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# Ectopia Lentis: Clinical profiles in a large cohort of children from a Tertiary Eye Care network in India

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## Abstract:

**PURPOSE:** To study the clinical presentations, visual, and refractive profiles of children with congenital ectopia lentis in a large cohort of patients from a tertiary eye care network in India.

**MATERIALS AND METHODS:** A retrospective review of electronic medical records from December 2012 to December 2020 was conducted. Two hundred and ninety-seven consecutive children  $\leq 18$  years of age at presentation were identified and analyzed for demographic details, patient distribution, lens subluxation, visual, and refractive profiles before and after the interventions.

**RESULTS:** Five hundred and ninety-four eyes of 297 (male 56%;  $n = 166$ ) patients were analyzed. The mean age at presentation was  $8.74 \pm 3.89$ . Best-corrected visual acuity (BCVA) at presentation ranged from 0.3 logMAR to 3.5 logMAR; (Snellen: 6/9 – close to face [CF]) (mean  $0.89 \pm 0.68$ ). High myopia ( $n = 201$ ; 33.83%) and mild astigmatism ( $n = 340$ ; 57.23%) were more frequent. Temporal ( $n = 108$ ; 18.18%) subluxation was most common followed by superior. Lensectomy with limited vitrectomy was performed in 243 eyes of 127 patients (40.90%). Median preoperative BCVA was 1.0 (range: 0.3–3.5 logMAR; 20/40 - CF). Median postoperative BCVA was 0.5 logMAR (6/18) in the pseudophakic group and 0.6 logMAR (6/24) in the aphakic group. Spherical equivalent in myopic children reduced from  $-12.06 \pm 6.84D$  to  $-1.57D$  ( $-0.25D$  to  $-5.5D$ ) in the pseudophakic group and  $+9.3D$  ( $+5.5D$  to  $15.5D$ ) in the aphakic group.

**CONCLUSION:** This study is a large cohort of children presenting with ectopia lentis. Following intervention, an improvement in the median BCVA and refractive correction was noted in the entire cohort.

## Keywords:

Astigmatism, ectopia lentis, lensectomy, Marfan's syndrome, visual rehabilitation

## Introduction

Congenital ectopia lentis is a subluxation of the natural crystalline lens from its normal position. It is often bilateral but could vary in degree of subluxation and may be associated with inherited connective tissue disorders.<sup>[1-4]</sup> The most common etiologies include Marfan's syndrome, familial or idiopathic ectopia lentis, and homocystinuria.<sup>[3]</sup> The patient may present to the ophthalmologist either as part of multisystem involvement or

ocular manifestation alone. Familial cases sometimes tend to report earlier.<sup>[5]</sup>

Ectopia lentis poses diagnostic and management challenges, especially in very young children especially with regard to the timing of treatment and variations in vision and refractive correction through the phakic and aphakic portions of the pupil.<sup>[6]</sup> Both conservative and surgical management for visually significant subluxation of the crystalline lens have been reported in these patients. Visual restoration may be conservative, such as spectacles or contact

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lenses, or surgery, which includes both limbal and pars plana approaches for lensectomy.<sup>[5]</sup>

Surgical options for intraocular lens (IOL) implantation in pediatric eyes with ectopia lentis include a posterior chamber in the bag implantation with Capsular tension ring (CTR) or segment (CTS), anterior chamber IOL, and iris-fixated or scleral-fixated posterior chamber IOL (PCIOL).<sup>[7]</sup> A tailored and individualized management helps attain the best possible vision in a given patient.

The current study is a large series of children with congenital ectopia lentis to understand the demographics, patient distribution, clinical profile, intervention, and visual and refractive outcomes.

## Materials and Methods

The present study was conducted at L. V. Prasad Eye Institute India Tertiary Eye Care network. The patients were identified after a systematic search of the electronic medical records (EMRs) database using the diagnostic codes "ICD H27.1" for ectopia lentis, congenital ectopia lentis, and familial ectopia lentis between December 2012 and December 2020. The study was approved by the Institutional Ethics Committee (LEC BHR-R-03-21-600) and adhered to the Declaration by Helsinki. The patient consent is waived by the Institutional Ethics Committee.

Patients  $\leq 18$  years of age at presentation and those diagnosed with congenital ectopia lentis as suggested by history and clinical features were included in the study. All patients older than 18 years at presentation and those with a history of trauma related to the possible etiology for the subluxation were excluded. The patients were subdivided into three age groups (<6 years, 7–12 years, and 13–18 years) to evaluate the differences in the visual acuity outcome following the intervention.

### Basic data

The EyeSmart EMR data were extracted and analyzed. We collected the following data from the medical case records: History of onset of vision loss, other presenting complaints, type of vision loss (simultaneous vs. sequential), age at presentation, gender, family history of similar complaints or ocular surgery, best-corrected visual acuity (BCVA), refractive status and the anterior segment including the lens findings of subluxation direction, and degree and/or spherical nature of the crystalline lens and retinal findings. In addition, systemic features where relevant were documented. Data collected also included managing individual cases either conservatively or surgically and their outcomes.

## Ocular examinations

The BCVA of each eye was recorded with Snellen's chart under the standard lighting conditions and converted to LogMAR. Visual impairment was graded as per the WHO criteria, mild (Snellen 20/40 or better;  $>0.3$  LogMAR), moderate (Snellen 20/50–20/160; 0.40–0.90 LogMAR), severe (Snellen 20/200–20/600; 0.1–1.50 LogMAR), and profound (worse than Snellen visual acuity of 20/600; 1.60–2.40 LogMAR and close to face (CF) as 3.5 logMAR). For those children who could not comprehend, the visual acuity measurement was deferred to a later date. The direction of subluxation was noted as documented in the medical records, where possible, with photographic evidence.

## Refractive status

The patients were subdivided into emmetropia, myopia (mild:  $-0.25$  to  $-3.0D$ , moderate:  $-3.25$ – $-6D$ , severe  $>-6.0D$ ), hyperopia (low  $<+2.0D$ , moderate  $+2.25$  to  $+5.0D$ , and high  $>+5.0D$ ) or with astigmatism (mild 0.50 to 2D, moderate 2D–4D, severe  $>4D$ ). The spherical equivalent (SE) of the BCVA at presentation and at follow-up visit 6 months after intervention was evaluated.

## Ancillary tests

These included A-scan biometry in determining the axial length and the keratometry values, where deemed necessary, an ultrasound B-scan (suspected posterior subluxation), ultrasound biomicroscopy, and Pentacam in suspected corneal astigmatism.

## Statistical analysis

The collected data were analyzed using Microsoft Excel 2019 (Microsoft Corporation Inc, Richmond, USA) version 16.0.6742.2048.

## Results

The analyzed data included 594 eyes of 297 children in the defined study period.

### Demographics

There was a slight male preponderance, in our cohort, males ( $n = 166$ ; 55.89%) and females ( $n = 131$ ; 44.11%). School-going students included the vast majority ( $n = 269$ ; 90.57%). Children were from urban locations ( $n = 135$ ; 45.45%), rural location ( $n = 119$ ; 40.07%), and metropolitan suburbs ( $n = 43$ ; 14.48%). Bilateral ectopia lentis was present in 91.25% ( $n = 271$ ) children; unilateral involvement, the right or left eye, was 4.38% ( $n = 13$ ) each.

### Visual acuity

BCVA at initial presentation ranged from 0.3 logMAR to 3.5 logMAR (Snellen 20/40 – count fingers CF

mean ( $0.89 \pm 0.68$ ). Visual impairment was mild in 32.65% ( $n = 194/594$ ) eyes, moderate in 30.46% ( $n = 180$ ) eyes, severe in 8.75% ( $n = 52$ ), and profound in 17.84% ( $n = 106$ ) eyes. The patients were managed conservatively and surgically. Surgery was performed in 243 eyes. The preoperative BCVA ranged from 0.3 to 3.5 logMAR (median 1.0; Snellen 20/50 – CF), and following surgery, the visual acuity improved to 0.2–2.2 logMAR (median 0.35; Snellen 20/40 – CF). The median BCVA in aphakic eyes (53.5%;  $n = 130$ ) was 0.6 logMAR (Snellen 20/80; 6/24) and in pseudophakic eyes (46.5%;  $n = 113$ ) was 0.5 logMAR (Snellen 20/60; 6/18.) Among the three age groups (<6 years, 7–12 years, 13–18 years), the pre- and post-operative visual acuity was comparable [Table 1].

### Refractive status

The refractive errors included myopia (–0.75D to –30.0D), hyperopia (+1.25 to +16.0D), and astigmatism (+2.00 to –9.0D). In this cohort, 11.95% ( $n = 71/594$ ) eyes did not have a refractive error. Overall, high myopia and myopic astigmatism were more frequent. Myopia was mild in 5.05% ( $n = 30$ ), moderate in 8.24% ( $n = 49$ ), and high in 33.83% ( $n = 201$ ) eyes. Hyperopia was low in 2.02% ( $n = 12$ ), moderate 2.99% ( $n = 17$ ), and high in 15.48% ( $n = 92$ ) eyes. Astigmatism was mild in 57.23% ( $n = 340$ ), moderate in 20.30% ( $n = 120$ ), and severe in 0.70% ( $n = 4$ ) eyes. There was no astigmatism in 21.8% ( $n = 130$ ) patients. In the myopic group, the preoperative SE was  $-12.06 \pm 6.84$ D while the postoperative SE in the pseudophakic group was  $-1.57$ D (–0.25D to –5.5D) and in the aphakic group was  $+9.3$ D (+5.5D to 15.5D).

### Lens subluxation

The details of the direction of subluxation are shown in Figure 1. The most common direction was temporal, in 18.18% ( $n = 108$ ), followed by superior in 13.97% ( $n = 83$ ), and nasal in 11.44% ( $n = 68$ ). A smaller number of eyes had an anterior 2.18% ( $n = 13$ ) and posterior 1.51% ( $n = 9$ ) dislocation of the lens.

**Table 1: Visual outcomes in ectopia lentis (age groups)**

Age groups (years)	VA presentation (logMAR)	BCVA surgical (preoperative)	BCVA surgical (postoperative)
<6	$n=194$ eyes	$n=89$ eyes	$n=89$ eyes
	Mean: 0.92	Mean: 1.1	Mean: 0.52
	Median: 0.8	Median: 1.0	Median: 0.3
7–12	$n=300$ eyes	$n=115$ eyes	$n=115$ eyes
	Mean: 0.87	Mean: 0.94	Mean: 0.56
	Median: 0.7	Median: 0.8	Median: 0.4
13–18	$n=100$ eyes	$n=39$ eyes	$n=39$ eyes
	Mean: 0.71	Mean: 0.8	Mean: 0.55
	Median: 0.6	Median: 0.6	Median: 0.3

BCVA=Best corrected visual acuity, VA=Visual acuity, logMAR=Logarithm of the Minimum Angle of Resolution

Ocular comorbidities [Figure 2] included eyes with cataract ( $n = 32$ ; 4.45%), strabismus ( $n = 27$ ; 3.76%), glaucoma ( $n = 21$ ; 3.53%), and aniridia ( $n = 16$ ; 2.23%). The syndromic cases included suspected Marfan’s syndrome ( $n = 165$ ; 26.09%), Weil Marchesani syndrome ( $n = 4$ ; 0.67%), and homocystinuria ( $n = 1$ ; 0.16%). A confirmatory genetic test in Marfan’s syndrome for fibrillin gene was positive in three patients; but the parents of most of the suspected children did not opt for genetic testing.

### Intervention

Surgical intervention was performed in 243 eyes of 127 patients (243/568 eyes; 40.90%). An anterior approach lensectomy with limited vitrectomy was performed in  $n = 91$  (37.14%) eyes, and pars plana lensectomy and vitrectomy were performed in 152 (62.04%) eyes. Following surgery, 53.5% ( $n = 130$ ) eyes were aphakic and 46.5% ( $n = 113$ ) were pseudophakic.

### Discussion

Few studies have reported and discussed the patient demographics and the spectrum of visual acuity and refractive errors in children presenting with ectopia lentis and their outcomes.<sup>[1–4]</sup> The current study has a substantial number of cases evaluated [Table 2]. The mean age at presentation was  $8.74 \pm 3.89$ . One-third presented  $\leq 6$  years of age. School-going students accounted for most cases ( $n = 269$ ; 90.57%). About a quarter of our cohort had severe to profound vision impairment ( $n = 158$ ; 26.59%). High myopia with mild-to-moderate astigmatism was the most common refractive error; temporal and superior subluxations were more common.

Some of the earlier studies have added valuable knowledge in ectopia lentis. Nelson and Maumenee<sup>[2]</sup> reported amblyopia as a common cause of decreased vision; Neely and Plager reported a higher incidence of induced and myopic astigmatism;<sup>[3]</sup> and Jin *et al.* have reported that one-tenth (in their series, 10.13%; 31/306) had mild or no visual impairment. In Jin *et al.* series, the average age for surgery was  $9.24 \pm 4.83$  years.<sup>[6]</sup> The older age of the child at the time of surgery may influence the final visual acuity as amblyopia would play a major role in late intervention.<sup>[6]</sup> Surgical interventions and outcomes have been previously reported in several studies.<sup>[5–7,9,16]</sup> In our cohort, surgical intervention, lensectomy-vitrectomy, was performed in 243 eyes. In the surgical group, almost half of the patients were left aphakic and re-habilitated with spectacles or contact lenses; and the remaining patients received an IOL. Irrespective of the type of surgery, all patients

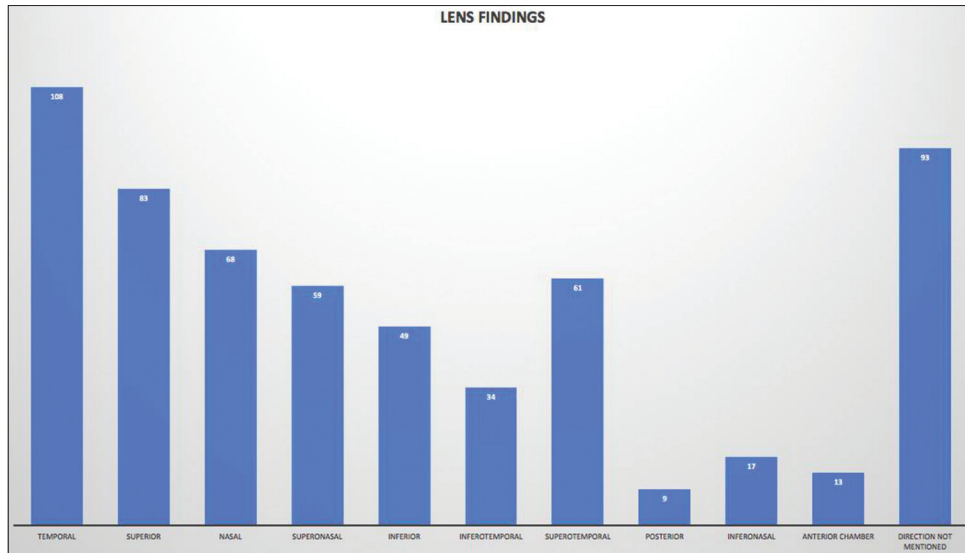


Figure 1: Direction of lens subluxation

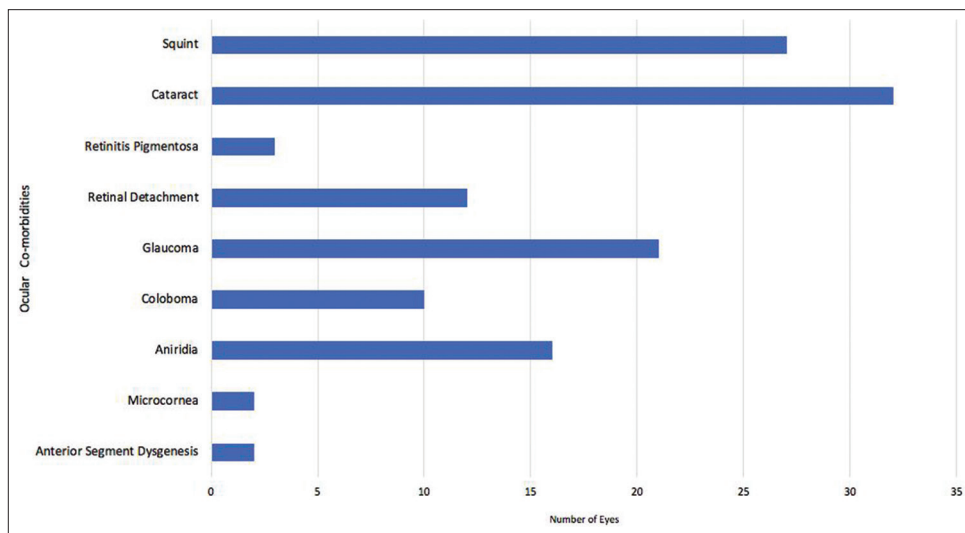


Figure 2: Ocular co-morbidities

had an improvement in vision, and the SE decreased after surgery.

Kemmanu *et al.* have reported the practice patterns of pediatric ophthalmologists for the management of lens anomalies in India.<sup>[16]</sup> In India, in cases of unilateral ectopia lentis, the most common procedure is the implantation of IOL assisted by a capsule-tension ring/CTS (44.68%) whereas in bilateral ectopia lentis, and lensectomy with limited anterior vitrectomy, without implantation of IOL and managed with spectacles correction (43.91%). The report further indicated that the Indian pediatric ophthalmologist's practice was comparable to the worldwide practice.<sup>[16]</sup>

After surgical intervention, visual rehabilitation and treatment of amblyopia are essential for patients within

the amblyogenic age group.<sup>[7,16]</sup> Çevik *et al.* studied the use of iris-claw IOL in children with ectopia lentis.<sup>[8]</sup> In their study of children with heritable ( $n = 21$ ) and nonheritable ( $n = 9$ ) ectopia lentis that received a postchamber artisan IOL, a postoperative target refraction value (SE of  $\pm 0.5D$ ) was achieved in 47% of the heritable form of ectopia lentis and 22% of the nonheritable ectopia lentis. Lalramhluni *et al.* studied the refractive and visual outcomes after scleral fixated IOL in 25 eyes of 15 children with nontraumatic ectopia lentis.<sup>[9]</sup> There was a significant improvement in visual acuity, reduction in SE, and achieving target refraction within 2D in 20 eyes. Similarly, Hsu *et al.* in their series of scleral fixated IOL achieved improvement in visual acuity postoperatively.<sup>[10]</sup> Studies have also reported the safe outcomes in children where a capsule-tension ring was used to stabilize the bag for

**Table 2: Comparison of ectopia lentis studies**

Publication	Number of cases	Principal intervention	Outcomes
Çevik <i>et al.</i> <sup>[8]</sup>	2 groups Group 1-11 patients (heritable disease) Group 2-6 patients (nonheritable disease)	Iris-claw IOL	Target refraction values achieved in 47% in group 1 and 22% in group 2
Lalramhluni <i>et al.</i> <sup>[9]</sup>	25 eyes of 15 children	Scleral fixated IOL	SE was within 2 D of the target refraction in 20/25 eyes
Hsu <i>et al.</i> <sup>[10]</sup>	13 eyes of 7 children	Scleral fixated IOL	Mean preoperative VA of 0.86±0.17 improved to 0.23±0.09 postoperatively
Konradsen <i>et al.</i> <sup>[11]</sup>	37 eyes of 22 children	Cioni modified CTR and conventional CTR	The median preoperative BCVA (22 eyes) 0.26 (range 0.015–0.65). The median postoperative BCVA (36 eyes) 0.59 (range 0.2–1.0)
Maharana <i>et al.</i> , 2022 <sup>[12]</sup>	78 eyes of 57 patients Mean age 14.73 years IOL implantation in 32 eyes (46.2%)	Lens aspiration±IOL	Improvement in VA Mean preoperative BCVA 0.85±0.55 logMAR Mean postoperative 0.44±0.29 logMAR Pseudophakic eyes fared better Patients age and degree of subluxation - no impact on final vision
Fuerst <i>et al.</i> <sup>[13]</sup>	76 eyes of 43 patients 56 eyes of 28 patients (long follow up) Mean age 11.1 years Mean follow up 3.59 years	Artisan iris-claw (anterior chamber lens)	BCVA improved from logMAR 0.36 (20/46) to logMAR 0.18 (20/30) Endothelial cell loss reported similar to PCIOL cases
Meng <i>et al.</i> <sup>[14]</sup>	47 eyes of 26 patients Age <9 years (mean age 61.6±22.3 months)	Scleral fixated IOL	Median BCVA preoperative 0.82 logMAR improved to 0.15 logMAR Sph equivalent improved from 9.00 to 0.75 Complications of pupil capture and IOL decentration (re-surgery) in 9%
Cai <i>et al.</i> <sup>[15]</sup>	101 eyes of 57 patients <8 years of age	PCIOL with Cioni-modified CTR	Significant improvement in postoperative BCVA.44 eyes had BCVA 0.9 or better. Better stability in the bag IOL placement. Re-surgery in 4 eyes for IOL decentration
Current study	243 eyes of 127 patients	Lensectomy/vitrectomy + IOL	Preoperative BCVA range 0.3–3.5 logMAR (median 1.0; Snellen 6/12 - CF) Postoperative the VA improved to 0.2–2.2 logMAR (Snellen 6/9 - CF). Aphakic eyes (53.5%; n=130) median BCVA 0.6 logMAR (Snellen 6/24). Pseudophakic eyes (46.5%; n=113) median BCVA 0.5 logMAR (Snellen 6/18)

SE=Spherical equivalent, BCVA=Best corrected visual acuity, VA=Visual acuity, IOL=Intraocular lens, CTR=Capsular tension ring, CF=Close to face, PCIOL=Posterior chamber IOL, logMAR=Logarithm of the Minimum Angle of Resolution

the IOL placement.<sup>[11]</sup> More recent studies by Maharana *et al.* including a cohort of 78 eyes concluded that pseudophakic eyes (32/78) in their series had better postoperative vision.<sup>[12]</sup> Fuerst *et al.* in a prospective series studied the artisan anterior chamber iris claw IOL outcomes in nontraumatic ectopia lentis in children. They reported that besides the improvement in postoperative BCVA, endothelial cell loss was comparable to a PCIOL implant, the mean age in their study being 11.1 years with a mean follow-up of 3.59 years.<sup>[13]</sup> Meng *et al.* studied scleral fixated IOL implantation in their series of ectopia lentis, they reported an improvement of median BCVA of 0.82 logMAR (preoperative) to 0.15 logMAR (postoperative) and an improvement of SE; however, a re-surgery rate of 9% for IOL decentration was reported.<sup>[14]</sup> Cai *et al.* report in their study of 101 eyes of 57 children under 8 years of age who received a cioni-modified capsular ring tension ring and in the bag IOL placement that in the bag placement offers better stability and the reported improvement in postoperative visual outcomes was significantly better, although re-surgery was needed in a very small number

of cases (4/101 eyes) for IOL decentration and anterior capsular contraction.<sup>[15]</sup>

The present study is an analysis of a large cohort of children with congenital ectopia lentis spanning over nearly a decade. It was a retrospective analysis with all inherent limitations attributable to a retrospective study. As a significant majority were school-going children at the time of presentation, we believe that amblyopia may have contributed to the final postintervention BCVA. Amblyopia management details were not adequately documented. Further, genetic testing although advised was not performed in the vast majority of patients due to economic and logistic constraints.

## Conclusion

The current study includes a large cohort of children presenting with ectopia lentis in the first two decades. Overall, an improvement in the median BCVA and SE was noted after intervention.

### Data availability statement

The datasets generated during and/or analyzed during the current study are available from the corresponding author on reasonable request.

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This study was financially supported by Hyderabad Eye Research Foundation.

### Conflicts of interest

The authors declare that there are no conflicts of interest in this paper.

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