A Case of Usher Syndrome Presenting with a Right Lamellar Hole

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Abstract

A 23-year-old lady presented to the Eye Clinic with poor night vision since childhood, worse in the right eye. There was an associated history of difficulty hearing noticed by her mother, which has progressively worsened. The presenting visual acuity was hand movement in the right eye and 6/60 in the left eye, respectively. She gave a history of recent and frequent involvement in domestic and road traffic accidents, which precipitated her presentation to the eye clinic. There were no concurrent systemic illnesses. Binocular indirect ophthalmoscopy revealed bilateral widespread bone spicule pigmentation, waxy disc pallor, and attenuated vessels with bilateral atrophic changes worse in the left eye. The optical coherence tomography scan revealed a lamella hole in the right eye. The otorhinolaryngologist made a diagnosis of sensorineural deafness. A diagnosis of Usher syndrome with a right lamellar hole was made in light of the clinical findings.

Keywords: Bony spicules, hearing deficit, lamellar hole, macula atrophy, retinitis pigmentosa

Introduction

Usher syndrome (USH) is a rare, inherited condition characterized by visual impairment, hearing deficit, and vestibular disorders.[1] It occurs in about 3 per 100,000 population and inheritance is more commonly autosomal recessive in nature. It is a syndromic retinitis pigmentosa (RP) disorder characterized by the typical fundal findings of bone spicule pigmentation, attenuated vessels, and a waxy pale disc in addition to the hearing loss and vestibular symptoms.[2] Approximately 50% of all hereditary disorders with both deafness and blindness are linked to USH, which is also responsible for 3% to 6% of diagnosis of hearing loss in childhood.[3-6]

Usher syndrome is classified into three categories namely types 1, 2, and 3. Type 1 is named USH 1 and has features of profound congenital deafness with vestibular dysfunction. Congenital moderate deafness with normal vestibular function is present in type 2 (USH 2) while type 3 (USH 3) consists of progressive hearing loss and occasional disturbances of vestibular function.[7,8] USH 2 is the most prevalent of the three types and the first for which the culpable gene was identified.[6,7,9] The first reported case of Usher syndrome in Nigeria was documented in a child about 42 years ago.[10] More recently, a report of two cases of USH has been documented from the eastern part of Nigeria.[11] This index case reports the rare occurrence of a lamella hole in a patient with Usher syndrome.

Case Report

A 23-year-old female petrol station attendant presented to the eye clinic with a 20-year history of poor vision, worse at night, noticed since childhood. There was an additional history of bumping into objects since childhood, especially at night, which has now progressively worsened. She had no antecedent history of trauma. There was no history of flashes, floaters, excessive glare, haloes around light, or metamorphopsia. She had no episodes of redness, discharge, or itching. The patient complained of poor night vision and difficulty with peripheral vision. There was no history of chronic use of medications, fever, or prior ocular infections.

She was accompanied to the eye clinic by her mother, who stated that the patient had had difficulty hearing since childhood and had to be tapped before responding to a call

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Received: 27 March 2023 **Revised:** 1 June 2023
Accepted: 3 July 2023 **Published:** 16 August 2023

How to cite this article: Babalola YO. A Case of Usher Syndrome Presenting with a Right Lamellar Hole. Niger J Ophthalmol 2023;31:69-72.

Access this article online

Quick Response Code:

Website: www.nigerianjournalofophthalmology.com

DOI: 10.4103/njo.njo_2_23

or instructions. A deficit in hearing was confirmed by our patient, but there was no accompanying history of ear discharge, otalgia, loss of balance, or vertigo. The patient was worried about her hearing deficit, as it was affecting her relationships with friends and colleagues at work. She stated that when her attention was needed and her name was called, she did not respond promptly due to her hearing deficit. Our patient was also disturbed by the fact that it was assumed she was being proud and was intentionally ignoring her co-workers. There was no history of skin lesions, difficulty breathing, or abdominal symptoms. She had never sought care for her ophthalmic and hearing issues until her presentation at the eye clinic. This presentation to the eye clinic was precipitated by progressive decline in vision and recent episodes of involvement in accidents.

Of note, she was knocked down by a motorcycle while crossing the road at night and lost a tooth in the process. She was unable to see the oncoming motorcycle hence the accident. There is no family history of poor night vision or hearing deficits. She had also been recently involved in a domestic accident at home when she bumped into a stool and sustained an injury to the right side of her face. She had never worn spectacles and had no concurrent systemic illnesses. Her genotype is unknown, and she has no known allergies. There was no family history of nyctalopia, similar ocular symptoms, or blinding eye disease.

The unaided visual acuity was hand movement and 6/60 in the right and left eyes, respectively. Her best corrected visual acuity improved to 6/9 with a subjective refraction of −2.75DS-1.00DC x 140° in the left eye, while there was no improvement in the right eye. The anterior segments were essentially normal, with clear leans. A right relative afferent pupillary defect was seen. Intraocular pressure by Goldmann applanation tonometry was 13 mmHg, respectively, in both eyes.

Binocular indirect ophthalmoscopy and slit lamp biomicroscopy with +78D lenses in both eyes revealed a cup disc ratio (CDR) of 0.3, disc pallor with peripapillary degeneration, attenuated vessels, and widespread bony spicule pigmentation more pronounced in the mid-periphery [Figure 1a and b]. A right lamellar hole just above the photoreceptor layer was present with bilateral atrophic changes at both maculae, worse in the right eye, was confirmed with an optical coherence tomography (OCT) scan (Optos). [Figure 2]

On general examination, she was afebrile, not pale, anicteric, good hydration status and had a scar on the right temporal area and no pedal oedema. Examination of the central nervous, cardiovascular, respiratory, and abdominal systems were essentially normal, with no abnormalities detected. There were no signs of vestibular dysfunction. She was referred to the otorhinolaryngologist for assessment of the hearing deficit. A diagnosis of high-frequency sensorineural hearing loss was made.

With the clinical findings of retinitis pigmentosa and sensorineural hearing loss, a diagnosis of syndromic retinitis pigmentosa, likely Ushers' syndrome (type 2) with right lamellar hole, was made. Our patient and her mother were counseled on the visual prognosis and course of the disease in the future. She was also referred to the low-vision clinic. Spectacles were prescribed and she was commenced on anti-oxidants and scheduled for routine follow-up and evaluation at the eye clinic. She is on follow-up with the otorhinolaryngologist.

Discussion

This patient had had poor vision since childhood, which is associated with hearing loss. Her presentation at the eye clinic was necessitated by a worsening deterioration in her vision, which was affecting her daily activities. The classical triad of fundal features of retinitis pigmentosa, such as bony spicule pigmentation, attenuation of vessels, and pale discs, were present.[1,2] In addition to the fundal findings, the diagnosis of sensorineural hearing loss by the otorhinolaryngologist pointed to the diagnosis of Usher's syndrome.[3,6] Differing from other reports from Nigeria, this index case presented with a right lamella hole in conjunction with the

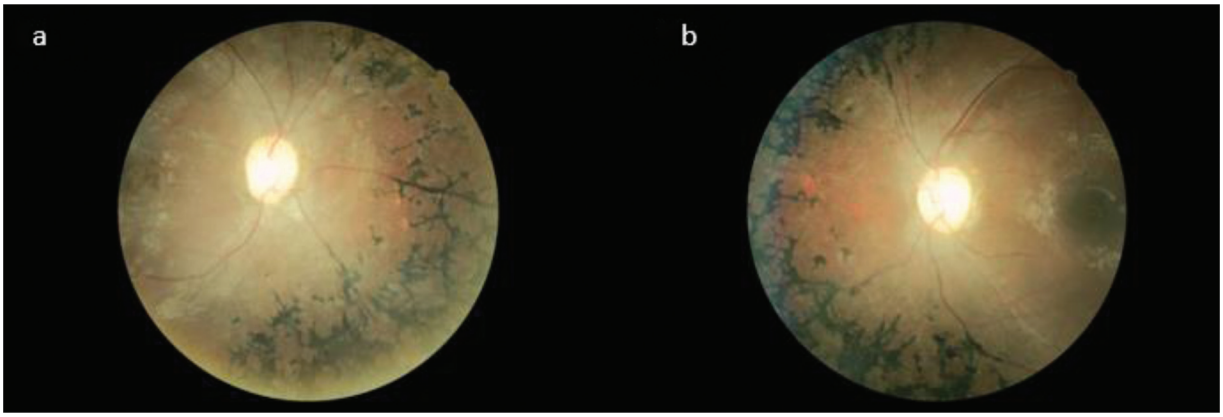

Figure 1: a and b are the fundus photos of the right and left eye showing waxy pale disc, bony spicule pigmentation, and attenuated vessels.

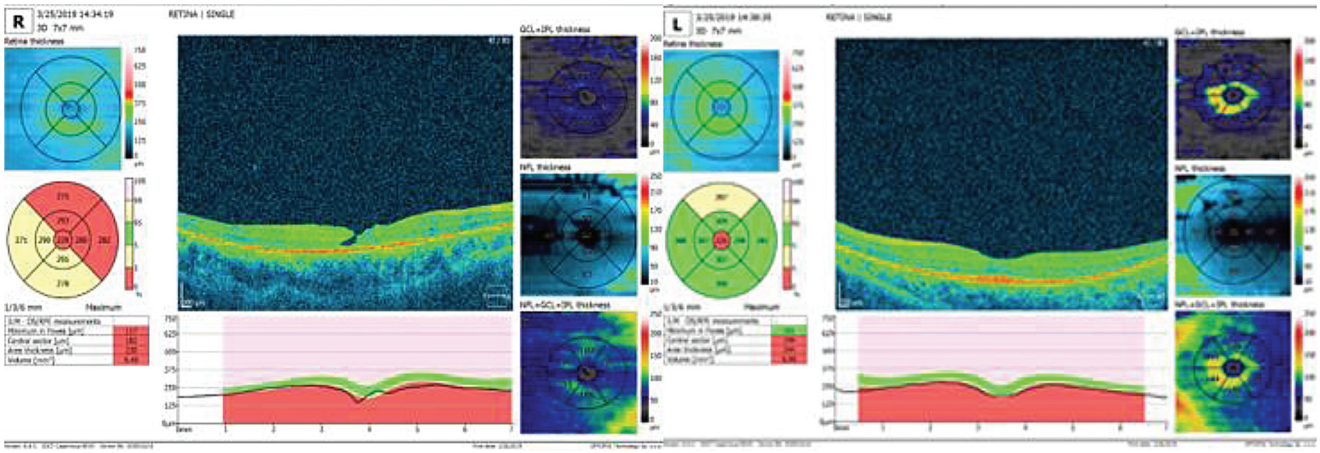

Figure 2: This is the optical coherence tomography scan of the right eye (R) showing a right lamella hole with associated disruption of the ellipsoid zone and atrophy, while the OCT scan of the left eye (L) shows a normal foveal contour and early atrophic changes.

typical ocular and hearing impairment associated with Usher syndrome.[10,11]

Macular lesions are frequent findings in patients with retinitis pigmentosa. The common maculopathies in RP include cystoid macula edema and atrophic maculopathy. Other less common macula lesions include epiretinal membranes, vitreomacular traction, and macula holes.[12-14] Of these macula lesions, cystoid macula edema is also one of the most common macula changes in RP worldwide.[14,15] A higher probability of developing atrophic maculopathy has been documented in those of African descent in comparison to Caucasians.[16] A right lamella hole with atrophic changes at each fovea was present on ocular examination in the reported case, which was confirmed by an OCT scan.

Lamellar holes and macula holes are thought to be less common macula lesions in RP compared to other typical macula pathology. [17] Nevertheless, lamellar holes and pseudoholes are thought to occur more commonly in patients with RP in comparison to the general population.[18] The prevalence of both lamellar and macula holes, respectively, has been documented as 1% in an Italian population, which corroborates the rarity of lamellar holes in RP.[15] The occurrence of bilateral lamellar holes in RP managed with pars plana vitrectomy with good visual and anatomical outcomes has been reported as well.[17] A right lamellar hole was the major finding in our patient, along with atrophic changes, which were also more pronounced in the right eye.

Mechanisms that have been postulated in the development of lamellar holes include contraction of epiretinal membranes and disruption and coalescense of cysts in macula oedema.[17] Cystoid macula edema in RP is believed to be due to inflammatory changes, retinal pigment epithelium (RPE) pump dysfunction, and vitreomacular traction. The OCT scan of our patient was negative for features of epiretinal membrane or cystoid macular edema. Hence, the right lamellar hole in this patient may be a sequela of prior cystoid macula oedema and degenerative changes.[17,19] The poor visual acuity in the right eye may be due to the marked atrophic macula changes, as lamellar holes typically do not cause marked visual loss, with the visual acuity remaining stationary unless it progresses into a full-thickness macula hole.[17,19]

Usher syndrome (USH) is the most frequent form of syndromic retinitis pigmentosa in practice. Based on the classification for Usher syndrome, with the fundal features and sensorineural hearing deficit in the absence of vestibular symptoms, our patient seamlessly fitted into the USH type 2 category.[7,8,20] This is not unusual, as USH type 2 is the milder form and most prevalent, accounting for 70% of cases.[8,21] It has been stated that for every patient diagnosed with USH 1, three cases of USH 2 will be diagnosed.[22] USH2a (1q41), USH2b (3p), USH2c, and (USH2d), of which the locus is unknown, are genes associated with USH 2.[21] Genetic testing to determine the responsible gene in this patient was not done due to the paucity of these facilities in our facility.[23]

Conclusion

Usher syndrome is a common cause of syndromic retinitis pigmentosa. All patients diagnosed with retinitis pigmentosa with some form of hearing impairment or difficulties with balance seen at the eye clinic should be promptly referred to otorhinolaryngology for a hearing assessment. A detailed examination and history are pertinent in ruling out syndromic RP disorders. Lamellar holes may be a rare presentation in retinitis pigmentosa; hence, detailed ocular examination and relevant multimodal imaging are crucial in the management of these patients for a precise diagnosis.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

References

1. Mathur P, Yang J. Usher syndrome: Hearing loss, retinal degeneration and associated abnormalities. Mol Basis Dis 2015;1852:406–20.
2. Boughman JA, Vernon M, Shaver KA. Usher syndrome: definition and estimate of prevalence from two high-risk populations. J Chron Dis 1983;36:595–603.
3. Vernon M. Usher syndrome: deafness and progressive blindness. J Chron Dis 1969;22:133–51.
4. Eberman I, Scholl HP, Charbel IP, *et al.* A novel gene for Usher syndrome type 2: mutations in the long isoform of whirlin are associated with retinitis pigmentosa and sensorineural hearing loss. Hum Genet 2007;121:203–11.
5. Bonnet C, Grati M, Marlin S, *et al.* Complete exon sequencing of all known Usher syndrome genes greatly improves molecular diagnosis. Orphanet J Rare Dis 2011; 6:21.
6. Whatley M, Francis A, Ng ZY, Khoh XE, Atlas MD, Dilley RJ, Wong EYM. Usher syndrome: genetics and molecular links of hearing loss and directions for therapy. Front Genet 2020;11:565216
7. Yang J, Wang L, Song H, Sokolov M. Current understanding of usher syndrome type II. Front Biosci 2012;17:1165–83.
8. Hartong DT, Berson EL, Dryja TP. Retinitis pigmentosa. Lancet 2006;368:1795–809.
9. Kimberling WJ, Orte D, Pieke-Dahl S. Genetic heterogeneity of usher syndrome. Genet Otorhinolaryngol 2000;56:11–8.
10. McMoli TE, Ijaduola GT. Usher's syndrome in a Nigerian child. East Afr Med J 1981;58:967–70.
11. Nwosu SNN, Ndulue CU, Ndulue OI, Uba-Obiano CU. Usher syndrome in adult Nigerians: report of two cases. Orient J Med 2022;34:3–4
12. Hajali M, Fishman G. The prevalence of cystoid macular oedema on optical coherence tomography in retinitis pigmentosa patients without cystic changes on fundus examination. Eye 2009;23:915–9.
13. Flynn MF, Fishman GA, Anderson RJ, Roberts DK. Retrospective longitudinal study of visual acuity change in patients with retinitis pigmentosa. Retina 2021;21:639–64.
14. Hagiwara YS, Ogata K, Sugawara T, Hiramatsu A, Shibata M, Mitamura Y. Macular abnormalities in patients with retinitis pigmentosa: prevalence on OCT examination and outcomes of vitreoretinal surgery. Acta Ophthalmologica 2011;89:e122–5.
15. Testa F, Rossi S, Colucci R, *et al.* Macular abnormalities in Italian patients with retinitis pigmentosa. Br J Ophthalmol 2014;98: 946–50.
16. Fishman GA, Lam BL, Anderson RJ. Racial differences in the prevalence of atrophic-appearing macular lesions between Black and White patients with retinitis pigmentosa. Am J Ophthal 1994;118:33–8.
17. Rush RB, Rush SW. Bilateral lamellar macular hole surgery in retinitis pigmentosa. Ret Cases Brief Rep 2016;10:83–5.
18. Gaudio AR, Berson EL, Sandberg MA. Macular pseudoholes in retinitis pigmentosa. Inv Ophthal Vis Sci 2007;48:3726.
19. Coassin M, Mori T, Di Zazzo A, Sgrulletta R, Varacalli G, Bonini S. Lamellar macular holes: monitoring and management strategies. Clin Ophthalmol. 2019;13:1173–82.
20. Fuster-Garcia C, Garcia-Bohorquez B, Rodriguez-Munoz A, *et al.* Usher syndrome: genetics of a human ciliopathy. Int J Mol Sci 2021;22:6723.
21. Spandau UH, Rohrschneider K. Prevalence and geographical distribution of Usher syndrome in Germany. Graefes Arch Clin Exp Ophthalmol. 2002;240:495–8.
22. Kitamura K, Steel KP. Genetic heterogeneity of Usher syndrome. Adv OtoRhino Laryngol 2000;56:11–8.
23. Adeyemo AA, Amodu OK, Ekure EE, Omotade OO. Medical genetics and genomic medicine in Nigeria. Mol Genet Genomic Med 2018;6:314–21.