Retinitis Pigmentosa and USH2A gene identified: A Case Report.

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PURPOSE

Describe a case of retinitis pigmentosa with USH2A gene identified

INTRODUCTION

Retinitis pigmentosa (RP) is one of the most common incurable eye diseases, and has clinical and genetic heterogeneity. The inheritance patterns are autosomal recessive (50–60%), autosomal dominant (30–40%), and X-linked (5–15%). Rod cells are mainly affected, result in poor night vision, night blindness (nyctalopia), and gradual peripheral vision loss

METHODS

This is a retospective case chart review study, , patient treated in 2022, at the Suel Abujamra Institute, in São Paulo-SP

RESULTS

Male, 56 years old, previously healthy, complained of worsening visual acuity in both eyes for 3 months. He also presents night blindness (nyctalopia) a few years ago. In the ophthalmologic examination, he had best corrected visual acuity (BCVA) in the right eye: 20/40, in the left eye: 20/50. . Anterior segment and intraocular pressure did not have significant findings in both eyes. Funduscopy revealed mild pallor of the papilla, free macula, severe arteriolar narrowing and bone spicules in the middle and periphery in both eyes.

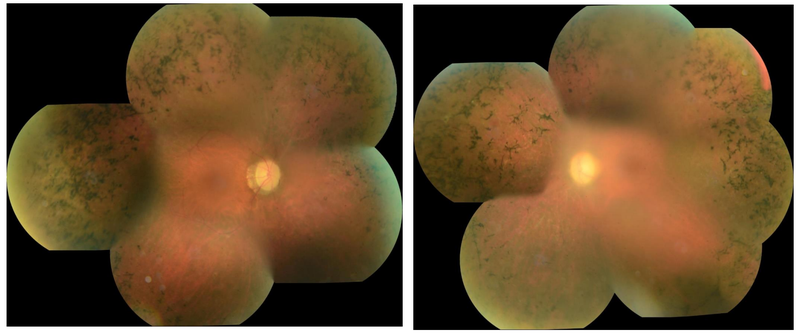

Fig. 1 and 2: retinography of the both eyes of the patients.

RESULT: POSITIVE

Two Pathogenic variants 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	c.13547G>C (p.Gly4516Ala)	homozygous	PATHOGENIC
AHI1	c.362A>G (p.Lys121Arg)	heterozygous	Uncertain Significance
CA4	c.705G>C (p.Trp235Cys)	heterozygous	Uncertain Significance
CA4	c.761A>C (p.Gln254Pro)	heterozygous	Uncertain Significance
IFT80	c.869A>G (p.Asn290Ser)	heterozygous	Uncertain Significance
MTPAP	c.1718G>A (p.Ser573Asn)	heterozygous	Uncertain Significance
SDCCAG8	c.968G>A (p.Arg323Lys)	heterozygous	Uncertain Significance

Fig. 3: Genetic test.

The patient did not present hearing loss and no relatives with the disease. The diagnostic hypothesis of Retinitis Pigmentosa was proposed, so a simple retinography exam was performed for documentation and salivary secretion was collected for genetic testing. The test identified two pathogenic variants, c.13547G>C (p. Gly4516Ala) homozygous, in the gene USH2A, that is a gene associated with autossomal recessive Usher syndrome and isolated retinitis pigmentosa.

DISCUSSION

Two major genes are responsible for autosomal recessive (AR) inheritance in RP, USH2A and EYS. *USH2A* causes 10–15% of AR cases and 30–40% of Usher syndrome type 2 cases, including moderate to severe congenital deafness and progressive RP starting in adolescence. In this case report, missense mutations in the *USH2A* cause RP isolated, without hearing loss..This presentation is the least common for this gene, but is currently widely described in cases of AR RP, which reinforces the importance of genetics to better understand the disease, its familial genetic programming and studies of possible treatments in the future.

BIBLIOGRAPHY

•Newton F, Megaw R. Mechanisms of photoreceptor death in retinitis pigmentosa. Genes (Basel). 2020;11(10):1120.

•Jauregui R, Park KS, , . Quantitative progression of retinitis pigmentosa by optical coherence tomography angiography. Nature. 2018;8:13130.

•Bocquet B, et al. mapping in autosomal recessive retinitis pigmentosa families detects novel mutations. Mol Vis. 2013;19:2487–2500