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School Difficulties Revealing Isolated Bilateral Crystalline Ectopia, About Two Cases And Review Of The Literature

Amadou Bouba Traoré H

Faculté des sciences de la santé, Université Dan Dicko Dan Koulodo de Maradi, Service d'ophtalmologie, Hôpital régional de Maradi, Niger

Aghali Hama N

Faculty of Health Sciences, Dan Dicko Dan Koulodo University, Maradi, Histo-Embryology and Cytogenetics Department, Maradi Referral Hospital, Niger

Laminou L

Faculty of Health Sciences, André Salifou University, Zinder

Issiaka Boukari M

Makkah Ophthalmological Complex, Maradi, Niger

Abdou A

Faculty of Health Sciences, Abdou Moumouni University, Niamey, Ophthalmology Department, Amirou Boubacar Diallo National Hospital, Niamey, Niger

Abstract: ***Crystalline ectopia is a congenital malformation resulting from displacement of the lens from its normal position due to asymmetric stretching of the zonular fibers.***

It is a rare, progressive anomaly with serious complications. Lens ectopia is an autosomal dominant inherited disorder that may be associated with other systemic diseases, such as Marfan syndrome, Weill Marchesani syndrome and homocysteinuria.

We report two cases of isolated bilateral lens ectopia in male children, resulting in reduced visual acuity and difficulties at school. After an improvement in visual acuity following surgery and optical correction of the aphakia, they returned to school without difficulty.

Keywords: ectopia of the lens, Marfan, Zonule

I. INTRODUCTION

Crystalline ectopia is a congenital displacement of the lens, due to an anomaly of the zonule. [1] About half of them are frequently associated with a general syndrome, Marfan's syndrome, Weill Marchesani syndrome and homocysteinuria being the best known [2]. Berryat described the first case of lens dislocation in 1749, and Stellwag coined the term lens ectopia in 1856 [3]. It is a rare condition, accounting for 0.3% of all congenital ocular anomalies [4].

We report two cases of crystalline ectopia revealed by school difficulties in two adolescents seen in consultation in our ophthalmology unit.

II. CASE REPORTS

CLINICAL OBSERVATION 1

This is a 12-year-old male patient referred by a family member for bilateral decreased visual acuity. He was the first

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of a sibling group of four (4) children, all alive and well; there was no parental consanguinity, and the pregnancy was well attended with a vaginal delivery. He showed no psychomotor developmental delay. General examination revealed normal height and weight for his age, and the absence of a long skeleton.

At school age, he had difficulty keeping up with classes at the start of attendance. His visual acuity was reduced to counting fingers at one (1) meter in the right eye and finger movement in the left eye; orbito-facially, he presented a divergent strabismus in the right eye.

Ophthalmological examination revealed the following in both eyes: anterior segment with clear cornea and increased anterior chamber depth; supra-temporal lens apposition in both eyes, indicating crystalline ectopia (figure 1); and normal ocular tone of 12 mmHg. Fundus examination revealed a physiologically excavated papilla with good foveolar reflex; a flat retina in both eyes; and no posterior pole lesions. The patient's general examination by the paediatrician revealed no obvious abnormalities, and an extension work-up was requested, including ultrasound of the heart and vessels, with no cardiac abnormalities found. Amino acid electrophoresis for homocystinuria in urine was negative.

CLINICAL OBSERVATION 2

This 13-year-old male patient was brought to the clinic by his aunt. He is the seventh of seven (7) children, all alive and well; no parental consanguinity, well monitored pregnancy and vaginal delivery, followed by the mother's death after delivery; no psychomotor development delay. At school age, he had difficulty seeing in the dark, which is why he was referred to our ophthalmology unit.

His visual acuity was reduced to one finger movement in the right eye and counting fingers at one (1) meter in the left eye; the rest of the ophthalmological examination was normal except for crystalline ectopia in both eyes in the superotemporal position (figure 2).

General examination showed normal size for his age, with no long skeleton.

Cardiovascular work-up and homocystinuria tests were unremarkable.

The management of our two cases consisted of phacophagy through a scleral tunnel in a single operation on both eyes, alternating with anterior vitrectomy without implant placement, as there was no capsular support at the time of surgery. The early post-operative course was straightforward, marked by slight corneal edema which quickly resolved by the third post-operative day.

Our two patients were followed up after surgery with a combination of antibiotic and local corticosteroid therapy (DEXAMETHASONE PHOSPHATE 100 mg/100ml, neomycin sulfate 350000 IU/100ml) four (4) times a day in both eyes, and a local anti-inflammatory agent, INDOCOLLYRE 0.1% eye drop solution and single-dose container. After one month post-operatively, the patients had their Aphakia corrected with +10 diopter glasses, which improved visual acuity to 1/10 in both eyes for case N°1 and 2/10 for the second. With the optical correction of Aphakia, both children have returned to school and continue their education in much better conditions than before their consultation in our ophthalmology unit (Figures 3A-B).

III. DISCUSSION

Crystalline ectopia is a congenital malformation resulting from displacement of the lens from its normal position due to asymmetric stretching of the zonular fibers [4]. It is a rare, progressive anomaly with serious complications [4]. Most often unnoticed at birth, it becomes obvious and disabling at a variable age, usually between 5 and 10 years [2].

The most common etiologies of lens ectopia are Marfan syndrome, simple ectopia, Weill Marchesani syndrome and homocystinuria [5]. Superior-temporal dislocation is the most common, but all directions are possible [5].

In the majority of cases, crystalline ectopia is autosomal dominant, with no associated systemic abnormality [4]. The literature notes a high frequency in boys in the occurrence of lens ectopia. The cases we report here are in line with those observed by several authors. Recessive mutations in the ADAMTSL4 gene (1q21.2) and dominant mutations in the FBN1 gene(15q21.1) have been identified as causative. [7]

Moustaine and associates in Casablanca, Morocco, found an 80% male predominance in their series [4]. Sorath N and associates in their series found that out of 50 patients, 26 were male and 24 female [3]. Wen Y in his series found the same observation with (60%) males [5]. Decreased acuity or poor visual behavior is responsible for poor school results or difficulties, hence the reason for consultation in this series of observations. Lens ectopia is most often bilateral, as was the case in our series.

Diagnosis is based on ophthalmological examination data and can be confirmed by molecular cytogenetics by locating the mutated gene. [7]

In our series, molecular cytogenetics was not performed because of the unavailability of the laboratory and the family's extremely precarious situation. However, genetic counselling was strongly recommended.

Indeed, displacement and deformation of the ectopic lens are responsible for amblyogenic and progressive severe ametropia (severe myopia in 48% of cases or severe hyperopia in 16.5% of cases) [4]. Visual prognosis depends on the degree of lens dislocation, age at onset of disease, age at initiation of treatment, and complications. [7]

Treatment is surgical, targeting major displacements. Elsewhere, surveillance is required for dislocation, secondary glaucoma or retinal detachment. [1]

IV. CONCLUSION

Lens ectopia is a rare pathology; it is easy or simple to diagnose; its management must be early, given the amblyogenic nature of the disease.

Awareness of the occurrence and evolution of this pathology is essential.

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CONSENT

Patients have given their Oral and Informed Consent for the publication of these case reports and accompanying images.

AUTHORS' CONTRIBUTIONS

All authors have contributed to the work. They also declare to have read and approved, and to have made amendments to the final version of the manuscript.

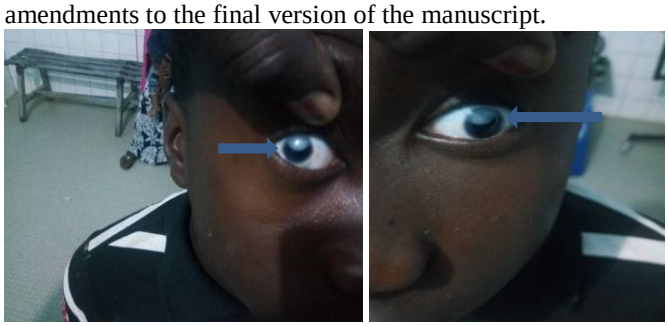

Figure 1: Bilateral lens ectopy in superotemporal position

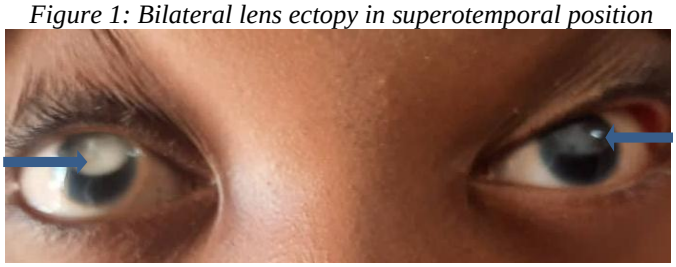

Figure 2: Bilateral lens ectopy in superotemporal position

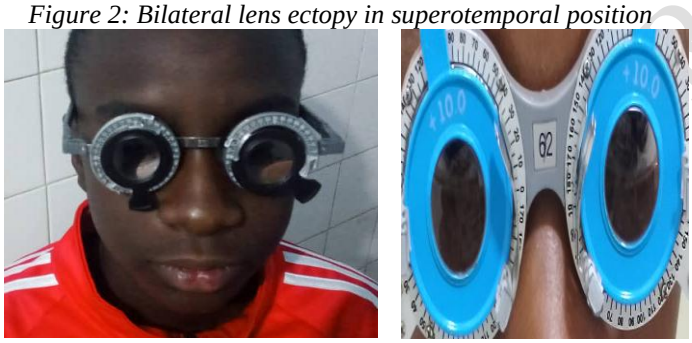

Figure 3: a and b correction of Aphakia after surgery

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