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ANTERIORLY DISLOCATED LENS IN WEILL MARCHESANI SYNDROME

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Abstract: Ectopia lentis has many systemic associations, Weill Marchesani syndrome being one of them. It is a rare connective tissue disorder, characterised by a small, spherical lens (Microspherophakia); short fingers (Brachydactyly) and short stature (Brachymorphia). We present a case of the same, in a 55 year old woman who presented with painful diminution of vision and her son noticed cataract in her left eye. She had undergone cataract surgery for the right eye previously. On examination, the best corrected visual acuity was 6/6 in right eye, and perception of hand movements in left eye. Anterior segment of left eye showed mild central corneal edema, anterior chamber reaction and anteriorly dislocated spherical cataractous lens. She had short stature and short stubby fingers. A clinical diagnosis of Weill Marchesani syndrome was made. She underwent lens extraction with ACIOL implantation. Post operatively, corneal edema resolved and visual acuity was 6/9. Thus, prompt lens extraction is needed to prevent corneal endothelial damage. We report a case of anterior dislocated cataractous lens in an elderly lady.

Key Words: Weill Marchesani syndrome, Anterior dislocation of lens, microspherophakia, ectopia lentis

INTRODUCTION

Ectopia lentis can be due to inherited and acquired causes. Inherited causes and predisposing factors include connective tissue disorders like Marfan syndrome, Homocystinuria, Weill Marchesani syndrome, Ehlers Danlos syndrome, hyperlysinemia and Sulfite oxidase deficiency.

Described first in 1930s by Weill[1] and Marchesani[2], separately, Weill Mrachesani syndrome is a rare connective tissue disorder characterized by a triad of features: Bradymorphia, brachydactyly and microspherophakia. It has autosomal dominant as well as autosomal recessive inheritance.[3-5] These patients usually report in the second or third decade with lenticular myopia and pupillary block. We present a case of a woman in her sixth decade, who presented with an anteriorly dislocated cataractous lens.

Case description

A 55 year old lady was brought to the outpatient department by her son with history of noticing cataract in her left eye which he noticed one week back. She had history of pain and redness since around two weeks followed by progressive, diminution of vision. She underwent cataract surgery in the right eye 2 years back for similar complaints. There was no history of trauma. On examination, best corrected visual acuity 6/6 in right eye and perception of hand movements in the left eye. On examination, right eye had a PCIOL insitu. Left eye showed mild circumcorneal congestion, mild central corneal edema due to corneolenticular touch, a deep anterior chamber with cells and flare of grade 3+ was present and it contained a dislocated spherical cataractous lens of diameter 5mm. [Fig 1] Intraocular pressures were 12mm and 14mm Hg. Fundus in the right eye was unremarkable, while that in left eye could not be viewed. General examination revealed short stubby fingers and her height was 143cm. [Fig 2 and 3] Axial length of right eye was 21.87mm and left eye was 22.01mm.

Fig. 1: Anteriorly dislocated spherical, small, cataractous lens with corneal edema

Fig. 2: Short and stubby fingers

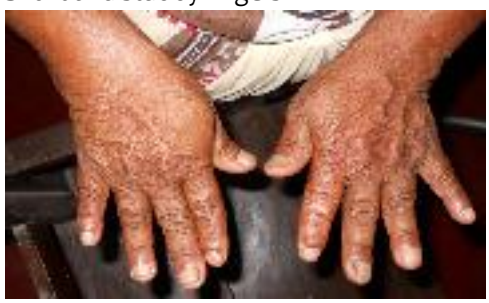

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Fig. 3: Short stature

She was started on topical steroids preoperatively. She underwent lens extraction, anterior vitrectomy with ACIOL implantation of 20D. She was continued postoperatively on topical steroids and tapered at the end of 6 weeks. Post operatively, her vision improved to 6/9. Fundus was normal, there was no evidence of glaucomatous cupping.

DISCUSSION

The diagnostic criteria for Weill Marchesani syndrome include, (i) short stature, (ii) Brachydactyly and (iii) microspherophakia and/or ectopia lentis.[6] other systemic associations are joint stiffness, mental retardation and heart defects.

Inheritance can be autosomal recessive, autosomal dominant or sporadic. Inversion of long arm of chromosome 15 has been reported with the incidence of Weill Marchesani syndrome.[7] It is thought to be a developmental anomaly affecting the ciliary body and the zonules. Mutations can affect the protein fibrillin-1 (FBN 1) on the long arm of chgromosome 15.[8] Our patient's height was 143cm, and also, her son had similar morphological features. His ocular examination was unremarkable.

It usually presents in the first or second decade, with progressive lenticular myopia and angle closure glaucoma due to pupillary block by the microspherophakic lens. The hypoplastic ciliary body, elongated zonules and spherophakic crystalline lens can be demonstrated by using a Scheimpflug imaging.[7] A pupil block can be induced by using mydriatics like cyclopentolate which does not reverse with pilocarpine induced miosis[9]. Use of miotics causes forward displacement of lens iris diaphragm and cause inverse glaucoma in such patients.

Treatment depends on the clinical condition. In case of increasing lenticular myopia only, prophylactic iridotomy can be performed in both the eyes provided the angles are open and there is no raised IOP. In case of occludable angles and pupil block, phacoemulsificaton of the lens with PCIOL implantation can be done using capsule tension rings. The intraocular pressure has to be brought down before performing a surgery. In case of chronic angle closure glaucoma, cataract extraction should be accompanied by a trabeculectomy or glaucoma drainage devices.[10]

CONCLUSION

The main ocular complications of Weill Marchesani syndrome are corneal endothelial dysfunction and pupillary block glaucoma. To avoid these complications, prophylactic iridotomy and prompt lensectomy are advisable.

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