, p.(Cys759Phe) | c.7132\_7133del, p.(Tyr2378Hisfs\*39) | nsRP-enriched | unknown | 1 | N |
| Pierrache et al 2016 | Classic USH2 | c.2276G>T, p.(Cys759Phe) | c.14803C>T, p.(Arg4935\*) | nsRP-enriched | non-specific | 1 | N |
| Pierrache et al 2016 | nsRP | c.2296T>C, p.(Cys766Arg) | c.4732C>T, p.(Arg1578Cys) | non-specific | non-specific | 1 | N |
| Pierrache et al 2016 | nsRP | c.2299del, p.(Glu767Serfs\*21) | c.4133T>C, p.(Leu1378Pro) | non-specific | non-specific | 1 | N |
| Pierrache et al 2016 | nsRP | c.2299del, p.(Glu767Serfs\*21) | c.15433G>A, p.(Val5145Ile) | non-specific | non-specific | 1 | N |
| Pierrache et al 2016 | nsRP | c.10525A>T, p.(Lys3509\*) | c.13274C>T, c.(Thr4425Met) | non-specific | non-specific | 1 | N |
| This study (MCGM) | nsRP | c.10073G>A p.(Cys3358Tyr) | c.1111\_1112delATp.(Ile371Phefs\*) | nsRP-enriched | unknown | 1 | Y |
| This study (MCGM) | nsRP | c.10342G>Ap.(Glu3448Lys) | c.2276G>Tp.(Cys759Phe) | nsRP-enriched | nsRP-enriched | 1 | Y |
| This study (MCGM) | nsRP | c.13441A>G p.(Arg4481Gly) | c.7595-3C>G, p.(Pro2533Asnfs\*5) | Unknown (novel) | non-specific | 1 | Y |
| This study (MCGM) | nsRP | c.2276G>T p.(Cys759Phe) | c.13283G>A p.(Gly4428Asp) | nsRP-enriched | Unknown (novel) | 1 | Y |
| This study (MCGM) | nsRP | c.14791+2T>A p.? | c.2276G>T p.(Cys759Phe) | Unknown (novel) | nsRP-enriched | 1 | Y |
| This study (MCGM) | Classic USH2 | c.1558delTp.(Cys520Alafs\*71) | c.1558delTp.(Cys520Alafs\*71) | unknown | unknown | 1 | Y |
| This study (MCGM) | Classic USH2 | c.1859G>T p.(Cys620Phe) | c.2139C>T, p.(Gly713Gly) | Unknown (novel) | unknown | 1 | Y |
| This study (MCGM) | nsRP | c.2276G>Tp.(Cys759Phe) | c.2299delG, p.(Glu767Serfs\*21) | nsRP-enriched | non-specific | 1 | Y |
| This study (MCGM) | Classic USH2 | c.2299delG p.(Glu767Serfs\*21) | c.4321G>T (p.Glu1441X) | non-specific | unknown | 1 | Y |
| This study (MCGM) | Classic USH2 | c.4474G>T p.(Glu1492\*) | c.5614delins12 p.(Ala1872Leufs\*64) | non-specific | unknown | 1 | Y |
| This study (MCGM) | Classic USH2 | c.4645C>Tp.(Arg1549\*) | c.3158-2A>G p.? | non-specific | unknown | 1 | Y |
| This study (MCGM) | nsRP | c.9413G>A, p.(Gly3138Asp) | c.1804G>A, p.(Gly602Arg) | unknown | Unknown (novel) | 1 | Y |
| This study (MCGM) | nsRP | c.4645C>T p.(Arg1549\*) | c.4106C>T, p.(Ser1369Leu) | non-specific | unknown | 1 | Y |
| This study (MCGM) | nsRP | c.10342G>A p.(Glu3448Lys) | c.10342G>A, p.(Glu3448Lys) | nsRP-enriched | nsRP-enriched | 1 | Y |
| This study (MCGM) | nsRP | c.2276G>T p.(Cys759Phe) | c.9371+1G>C p.? | nsRP-enriched | non-specific | 1 | Y |
| This study (MCGM) | Classic USH2 | c.6862G>T p.(Glu2288\*) | c.3407G>A p.(Ser1136Asn) | non-specific | non-specific | 1 | Y |
| This study (MCGM) | nsRP | c.2276G>T p.(Cys759Phe) | c.12574C>T p.(Arg4192Cys) | nsRP-enriched | non-specific | 1 | Y |
| This study (MCGM) | nsRP | c.2276G>T p.(Cys759Phe) | c.13331delC p.(Pro4444Glnfs\*17) | nsRP-enriched | Unknown (novel) | 1 | Y |
| This study (MCGM) | Classic USH2 | c.5776+1G>A p.? | exon57to60 3 copiesp.? | non-specific | unknown | 1 | Y |
| This study (MCGM) | nsRP | c.9974G>A p.(Gly3325Glu) | exon10to14 del hetp.(Cys549Metfs\*5) | Unknown (novel) | unknown | 1 | Y |
| This study (MCGM) | nsRP | c.3485C>Ap.(Ser1162\*) | c.2276G>Tp.(Cys759Phe) | Unknown (novel) | nsRP-enriched | 1 | Y |
| This study (MCGM) | Classic USH2 | c.2299delG p.(Glu767Serfs\*21) | c.6446C>Ap.(Pro2149Gln) | non-specific | unknown | 1 | Y |
| This study (MCGM) | nsRP | c.4222C>Tp.(Gln1408\*) | c.4222C>Tp.(Gln1408\*) | non-specific | non-specific | 1 | N |

**nsRP non-syndromic retinitis pigmentosa, USH2: Usher syndrome type IIa; Y valid model, N not valid model; MCGM Manchester Centre for Genomic Medicine**