USHER SYNDROME WITH ATYPICAL RETINITIS PIGMENTOSA: A RARE CASE REVIEW.

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- THERE ARE NO FINANCIAL INTERESTS.
- INSTITUTIONAL ETHICS COMMITTEE APPROVAL HAS BEEN TAKEN.

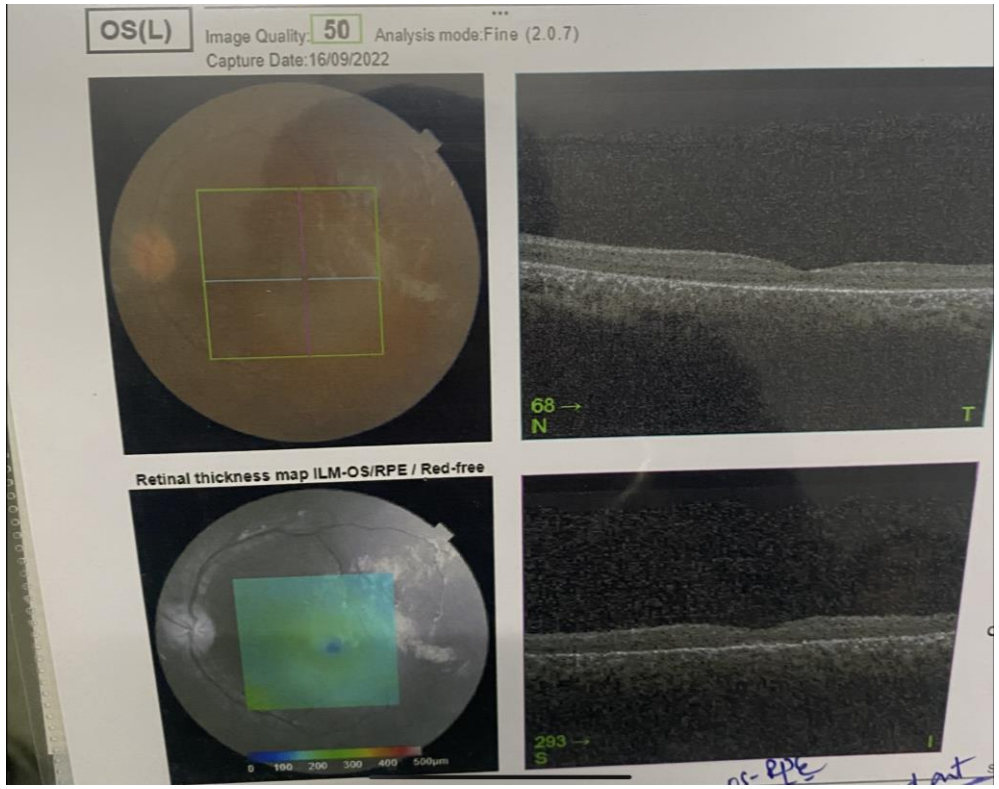
INTRODUCTION

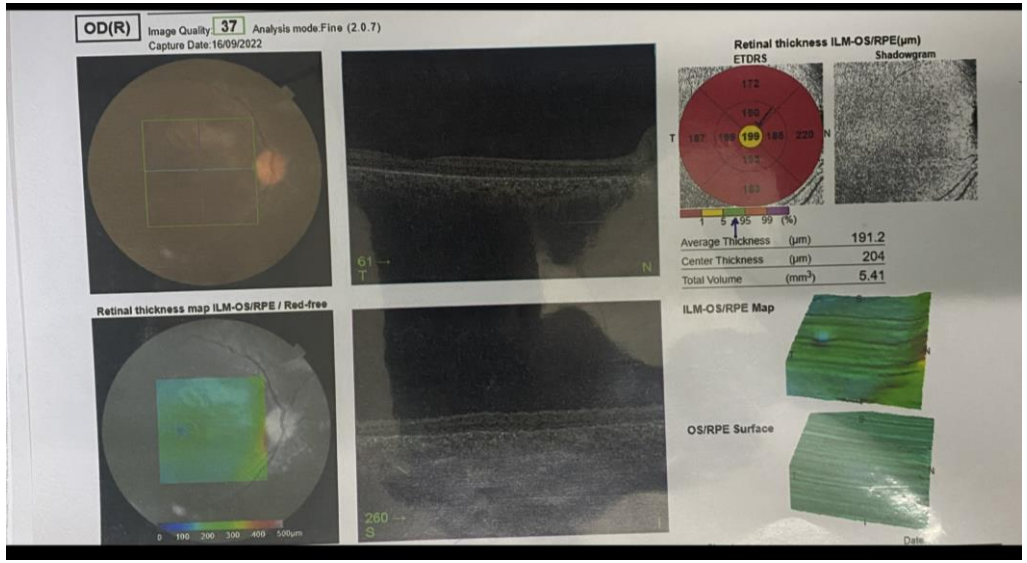
- Usher Syndrome, also known as Hallgren syndrome, is a rare genetic disorder characterised by progressive vision and hearing loss.
- It is classified into three subtypes: I, II and III.
- Usher Syndrome Type I is the most severe subtype. It is characterized by profound congenital hearing loss, retinitis pigmentosa(RP), and absent vestibular function.
- Usher Syndrome Type II is less severe. Patients with this subtype have moderate-to-severe congenital hearing loss, RP, and normal vestibular function.
- Usher Syndrome Type III involves progressive hearing loss, RP, and varying degrees of vestibular dysfunction. Onset for type III is typically within the second to fourth decades of life. These patients also tend to have better vision than the other subtypes.[1]

MATERIALS AND METHODS

- A 35 year old female came to ophthalmology OPD for visually handicapped certificate.
- She was found to have visual acuity of 6/36 in both eyes which was not improving with pin hole
- Rest anterior segment was found to be normal
- On fundus examination OU optic disc had waxy pallor and severe arteriolar attenuation with no pigmentory changes.

- Perimetry and OCT was also done for the patient
- On perimetry she was found to have constricted visual fields with central vision spared.
- OCT showed thinning of outer nuclear layer and loss of delineation between ellipsoid zone, cone outer segment tips and RPE.
- Patient gave history of hearing loss since birth.
- She was referred to ENT for hearing assessment and was found to have severe sensorineural hearing loss.
- The parents had a history of consanguinity.



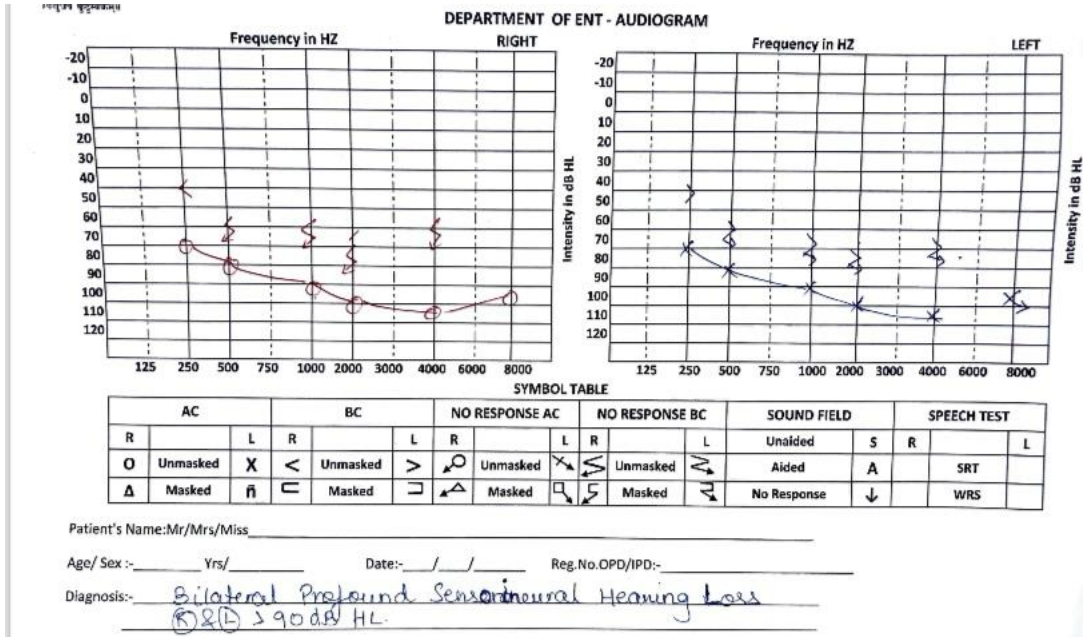


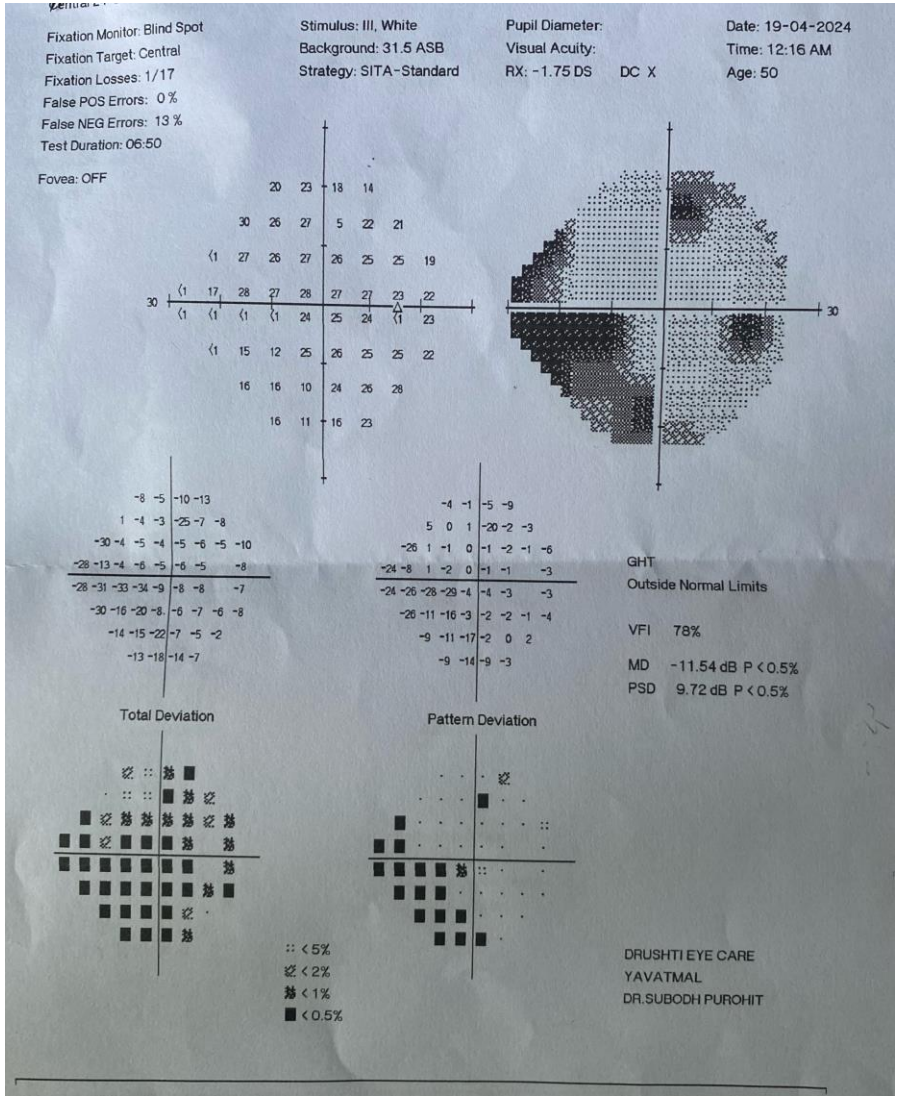
Left: OCT of left eye showing thinning of Outer nuclear layer

Right: OCT of right eye showing thinning of outer nuclear layer

Left: fundus photo of right eye showing waxy pallor of optic disc arteriolar attenuation and no bony spicules.

Right:fundus photo of left eye showing waxy pallor of optic disc arteriolar attenuation and no bony spicules.





Left : audiogram of the patient showing profound

SNHL

Right:perimetry of right showing field constriction, similar findings were noted in left eye

DISCUSSION

- Based on above findings of waxy pale optic disc, severe arteriolar attenuation and no pigmentory changes in peripheral fundus and also based on OCT and perimetry findings, diagnosis of Atypical retinitis pigmentosa was made.
- Combined with her above mentioned sensorineural hearing loss, a diagnosis of Usher Syndrome was made.
- The mode of inheritance for Usher Syndrome is autosomal recessive. 13 genes and 16 loci have been found to contribute to Usher Syndrome.[2]
- Hearing loss in Usher Syndrome is due to a defect in the inner ear hair cells. Mutations in the USH genes cause deficits in hearing and vestibular function. Additionally, transmission of neuronal signals from inner ear hair cells is potentially compromised by USH mutations.
- Vision loss results from retinitis pigmentosa (RP) which involves the degeneration of retinal cells. Research on the exact mechanism of vision loss from Usher Syndrome is ongoing, but current literature revolves around the interface of the inner and outer segments of photoreceptors.
- Current theories propose that defects in the USH1 protein complex leads to disruption in the organization of the pericilieary membrane complex(PMC).[3] USH1C mutation is also conjectured to produce truncated harmonin protein in abnormal splicing that causes retinal degeneration.

CONCLUSION

- Usher I and II have congenital onset of hearing loss. As a result, hearing aids provide limited benefit. Because of the gradual progression of hearing loss in Usher Syndrome Type III hearing aids are often used successfully. Usher types I, II, and III have progressive loss of visual acuity by midlife, which is at present not curable, low vision aids may be helpful in later stages.
- Usher syndrome is a rare genetic condition that involves various degrees of hearing and vision loss over time. It is classified into three subtypes (I, II, and III) according to onset and severity of symptoms. Type II does not have vestibular dysfunction. It is at present incurable. However, gene therapy is one of the potential treatment modalities that are being researched and developed for potential future cures.[4]

REFERENCES

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