Non-traumatic Ectopia Lentis in a Pediatric Ophthalmology Practice, Ibadan, Nigeria

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Abstract

PURPOSE: The aim of this study is to describe the clinical profile of patients with non-traumatic Ectopia lentis in a pediatric ophthalmology clinic in Ibadan, Nigeria.

METHODS: The clinical records of children ≤ 16 years diagnosed with non- traumatic ectopia lentis at the Pediatric Ophthalmology Clinic, University College Hospital Ibadan, from May 1, 2015 to Dec 31, 2019 were retrospectively reviewed. Information on demographic data, family history, visual acuity (VA), mean refractive error (spherical equivalent), and management was retrieved.

RESULTS: Clinical records of 25 patients were reviewed. The mean age was 8.9 (±3.41) years with a range of 2-15 years. Sixteen (64%) patients were males. All the patients had bilateral involvement. A positive family history of ectopia lentis was elicited in 6 (24%) patients. Thirteen (52%) patients had a Marfanoid habitus and superior displacement of the lens was observed in 26(52%) eyes. The presenting visual acuity was ≤ 6/36 in 38 (76%) eyes. The spherical equivalent of the refractive errors ranged from -20.00DS to +13.25DS. Twenty (40%) eyes underwent surgery within the period of the study and the best corrected postoperative visual acuity improved by 2 or more lines in 12 (60%) of operated eyes.

CONCLUSION: Severe visual morbidity was common in this cohort of patients with Ectopia lentis in our practice. Treatment provided some improvement in vision which highlights the need to encourage early presentation for care. Detailed family history is important as a few of the patients were diagnosed on our request to examine siblings with visual impairment.

Introduction

Ectopia Lentis is a hereditary or acquired displacement of the lens from its normal position.[1] In 1856, Karl Stellwag, an Austrian Ophthalmologist first introduced the term 'Ectopia Lentis' to describe an abnormal positioning of the crystalline lens relative to its natural position in the eye. Since then, it has been recognized as a hallmark of many systemic diseases; some of which have life threatening manifestations.[2] Hereditary Ectopia Lentis is frequently bilateral but rare case reports of familial unilateral ectopia lentis have been described.[3]

Congenital ectopia lentis is a rare ocular disorder. Point prevalence is reported as 0.83/10,000 live births in the Danish population.[4] The prevalence in the Nigerian population is not known, however, a few cases have been reported in the country.[5–8] Non-traumatic ectopia lentis, may occur in isolation but it is commonly associated with syndromes such as Marfan syndrome, homocystinuria and Weill–Marchesani syndrome.[9] When it occurs in isolation it may be sporadic or familial. This makes genetic testing a very important tool in the management of ectopia lentis. In view of these syndromic associations which may sometimes be life-threatening, a detailed systemic examination is pertinent for proper diagnosis, management and prognostication.

Ocular features of ectopia lentis commonly include: impaired vision, which could range from mild to severe; double vision; strabismus and refractive errors. Other clinical features such as amblyopia, secondary glaucoma, and retinal detachment can markedly impair quality of vision with profound visual morbidity.[10] Irrespective of the severity at the initial presentation, the degree of the ectopia lentis and the disease progression with the course of time vary among affected individuals.[11]

Management options may be conservative and these include the use of spectacles or contact lens. Management could also involve surgical methods which have been well described in the literature.[9,12] Classical indications for surgical therapy include situations when the lens edge is bisecting the pupil, making it impossible to achieve a good optical correction; and an anteriorly displaced lens causing secondary glaucoma.[11] Lensectomy is the most commonly performed surgical intervention with the approach (pars plana versus limbal) depending on the surgeon's expertise, as no study has shown superiority of either approach.[13]

Visual rehabilitation following surgery can be challenging and may require multiple interventions and long term follow up. Options for visual rehabilitation include iris fixated intraocular lens (IOL), Scleral fixated IOL, as well Posterior chamber IOL insertion with the use of Capsular tension rings (CTR).[12–15] Contact lenses and aphakic spectacles are alternatives to IOLs but also have significant challenges.[11,15] The management and outcome of ectopia lentis in children have not been reported in sub-Saharan Africa to the best of our knowledge. Therefore, the aim of this study is to describe the clinical profile of patients with non-traumatic Ectopia lentis in a pediatric ophthalmology clinic in Ibadan, Nigeria.

Methods

This study was a retrospective review of children aged 0 – 16 years managed for ectopia lentis at the Pediatric Ophthalmology and Strabismus Clinic, University College Hospital (UCH), Ibadan, from May 1, 2015 to Dec 31, 2019. Ethical approval was obtained from the University of Ibadan/ UCH Ethical Review Committee.

Children, aged 16 years and below, diagnosed with ectopia lentis who presented to the clinic during the study period were included in the study. Patients with ectopia lentis due to trauma as an underlying cause for subluxation as well as those with missing records were excluded from the study.

Information on demographic data, family history of ectopia lentis, visual acuity (VA), mean refractive error (spherical equivalent), and management was retrieved from the medical records. Other information collected included age at the time of surgery, and postoperative refraction. A 2-line improvement in Snellen visual acuity following management was considered a successful outcome.

Patients were referred to the pediatrician for systemic evaluation which comprise a physical examination and investigations. A cardiac evaluation with electrocardiography was done for all patients while a few had echocardiograph.

For the anterior segment approach to lensectomy, the patient was placed under anesthesia, aseptic routine cleaning and draping was done. A scleral incision of 3 mm was made and tunneled into the clear cornea. A side port 2-3mm was created in the temporally. Following anterior chamber entry, a high-molecular-weight viscoelastic material was injected into the anterior chamber, and anterior capsulorhexis was carried out. Aspiration of all lens matter from within the intact capsular bag was performed by a simcoe canula. The vitrectomy instrument was inserted into the anterior chamber and cutting mode was activated to remove the lens capsule, zonules and the anterior vitreous making sure no vitreous extended to the wounds. On completion of the procedure wounds were closed with 10/0 nylon suture. Subconjunctival antibiotic and steroid combination was given.

For the pars plana approach to lensectomy, the patient was placed under anesthesia, aseptic routine cleaning and draping was done. Three 25-gauge trocars were inserted obliquely at the pars plana, 3.5 mm from the limbus, with one trocar for an infusion cannula in the inferotemporal quadrant, and with the other two trocars for the vitreous cutter and the light source in the superonasal and superotemporal quadrant. Lensectomy ensuring complete capsular removal, and anterior vitrectomy was performed using the vitreous cutter. Trocars were removed and the entry ports were ensured to be sealed. Subconjunctival antibiotic and steroid combination was given.

The data was entered into an Excel® spreadsheet (Microsoft Corp., Redmond, WA, USA). Statistical Package for the Social Studies software (SPSS 20 IBM Corp., Armonk, NY, USA) was used to perform the analyses. For qualitative variables, the frequencies and percentage proportions were calculated. For quantitative variables, the mean and standard deviations were calculated.

Results

Fifty eyes of 25 children met the inclusion criteria. There were 16 (64%) males and 9 (36%) females. The mean age at presentation was 8.9 (±3.41) years with a range of 2-15 years. Only 8 (32%) of the patients presented before the age of 8 years.

The presenting visual acuity (VA) ranged from 6/18 to No light perception (NLP) and was equal to or worse than 6/36 in 38 (76%) eyes. Objective visual acuity measurement was not obtained for 3(12%) children. Refraction in 19 eyes showed high refractive errors with the spherical equivalent ranging from -20.00DS to +13.25DS. Glaucomatous cupping was present in both non-operated eyes of 1 (4%) patient and elevated intraocular pressure without disc cupping was found in both non-operated eyes of 1 (4%) patient. At presentation, there was superior displacement of the lens in 26 (52%) eyes, inferior displacement in 12 (24%), horizontal displacement with no vertical component in 4 (8%) while the direction of displacement was not documented in 8 (16%) eyes. Lens opacities were seen in 12 (24%) eyes and 3 (6%) eyes had pre-existing retinal detachment at presentation.

Systemic examination revealed a Marfanoid habitus in 13 (52%) of the patients. There was cardiac pathology in 4 (16%) patients. The cardiac anomalies included aortic root dilatation (2 patients), atrial

septal defect (1 patient) and cardiac arrhythmias (1 patient). One patient (4%) had a diagnosis of Weill Marchesani syndrome and 1 (4%) patient was diagnosed with Ectopia lentis et pupillae. A history of poor vision was elicited in the relatives of 12(48%) patients and a diagnosis of ectopia lentis was confirmed in relatives of 6 (24%) patients. Figure 1 shows the pedigree charting of 5 patients in our series who belong to 2 generations of the same family.

Twenty (40%) eyes of 13 patients underwent surgery within the study period. The mean age at surgery was 9.2 (±3.76) years. There were 11 (55%) right eyes and 9 (45%) left eyes. Lensectomy with anterior vitrectomy was performed in 19 (95%) eyes; via the limbal route in 16 (80%) eyes and via the pars plana route in 3 (15%) eyes. These eyes were left aphakic. One (5%) eye had Small Incision Cataract Surgery (SICS) with insertion of a Posterior Chamber intraocular Lens (PCIOL). This eye had zonular dialysis limited to only one quadrant.

The preoperative uncorrected VA ranged from 6/18 to Hand Motions (HM) and only 1 (5%) eye had a VA of 6/18. The best corrected postoperative visual acuity was ≥ 6/18 in 9 (45%) of operated eyes. Vision improved by two or more lines in 12 (60%) eyes (Figure 2). Retinal detachment as a complication of lensectomy, was observed in 2 (10%) of the eyes operated in this series. One of these eyes underwent Scleral buckle surgery with initial visual outcome of 6/36, however there was a re-detachment in the same eye after 19 months and final VA was HM. The other patient with postoperative retinal detachment was still awaiting surgery as at the time of this report, on account of lack of funds. Two (10%) eyes of different patients had elevated intraocular pressure (IOP) post operatively and were controlled on antiglaucoma medications.

Discussion

This is perhaps the largest series of non-traumatic Ectopia Lentis reported thus far in African children. The mean age at presentation was 8.9 years and the age range was similar to the range of 5-12 years reported by Shafique et al[16] in Pakistan. The youngest child in our cohort was 2 years of age and this buttresses the findings of a study in China which showed that greater than 50% of patients with Marfan Syndrome acquired Ectopia lentis before 5 years of age or during early childhood.[10] However, the majority of patients in our series presented at an age when amblyopia is likely to have developed. The mean age at surgery was 9.2 (±3.76) years and this was higher than 70.3months (5.86years) reported by Hsu et al.[13] Some other studies have reported younger median age of 52 months (4.33years) and 4.8years.[12,14]

The best corrected visual acuity (BCVA) at presentation was poor in all our patients. This is similar to reports by Hsu et al[13] in USA, Wu-Chen et al [17] in USA and Noorani et al[18] in Pakistan. This we believe is due to the combination of uncorrected high refractive errors and amblyopia due to the late presentation we observed in our patient population.

More than half of the eyes in our series had superior displacement of the lens. This is contrary to findings by Shafique et al[16] who reported 75% inferior subluxation. This could be explained by the fact that more

patients in our series had features suggestive of Marfan syndrome in contrast to homocystinuria reported in their study.

A significant proportion of our patients had high refractive errors at presentation. This is similar to the high myopic astigmatism reported by Hsu et al.[13] Also, lens opacities were seen in 12 (24%) eyes of our patients; which is quite similar to the 20% reported by both Shafique et al[16] and Noorani et al.[18] A combination of these, in addition to the older age at presentation and the extent of lens subluxation with respect to the visual axis would increase the risk of amblyopia in these patients.

The pedigree charting of 5 patients in our cohort who belong to 2 generations of the same family underscores the need for a detailed family history and examination of all family members. The proband, aged 13 years, had been enrolled in the school for the blind due to poor vision and was referred to our clinic from the school. Following satisfactory visual improvement with surgery and aphakic spectacles, her sister, brother, mother, nephew and niece also presented to the eye clinic for treatment. All these relatives except her mother are among the patients in this case series. Similarly, Noorani reported that a third of their patients had familial ectopia lentis.[18]

Management of patients with Ectopia Lentis should be comprehensive with respect to diagnosis, detection of systemic comorbidities, counselling and examination of at-risk relatives, indication and timing of surgery, surgical approach, visual rehabilitation and need for long-term follow up. In view of the progressive nature of the disease and the variability among patients, it is challenging to establish an absolute standard of care. There are still controversies as to the adequacy of conservative management.[11] A lack of improvement in vision with conservative approach is the commonest indication for surgery.[15]

For the patients who had surgical management in our series, lensectomy with anterior vitrectomy was performed in 95% of the eyes and the limbal route was used in majority of the eyes. This was largely due to surgeons' preference and lack of facilities for posterior vitrectomy in our center which is the scenario in most resource poor settings. These eyes were all left aphakic. Some previously reported postoperative complications of aphakia include transient ocular hypertension, transient vitreous hemorrhage, wound dehiscence and glaucoma secondary to pupillary block.[17]

Postoperatively, the best corrected visual acuity was ≥6/18 in only about half of the eyes in our series. This outcome could be attributed to amblyopia as they had high ametropia and late presentation. Even in the absence of any of these, ectopia lentis in a child is a known risk factor for amblyopia.[2,17] Furthermore, the sub-optimal optical correction of surgical aphakia with aphakic spectacles could be contributory.

Noorani et al[18] in their large cohort of 54 surgical eyes also corrected surgical aphakia using aphakic spectacles and contact lenses but noted significant visual improvement with 71% achieving ≥6/18. They attributed the poor postoperative vision of 6/60 - HM to preoperative glaucoma present in half of their patients and irreversible amblyopia in a about third[18] Other identifiable factors responsible for the poorer

BCVA include older age at surgery, short follow up duration, mild visual axis opacification, and residual refractive errors.[12]

On the other hand, better visual outcomes have been recorded in some studies.[9,17,19] Even though 78% of their patients were >5years at the time of surgery and there was no IOL implantation, Halpert and BenEzra[9] were able to achieve VA ≥ 20/40 (6/12) in 80% (47) of eyes. Wu-Chen et al[17] also recorded VA improvement in all cases and BCVA was ≥20/30 (6/9) in 82.4% despite a mean age of 7.7 years and no IOL insertion. Their longer mean follow-up duration of 4.6years and 8.5years respectively could have been a factor in this. Even with shorter duration of follow up of 1 year Nb et al[19] documented BCVA of ≥20/30 in 84% postoperatively. This may be attributed to use of intraocular lenses in their cohort which allows for better visual rehabilitation.

Postoperative retinal detachment observed in this series is not uncommon and has been reported in other studies.[9,18] Retinal detachment was reported up to 2 years post lensectomy in a previous series with improved visual acuity post repair.[9] Postoperative elevated IOP was also noted in two eyes in our study. Even with a mean follow-up of 4.6years Halpert and BenEzra[9] recorded normal IOP in the range of 12-18mmHg throughout the follow up period in all their patients. Long-term follow-up to monitor for glaucoma is required for children with ectopia lentis.

There are some limitations to our study. The diagnosis of Marfan syndrome in our series was difficult because of the absence of genetic diagnostic facilities to detect gene mutations. Notwithstanding, Ectopia lentis is an ocular manifestation that is considered a major diagnostic criterion and other suggestive clinical features were present leading to a clinical diagnosis of Marfan syndrome in about half of our patients. The retrospective nature of the study made retrieval of data a challenge with some missing data. Furthermore, some patients were lost to follow-up.

Conclusion

Ectopia lentis, though rare, causes profound visual morbidity. Management of ectopia lentis in a tertiary eye hospital in Nigeria provides some improvement in vision and patients should be encouraged to access care early. Detailed systemic examination and family history are important and should be routinely evaluated. The complications of our surgical approach were few, and most of such complications could be managed medically and surgically. There is a need for public health interventions to encourage early diagnosis and uptake of care.

Declarations

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Authors' contributions

M.O Ugalahi: Conceptualization, Methodology, Writing, Reviewing, Editing, Visualization

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Figures

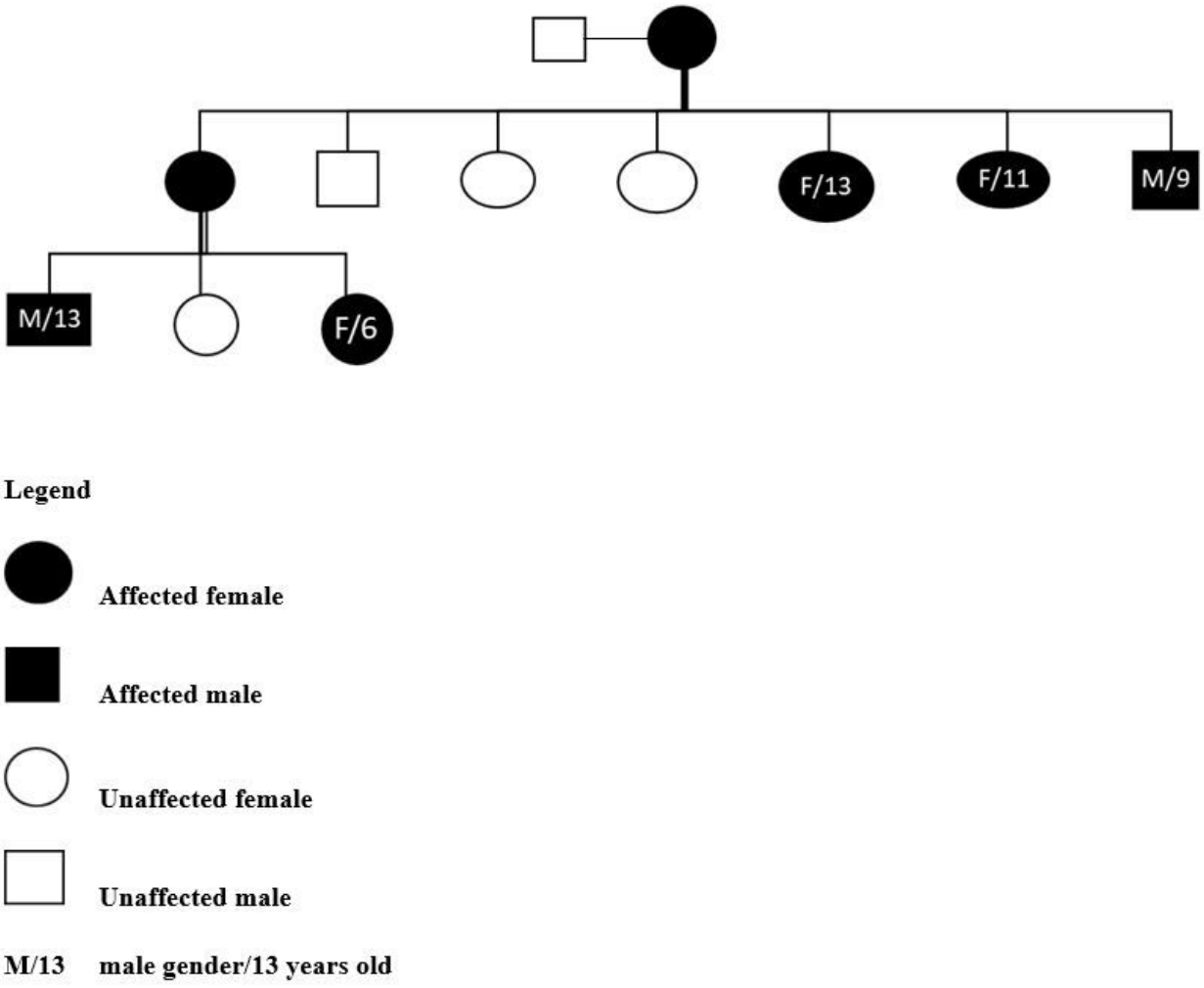

Figure 1

Pedigree charting of 5 patients in this series who belong to two generations of the same family

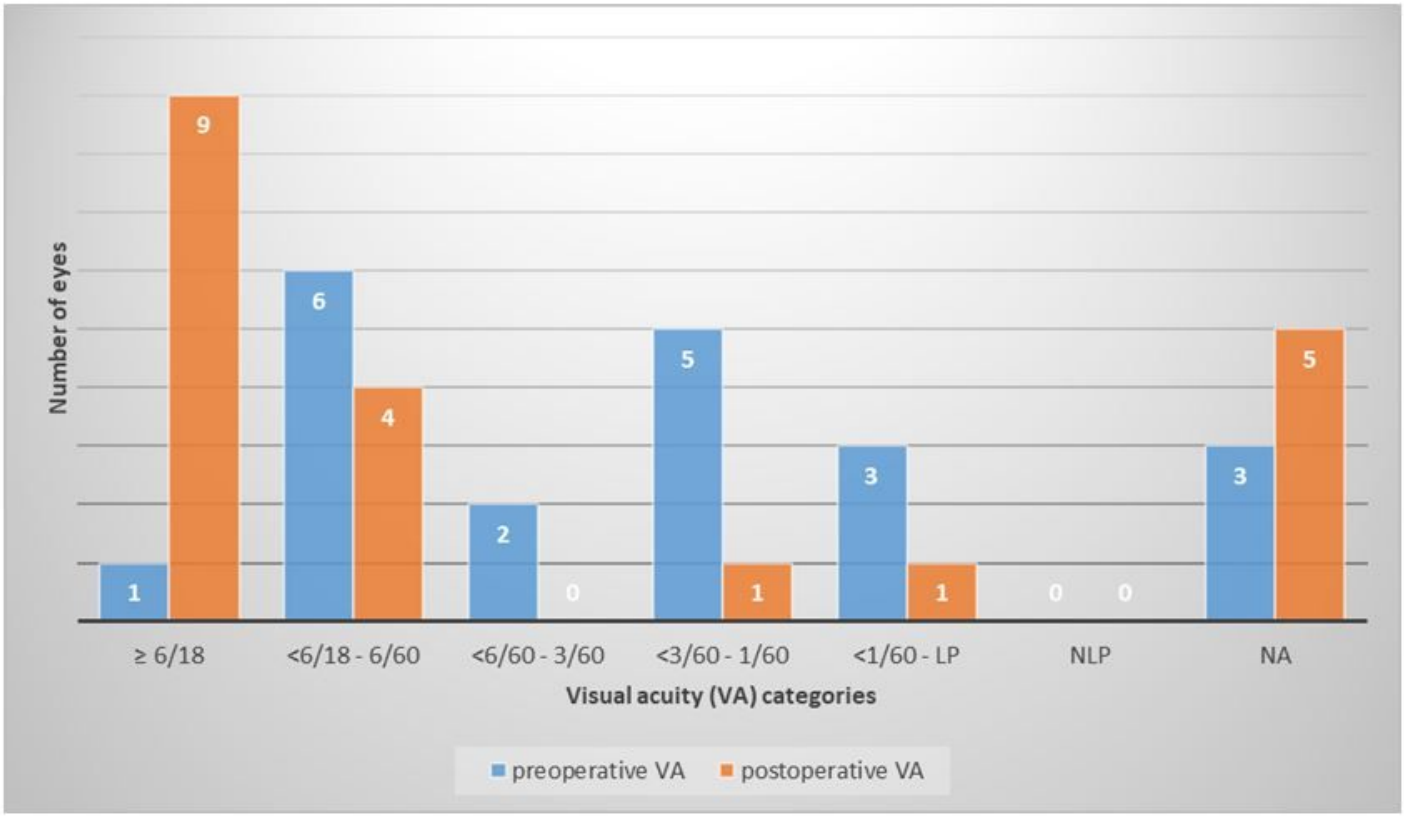

Figure 2

Preoperative and postoperative visual acuity of 20 eyes that underwent surgery for Ectopia lentis NA- Visual acuity not assessed objectively LP – Light perception NLP – No light perception VA – Visual acuity