ORIGINAL RESEARCH PAPER
Volume - 14 | Issue - 06 | June - 2025 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

# A FASCINATING INSTANCE OF ECTOPIA LENTIS ET PUPILLAE (ELeP): A RARE CASE REPORT

Medical Science

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## ABSTRACT

**Purpose-** To report a rare case of Ectopia Lentis et Pupillae with associations like axial myopia, and aqueous deficient dry eye. **Method-** A 32yr old male came to the OPD with complaints of defective vision and dry eye symptoms. Patient history taken using a symptom-oriented questionnaire. Tear film break-up time and Ocular surface staining with fluorescein, Schirmer test, to evaluate type of dry eye. Slitlamp, fundus examination, gonioscopy, pachymetry ,keratometry ,GAT done. 2D echocardiography and serum homocysteine tests were done to rule out marfan's, homocystinuria including a complete systemic examination. **Result-** O/E visual acuity- RE-CF 1metre,LE-6/18, not improving with glasses. S/L findings are loss of conjunctival sheen(lipcofs), iridodonesis, ectropion uvea, with corectopia, eccentric pupils poorly dilating to light. Right pupil displaced superotemporally, left pupil displaced superiorly. fundus examination-tesselated fundus with peripapillary atrophy, B-scan revealed PVD and posterior staphyloma in RE and both lenses were lying inferiorly in the vitreous, pachymetry and keratometry normal. Gonioscopy -pigment deposition in angles. IOP normal. Marfan's ruled out on a complete cardiac workup. serum homocysteine-12.3µmol. **Conclusion-** Ectopia lentis et pupillae (ELeP) is a rare syndrome inherited as an autosomal recessive trait. Its pathogenesis includes a neuroectodermal defect or persistence of fetal vasculature. Patient treated for dry eye and followed up regularly to prevent complications like glaucoma and RD.

## KEYWORDS

Ectopia lentis, corectopia, Dry eye, myopia, Marfan's, autosomal recessive

## INTRODUCTION

Ectopia lentis is characterized by the abnormal positioning of the natural crystalline lens either due to genetic factors or from external causes, resulting in displacement from the patellar fossa(1). This condition can either be inherited or acquired. The lens may be either partially displaced (subluxated) or completely dislocated from its normal position. Various factors, including trauma or underlying medical conditions, can lead to this lens displacement. The lens may appear in the anterior chamber, within the vitreous body, or on the surface of the retina(2). Ectopia lentis can serve as an initial indication of an underlying systemic condition, prompting further investigation into potential systemic issues. This disorder has multiple possible causes.

It is essential to gather a comprehensive past medical and systemic history for each patient. Inquiries should be made about any underlying cardiovascular, skeletal, or ocular irregularities. Conducting an extensive ocular and systemic examination is vital in confirming the diagnosis. Family members should also be assessed to identify any genetic links. Additionally, pediatricians have a crucial role in excluding hereditary disorders. The visual prognosis for the patient is influenced by the severity and nature of the lens displacement, as well as whether any complications are present or absent.

The etiology of ectopia lentis is broadly grouped as congenital, traumatic, metabolic/syndromic, and consecutive/spontaneous.

Ectopia Lentis et Pupillae (ELeP) is a rare autosomal recessive congenital disorder where in the pupil and the lens are displaced in the opposite directions (3). The pupil becomes asymmetric, eccentric, oval, and ectopic. There is minimal pupillary dilatation, and the lens displacement is usually bilateral and asymmetric. The incidence of this condition has been reported to be approximately 7% of all cases of ectopia lentis without systemic abnormalities.

### Pathophysiology Of Ectopia Lentis Et Pupillae (ELeP) :

- **Zonular Fibers:** It is believed that a defect in the zonular fibers, which are fine strands that secure the lens in the eye, contributes to the condition.
- **Genetic Mutations:** This condition is particularly associated with mutations in the ADAMTSL4 gene (4).
- **Developmental Abnormalities:** The issue with zonular fibers may result in the lens becoming displaced and the pupil being misaligned during the eye's developmental phase (5).

### Patients And Methods

A 32yr old male came to the opd with complaints of defective vision and dry eye symptoms. Patient history taken using a symptom-oriented questionnaire. Tear film break-up time and Ocular surface staining with fluorescein, Schirmer test, to evaluate type of dry eye.

Slit lamp, fundus examination, B-scan ultrasonography, gonioscopy , pachymetry, keratometry , GAT were done. 2D echocardiography and serum homocysteine tests were done to rule out Marfan's, homocystinuria including a complete systemic examination.

## RESULTS

- O/E visual acuity- RE-CF 1metre, LE-6/18, not improving with glasses.
- S/L findings are loss of conjunctival sheen(lipcofs), iridodonesis, ectropion uvea, with corectopia , eccentric pupils poorly dilating to light. right pupil displaced superotemporaly, left pupil displaced superiorly (figure 1).
- Fundus examination-tesselated fundus with peripapillary atrophy (figure 2), B-scan revealed PVD and posterior staphyloma in RE and both lenses were lying inferiorly in the vitreous (figure 3). Pachymetry, GAT, Keratometry normal.
- Gonioscopy -pigment deposition in angles. IOP is 17mmhg in RE &12mmhg in LE.
- Marfan's ruled out on a complete cardiac workup. serum homocysteine-12. 3µmol. Systemic examination ruled out associated syndromes.
- Schirmer test revealed Aqueous deficient Dry eye, patient was treated for same, and followed up regularly to prevent complications like Glaucoma and retinal detachment.

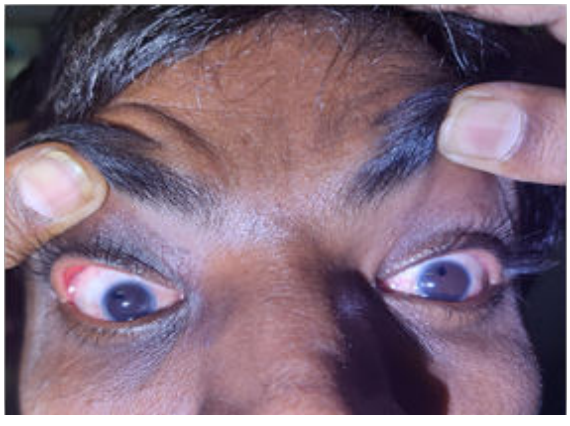

**Figure 1-** Showing right pupil displaced superotemporaly, left pupil displaced superiorly.

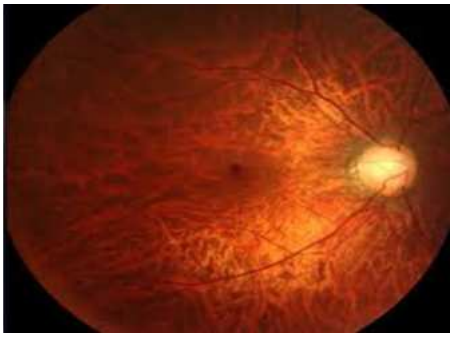

**Figure 2-** Tesselated Fundus With Peripapillary Atrophy

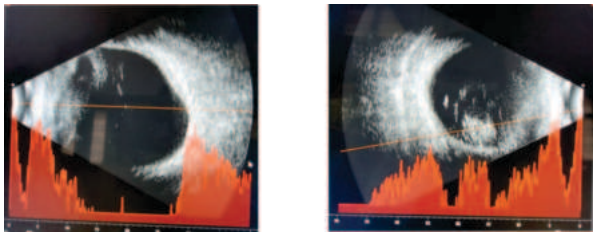

**Figure 3- RE B Scan-** A. Posterior Staphyloma B. Lens lying in vitreous

## CONCLUSION

The patient's prognosis depends on the presenting visual acuity, the extent of ocular and systemic involvement, degree of subluxation of the lens, age of onset of ectopia lentis, and associated systemic complications. Traumatic ectopia lentis patients usually have a very poor visual outcome. Complications associated with ectopia lentis includes Amblyopia, Corneal endothelial damage, Corneal decompensation, Chronic iridocyclitis, Pupillary block glaucoma, Angle recession glaucoma, Subluxated lens, Dislocated lens, Retinal detachment.

In summary, ectopia lentis is an uncommon condition, either familial or acquired, characterized by the displacement of the crystalline lens from the patellar fossa. The lens that is subluxated or dislocated may be found in either the anterior chamber or the posterior vitreous cavity. Patients may experience impaired vision, reduced accommodation, monocular diplopia, discomfort, and inflammation. A thorough assessment of both the anterior and posterior segments, along with a systemic evaluation, is essential for every individual case. Treatment options are determined by the age of onset, visual acuity, the degree of lens subluxation or dislocation, and the presence of any associated ocular or systemic issues. It is important for all patients to undergo familial screening for ectopia lentis to eliminate the possibility of syndromic associations.

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