RESEARCH PAPER

Familial Blepharophimosis Syndrome with Associated Ocular Anomalies: A Case Series

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ABSTRACT
Blepharophimosis syndrome is a rare congenital eyelid disorder characterized by ptosis, telecanthus, and epicanthus inversus, commonly inherited in an autosomal dominant pattern due to mutations in the FOXL2 gene. We report a familial cluster with varying degree of presentation involving three individuals, a 55-year-old female(mother) and her offsprings (21-year-old male and 18-year-old male) —presenting with a myriad of ocular manifestations which include congenital ptosis , iris coloboma, aniridia, microcornea, and corneal opacity. Despite appropriate interventions, visual outcomes were suboptimal due to associated structural abnormalities. This case series highlights the phenotypic variability of familial blepharophimosis syndrome, an additional factor noted being consanguinity and it highlights the importance of early diagnosis, timely intervention, genetic counselling and a multidisciplinary approach to optimize functional and cosmetic outcomes.

Keywords: Blepharophimosis, Ptosis, Aniridia, Iris coloboma, Microcornea, FOXL2

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INTRODUCTION

Blepharophimosis syndrome is a rare congenital condition characterized by a classical tetrad of blepharophimosis, ptosis, epicanthus inversus, and telecanthus. It is most commonly inherited in an autosomal dominant pattern and is associated with mutations in the FOXL2 gene located on chromosome 3q23.

Two clinical subtypes are recognized:

Type I: Associated with premature ovarian insufficiency

Type II: Limited to eyelid abnormalities

Although primarily an eyelid disorder, association with additional ocular anomalies such as iris coloboma, aniridia, and microcornea is extremely rare or negligible and can significantly affect visual prognosis.

Case Presentation

Case 1

A 21-year-old male presented with progressive, painless diminution of vision in both eyes since childhood. He had a history of cataract surgery in the left eye 8 months prior. There was a positive family history of similar eyelid deformities, and the parents had a second degree consanguineous marriage.

Examination findings included severe bilateral ptosis with poor levator function, frontalis overaction, and nystagmus. Visual acuity was 6/36 in the right eye and 6/60 in the left eye,

Anterior segment findings showed ectropion uvea , posterior synechiae ,superficial puctate keratitis inferiorly in the right eye, and microcornea, inferior iris coloboma, and PCIOL with posterior capsular opacification in the left eye. Fundus was within normal limits.

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ANTERIOR SEGMENT	OD	OS
	Ptosis	Ptosis
	Horizontal nystagmus	Horizontal nystagmus
	Ectropion uvea, posterior synechiae	Inferior iris coloboma
	Superficial punctate keratitis inferiorly	Microcornea with 360 degree superficial vascularization
	Cataractous lens (posterior subcapsular cataract)	PCIOL with posterior capsular opacification

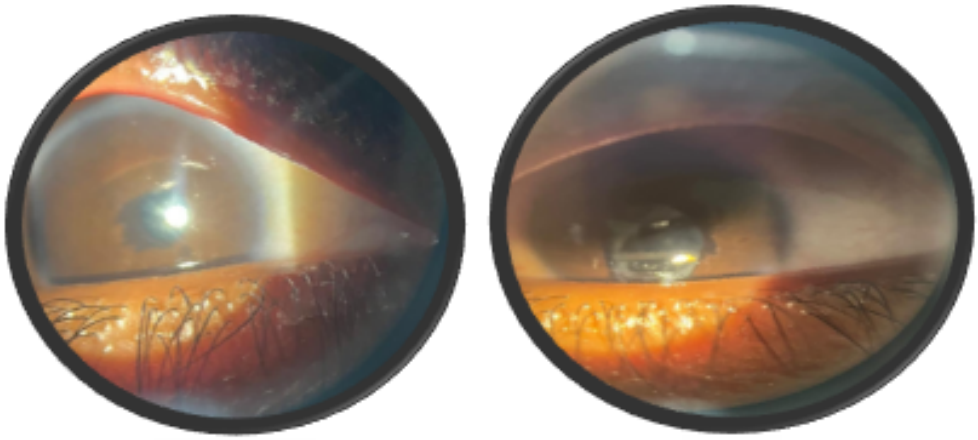
PTOSIS EVALUATION	OD	OS
MRD1	1mm	0mm
MRD2	4mm	3mm
MRD3	2mm	2mm
LPS	4mm (poor function)	4mm(poor function)

DIAGNOSIS-

Right eye moderate dry eye with posterior subcapsular cataract with nystagmus with blepharophimosis syndrome

Left eye moderate dry eye disease with typical iris coloboma with nystagmus with pseudophakia with posterior capsular opacification with blepharophimosis

The patient underwent Nd:YAG laser posterior capsulotomy in the left eye and received topical treatment for dry eye disease. At the subsequent follow-up, the BCVA was 6/36 in the right eye and 6/24 in the left eye.



Case 2

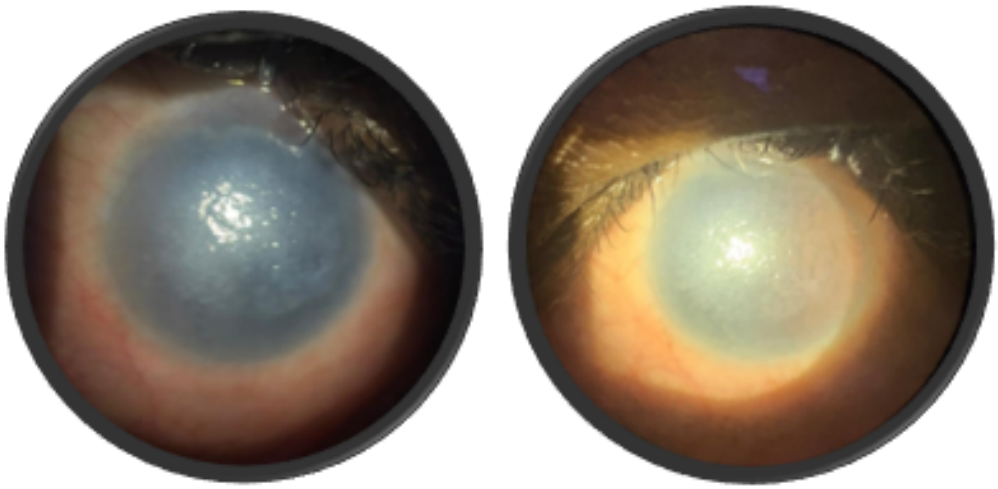
An 18-year-old male presented with bilateral diminution of vision since childhood, associated with congenital non-progressive ptosis and photophobia. Family history was positive.

Systemic findings included growth retardation(Height 148 cm , weight 38 kg with BMI 17.3kg/m^2) , dental crowding with enamel hypoplasia, and a high-arched palate.

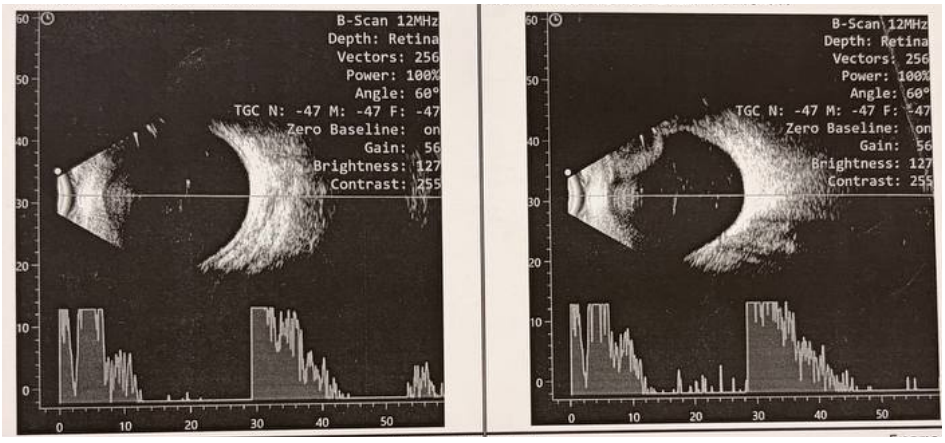
Patient had underwent right eye cataract surgery 12 years ago

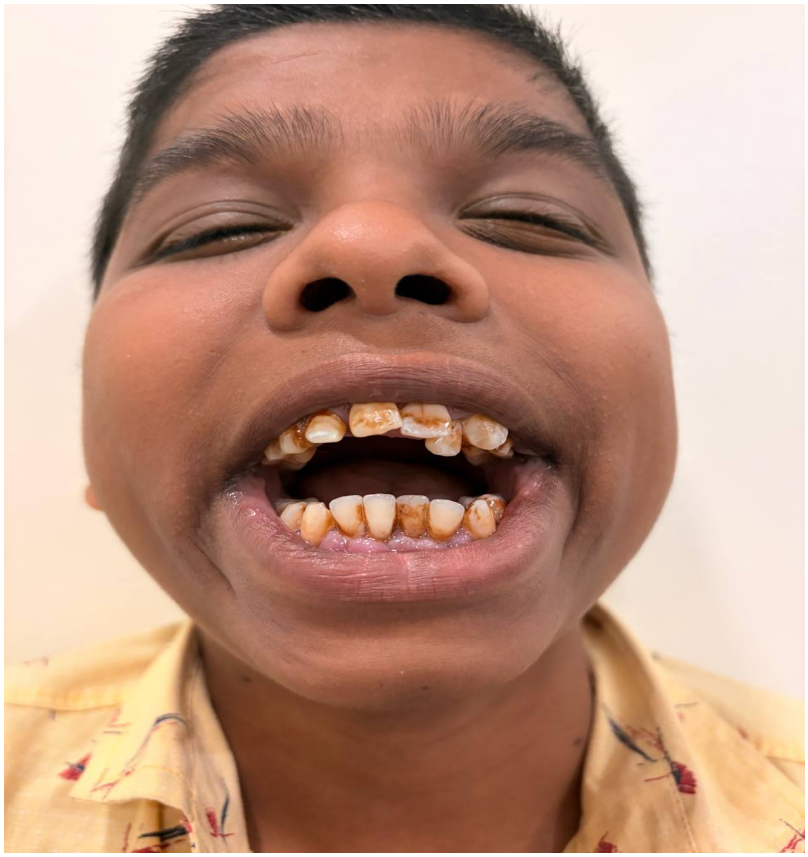
Ocular findings included severe bilateral ptosis, with mebomian gland dysfunction ,bilateral superficial punctate keratitis, and aniridia . Visual acuity was CF 3 m in right eye and hand movements close to face in left eye with BCVA 6/36 in right eye and HMCF in left eye

ANTERIOR SEGMENT	OD	OS
	Ptosis	Ptosis
	Horizontal nystagmus	Horizontal nystagmus
	Aniridia	Aniridia
	Superficial punctate keratitis , 360 degree corneal vascularisation	Superficial punctate keratitis , 360 degree corneal vascularisation
	PCIOL with posterior capsular opacification	Cataractous lens



B scan of both the eyes was within normal limits





Diagnosis-Right eye with severe Mebomian gland dysfunction with severe dry eye disease with pseudophakia with posterior capsular opacification with nystagmus with aniridia with blepharophimosis syndrome

Left eye severe Mebomian gland dysfunction with severe dry eye disease with mature cataract with aniridia with nystagmus with Blepharophimosis syndrome

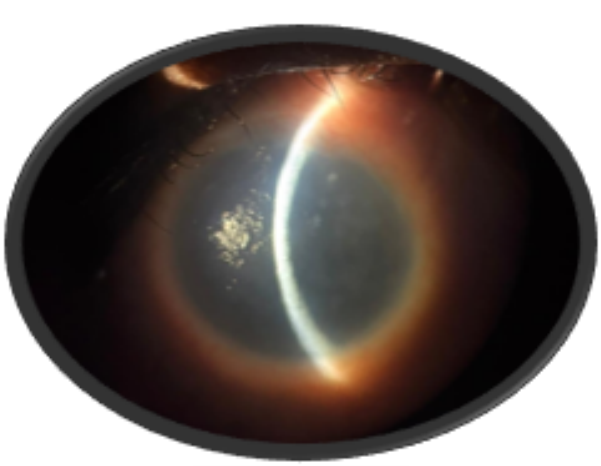
Patient was started on topical medications for dry eye disease and was advised left eye cataract surgery

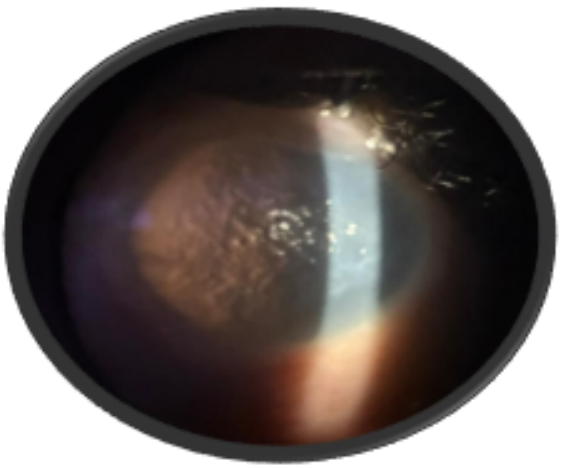
Case 3

A 55-year-old female presented with ptosis and progressive visual impairment since childhood.

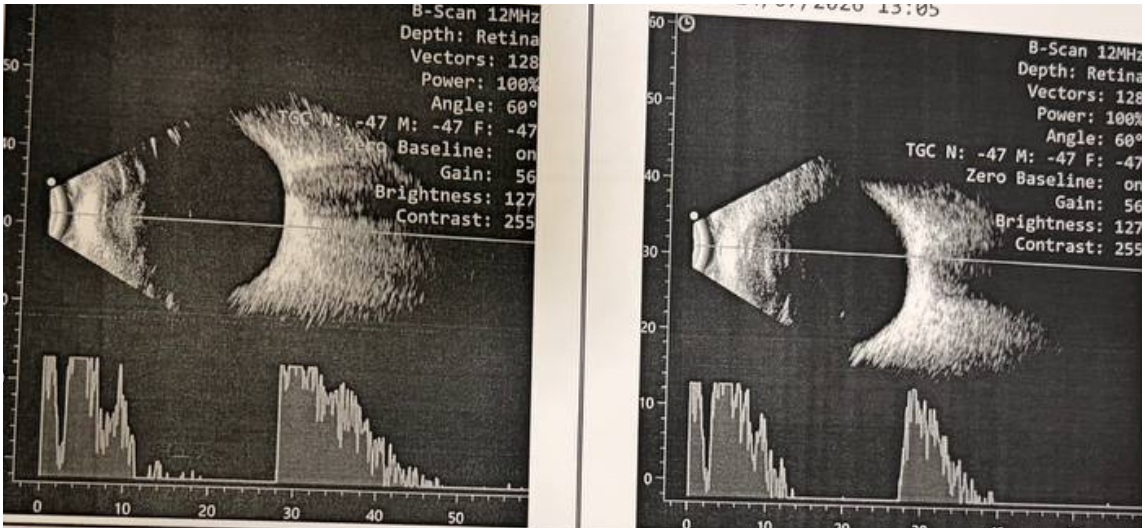
On examination, the patient had severe ptosis in the right eye and moderate ptosis in the left eye, along with bilateral corneal opacity, aniridia, and nystagmus. The patient had undergone bilateral cataract surgery 16 years previously. There was a history of trauma to the left eye one year prior to presentation. At presentation, the visual acuity was counting fingers at 3 m (CF 3 m) in the right eye and perception of light positive (PL+) with accurate projection of rays in all quadrants in the left eye. Following management, the best-corrected visual acuity (BCVA) improved to 6/24 in the right eye.

ANTERIOR SEGMENT	OD	OS
	Ptosis	Ptosis
	Horizontal nystagmus	Horizontal nystagmus
	Aniridia	Aniridia
	Superficial punctate keratitis , 360 degree corneal vascularisation	Superficial punctate keratitis , 360 degree corneal vascularization with anterior stromal dystrophy
	PCIOL	Dislocated PCIOL





B scan of both eye was within normal limits.



Diagnosis-Right eye severe meibomian gland dysfunction with severe dry eye disease with Aniridia with pseudophakia with nystagmus with blepharophimosis syndrome

Left eye eye severe meibomian gland dysfunction with severe dry eye disease with Aniridia with pseudophakia with nystagmus with blepharophimosis syndrome

Patient was started on topical treatment for dry eye disease

DISCUSSION

This case series demonstrates autosomal dominant inheritance with intrafamilial phenotypic variability. In addition to classical eyelid features, rare ocular anomalies such as iris coloboma, aniridia, microcornea, dry eye disease , with evidence of cataract were observed.

Iris coloboma results from defective closure of the embryonic fissure and is often associated with other ocular malformations. These anomalies significantly limit visual potential.

Early-onset ptosis increases the risk of amblyopia, and associated anomalies further worsen prognosis. Consanguinity may contribute to increased phenotypic severity.

MANAGEMENT

Management is multidisciplinary and staged. It includes ptosis correction (frontalis sling or levator resection), amblyopia therapy, and cataract surgery when indicated.

Surgical sequencing involves early correction of epicanthus and telecanthus followed by ptosis correction. In adults, management focuses on functional and cosmetic rehabilitation.

CONCLUSION

Familial blepharophimosis syndrome shows significant phenotypic variability and may be associated with vision-limiting ocular anomalies. Early diagnosis, timely intervention, and genetic counseling are essential. Visual prognosis remains guarded due to associated structural abnormalities.

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