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Ectopia Lentis in Three Successive Generations: A Case Series Study

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Research Article

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Abstract

Aim: To describe the clinical profiles of patients of three successive generations of a Bangladeshi family affected by familial ectopia lentis.

Methods: A case series study was conducted in a tertiary eye hospital in Bangladesh from 2015 to 2019. We evaluated twenty-three people of the same family of three successive generations. Among them, thirteen patients were affected by familial ectopia lentis. Several patients from all three generations underwent elaborate general examinations to rule out systemic and metabolic disorders.

Results: Among thirteen patients, eleven had bilateral Ectopia lentis. Among six adult patients, three had bilateral cataracts. ICCE was done on one patient from 2nd generation (pedigree 7) in another eye hospital. The other two patients (Pedigree-2 and 9) were blind; among seven child patients, four developed cataracts. One was visually significant and underwent PPL with AVT (Pedigree-14). Visual rehabilitation was done with aphakic glass. The other three children had cataracts but not in the pupillary area - not visually significant. No systemic associations were found except one who was suspected of Marfan syndrome.

Conclusions: Not only many patients in one family but also the occurrence in three generations is exceptional for Ectopia Lentis Syndrome. The autosomal recessive mode was found in other studies, but this family pedigree analysis favoured an autosomal dominant inheritance. Emphasis is needed on the importance of regular follow-up and management.

Keywords: Ectopia Lentis; Hypermetropia; Subluxation; Aphakic; Pars Plana Lensectomy

Introduction

Ectopia lentis is the displacement of the natural lens of the eye. The lens is considered subluxated when partially displaced but remains in the lens space [1]. Ectopia lentis may be congenital, traumatic, metabolic, and consecutive or spontaneous [2,3]. It may be associated with different syndromes. It is an isolated or Autosomal dominant or autosomal recessive pattern. Autosomal recessive families. Traumatic ectopia lentis usually manifests at birth or at any age [3,4]. It is essential to find out the cause of ectopia lentis for proper management of the patient. Ectopic lentis may be associated with major complications, including significant refractive transformation, pupillary block glaucoma, retinal pathology, and blindness [5].

Simple ectopia lentis can be a congenital or spontaneous disorder that is hereditary and can be inherited in an autosomal dominant or recessive pattern [4] Mutations to the ADAMTSL4 gene located on chromosome 1 in autosomal recessive inheritance pattern ectopia lentis, and the mutations occur on the FBN1 gene (Marfan syndrome) located on chromosome 15 of autosomal dominant inheritance pattern ectopia lentis [6]. Bilateral superotemporal dislocation of the lens usually occurs in simple ectopia lentis [7]. The asymmetric oval and often dilated eccentric pupil is associated in the opposite direction of the dislocated lens on slit lamp examination in ectopia lentis et pupillae, which is an uncommon congenital autosomal recessive disorder [6,8]. Marfan syndrome is one of the most common causes of inherited ectopia lentis, and bilateral ectopia lentis occurs in approximately 75% of patients [9,10]. The reduced visual acuity is the most significant symptom of ectopia lentis. The usual visual problem is the shifting of the refractive status of the eyes. Near vision difficulties can occur due to loss of accommodation and poor distant vision due to astigmatism or myopia [11]. Medical by an internist or paediatrician. Genetic counselling is indicated for predisposing hereditary conditions.Surgical correction of ectopia lentis remains a challenge; often, patients are managed with lensectomy/vitrectomy followed by optical rehabilitation with a contact lens/spectacle. Multiple surgical techniques may be required for ectopia lentis, each with associated complications [11-13]. This study attempted to report a series of patients with an autosomal dominant pattern of ectopia lentis of three successive generations of a family.

Patients and Methods

This case series study was conducted at Bangladesh Eye Hospital and Institute, Dhaka, Bangladesh, from 2015 to 2019. We studied twenty-three people of the same family of three successive generations. Among them, thirteen patients suffered from familial ectopia lentis. The ophthalmic evaluation for ectopia lentis should consist of checking visual status, an external ocular exam by torchlight, a slit lamp biomicroscopic evaluation for the anterior segment, an assessment of corneal diameter to rule out megalocornea (a feature of Marfan syndrome), retinoscopy and autorefraction, which is needed, and finally, a dilated posterior segment examination to rule out retinal detachment which is a consequence of ectopia lentis. General and systemic examinations of all patients from all three generations were evaluated thoroughly to rule out systemic and metabolic disorders. There was no history of marriage consanguinity among the brothers. Spouses found normal. A family pedigree chart (Figure 1) was made and analysed thoroughly. An informed written consent form was obtained from the patients or patients' guardians for the management of the patients and consent for the research.

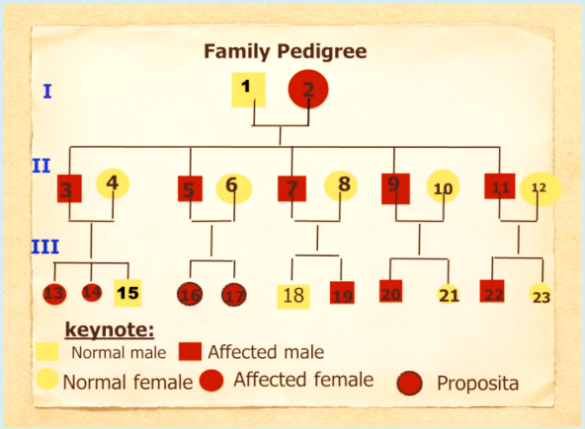

Figure 1: Family pedigree chart.

Results

Thirteenof twenty-three family members of three successive generations of a Bangladeshi family were affected by familial ectopia lentis. Among the affected patients, 6 were adults, and seven were children. Among 13 patients, eight were male and five female. Among seven child patients, four developed cataracts. One was visually significant and underwent pars plana lensectomy (PPL) with anterior vitrectomy (AVT) (Pedigree 14). Visual rehabilitation was done with aphakic glass. The other three children had cataracts but not in the pupillary area - not visually significant. The first presenting patients were two sisters (Figure 2) aged 8 and 10 (Pedigree 16 and 17). They presented with a dimness of vision. The complete ophthalmological evaluation revealed high hypermetropia with bilateral ectopia lentis. The undilated pupillary part was aphakic, as subluxation was superior. The cataract was developed and evaluated during the follow-up time. The Cataract doesn't hamper their vision in a non-dilated pupillary position. The vision was corrected by aphakic glass. Most of the family members depict similar eye problems. Among 13 patients, 11 had bilateral Ectopia lentis (Table 1). Among six adult patients, three patients had bilateral cataracts. ICCE was done in 1 patient from 2nd generation (pedigree 7) in another institute. The other two patients (Pedigree 2 and 9) were blind when we examined them. Cousins from 3rd generation with bilateral ectopia lentis. Patients were presented with a variety of ocular presentations. The patient (Pedigree 20) has bilateral superonasal ectopia lentis (Figure 3) in a dilated pupil. The aphakic part doesn't hamper his vision in a dilated position. However, the patient has high Myopia with myopic fundus. The best corrected visual acuity (BCVA) was 6/24. A girl from third-generation pedigree no 14 had cataractous ectopia lentis (Figure 4). No other systemic association was found. Pars plana lensectomy and vitrectomy were done, aphakic glass was given, and BCVA was 6/36. There were no systemic associations found, except one boy from 3rd

generation (pedigree 19) had Marfanoid-like features (tall stature, elongated digits). His height was 165 cm, upper to lower segment ratio was 1:1, and he had no dysmorphic features. Cardiac and skeletal anomalies were absent. The echocardiogram & chest X-ray, Urine Amino acids, and liver function test were normal.

Pedigree No	Sex/Age	Eye Involve	Direction	BCVA	Complication
2	F/65	OU	SN	Blind	Cataract & Glaucoma
3	M/49	OU	SN	HM	Cataract
5	M/44	OU	S	FC	Hyperopia
7	M/40	OS	S	Blind	Cataract & Glaucoma
9	M/38	OS	ST	HM	Hyperopia
11	M/35	OU	SN	Jun-36	Hyperopia
13	F/16	OU	SN	Jun-60	Hyperopia/Cataract
14	F/10	OU	S	Jun-36	Hyperopia/Cataract
16	F/10	OU	S	Jun-36	Hyperopia/Cataract
17	F/8	OU	ST	24-Jun	Hyperopia/Cataract
19 20	M/12 M/10	OU	SN	6/60 6/24	Myopia
22	M/9	OU	ST	18-Jun	Hyperopia

Table 1: Shows the pedigree no. demography and ocular features of the patients.

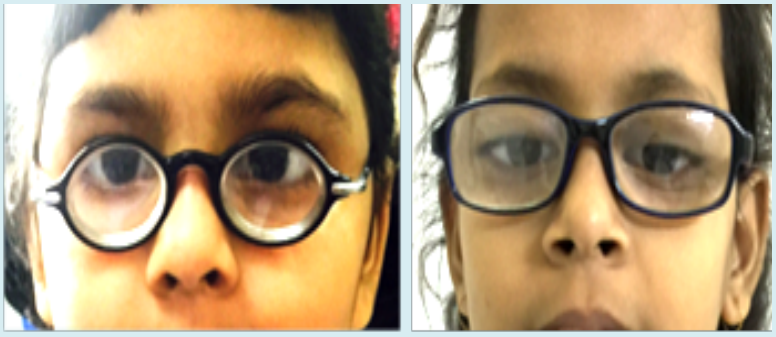

Figure 2: Two siblings (pedigree 16 and 17) from 3rd generation presented high hypermetropia with bilateral ectopia lentis. Subluxation corrected by aphakic lens.

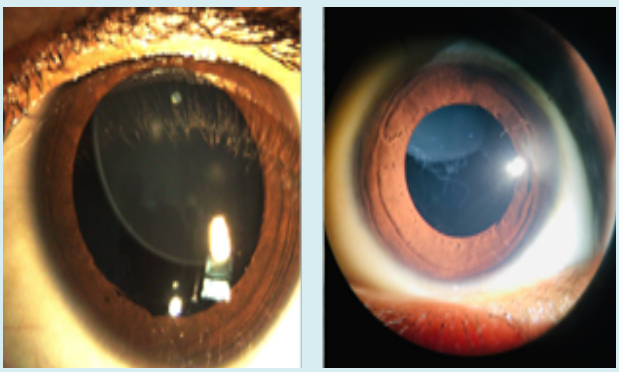

Figure 3: Bilateral Superonasal ectopia lentis.

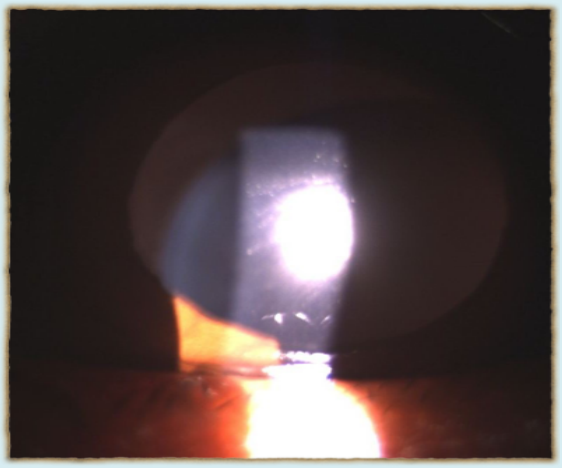

Figure 4: Ectopia lentis with cataract.

Discussion

Different case studies show familial ectopia lentis might be present in successive generations, which is like this case. Similarly, in their case studies, Casper DS, et al. [14] and Cruysberg JR, et al. [15] showed familial ectopia lentis asAutosomal dominant. However, Colley A, et al. [16] showed his case as Autosomal recessive [16]. Cataract was the most common ocular complication in Cruysberg JR, like the present case study, but Casper DS, et al. [14] reported glaucoma as the commonest, unlike the present case [14]. In our study, no systemic associations were found- Except one that was suspected. One boy from 3rd generation (pedigree-19) has Marfanoid-like features (tall stature, elongated digits). His height was 165 cm, upper to lower segment ratio was 1:1, and he had no dysmorphic features. Cardiac /skeletal anomalies were absent. The echocardiogram, chest X-ray, urine amino acid, and liver function test were normal. Found to have no Marfan syndrome or connective tissue disorder. Biochemical correlations could not be correlated with the clinical history. Genetic analysis wasn't possible. A Danish study found 396 cases of Ectopia Lentis (ECL). The estimated prevalence rate of ECL was 6.4 per 100,000. ECL was observed in 21.2% of cases as Ectopia Lentis et Pupillae, 8.0% of cases as Simple Dominant Ectopia Lentis, and 1.1% of cases as Homocystinuria [17,18].

ECL patients have a dislocated eye lens due to systemic disease. A thorough evaluation, including physical examination, metabolic screening, and echocardiography, is necessary to diagnose and prevent systemic complications Drack AV, et al. [17] Ectopia lentis can be part of different syndromes, with Marfan syndrome and Homocystinuria being common. It can also occur on its own. Simple ectopia lentis can be inherited autosomal dominant or autosomal recessive. In ectopia lentis et pupillae, pupils are oval and slit-shaped and inherited in an autosomal recessive pattern [19]. Ectopia Lentis et Pupillae Syndrome was diagnosed in nine family members across three generations. This syndrome is characterized by lens and pupil displacement, persistent pupillary membrane, iris transillumination, and poor pupillary dilatation. All patients developed cataracts before age 40, and two had intraocular hypertensive crises [20].

This study reports many patients across three generations exhibiting the ectopia lentis et pupillae syndrome, which differs from other studies. Despite the syndrome being previously identified as an autosomal recessive pattern, this family showed a mother-to-five-sons inheritance pattern from the first to the second generation and a father-to-son and daughter inheritance pattern from the second to the third generation. Based on the pedigree analysis, it was suggested that the syndrome may be inherited in an autosomal dominant pattern with reduced penetrance.

Conclusion

Not only many patients in one family but also the occurrence in three generations is very exceptional for Ectopia Lentis Syndrome. The autosomal recessive mode was found in other studies, but this family pedigree analysis favoured an autosomal dominant inheritance. No systemic association was found. Emphasis is needed on the importance of regular follow-up and management. Genetic counselling was done.

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