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A Congenital Monocular Aniridia - A Rare Case Report

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Abstract
Aniridia is rare genetic disorder characterized by partial or complete absence of iris, associated with other ocular features like cataract, glaucoma, corneal opacification, fovealhypoplasia; nystagmus.It commonly has autosomal dominance inheritance due to mutation of PAX6 gene on chromosome 11_p13. Congenital aniridia isusually bilateral in presentation, but my paper reports a rare case of unilateral aniridia in 17 years old female child based on the clinical features.

Key Words: *UnilateralAniridia, congenital,cataract, PAX6, strabismus,Enophthalmos*

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I. Introduction

Congenital aniridia is described as partial or total absence of iris, but gonioscopy almost always reveals an iris stump[1].Aniridia is due to mutations in the following genes –PAX6, FOXC1, PITX2, CYP1B1, FOXD3, and TRIM44. Aniridia is seen approximately 1.8/100000 live births. No gender or racial predilection has been described[1].It usually presents in early infancy .Aniridia occurs either as isolated ocular abnormality or with systemic involvement as WAGR syndrome (Wilmstumour, Aniridia, Genitourinary anomalies) and Gilliespie syndrome[2].

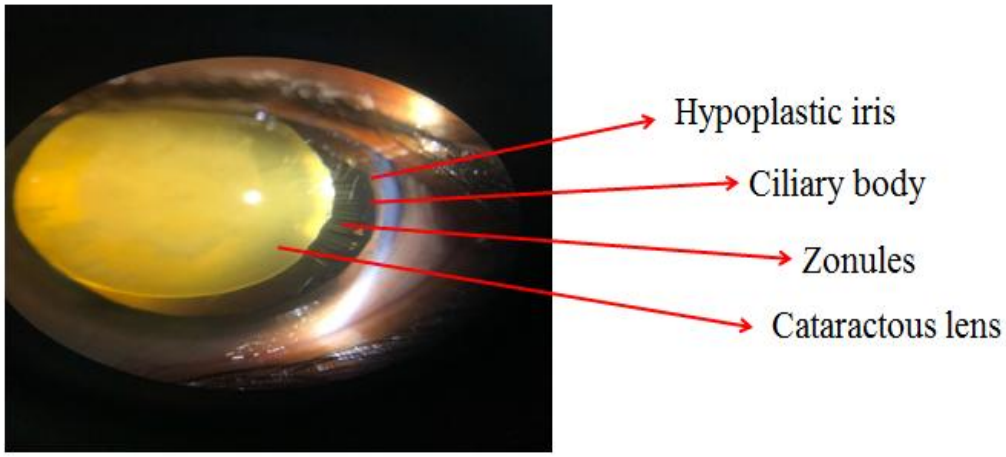
II. Case Report

A 17 years old female child presented to departmental OPD with progressive painless loss of vision in left eye since childhood.Detailed family and medical history was taken which revealed that she was born out of a non-consanguineous marriage.In family, sibblings and near relatives are normal. General examination is normal. Ocular examination:

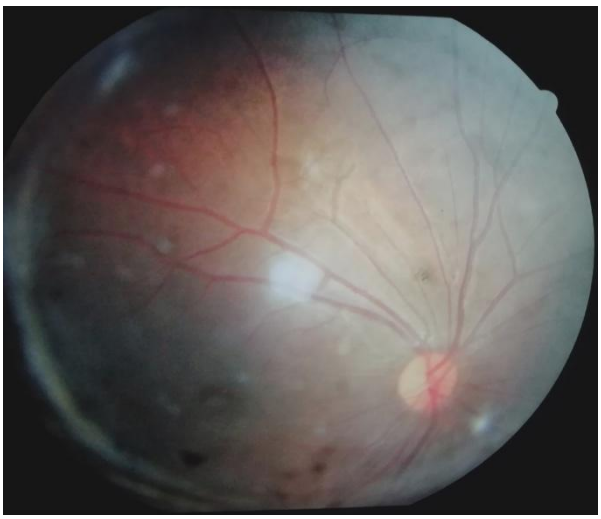
Gonioscopy : RE open angles. LE Rudimentary iris stump +

	OD	OS
Ocular alignment	Orthophoric, exophthalmometry reading-20mm	Exotropia 15^o, Enophthalmos+ with exophthalmometry reading 11mm
UCVA	6/6	NO PL ,PR
Cornea	Clear	Hazy
Anterior chamber	Normal depth	VH grade 2
Iris	Normal colour ,pattern	Hypoplastic iris stump
Lens	Clear	Cataract
Pupil	Normal ,reacting to light	Dilated ,fixed
Fundus	Normal	Details not seen due to hazy media
IOP	18.7mmHg	7.5mmHg
B scan	Normal	Hyperechoic lesions entire vitreous (VH)

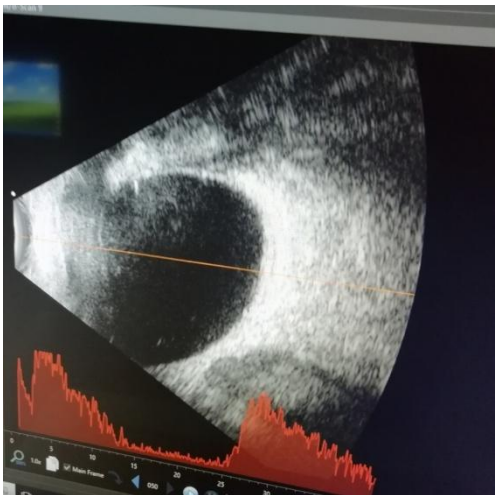
USG Abdomen showed no evidence of Wilms tumour.

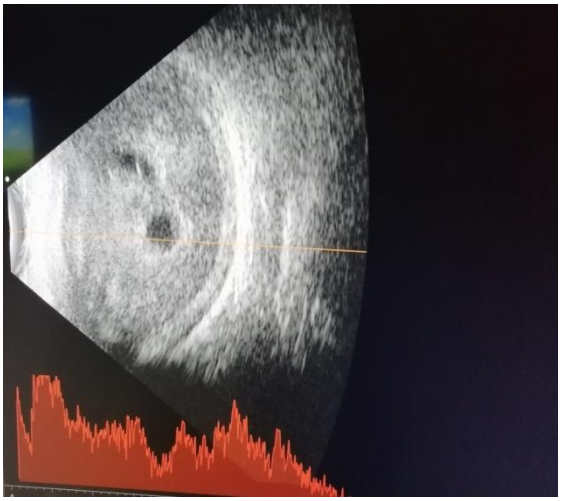


FUNDUS PICTURE: RE –Normal , LE – media hazy due to cataract and details not madeout



B SCAN: OD OS





III. Discussion

Aniridia is a rare, bilateral, hereditary absence of the iris and usually presents with a rudimentary stump of iris.Three phenotypes are recognised, of which autosomal dominant aniridia is most common (85%) and is not associated with systemic manifestations. The second phenotype is congenital sporadic aniridia, found in association with Wilms tumour, genitourinary anomalies and Mental retardation (WAGR syndrome). Third type is Autosomal recessive aniridia associated withcerebellar ataxia and mental retardation (Gillespie syndrome). Ocular features include pendular nystagmus, decreased vision, stabismus ,rudimentary iris, cataract,ectopia lentis,glaucoma,microphakia,corneal opacification, foveal andoptic nerve head hypoplasia[3] .The visual function in aniridia varies from normal to blindess.The more serious cases where blindness occurs are due to associated conditions like cataract,glaucoma,foveal hypoplasia,corneal opacity[4]. No specific treatment for aniridia except for glaucoma and cataract management.

IV. Conclusion

Congenital aniridia is a complex disease affecting the various segments of the eye.The visual prognostic of patients with aniridia is poor from early ages.Most of patients with aniridia presents with foveal hypoplasia since birth, associated with pendular nystagmus and strabismus[5].In my case patient had total loss of vision in left eye probably secondary tosecondary glaucoma, Retinal detachment with proliferative vitreoretinopathy leading to pthisis bulbi. Congenital aniridia is usually bilateral, but my case report concluded that congenital aniridia can also presents as unilateral isolated form which is rare.

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