

Ocular presentation of Walker-Warburg syndrome with *POM2* mutation

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Access this article online	
Quick Response Code:	Website: www.ijo.in
	DOI: 10.4103/ijo.IJO_2128_21

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Received: 14-Aug-2021

Revision: 16-Oct-2021

Accepted: 27-Oct-2021

Published: 30-Jun-2022

Key words: Ocular manifestations, Walker-Warburg, foetal ultrasound

Walker-Warburg syndrome (WWS) is an autosomal recessive muscular dystrophy which is fatal in the first year of life and death often occurs in the first month of life.^[1,2] It is characterized by congenital muscular abnormalities, ocular anomalies, and brain malformation. The common ocular features include microphthalmos, cataract, microcornea, retinal dysgenesis, and glaucoma.^[1] However, this condition closely resembles other dystrophies like Fukuyama muscle dystrophy and muscle eye brain disease.^[3] The ocular manifestations help in establishing a diagnosis along with genetic analysis.

A 9-month-old male presented with enlarged eyes and a white reflex in both eyes which according to the parents was present since birth. On examination, the child had frontal

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Cite this article as: Sukhija J, Isher HK, Kaur S, Korla S, Kaur A, Raj S. Ocular presentation of Walker-Warburg syndrome with *POM2* mutation. Indian J Ophthalmol 2022;70:2626-7.



Figure 1: Frontal bossing is seen with wide intercanthal distance, upturned cilia, large cornea, and cataract

bossing low-set ears and large eyes and white reflex in both eyes [Fig. 1]. The ocular examination revealed a wide intercanthal distance and large corneal diameter (16 mm × 16 mm in the right eye and 13 mm × 12 mm in the left eye). In the right eye, the keratometry reading was 40.4 D × 34.9 D (avg 37.7 D) and in the left eye 40.2 D × 38.6 D (avg 39.4 D). The axial length was 30.9 mm in the right eye and 28.8 mm in the left eye. The anterior chamber depth was 4.86 mm and 4.80 in the right and left eyes, respectively. The lens thickness was 4.99 in the right eye and 4.93 in the left eye. The corneal thickness was 436 μm in the left eye and 555 μm in the right eye. The intraocular pressure was 40 mmHg in the right eye and 32 mmHg in the left eye. The axial length in the right was 30.9 mm and in the left eye was 28.8 mm. All these features were suggestive of buphthalmos. There was a total cataract in both eyes. Ultrasound of the posterior segment of the eye was unremarkable with no evidence of vitreous hemorrhage or retinal detachment. Previous records showed lissencephaly (prenatal ultrasound and foetal Magnetic Resonance Imaging) and features suggestive of the Dandy-Walker malformation. Ultrasound of the abdomen was normal. The child had severely delayed milestones and was born of consanguineous marriage. The genetic analysis revealed mutation c.1484G>A in the POM2 gene.

Discussion

Walker-Warburg syndrome is a clinical diagnosis which can be suspected prenatally and confirmed by genetic analysis. Our patient has survived 9 months and there was a delay in presentation to us because of the Coronavirus disease-19 (COVID-19) lockdown. Given the average survival time of such infants, it is rare to find such a case at 9 months. Detailed ocular findings have never been reported before. The ocular parameters measured in our case would help in narrowing the phenotype as there is an overlap among the other muscle

dystrophies. This differentiation helps in prognosticating life expectancy.^[4]

Buphthalmos, and that too bilateral, is an unusual manifestation of this syndrome as compared to microphthalmos which is seen in 38–62% of the affected infants.^[5-7] It is related to the arrest of the anterior chamber angle development after the fourth month of gestation. Posterior fossa anomalies in association with hydrocephalus detected during the gestation are not specific but coupled with ocular findings do help in arriving at a diagnosis. Other ocular features include cataract, colobomas and retinal dysplasia, retinal detachment, and optic nerve hypoplasia. We could not visualize the retina because of a significant cataract. The child last seen was awaiting clearance for surgery.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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