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A Rare Case of Bilateral Congenital Aniridia

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Abstract: *Aniridia is a rare congenital bilateral condition that occurs as a result of abnormal neuroectodermal development secondary to a mutation in the PAX6 gene on chromosome 11p13 adjacent to WT1 gene mutation of which predisposes to Wilm's tumor. Our paper reports the diagnosis of this rare case in a 26 year old male patient based on clinical features.*

Key Words: *ANIRIDIA, MICROTIA, NYSTAGMUS*

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I. Introduction

Aniridia occurs as an Autosomal dominant disorder in 85% of cases and is not associated with any systemic manifestations.Sporadic cases accounts for 13% and includes WAGR(Wilm's tumour, Aniridia, Genitourinary abnormalities, Mental retardation)**.** It occurs as anAutosomal recessive disorder in 2% of casesassociated with cerebellar ataxia and mental retardation (Gillespie syndrome).

II. Case Report

A 26year old male patient presented with complaints of abnormal eye movements since birth, gradual outward deviation of the left eye for 15 years and decreased vision since a few months. There was a history of using spectacles for 15 years. There were similar complaints in father and lost vision in both eyes at 50 years. There were similar complaints in elder brother also.On general examination left microtia observed.The systemic examination was within normal limits. On local examination, Head posture wasnormal, and the face was symmetrical. Ocular alignment – HBT 35 degree and Cover test 75 PD exotropia. Ocular motility of both eyes: Ductions and versions were normal. There was horizontal jerky nystagmus with moderate amplitude and frequency.

	OD	OS
VA- UCVA	20/1200	20/1200
BCVA	20/125	20/125
NV	N6	N6
	OD	OS
1.Eyelids and adnexa	Normal	Normal
2.Conjunctiva and sclera	Normal	Normal
3.Cornea	Clear	Clear
4.Anterior chamber	Normal in-depth and contents PACD=1CT	Normal in-depth and contents PACD=1CT
5.Iris	Total hypoplasia	Total hypoplasia
6.Pupil	9mm	9mm
7.Lens	Blue dot cataract	Blue dot cataract
8.IOP(GAT at11:30 A.M)	19mm of Hg	19mm of Hg
9.Gonioscopy	Remnants of iris tissue in superior and temporal quadrants	Remnants of iris tissue in the nasal quadrant
10.FUNDUS		
Optic disc	Size, shape, color normal,Margins distinct, C:D 0.3:1 NRR healthy	Size, shape, color –normal Margins distinct,C:D 0.5:1 NRR healthy
Vessels	Arteries and veins normal A:V 2:3	Arteries and veins normal A:V 2:3
Macula	Foveal reflex dull	Foveal reflex dull
Background retina	Highly tessellated	Highly tessellated

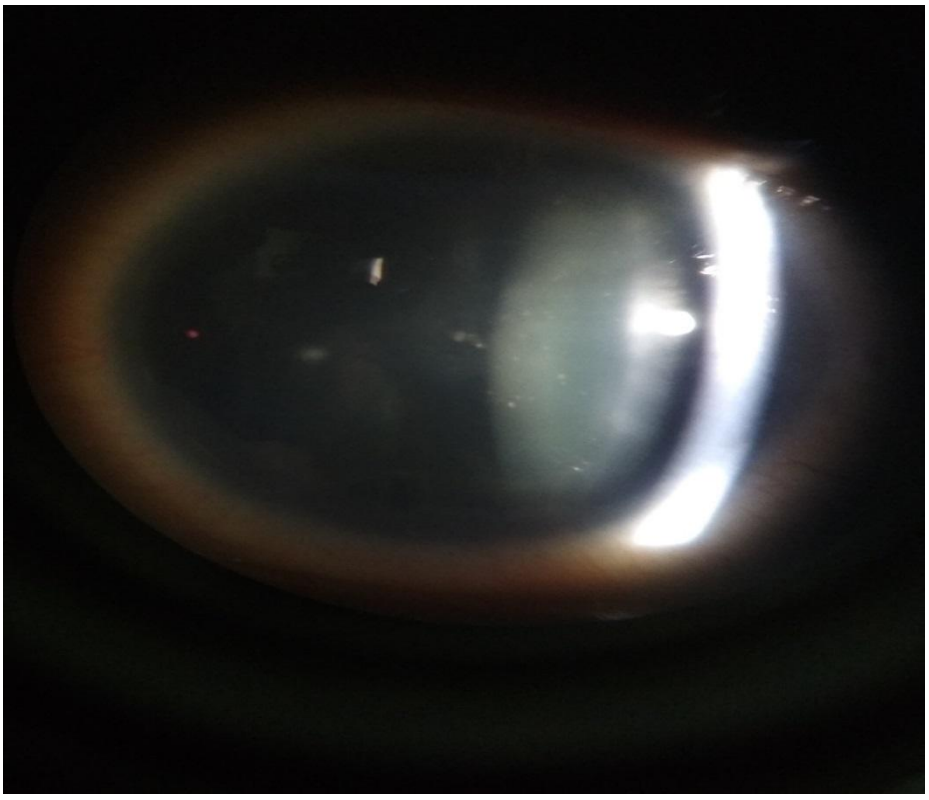


FIGURE 1: RIGHT EYE ANIRIDIA AND BLUE DOT CATARACT

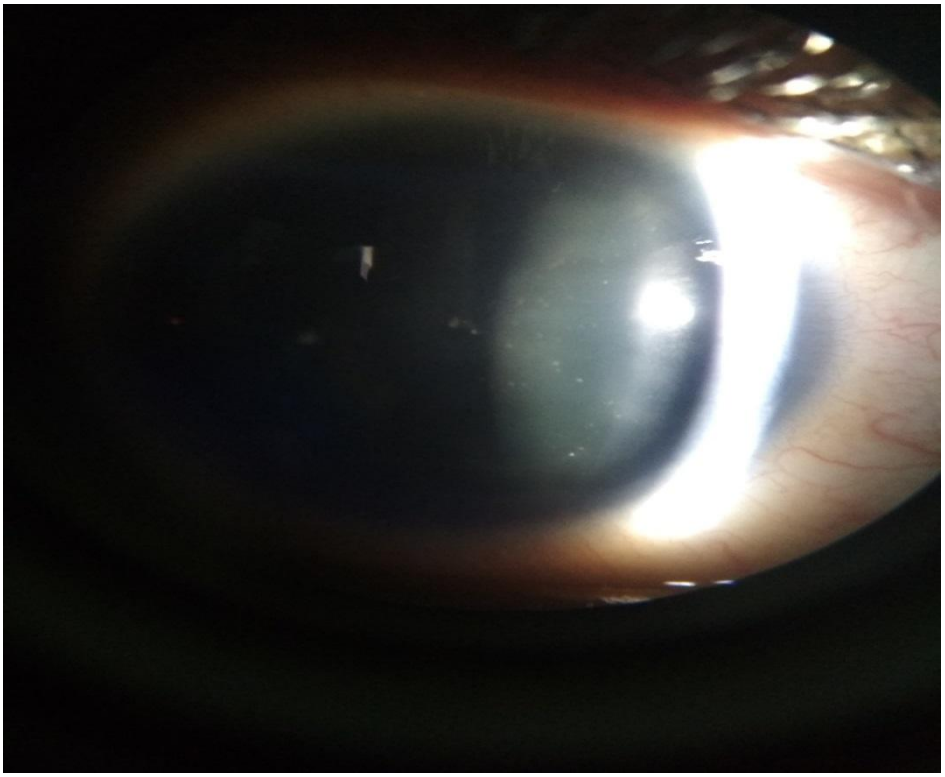


FIGURE 2: LEFT EYE ANIRIDIA AND BLUE DOT CATARACT

FIGURE 3: VISUAL FIELDS OF RIGHT EYE

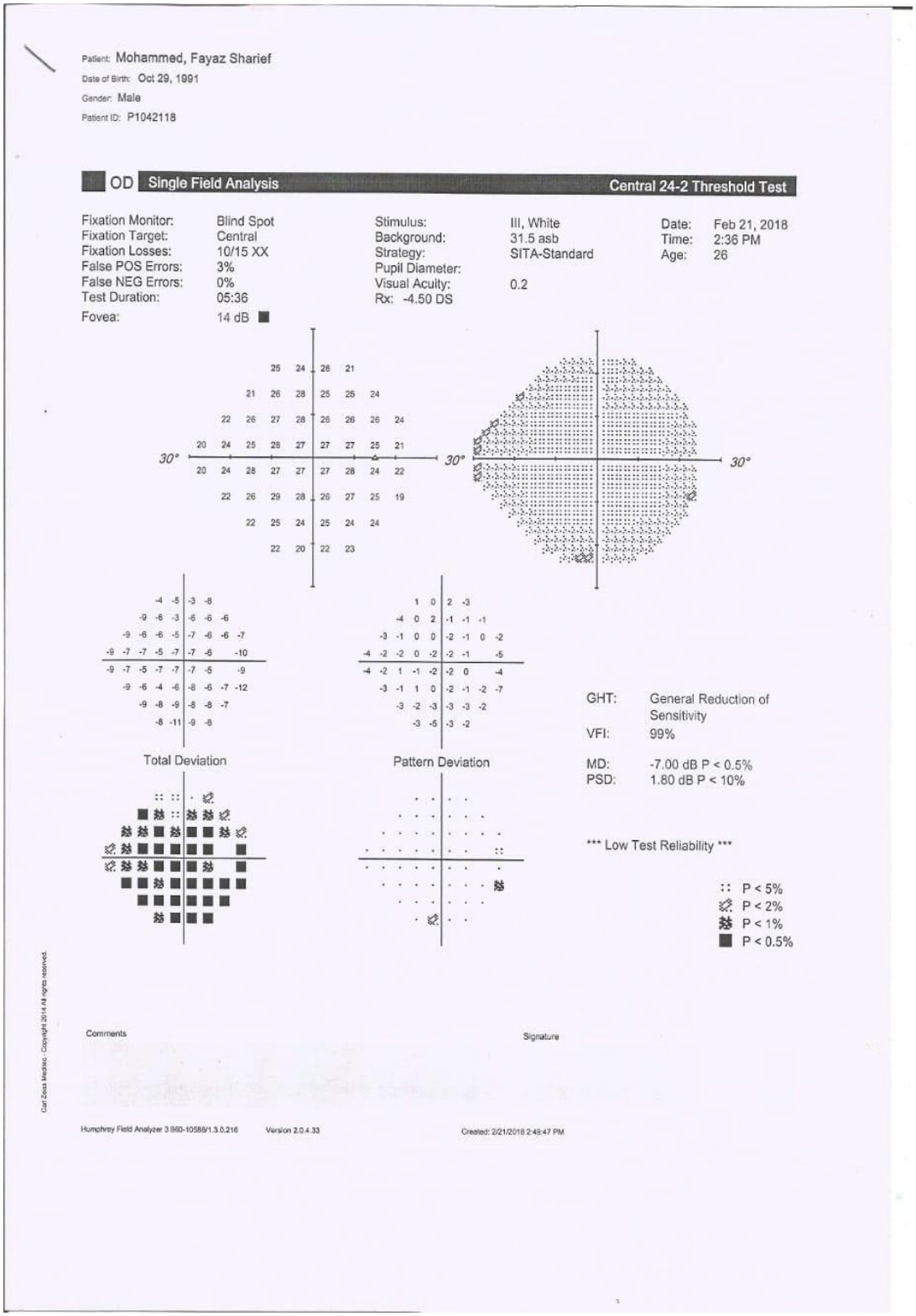


FIGURE 4: VISUAL FIELDS OF LEFT EYE

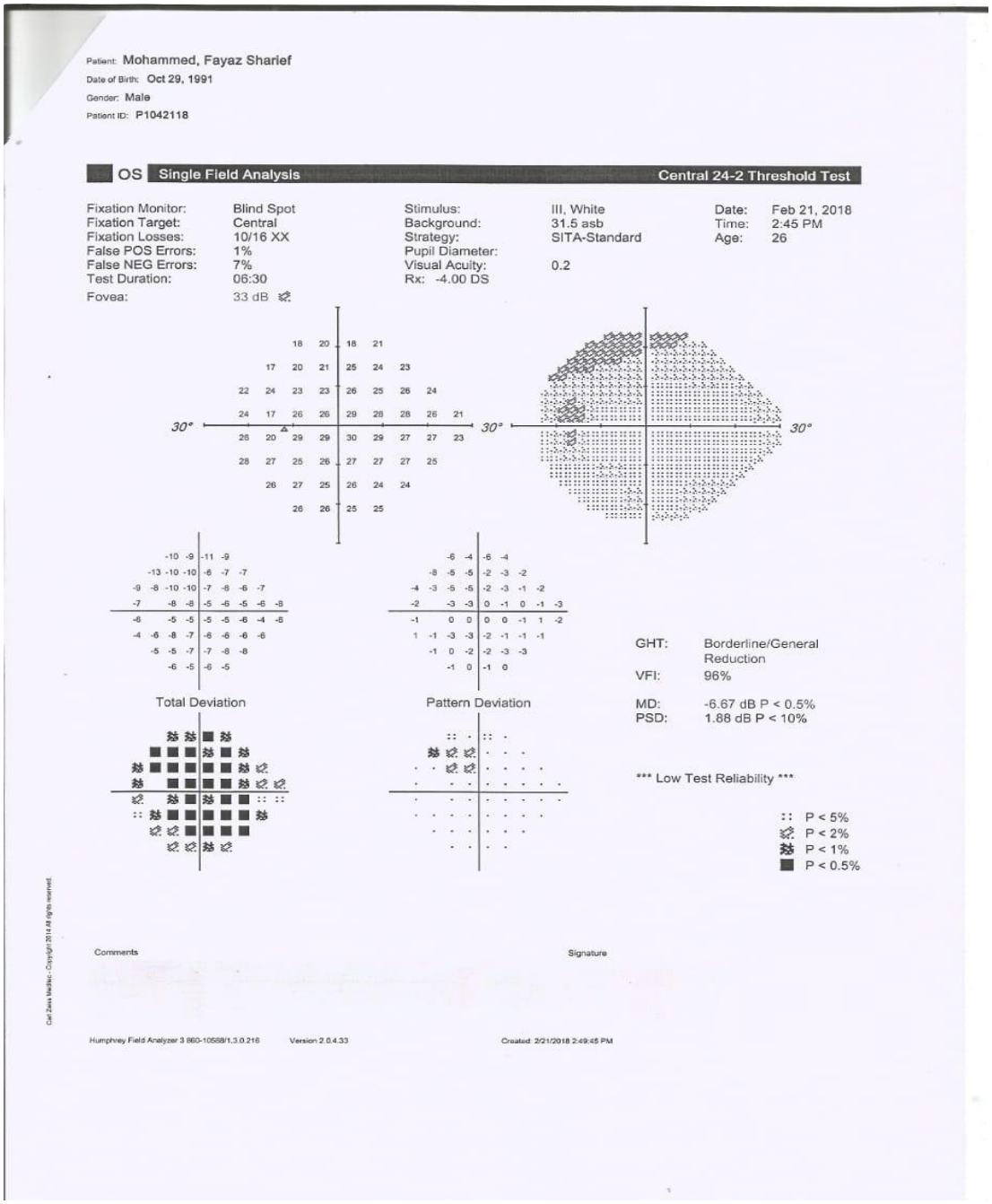
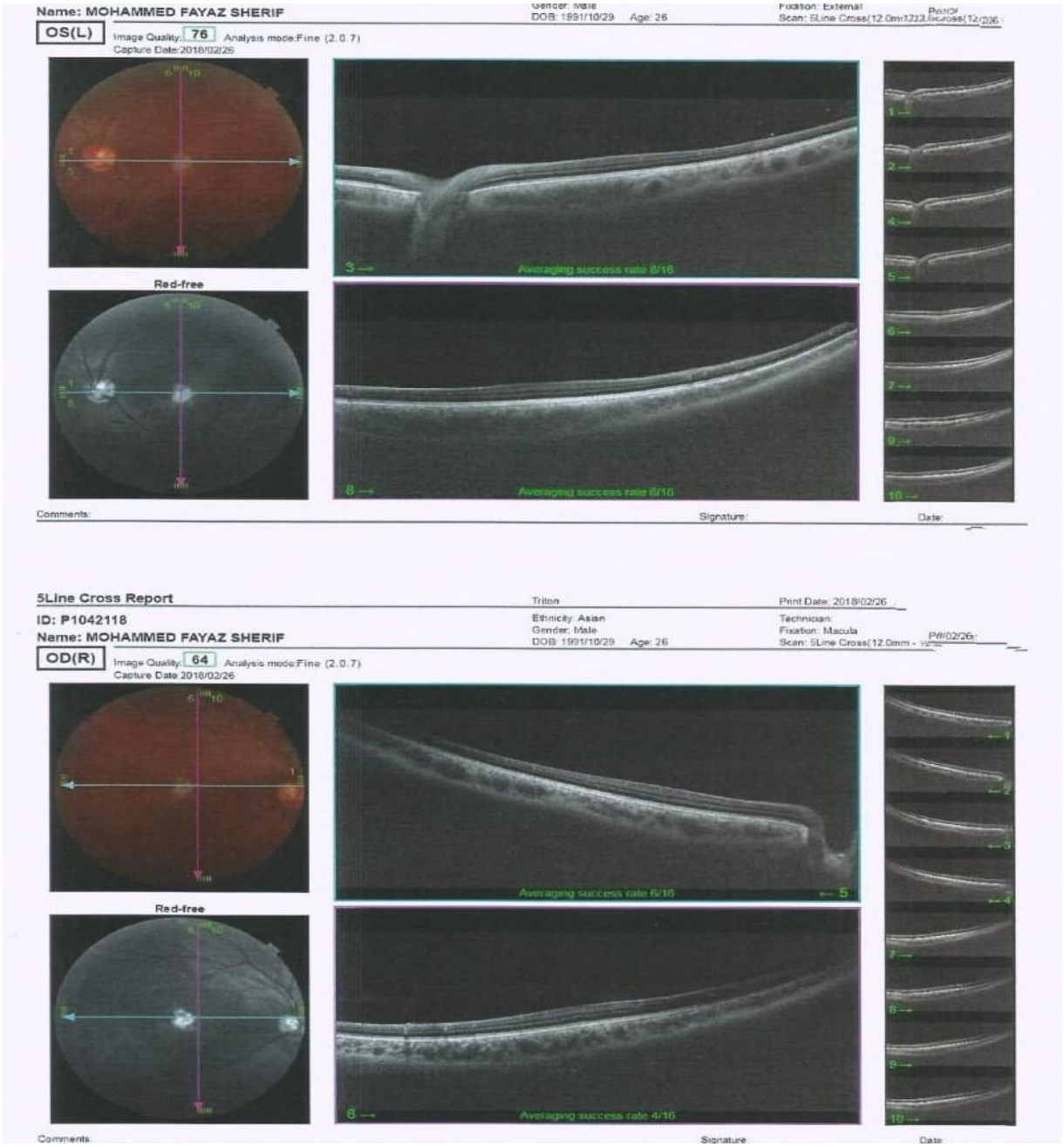


FIGURE 5: FUNDUS AND OCT PHOTOGRAPHS SHOWING FOVEAL HYPOPLASIA



The patient was started on Antiglaucoma medication. Contact lens trial was explained. Periodic follow up was explained to prevent for further progression of glaucoma

III. Discussion

Aniridia is a rare bilateral disorder characterized by the congenital absence of the normal iris.Ocular manifestations includemeibomian gland dysfunction, Tear film instability, dry eye, epithelial defects, limbal stem cell deficiency leading to conjunctivalisation of cornea, total corneal central stromal scarring and vascularization[1]. Cataract and subluxation of the lens or congenital absence or reabsorption of lens [2,3,4].Foveal and optic nerve hypoplasia, choroidal coloboma.Hypoplastic or rudimentary frill of the iris in total aniridia.Systemic associations include WAGR (Wilm's tumor, Aniridia, Genitourinary abnormalities) and cerebellar ataxia[5]. Glaucoma presents in late childhood or adolescence due to synechial angle closure secondary to rudimentary iris tissue contraction [6,7].Management of ocular manifestations include, Glaucoma by medical management which is ineffective in most of the cases. Goniotomy before the development of irreversible angle-closure[7]. Trabeculectomy or combined trabeculectomy– trabeculotomy or Glaucoma drainage devices are successful [8]. Diode laser cycloablation if all measures fail[9]. Painted contact lenses to create artificial pupil which

improves vision and cosmesis. Lubricants frequently for associated keratopathy. In cataract surgery tinted artificial lens implant to improve photophobia[10]. Prosthetic iris implant used in pseudophakic aniridia but worsens glaucoma. Limbal stem cell transplantation may be required. Refractive errors, amblyopia, squint should be managed aggressively.

IV. Conclusion

Aniridia is a rare congenital bilateral condition in which glaucoma and its surgical complications are the main causes of blindness. A prophylactic modified goniotomy has been advocated to prevent secondary glaucoma in young patients with aniridia. Our patient showed characteristic ocular manifestations of aniridia nystagmus, refractive error, strabismus, blue dot cataract, increased intraocular pressures, Foveal hypoplasia with positive family history and no systemic manifestations most probably of autosomal dominant type and was managed.

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