Usher Sydrome: A Case Report.

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PURPOSE

Describe a case of usher syndrome

INTRODUCTION

Usher Syndrome was first described by Aldbrecht von Graefe in 1858 and later named by Charles Usher. It represents an autosomal recessive disease characterized by the combination of profound congenital sensorineural deafness and blindness due to retinitis pigmentosa, which is a hereditary retinal dystrophy caused by rod dysfunction. There are 3 subtypes of Usher Syndrome according to their clinical manifestations, with type II being responsible for 85% of cases. The main symptoms include difficulty adapting to night vision, decreased peripheral vision and changes in color perception.

METHODS

Medical records review

RESULTS

A 21-year-old boy complaining of visual loss for 3 years, mostly reporting peripheral vision loss. He denied family history. Patient with a history of hearing impairment since childhood.
On ophthalmological examination, he presented best-corrected visual acuity of 20/40 in both eyes, on fundus examination, the presence of a pale optic disc, vascular thinning and bone spicules. In fluorescein angiography, areas of hyperfluorescence are observed interspersed with areas of hypofluorescence in the periphery, showing bone spicules.

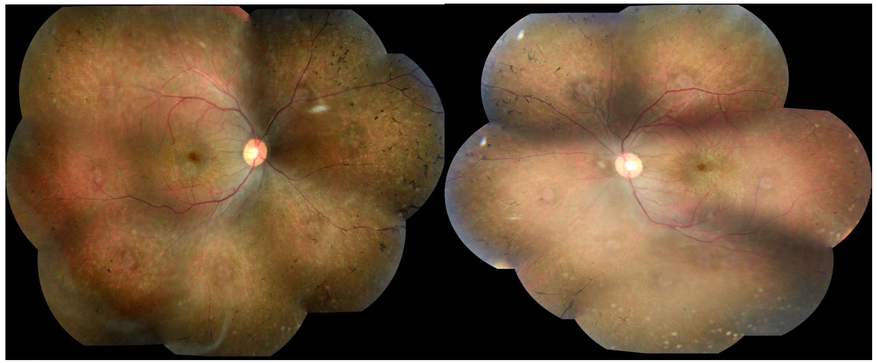


Fig. 1 and 2: retinography of both eyes of the patient.

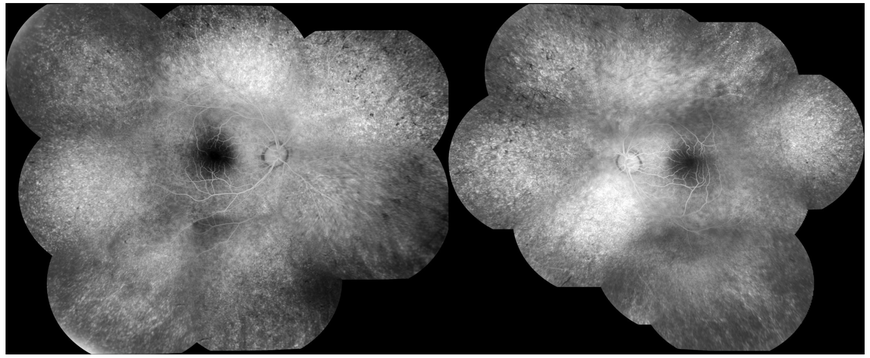
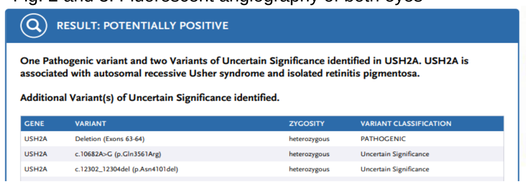


Fig. 2 and 3: Fluorescent angiography of both eyes



RESULT: POTENTIALLY POSITIVE

One Pathogenic variant and two Variants of Uncertain Significance identified in USH2A. USH2A is associated with autosomal recessive Usher syndrome and isolated retinitis pigmentosa.

Additional Variant(s) of Uncertain Significance identified.

GENE	VARIANT	ZYGOSITY	VARIANT CLASSIFICATION
USH2A	Deletion (Exons 63-64)	heterozygous	PATHOGENIC
USH2A	c.10682A>G (p.Gln3561Arg)	heterozygous	Uncertain Significance
USH2A	c.12302_12304del (p.Asn4101del)	heterozygous	Uncertain Significance

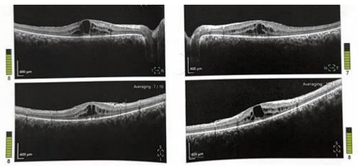


Fig. 4 and 5: Genetic Test and OCT of the right and left eye.

Optical coherence tomography of both eyes showed areas of intraretinal hyporeflectivity characterizing macular edema that was being treated with a carbonic anhydrase inhibitor.
Genetic testing confirmed the diagnosis through the pathogenic mutation of the USH2A gene (exons 63-64) in heterozygosis. The patient is still undergoing treatment and follow-up at this service.

DISCUSSION

Nine genes causing Usher Syndrome have been identified so far: MYO7A, USH1C, CDH23, PCDH15 and SANS for Usher type 1; USH2A, ADGRV1 and WHRN for Usher type 2; CLRN1 for Usher type 3. Deficiencies such as visual and hearing loss lead to important consequences in the socio-environmental scope of an individual, impairing the quality of life. It has already been observed that the gradual loss of vision simultaneously accompanies behavioral and psychological symptoms. Thus, early diagnosis is essential and thus clinical support for these patients and their families in a multidisciplinary way.

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