

Review Articles

Ectopia Lentis: Weill Marchesani Syndrome

HL Trivedi*, Ramesh Venkatesh**

Abstract

A 20 yr old boy came to our OPD with decreased vision since 3 yrs. He complained of double vision in both the eyes. There were no other ocular or systemic complaints. On systemic examination, the boy had a short stature compared to his age, short fingers and limbs. On ophthalmic examination, Vn in RE was 20/200 and LE was finger counting 5 ft. Cornea and other ocular adnexa were normal. The lens was spherical in shape and dislocated in the anterior chamber. There were no signs of iridocyclitis. Intraocular tension in both eyes was 20.6 mm Hg. Posterior segment evaluation was normal.

Introduction

Ectopia lentis is defined as displacement or malposition of the crystalline lens of the eye. The lens is considered dislocated or luxated when it lies completely outside the lens patellar fossa, in the anterior chamber, free-floating in the vitreous, or directly on the retina. The lens is described as subluxed when it is partially displaced but contained within the lens space. In the absence of trauma, ectopia lentis should evoke suspicion for concomitant hereditary systemic disease or associated ocular disorders.

Weil Marchesani syndrome is also known as Marchesani's Syndrome.

The condition is named after Oswald Marchesani and Georges Weill.

Pathophysiology

Disruption or dysfunction of the zonular fibres of the lens, regardless of cause (trauma or heritable condition), is the underlying pathophysiology of ectopia lentis. The degree of zonular impairment determines the degree

of lens displacement.

Frequency

United States

Ectopia lentis is a rare condition. Incidence in the general population is unknown. The most common cause of ectopia lentis is trauma, which accounts for nearly one half of all cases of lens dislocation.

Mortality/Morbidity

Ectopia lentis may cause marked visual disturbance, which varies with the degree of lens displacement and the underlying aetiologic abnormality.

Sex

Males appear more prone to ocular trauma than females; therefore, a male preponderance has been reported. Male and female frequency varies with the aetiology of the lens displacement.

Age

Ectopia lentis can occur at any age. It may be present at birth, or it may manifest late in life.

Inheritance Pattern : Autosomal recessive

*Associate Professor; **Resident, Department of Ophthalmology, TNMC and BYL Nair Hospital, Mumbai 400 008.

Clinical

History

A 20 yr old boy came to our OPD with decreased vision since 3 yrs. He complained of double vision in both the eyes. There were no other ocular or systemic complaints. On systemic examination, the boy had a short stature compared to his age, short fingers and limbs (Figs. 1-3). On ophthalmic examination, Vn in RE was 20/200 and LE was finger counting 5 ft. Cornea and other ocular adnexa were normal. The lens was spherical in shape and dislocated in the anterior chamber. There were no signs of iridocyclitis. Intraocular tension in both eyes was 20.6 mm Hg. Posterior segment evaluation was normal.

Patient was operated in the left eye with lensectomy through the limbal route and scleral fixated intraocular lens implantation in the same sitting. Post operatively patient

Fig. 2 : Short toes.

Fig. 3 : Short fingers.

vision improved to 20/60 in the left eye with no other complications.

- Common presenting symptoms (visual disturbance)
 - Decreased distance visual acuity (secondary to astigmatism or myopia)
 - Poor near vision (loss of accommodative power)
 - Monocular diplopia
- Obtain a detailed history investigating possible systemic disease associations.
- Cardiovascular disease (e.g., Marfan syndrome)

Fig. 1 : Boy with short stature.

- Skeletal problems
 - Marfan syndrome
 - Weil-Marchesani syndrome
 - Homocystinuria
- Pertinent family history
 - Consanguinity
 - Mental retardation
 - Unexplained deaths at young age (e.g., autosomal recessive conditions, including homocystinuria, hyperlysinaemia, ectopia lentis et pupillae, or sulphite oxidase deficiency)

Physical

A paediatrician or an internal medicine physician should perform a comprehensive physical examination of patients with ectopia lentis of undetermined aetiology because of the commonly associated hereditary systemic disorders. The ocular examination should include the following:

- Vision
 - Ectopia lentis is potentially visually debilitating.
 - Visual acuity varies with the degree of malpositioning of the lens.
 - Amblyopia is a common cause of decreased vision in congenital ectopia lentis and is preventable and treatable.
- External ocular examination
 - Attention to orbital anatomy is important to evaluate for hereditary malformations (eg, enophthalmos with facial myopathic appearance seen in patients with Marfan syndrome).
 - Measure corneal diameter (megalocornea is associated with Marfan syndrome).
 - Strabismus is not uncommon

(secondary to amblyopia).

- Retinoscopy and refraction
 - Careful retinoscopy and refraction is essential, often revealing myopia with astigmatism.
 - Keratometry may help ascertain degree of corneal astigmatism.
- Slit lamp examination
 - Evaluate lens position, and identify phacodonesis or cataract.
 - Measure intraocular pressure. Elevation may indicate secondary glaucoma.
 - Causes of glaucoma in ectopia lentis include the following: (1) pupillary block, (2) phacoanaphylaxis or phacolytic, (3) posttraumatic angle recession, (4) poorly developed angle structures, and (5) lens in the anterior chamber.
- Dilated fundus examination: Retinal detachment is one of the most serious consequences of a dislocated lens.
- Echography: Axial length measurement may be of benefit (patients with Marfan syndrome have large globes).

Causes

The numerous causes of ectopia lentis can be classified as follows:

- Traumatic dislocation (most common cause)
- Hereditary ectopia lentis without systemic manifestations
 - Single (isolated) ectopia lentis is characterized by autosomal dominant inheritance with the genetic defect located on chromosome 15, causing a dysfunctional zonular apparatus. Microspherophakia is common. Although most often present at birth,

- late onset has been described. Typically, the lens is displaced supertemporally.
- Ectopia lentis et pupillae is characterized by asymmetric eccentric pupils that are displaced in the opposite direction of the lens dislocation (toward the most dysfunctional zonular fibres). The condition usually is bilateral and typically autosomal recessive. The irides often appear atrophic with transillumination defects on slit lamp examination. Cataracts commonly are seen.
 - Systemic conditions commonly associated with ectopia lentis
 - Marfan syndrome
 - Homocystinuria
 - Weil-Marchesani is a rare syndrome characterized by skeletal malformations (e.g., short stature, brachycephaly, limited joint mobility, well-developed muscular appearance) and ocular abnormalities (e.g., ectopia lentis, microspherophakia, lenticular myopia). Small shallow orbits (Fig. 4), mild maxillary hypoplasia, narrow palate, small spherical crystalline lenses, myopia with or without glaucoma, frequent ectopia lentis, occasional blindness, malformed and malaligned teeth, and cardiac defects are the features of Weil Marchesani syndrome. Late ossification of the epiphyses is a constant feature. The inheritance pattern is not well understood; usually autosomal recessive. Microspherophakia is the most prominent feature of this syndrome. High incidence of lens subluxation occurs inferiorly, often



Fig. 4 : Small shallow orbits.

- progressing to complete dislocation. Pupillary block glaucoma is common; therefore, prophylactic laser peripheral iridotomies are recommended.
- Sulphite oxidase deficiency
- Hyperlysinaemia.
- Primary ocular disorders associated with ectopia lentis
 - Congenital glaucoma/buphthalmos
 - Pseudoexfoliation syndrome
 - Syphilis/chronic uveitis
 - Retinitis pigmentosa
 - Megalocornea
 - Aniridia
 - Hypermature cataract
 - Intraocular tumour
 - High myopia
- Systemic diseases rarely associated with ectopia lentis
 - Ehlers-Danlos syndrome
 - Crouzon disease
 - Refsum syndrome
 - Kniest syndrome
 - Mandibulofacial dysostosis
 - Sturge-Weber syndrome
 - Conradi syndrome
 - Pfaundler syndrome
 - Pierre Robin syndrome
 - Wildervanck syndrome
 - Sprengel deformity

Workup

Lab Studies

- Perform appropriate diagnostic and laboratory evaluation, if a hereditary condition is suspected (e.g., cardiac evaluation for Marfan syndrome, check serum and urine levels of homocysteine or methionine for homocystinuria).

Treatment

Medical Care

Without an antecedent history of trauma, patients with ectopia lentis may possess a systemic disease with potentially deleterious effects; therefore, comanagement with the patient's paediatrician or internist is essential. Dietary restriction may be partially effective in patients with homocystinuria. Repair of an impending dissecting aortic aneurysm in Marfan syndrome may be life saving. If a hereditary condition is discovered, appropriate genetic counselling should be given. Moreover, all relatives with potential risk should be examined.

- Treatment of glaucoma is dependent on the aetiologic mechanism.
 - In pupillary block (e.g., patients who have Weil-Marchesani with microspherophakia), laser peripheral iridotomy or iridectomy should be performed, and intraocular pressure elevation should be treated medically. Prophylactic laser iridotomy in patients with microspherophakia is beneficial.
 - Treatment of a lens dislodged into the anterior chamber is initially pharmacological with mydriasis/cycloplegia (to permit posterior migration of the lens behind the iris) in conjunction with ocular massage through a closed lid to promote this

posterior migration. Surgical treatment will then be needed to prevent further complications.

- Treatment of a dislocated lens in the vitreous is surgical; however, many vitreoretinal surgeons may advocate observation if no visual disturbance or impending retinal complication is apparent.

Surgical Care

Lens surgery in ectopia lentis is technically challenging, and the numerous techniques and strategies are beyond the scope of this article.

- Indication for lensectomy
 - Lens in the anterior chamber
 - Lens-induced uveitis
 - Lens-induced glaucoma
 - Lenticular opacity with poor visual function
 - Anisometropia or refractive error not amenable to optical correction (e.g., in a child to prevent amblyopia)
 - Impending dislocation of the lens (Fig. 5).

Follow-up

Further Outpatient Care

- Close follow-up care with a full ocular



Fig. 5 :

examination including tonometry (intraocular pressure check) and dilated fundus examination is important.

In/Out Patient Medications

- Topical drops may be necessary to lower the intraocular pressure or help decrease inflammation.

Deterrence/Prevention

- Early diagnosis of ectopia lentis with appropriate optical correction can prevent amblyopia.

Complications

- The most common ocular complications of ectopia lentis include amblyopia, uveitis, glaucoma, and retinal detachment; appropriate treatment for these specific entities should be implemented.

Prognosis

- Depending on the degree of lens dislocation, the age of onset, and its associated secondary complications, most patients do well.
- Those patients who have trauma-associated ectopia lentis may have other more life-threatening complications (depending on the severity of the trauma).
- Patients with heritable conditions associated with ectopia lentis may have other systemic complications.

Patient Education

- Patients with ectopia lentis associated with a heritable condition need to be educated on the importance of following up with a primary care physician to rule out life-threatening disorders.
- Safety glasses are advocated when risk

of eye injury is possible.

Miscellaneous

Medical/Legal Pitfalls

- The gravity of ectopia lentis when associated with a heritable disorder must be recognized and referred to the primary care physician as soon as possible. The primary care doctor may not be aware of this clinical entity and its ramifications; thus, it is imperative for the ophthalmologist to ensure the appropriate workup for systemic conditions such as Marfan syndrome or homocystinuria has been completed.
- Provide close follow-up care with an ocular examination documenting vision, intraocular pressure, degree of lens dislocation, and any retinal pathology (full dilated fundus examination).

References

1. Jones KL. Smith's Recognizable Patterns of Human Malformations, 4th edn. Philadelphia. WB Saunders Co., 1988; 397 : 714-15.
2. Jensen AD, Cross HE, Paton D. Ocular 926 complications in the Weill-Marchesani syndrome. *Am J Ophthal* 1974; 77 : 261-69.
3. William H, Spencer MD. Ophthalmic pathology. An Atlas and Text Book, Philadelphia, WB Saunders Co. 1985; 1 : 436-37.
4. Duke-Elder S. Summary of system of ophthalmology. London, Henry Kimpton Publishers 1876: 96.
5. Fujiwara H, Takigawa Y, Ueno S, Okuda K. Histology of the lens in the Weill Marchesani syndrome. *Br J Ophthalmol* 1990; 74 : 631-34.
6. Verloes A, Hermia JP, Galand A, Koulischer L, Dodinval P. Glaucomalens ectopia - microspherophakia - stiffness shortness (GEMSS) syndrome: a dominant disease with manifestations of Weill Marchesani syndromes. *Am J Med Genet* 1992; 44 : 48-51.