**Table S1. Allele classification as per the *USH2A* allelic hierarchy model**

|  | **Nucleotide change** | **Amino acid change** | **Key references**  |
| --- | --- | --- | --- |
| **nsRP-enriched *USH2A* alleles** | c.9882C>G | p.(Cys3294Trp) | (1,2)  |
| c.9571-2A>G | p. ? | (2,3) |
| c.4027A>C | p.(Asn1343His) | (2,4)  |
| c.3902G>T | p.(Gly1301Val) | (2,5) |
| c.2276G>T | p.(Cys759Phe) | (4,6–19) |
| c.13335\_13347delGAACATGGACTCTinsCTTG | p.(Glu4445\_Ser4449delinsAspLeu) | (2,4,20) |
| c.13331C>T |  p.Pro4444Leu | (2) |
| c.13126T>G | p.(Trp4376Gly) | (2) |
| c.12874A>G |  p.(Asn4292Asp) | (2,21) |
| c.12575G>A |  p.(Arg4192His) | (4,5,11,20,22,23) |
| c.10342G>A |  p.(Glu3448Lys) | (2,3,20,24–26) |
| c.10073G>A |  p.(Cys3358Tyr) | (2,5,11,19,20,22,23,27–29) |
| **non-specific *USH2A* alleles** | c.9815C>T |  p.(Pro3272Leu) | (18,26,30–35) |
| c.949C>A |  p.(Tyr318Cysfs\*17) | (6,18,32,33,36–40) |
| c.9371+1G>C |  p.(?)  | (5,22,33,41) |
| c.920\_923dupGCCA |  p.(His308Glnfs\*16) | (9,12,17,18,33,36,39,42–46) |
| c.8981G>A |  p.(Trp2994\*) | (2,47) |
| c.820C>G |  p.(Arg274Gly) | (2,18,19) |
| c.8079G>A |  p.(Trp2876\*) | (27,48) |
| c.7950dup |  p.(Asn2651Glnfs\*10) | (27,33) |
| c.7595-2144A>G  | p.(Lys2532Thrfs\*56) | (2,18,22,27,33,38,44,49–52) |
| c.7595-3C>G | p.(Pro2533Asnfs\*5) | (5,18,22,50,53–55) |
| c.7121-8313\_11048-962delins12 | p.? | (27) |
| c.6926G>T |  p.(Cys2309Phe) | (4,27,33) |
| c.6862G>T |  p.(Glu2288\*)  | (22,27,33,47) |
| c.6722C>T |  p.(Pro2241Leu) | (27,56) |
| c.653T>A |  p.(Val218Glu) | (2,4,6,18,40,46,50,53) |
| c.5776+1G>A |  p.(?)  | (4,5,7,29,32,33,39,45,49) |
| c.4957C>T |  p.(Arg1653\*) | (18,23,27,33,39) |
| c.486-14G>A |  p.? | (18,23,27,53,57) |
| c.4732C>T |  p.(Arg1578Cys) | (18,22,27) |
| c.4645C>T |  p.(Arg1549\*) | (7,18–20,22,27,33,38) |
| c.4474G>T |  p.(Glu1492\*)  | (16,18,19,33,52) |
| c.4222C>T |  p.(Gln1408\*) | (17,33,58) |
| c.4133T>C |  p.(Leu1378Pro) | (18,22,27) |
| c.3407G>A |  p.(Ser1136Asn)  | (22,33), this study |
| c.3395G>A |  p.(Gly1132Asp) | (2,59) |
| c.3368A>G |  p.(Tyr1123Cys) | (27,53,59) |
| c.280T>G |  p.(Cys934Trp) | (2,5,33,35,60,61) |
| c.2522C>A |  p.(Ser841Tyr) | (2,18,19,22,45) |
| c.2391\_2392del |  p.(Cys797\*) | (18,27) |
| c.2299delG | p.(Glu767Serfs\*21) | (5,7–9,12,13,15–19,22,31–33,36–38,40,42,45,46,49,50,52,59,62–72) |
| c.2296T>C |  p.(Cys766Arg) | (4,27,33,52) |
| c.2242C>T |  p.(Gln748\*) | (27,36) |
| c.2168-1G>C |  p.(Leu724Valfs\*31) | (18,38,71) |
| c.1978G>A |  p.(Gly660Arg) | (18,27,33) |
| c.187C>T |  p.(Arg63\*) | (10,12,22,48,49,59,71) |
| c.1876C>T |  p.(Arg626\*) | (12,17,18,27,33,39,46,48,50,61,65,66,73) |
| c.1606T>C |  p.(Cys536Arg) | (11,12,22,27,36,39,40,59) |
| c.15433G>A |  p.(Val5145Ile) | (2,4,18,20,22,27) |
| c.15089C>A |  p.(Ser5030\*) | (27,33,40,50) |
| c.14977\_14978del |  p.(Phe4993Profs\*7) | (18,27,33,49) |
| c.14803C>T |  p.(Arg4935\*) | (6,8,18,20,27,33,50,53,65) |
| c.14426C>T |  p.(Thr4809Ile) | (2,5,18,33,40,65) |
| c.14131C>T |  p.(Gln4711\*) | (7,27,33,44) |
| c.13316C>T |  p.(Thr4439lle) | (2,5,18,22,27,33,39) |
| c.1328+1G>A |  p.? | (19,27) |
| c.13274C>T |  p.(Thr4425Met) | (2,23,27,33,37,50) |
| c.12954C>A |  p.(Tyr4318Ter) | (2,20,33,39) |
| c.12574C>T |  p.Arg4192Cys | (2,6,33,34,43,74), this study |
| c.1256G>T |  p.(Cys419Phe) | (5,17,18,22,27,31,36,42,75) |
| c.12505A>G |  p.(Thr4169Ala) | (2,18,22) |
| c.1227G>A |  p.(Trp409\*) | (27,36) |
| c.11875\_11876delCA | p.(Gln3959Asnfs\*53) | (2,18,22,33,39,47,48) |
| c.11864G>A |  p.(Trp3955\*) | (2,27,48) |
| c.11819A>C |  p.(Tyr3940Ser) | (4–7,17,18,20,22,30,32,33,37–40,44,47,49,59) |
| c.11156G>A |  p.(Arg3719His) | (2,5,20,33,76) |
| c.10561T>C |  p.(Trp3521Arg) | (2,4,20,22–24,27,33,39,48) |
| c.10525A>T |  p.(Lys3509\*) | (23,27,48) |
| c.1055C>T | p.(Thr352Ile) | (2,6,32,33,39,40,50) |
| c.1039G>C |  p.(Asp347His) | (27) |
| c.1036A>C |  p.(Asn346His) | (12,18,22,27,33,36,39,44,50,66) |
| c.100C>T |  p.(Arg34\*) | (2,12,16,22,33,44,45,77) |
| **Unknown and/or novel *USH2A* alleles** | Exon 57 to 60 duplication |  | (5) |
| Exon 33-34 deletion |  | - |
| Exon 12-13 deletion |  p.(Ile658Phefs\*23) | (27) |
| Exon 10-14 deletion |  p.(Cys549Metfs\*5) | - |
| Deletion 1:216259365-216318209 |  | - |
| Deletion 1:216240159-222780953 |  | - |
| Deletion 1:216009683-216011948 |  | - |
| Deletion 1:215836170-215851932 |  | - |
| c.9974G>A |  p.(Gly3325Glu)  | - |
| c.9860\_9873delATGATGGCCATGGC |  p.(His3287ProfsTer54) | - |
| c.9785G>T |  p.(Gly3262Val) | (2) |
| c.9433C>T |  p.(Leu3145Phe) | (26) |
| c.9413G>A |  p.(Gly3138Asp) | (56) |
| c.9372-?\_9570+? |  | - |
| c.926C>T |  p.(Pro309Leu) | (2,33) |
| c.9258G>T |  p.(Gln3086His) | (27) |
| c.917\_918insGCTG | p.(Ser307Leufs\*17) | (23) |
| c.8954delG |  p.(Gly2985Alafs\*3) | (19) |
| c.8723\_8724del |  p.(Val2908Glyfs\*29) | (27,37) |
| c.8628G>A |  p.(Trp2876\*) | (27,49) |
| c.8223+1G>C |  p.(?) | - |
| c.7931G>A |  p.(Trp2644\*) | (18,27) |
| c.7853G>A |  p.(Trp2618\*) | (27) |
| c.785-?\_+5572+?dup deletion exon 5-27 |  | (27) |
| c.7501C>T |  p.(Gln2501\*) | (27) |
| c.7358T>A |  p.(Val2453Asp) | (2) |
| c.7334C>T |  p.(Ser2445Phe) | (2) |
| c.7301-?\_10939+?39-55indel |  | (27) |
| c.7187G>A |  p.(Trp2396\*) | (27) |
| c.7132\_7133del |  p.(Tyr2378Hisfs\*39) | (27) |
| c.7121-?\_11047+? |  p.(?) | (27) |
| c.7054C>T |  p.(Pro2352Ser) | (27,56) |
| c.6967C>T \* |  p.(Arg2323\*) | (27) |
| c.6670G>T |  p.(Gly2224Cys) | (2,78) |
| c.6658-2A>G |  p.? | (27) |
| c.6544\_6548dup |  p.(His2183fs) | (27) |
| c.6446C>A |  p.(Pro2149Gln) | (2); this study |
| c.6050-1G>A |  p.? | (2) |
| c.5614delins12 |  p.(Ala1872Leufs\*64)  | - |
| c.5603T>G |  p.(Phe1868Cys) | (2,22) |
| c.5576T>G |  p.(Phe1859Cys) | (23,27) |
| c.5516T>A |  p.(Val1839Glu) | (27) |
| c.5012G>A |  p.(Gly1671Asp) | (2,22) |
| c.4810G>A\* |  p.(Asp1604Asn) | (27) |
| c.4773del |  p.(Val1592\*) | (27) |
| c.4510dupA |  p.(Arg1504Lysfs\*26) | (2,22) |
| c.4405C>T |  p.(Gln1469\*) | (36) |
| c.4362\_4367delinsACTC |  | (27) |
| c.4321G>T |  p.(Glu1441\*) | - |
| c.4106C>T |  p.(Ser1369Leu) | (27) |
| c.4056G>A |  p.(Trp1352\*) | (27) |
| c.3831\_3834delACTAinsG |  p.(Leu1278del) | - |
| c.3485C>A |  p.(Ser1162\*) | - |
| c.3158-6A>G |  p.(?) | (2) |
| c.3045C>G |  p.(His1015Gln) | (27) |
| c.2994A>T |  p.(Arg998Ser)  | (2) |
| c.895delC |  p.(Gln299Asnfs\*37) | (2) |
| c.2710\_2720dup |  p.(Leu908Profs\*63) | (27) |
| c.2555-1G>C |  p.? | (27) |
| c.2140C>T |  p.(Gln714\*) | - |
| c.2139C>T |  p.(Gly713Gly)  | - |
| c.2081G>A |  p.(Cys694Tyr) | (2,5) |
| c.2014C>T |  p.(Gln672\*) | (27) |
| c.1965T>G |  p.(Cys655Thr) | (27) |
| c.1859G>T |  p.(Cys620Phe)  | - |
| c.1808G>A |  p.(Gly603Glu) | (27) |
| c.1804G>A |  p.(Gly602Arg)  | - |
| c.1558delT |  p.(Cys520Alafs\*71) | - |
| c.15053-2A>T \* |  p.(?) | (27) |
| c.14802C>G |  p.(Tyr4934\*) | (27) |
| c.14791+2T>A |  p.(?) | - |
| c.14545T>C |  p.(Trp4849Arg) | (27) |
| c.14289del |  p.(Ile4764Serfs\*42) | (27) |
| c.14174G>A \* |  p.(Trp4725\*) | (27) |
| c.13576C>T |  p.(Arg4526\*) | (2,79) |
| c.13508\_13523delinsAG | p.(Val4503Glufs\*54) | (27) |
| c.13441A>G |  p.(Arg4481Gly)  | - |
| c.13396C>T |  p.(Pro4466Ser) | (2) |
| c.13283G>A |  p.(Gly4428Asp)  | - |
| c.13262T>C |  p.(Leu4421Pro) | (27) |
| c.12992A>G |  p.(Tyr4331Cys) | (27) |
| c.12819T>A |  p.(Tyr4273\*) | (2) |
| c.12806C>A |  p.(Pro4269His) | (18,27) |
| c.12729G>A |  p.(Trp4243\*) | (27) |
| c.12394del |  p.(Leu4132Trpfs\*35) | (27) |
| c.12309delC |  p.(Phe4103Leufs\*11) | (2) |
| c.12145G>A |  p.(Ala4049Thr) | (2,18) |
| c.1206G>T |  p.(Lys402Asn) | (27) |
| c.11875C>T |  p.(Gln3959\*) | (27) |
| c.11699A>G |  p.(Tyr3900Cys) | - |
| c.11694delC | p.(Asn3899ThrfsTer34) | (2) |
| c.11676del |  p.Lys3892Asnfs\*41 | (27) |
| c.11507C>T |  p.(Pro3836Leu) | (2) |
| c.1111\_1112delAT |  p.(Ile371Phefs\*) | (7,80) |
| c.11007C>A |  p.(Ser3669Arg) | (23) |
| c.10901A>C |  p.(His3634Pro) | (27) |
| c.10689T>A |  p.(Tyr3563\*) | (27) |
| c.10387+2T>C p.(?) | p.? | (27) |
| c.13331delC  | p.(Pro4444Glnfs\*17) | - |

**Table S1 References:**

1. Nishiguchi KM, Tearle RG, Liu YP, Oh EC, Miyake N, Benaglio P, et al. Whole genome sequencing in patients with retinitis pigmentosa reveals pathogenic DNA structural changes and NEK2 as a new disease gene. Proc Natl Acad Sci U S A. 2013 Oct 1;110(40):16139–44.

2. Carss KJ, Arno G, Erwood M, Stephens J, Sanchis-Juan A, Hull S, et al. Comprehensive Rare Variant Analysis via Whole-Genome Sequencing to Determine the Molecular Pathology of Inherited Retinal Disease. Am J Hum Genet. 2017;100(1):75–90.

3. Comander J, Weigel-DiFranco C, Maher M, Place E, Wan A, Harper S, et al. The Genetic Basis of Pericentral Retinitis Pigmentosa-A Form of Mild Retinitis Pigmentosa. Genes (Basel). 2017 Oct 5;8(10).

4. Glöckle N, Kohl S, Mohr J, Scheurenbrand T, Sprecher A, Weisschuh N, et al. Panel-based next generation sequencing as a reliable and efficient technique to detect mutations in unselected patients with retinal dystrophies. Eur J Hum Genet. 2014 Jan 17;22(1):99–104.

5. Lenassi E, Vincent A, Li Z, Saihan Z, Coffey AJ, Steele-Stallard HB, et al. A detailed clinical and molecular survey of subjects with nonsyndromic USH2A retinopathy reveals an allelic hierarchy of disease-causing variants. Eur J Hum Genet. 2015 Oct 4;23(10):1318–27.

6. Besnard T, García-García G, Baux D, Vaché C, Faugère V, Larrieu L, et al. Experience of targeted Usher exome sequencing as a clinical test. Mol Genet Genomic Med. 2014 Jan;2(1):30–43.

7. Sandberg MA, Rosner B, Weigel-DiFranco C, McGee TL, Dryja TP, Berson EL. Disease course in patients with autosomal recessive retinitis pigmentosa due to the USH2A gene. Invest Ophthalmol Vis Sci. 2008 Dec;49(12):5532–9.

8. Rivolta C, Sweklo EA, Berson EL, Dryja TP. Missense mutation in the USH2A gene: association with recessive retinitis pigmentosa without hearing loss. Am J Hum Genet. 2000;66(6):1975–8.

9. Aller E, Nájera C, Millán JM, Oltra JS, Pérez-Garrigues H, Vilela C, et al. Genetic analysis of 2299delG and C759F mutations (USH2A) in patients with visual and/or auditory impairments. Eur J Hum Genet. 2004 May 18;12(5):407–10.

10. Bernal S, Ayuso C, Antiñolo G, Gimenez A, Borrego S, Trujillo MJ, et al. Mutations in USH2A in Spanish patients with autosomal recessive retinitis pigmentosa: high prevalence and phenotypic variation. J Med Genet. 2003 Jan;40(1):e8.

11. Ávila-Fernández A, Cantalapiedra D, Aller E, Vallespín E, Aguirre-Lambán J, Blanco-Kelly F, et al. Mutation analysis of 272 Spanish families affected by autosomal recessive retinitis pigmentosa using a genotyping microarray. Mol Vis. 2010 Dec 3;16(July):2550–8.

12. Dreyer B, Tranebjaerg L, Rosenberg T, Weston MD, Kimberling WJ, Nilssen O. Identification of novel USH2A mutations: implications for the structure of USH2A protein. Eur J Hum Genet. 2000;8(7):500–6.

13. Aller E, Jaijo T, Beneyto M, Nájera C, Oltra S, Ayuso C, et al. Identification of 14 novel mutations in the long isoform of USH2A in Spanish patients with Usher syndrome type II. J Med Genet. 2006 Nov;43(11):e55.

14. Aller E, Larrieu L, Jaijo T, Baux D, Espinós C, González-Candelas F, et al. The USH2A c.2299delG mutation: dating its common origin in a Southern European population. Eur J Hum Genet. 2010;18(7):788–93.

15. Nájera C, Beneyto M, Blanca J, Aller E, Fontcuberta A, Millán JM, et al. Mutations in myosin VIIA (MYO7A) and usherin (USH2A) in Spanish patients with Usher syndrome types I and II, respectively. Hum Mutat. 2002 Jul;20(1):76–7.

16. Bernal S, Medà C, Solans T, Ayuso C, Garcia-Sandoval B, Valverde D, et al. Clinical and genetic studies in Spanish patients with Usher syndrome type II: description of new mutations and evidence for a lack of genotype-phenotype correlation. Clin Genet. 2005 Jul 25;68(3):204–14.

17. Seyedahmadi BJ, Rivolta C, Keene JA, Berson EL, Dryja TP. Comprehensive screening of the USH2A gene in Usher syndrome type II and non-syndromic recessive retinitis pigmentosa. Exp Eye Res. 2004 Aug;79(2):167–73.

18. Baux D, Blanchet C, Hamel C, Meunier I, Larrieu L, Faugère V, et al. Enrichment of LOVD-USHbases with 152 USH2A Genotypes Defines an Extensive Mutational Spectrum and Highlights Missense Hotspots. Hum Mutat. 2014 Oct;35(10):1179–86.

19. García-García G, Aparisi MJ, Jaijo T, Rodrigo R, Leon AM, Avila-Fernandez A, et al. Mutational screening of the USH2A gene in Spanish USH patients reveals 23 novel pathogenic mutations. Orphanet J Rare Dis. 2011;6:65.

20. McGee TL, Seyedahmadi BJ, Sweeney MO, Dryja TP, Berson EL. Novel mutations in the long isoform of the USH2A gene in patients with Usher syndrome type II or non-syndromic retinitis pigmentosa. J Med Genet. 2010 Jul;47(7):499–506.

21. Watson CM, El-Asrag M, Parry DA, Morgan JE, Logan C V., Carr IM, et al. Mutation Screening of Retinal Dystrophy Patients by Targeted Capture from Tagged Pooled DNAs and Next Generation Sequencing. den Hollander AI, editor. PLoS One. 2014 Aug 18;9(8):e104281.

22. Le Quesne Stabej P, Saihan Z, Rangesh N, Steele-Stallard HB, Ambrose J, Coffey A, et al. Comprehensive sequence analysis of nine Usher syndrome genes in the UK National Collaborative Usher Study. J Med Genet. 2012 Jan;49(1):27–36.

23. Neveling K, Collin RWJ, Gilissen C, Van Huet RAC, Visser L, Kwint MP, et al. Next-generation genetic testing for retinitis pigmentosa. Hum Mutat. 2012 Jun;33(6):963–72.

24. Eisenberger T, Neuhaus C, Khan AO, Decker C, Preising MN, Friedburg C, et al. Increasing the yield in targeted next-generation sequencing by implicating CNV analysis, non-coding exons and the overall variant load: The example of retinal dystrophies. Li T, editor. PLoS One. 2013 Nov 12;8(11):e78496.

25. Maranhao B, Biswas P, Duncan JL, Branham KE, Silva GA, Naeem MA, et al. ExomeSuite: Whole exome sequence variant filtering tool for rapid identification of putative disease causing SNVs/indels. Genomics. 2014 Feb;103(2–3):169–76.

26. Ge Z, Bowles K, Goetz K, Scholl HPN, Wang F, Wang X, et al. NGS-based Molecular diagnosis of 105 eyeGENE ® probands with Retinitis Pigmentosa. Sci Rep. 2015 Dec 15;5:18287.

27. Pierrache LHM, Hartel BP, Van Wijk E, Meester-Smoor MA, Cremers FPM, De Baere E, et al. Visual Prognosis in USH2A-Associated Retinitis Pigmentosa Is Worse for Patients with Usher Syndrome Type IIa Than for Those with Nonsyndromic Retinitis Pigmentosa. Ophthalmology. 2016;123(5):1151–60.

28. Zhao L, Wang F, Wang H, Li Y, Alexander S, Wang K, et al. Next-generation sequencing-based molecular diagnosis of 82 retinitis pigmentosa probands from Northern Ireland. Hum Genet. 2015 Feb 4;134(2):217–30.

29. Wang F, Wang H, Tuan H-F, Nguyen DH, Sun V, Keser V, et al. Next generation sequencing-based molecular diagnosis of retinitis pigmentosa: identification of a novel genotype-phenotype correlation and clinical refinements. Hum Genet. 2014 Mar 24;133(3):331–45.

30. Herrera W, Aleman TS, Cideciyan A V., Roman AJ, Banin E, Ben-Yosef T, et al. Retinal Disease in Usher Syndrome III Caused by Mutations in the Clarin-1 Gene. Investig Opthalmology Vis Sci. 2008 Jun 1;49(6):2651.

31. Leijendeckers JM, Pennings RJE, Snik AFM, Bosman AJ, Cremers CWRJ. Audiometric characteristics of USH2a patients. Audiol Neurotol. 2009;14(4):223–31.

32. Lenarduzzi S, Vozzi D, Morgan A, Rubinato E, D’Eustacchio A, Osland TM, et al. Usher syndrome: An effective sequencing approach to establish a genetic and clinical diagnosis. Hear Res. 2015;320:18–23.

33. Bonnet C, Riahi Z, Chantot-Bastaraud S, Smagghe L, Letexier M, Marcaillou C, et al. An innovative strategy for the molecular diagnosis of Usher syndrome identifies causal biallelic mutations in 93% of European patients. Eur J Hum Genet. 2016;24:1730–8.

34. Hettinga YM, van Genderen MM, Wieringa W, Ossewaarde-van Norel J, de Boer JH. Retinal Dystrophy in 6 Young Patients Who Presented with Intermediate Uveitis. Ophthalmology. 2016 Sep 1;123(9):2043–6.

35. Jiang L, Liang X, Li Y, Wang J, Zaneveld JE, Wang H, et al. Comprehensive molecular diagnosis of 67 Chinese Usher syndrome probands: High rate of ethnicity specific mutations in Chinese USH patients. Orphanet J Rare Dis. 2015;10(1).

36. Pennings RJE, Te Brinke H, Weston MD, Claassen A, Orten DJ, Weekamp H, et al. USH2A mutation analysis in 70 Dutch families with Usher syndrome type II. Hum Mutat. 2004;24(2):185.

37. van Wijk E, Pennings RJE, te Brinke H, Claassen A, Yntema HG, Hoefsloot LH, et al. Identification of 51 Novel Exons of the Usher Syndrome Type 2A (USH2A) Gene That Encode Multiple Conserved Functional Domains and That Are Mutated in Patients with Usher Syndrome Type II. Am J Hum Genet. 2004 Apr;74(4):738–44.

38. Vaché C, Besnard T, le Berre P, García-García G, Baux D, Larrieu L, et al. Usher syndrome type 2 caused by activation of an USH2A pseudoexon: Implications for diagnosis and therapy. Hum Mutat. 2012 Jan;33(1):104–8.

39. Dreyer B, Brox V, Tranebjærg L, Rosenberg T, Sadeghi AM, Möller C, et al. Spectrum of USH2A mutations in Scandinavian patients with Usher syndrome type II. Hum Mutat. 2008 Mar;29(3):451–451.

40. Bonnet C, Grati M, Marlin S, Levilliers J, Hardelin J-P, Parodi M, et al. Complete exon sequencing of all known Usher syndrome genes greatly improves molecular diagnosis. Orphanet J Rare Dis. 2011 May 11;6(1):21.

41. Shu H-R, Bi H, Pan Y-C, Xu H-Y, Song J-X, Hu J. Targeted exome sequencing reveals novel USH2A mutations in Chinese patients with simplex Usher syndrome. BMC Med Genet. 2015 Dec 16;16(1):83.

42. Weston MD, Eudy JD, Fujita S, Yao S, Usami S, Cremers C, et al. Genomic structure and identification of novel mutations in usherin, the gene responsible for Usher syndrome type IIa. Am J Hum Genet. 2000 Apr;66(4):1199–210.

43. Corton M, Nishiguchi KM, Avila-Fernández A, Nikopoulos K, Riveiro-Alvarez R, Tatu SD, et al. Exome Sequencing of Index Patients with Retinal Dystrophies as a Tool for Molecular Diagnosis. Stieger K, editor. PLoS One. 2013 Jun 14;8(6):e65574.

44. Krawitz PM, Schiska D, Krüger U, Appelt S, Heinrich V, Parkhomchuk D, et al. Screening for single nucleotide variants, small indels and exon deletions with a next-generation sequencing based gene panel approach for Usher syndrome. Mol Genet genomic Med. 2014 Sep;2(5):393–401.

45. Jaijo T, Aller E, García-García G, Aparisi MJ, Berna S, Ávila-Fernández A, et al. Microarray-based mutation analysis of 183 Spanish families with Usher syndrome. Investig Ophthalmol Vis Sci. 2010 Mar 1;51(3):1311–7.

46. Leroy BP, Aragon-Martin JA, Weston MD, Bessant DA, Willis C, Webster AR, et al. Spectrum of mutations in USH2A in British patients with Usher syndrome type II. Exp Eye Res. 2001 May;72(5):503–9.

47. Yan D, Ouyang X, Patterson DM, Du LL, Jacobson SG, Liu X-Z. Mutation analysis in the long isoform of USH2A in American patients with Usher Syndrome type II. J Hum Genet. 2009 Dec 30;54(12):732–8.

48. Hartel BP, Löfgren M, Huygen PLM, Guchelaar I, Lo-A-Njoe Kort N, Sadeghi AM, et al. A combination of two truncating mutations in USH2A causes more severe and progressive hearing impairment in Usher syndrome type IIa. Hear Res. 2016;339:60–8.

49. Sodi A, Mariottini A, Passerini I, Murro V, Tachyla I, Bianchi B, et al. MYO7A and USH2A gene sequence variants in Italian patients with Usher syndrome. Mol Vis. 2014;20:1717–31.

50. Baux D, Larrieu L, Blanchet C, Hamel C, Ben Salah S, Vielle A, et al. Molecular and in silico analyses of the full-length isoform of usherin identify new pathogenic alleles in Usher type II patients. Hum Mutat. 2007 Aug;28(8):781–9.

51. Steele-Stallard HB, Le Quesne Stabej P, Lenassi E, Luxon LM, Claustres M, Roux A-F, et al. Screening for duplications, deletions and a common intronic mutation detects 35% of second mutations in patients with USH2A monoallelic mutations on Sanger sequencing. Orphanet J Rare Dis. 2013;8(1):122.

52. Aparisi MJ, Aller E, Fuster-García C, García-García G, Rodrigo R, Vázquez-Manrique RP, et al. Targeted next generation sequencing for molecular diagnosis of Usher syndrome. Orphanet J Rare Dis. 2014 Dec 18;9(1):168.

53. Le Guédard-Méreuze S, Vaché C, Baux D, Faugère V, Larrieu L, Abadie C, et al. Ex vivo splicing assays of mutations at noncanonical positions of splice sites in USHER genes. Hum Mutat. 2010 Mar;31(3):347–55.

54. Sloan-Heggen CM, Bierer AO, Shearer AE, Kolbe DL, Nishimura CJ, Frees KL, et al. Comprehensive genetic testing in the clinical evaluation of 1119 patients with hearing loss. Hum Genet. 2016;

55. Wang J, Zhang VW, Feng Y, Tian X, Li FY, Truong C, et al. Dependable and efficient clinical utility of target capture-based deep sequencing in molecular diagnosis of retinitis pigmentosa. Investig Ophthalmol Vis Sci. 2014 Oct 6;55(10):6213–23.

56. van Huet RAC, Pierrache LHM, Meester-Smoor MA, Klaver CCW, van den Born LI, Hoyng CB, et al. The efficacy of microarray screening for autosomal recessive retinitis pigmentosa in routine clinical practice. Mol Vis. 2015;21:461–76.

57. Zhao Y, Hosono K, Suto K, Ishigami C, Arai Y, Hikoya A, et al. The first USH2A mutation analysis of Japanese autosomal recessive retinitis pigmentosa patients: a totally different mutation profile with the lack of frequent mutations found in Caucasian patients. J Hum Genet. 2014 Sep;59(9):521–8.

58. Tajiguli A, Xu M, Fu Q, Yiming R, Wang K, Li Y, et al. Next-generation sequencing-based molecular diagnosis of 12 inherited retinal disease probands of Uyghur ethnicity. Sci Rep. 2016 Feb 9;6:21384.

59. Vozzi D, Aaspõllu A, Athanasakis E, Berto A, Fabretto A, Licastro D, et al. Molecular epidemiology of Usher syndrome in Italy. Mol Vis. 2011;17(June):1662–8.

60. Zheng S-L, Zhang H-L, Lin Z-L, Kang Q-Y. Whole-exome sequencing identifies USH2A mutations in a pseudo-dominant Usher syndrome family. Int J Mol Med. 2015;36(4):1035–41.

61. Xu W, Dai H, Lu T, Zhang X, Dong B, Li Y. Seven novel mutations in the long isoform of the USH2A gene in Chinese families with nonsyndromic retinitis pigmentosa and Usher syndrome Type II. Mol Vis. 2011;17(January):1537–52.

62. Dreyer B, Tranebjaerg L, Brox V, Rosenberg T, Möller C, Beneyto M, et al. A common ancestral origin of the frequent and widespread 2299delG USH2A mutation. Am J Hum Genet. 2001 Jul;69(1):228–34.

63. Eudy JD, Weston MD, Yao S, Hoover DM, Rehm HL, Ma-Edmonds M, et al. Mutation of a gene encoding a protein with extracellular matrix motifs in Usher syndrome type IIa. Science (80- ). 1998;280(5370):1753–7.

64. Liu XZ, Hope C, Liang CY, Zou JM, Xu LR, Cole T, et al. A mutation (2314delG) in the Usher syndrome type IIA gene: high prevalence and phenotypic variation. Vol. 64, American journal of human genetics. 1999. p. 1221–5.

65. Ebermann I, Koenekoop RK, Lopez I, Bou-Khzam L, Pigeon R, Bolz HJ. An USH2A founder mutation is the major cause of Usher syndrome type 2 in Canadians of French origin and confirms common roots of Quebecois and Acadians. Eur J Hum Genet. 2009 Jan 30;17(1):80–4.

66. Ouyang XM, Yan D, Hejtmancik JF, Jacobson SG, Li AR, Du LL, et al. Mutational spectrum in Usher syndrome type II. Clin Genet. 2004 Apr 16;65(4):288–93.

67. Liquori A, Vaché C, Baux D, Blanchet C, Hamel C, Malcolm S, et al. Whole USH2A Gene Sequencing Identifies Several New Deep Intronic Mutations. Hum Mutat. 2016 Feb;37(2):184–93.

68. Kimberling WJ, Hildebrand MS, Shearer AE, Jensen ML, Halder JA, Trzupek K, et al. Frequency of Usher syndrome in two pediatric populations: Implications for genetic screening of deaf and hard of hearing children. Genet Med. 2010 Aug 6;12(8):512–6.

69. Lenassi E, Saihan Z, Bitner-Glindzicz M, Webster AR. The effect of the common c.2299delG mutation in USH2A on RNA splicing. Exp Eye Res. 2014 May;122:9–12.

70. Licastro D, Mutarelli M, Peluso I, Neveling K, Wieskamp N, Rispoli R, et al. Molecular Diagnosis of Usher Syndrome: Application of Two Different Next Generation Sequencing-Based Procedures. El-Maarri O, editor. PLoS One. 2012 Aug 29;7(8):e43799.

71. Pierrottet CO, Zuntini M, Digiuni M, Bazzanella I, Ferri P, Paderni R, et al. Syndromic and non-syndromic forms of retinitis pigmentosa: a comprehensive Italian clinical and molecular study reveals new mutations. Genet Mol Res. 2014 Oct 27;13(4):8815–33.

72. Schwartz SB, Aleman TS, Cideciyan A V., Windsor EAM, Sumaroka A, Roman AJ, et al. Disease Expression in Usher Syndrome Caused by *VLGR1* Gene Mutation ( *USH2C* ) and Comparison with *USH2A* Phenotype. Investig Opthalmology Vis Sci. 2005 Feb 1;46(2):734.

73. Adato A, Weston MD, Berry A, Kimberling WJ, Bonne-Tamir A. Three novel mutations and twelve polymorphisms identified in the USH2A gene in Israeli USH2 families. Hum Mutat. 2000 Apr;15(4):388–388.

74. De Castro-Miró M, Pomares E, Lorés-Motta L, Tonda R, Dopazo J, Marfany G, et al. Combined Genetic and high-throughput strategies for molecular diagnosis of inherited retinal dystrophies. Escriva H, editor. PLoS One. 2014 Feb 7;9(2):e88410.

75. Requena T, Gallego-Martinez A, Lopez-Escamez JA. A pipeline combining multiple strategies for prioritizing heterozygous variants for the identification of candidate genes in exome datasets. Hum Genomics. 2017 Dec 22;11(1):11.

76. Chen X, Sheng X, Liu X, Li H, Liu Y, Rong W, et al. Targeted next-generation sequencing reveals novel USH2A mutations associated with diverse disease phenotypes: Implications for clinical and molecular diagnosis. Dermaut B, editor. PLoS One. 2014 Aug 18;9(8):e105439.

77. Li J, Zhao X, Xin Q, Shan S, Jiang B, Jin Y, et al. Whole-exome sequencing identifies a variant in TMEM132E causing autosomal-recessive nonsyndromic hearing loss DFNB99. Hum Mutat. 2015;36(1):98–105.

78. Consugar MB, Navarro-Gomez D, Place EM, Bujakowska KM, Sousa ME, Fonseca-Kelly ZD, et al. Panel-based genetic diagnostic testing for inherited eye diseases is highly accurate and reproducible and more sensitive for variant detection, than exome sequencing. Genet Med. 2015 Apr 20;17(4):253–61.

79. Nakanishi H, Ohtsubo M, Iwasaki S, Hotta Y, Usami S-I, Mizuta K, et al. Novel USH2A mutations in Japanese Usher syndrome type 2 patients: marked differences in the mutation spectrum between the Japanese and other populations. J Hum Genet. 2011;56(10):484–90.

80. Cremers FPM, Kimberling WJ, Külm M, de Brouwer AP, van Wijk E, te Brinke H, et al. Development of a genotyping microarray for Usher syndrome. J Med Genet. 2007 Aug 11;44(2):153–60.