Indian Journal of Genetics and Molecular Research
Volume 3 Number 2, July - December 2014

Ectopia Lentis et Pupillae: A Rare Condition

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Abstract

Aims: To report a case of bilateral ectopia lentis et pupillae. *Settings and Design:* Case report *Methods and Material:* A 9 year old female with history of bilateral gradual painless diminution of vision underwent a thorough systemic, ophthalmic and serological examination. *Results:* Increased corneal diameters and bilateral superotemporally displaced pupils with inferonasally displaced lens were detected with normal systemic features and serology.*Conclusions:* A rare case of bilateral ectopia lentis et pupillae with megalocornea was diagnosed.

Keywords: Ectopia Lentis et Pupillae; Megalocornea; Congenital Ocular Abnormalities.

Introduction

Ectopia Lentis et Pupillae is a rare congenital anomaly which presents with lens displacement along with ectopic pupils present bilaterally but without any systemic disorder. Online Medelian inheritance in man (OMIM) has entry as #225200 with evidences that it can be due to mutations in ADAMTSL4 gene. The diagnosis of the condition is essential for optimal management and rule out associated conditions like Marfan syndrome

Case Report

The patient, 9 year old female student, resident of Jodhpur, Rajasthan presented with complaints of gradually progressive painless diminution of vision in both eyes for past 4 years. Her parents complained of the patient watching television from very near. There was no history of nyctalopia, excessive watering, head trauma, seizure episodes or febrile episodes. No family history of diabetes mellitus, hypertension or tuberculosis.

Patient was conceived out of consanguineous marriage. She was conceived 3 years after marriage. It was a twin pregnancy but the other child died on second post-natal day. Patient achieved developmental milestones normal for her age. She is the eldest of three siblings, the other two being a younger sister 5 years of age and a brother 3 years old. The siblings didn't have similar complaints.

General condition was found to be normal with no pallor, icterus, clubbing, cyanosis or lymphadenopathy. Patient weighed 26 kgs with height of 127.8 cm.

Systemic examination was found to be within normal limit.

On ophthalmic examination, diminution of vision in both eyes was observed. Snellen's Visual acuity was 6/18 OU, NVA- N6 OU. Colour vision was normal with Ishihara's plates OU. IOP was found to be 14.6 mm of Hg OU with Schiotz tonometer (5.5gm). Confrontation was found to be normal OU. Convergence was normal, up to nose. Anterior segment examination revealed increased corneal diameters in eyes, measuring 14mm vertically and 13mm horizontally. Persistent papillary membrane extending from 5-10 o'clock was present OS [Fig. 1]. Pupils were displaced supero-temporally in both eyes, towards 11 o' clock position in right eye and towards 1 o' clock position in left eye [Fig. 2, 3]. Pupils were obliquely oval in shape and slightly sluggish in reaction OU. Pupils dilated poorly with cyclopentolate eye drop 0.5%. After dilatation, it was

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Figures with legends are given below.

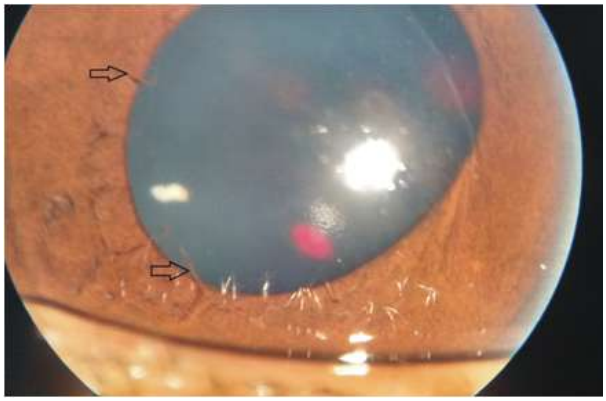

Fig. 1. Strands of persistent papillary membrane (black arrows) from 5-11 o'clock position in left eye

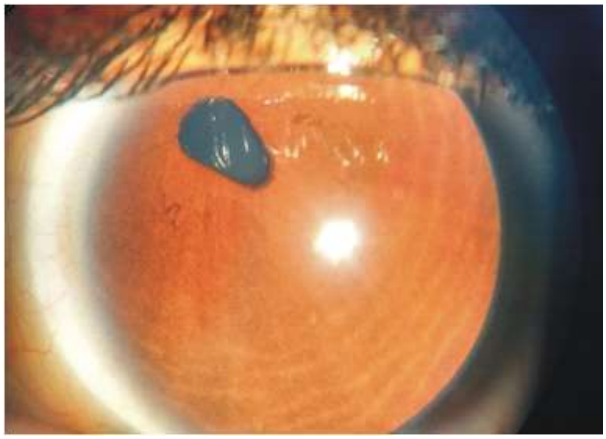

Fig. 2. Supero-temporally displaced pupil in right eye towards 11 o' clock position

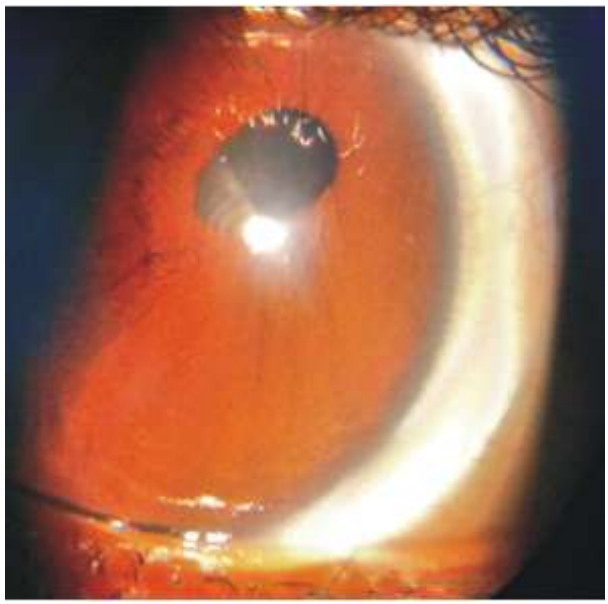

Fig. 3. Supero-temporally displaced pupil in left eye towards 1 o'clock position

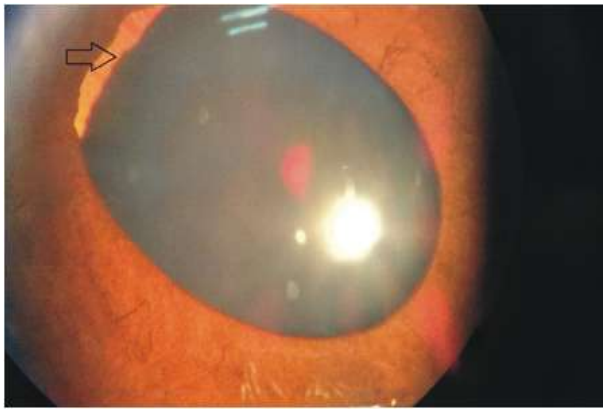

Fig. 4. Infero-temporally displaced lens (black arrow) in right eye with broken zonules supero-temporally

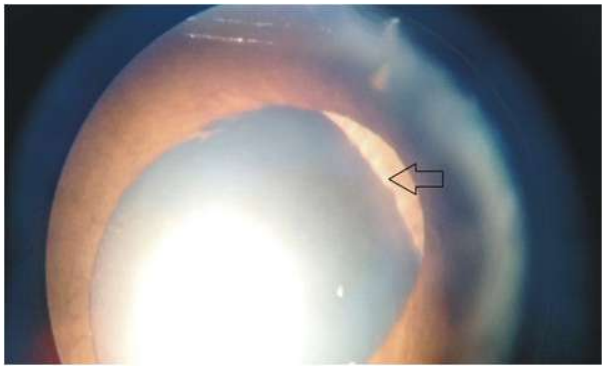

Fig. 5. Infero-temporally displaced lens (black arrow) in left eye with broken zonules supero-temporally

noted that the lens in both eyes were displaced infero-nasally by 4 clock hours, with broken zonules in the superotemporal quadrant [Fig. 4, 5]. Cycloplegic refraction revealed oblique myopic astigmatism. Neutralisation was achieved with -5DS along 50° axis, -7DS along 140° axis OD and -5DS along 140°, -7DS along 40° axis OS. The best corrected visual acuity of 6/9 was achieved with -6.0DS, -2DC at 50° OD and -6.0 DS, -2.0DC at 140° OS. Fundus examination was found to be normal with normal sized, round disc, distinct margins, C/D ratio 0f 0.2 and healthy pink neuro-retinal rim OU.

Serology, including complete haemogram and serum homocysteine levels, was found to be within normal range.

Bilateral megalocornea with persistent papillary membranes in left eye and bilateral displacement of pupils with ectopia lentis in diagonally opposite direction to papillary displacement was noted. Since the patient lacked any systemic and serological manifestations, a diagnosis of bilateral ectopia lentis et pupillae was made.

Discussion

Ectopia lentis et pupillae (ELeP) is a very rare autosomal recessively inherited condition characterized by displacement of both pupils and lenses without any systemic abnormalities. This fact has a prognostic bearing as survival is unaffected as opposed to other causes of ectopia lentis like homocystinuria, hyperlysinemia etc which affect the morbidity and mortality due to their systemic afflictions [1,2,3]. Mutations in ADAMTSL4 located on chromosome 1q21 have been implicated [4].

As described by Goldberg, the associated ophthalmic features of ELeP described in literature are severe axial myopia, poor vision, retinal detachment, cataract, iris transillumination defects, poor pupillary dilatation, iridohyaloid adhesions, and prominent iris processes in the anterior chamber angle [2]. Our case presented with additional features of increased corneal diameters with persistent pupillary membrane and was managed conservatively with spectacle correction.

ELeP is a very rare condition with less than 100 cases reported in literature, the largest studies being those done by Fuchs *et al* and Goldberg[2,3]. This case report brings into perspective a rare case of bilateral ectopia lentis et pupillae with increased corneal diameters and persistent papillary membrane unilaterally.

Investigating the largest Norwegian families Christensen *et al* revealed homozygous mutation in ADAMTSL4 gene on chromosome 1[4]. On the other hand, Chandra *et al* in a study on a patient found compound heterozygosis in same gene[6]. We could not perform the molecular testing in the child but are following the child for ophthalmic care.

The case illustrates a rare entity which needs a thorough workup to rule out other conditions with ectopia lentis.

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