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CRISTALINIAN ECTOPIA: Clinical Study And Therapeutic Difficulties About 50 Cases

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ABSTRACT

This study highlights the prevalence and clinical presentation of crystalline ectopia in a cohort of patients, predominantly from consanguineous marriages. Key findings include a mean age of diagnosis at 12 years, bilateral lens displacement, and associated myopia in 90% of cases. Effective management involved surgical interventions and amblyopia treatment, leading to improved visual acuity in 75% of patients. The study emphasizes the importance of early detection and comprehensive care in managing this condition.

Introduction :

Crystalline ectopia is a rare congenital malformation where the lens is displaced from its normal position due to uneven stretching of the zonular fibers. This condition is progressive and can lead to serious complications. Common symptoms include blurred vision, double vision, difficulty focusing, and refractive issues. Crystalline ectopia can also occur as a result of systemic diseases, and its transmission can be either autosomal dominant or autosomal recessive. Diagnosis is typically made based on clinical examination, without the need for specialized tests. Our study aims to explore the causes, prevalence, clinical presentation, and associated systemic conditions of this disease, with a focus on the important role of ophthalmologists in managing patients with crystalline ectopia.

Materiel et methodes

We report a retrospective study of 50 patients, collected between 2019 and 2023 at the ophthalmology department B of the specialty hospital in Rabat. Our study included children managed for crystalline ectopia . Patients presented with refractive disorders, diplopia, delayed schooling or at the stage of complications.

Resultats :

This study focuses on patients with genetic conditions linked to visual disorders, particularly those from consanguineous marriages. Specific conditions examined include Marfan syndrome, homocystinuria, and other forms of crystalline ectopia.

In our study, 65% of patients were from consanguineous marriages. Age at diagnosis ranged from 2 to 25 years, with a mean age of 12. The main symptoms observed are reduced visual acuity and poor visual behavior. Decline in visual acuity is progressive in 80% of cases and abrupt in 20%.

Ophthalmological examination on admission reveals visual acuity ≤ 1/10 in 85% of patients and ≥ 1/10 in 15%. Crystalline ectopia was bilateral in all patients, with anterior crystalline dislocation present in 5% of cases (5/100 eyes) on admission and posterior crystalline dislocation present in 2% of cases (2 eyes). Lens displacement varied from patient to patient (Fig. 2), with superior-temporal displacement predominating in 30% of cases and superior-nasal in 25% (Fig. 3), but also inferior or superior. Associated myopia was noted in 90% of patients.

Ophthalmological and pediatric diagnosis:

- Isolated crystalline ectopia: 10 patients

- Marfan syndrome: 29 patients

- Homocystinuria: 11 patients

In terms of complications, retinal detachment was noted in 5 patients and was treated surgically. Peripheral degenerative lesions were observed in 6 patients and were treated with Argon laser.

In terms of management, all patients underwent phacophagia and anterior vitrectomy. Thromboembolic complications were prevented in 11 patients with homocystinuria. Surgical treatment was complemented by the use of aphakic glasses and treatment of amblyopia.

Progression was marked by a final visual acuity greater than 2/10 in 75% of patients. Postoperative retinal detachment occurred in two eyes.

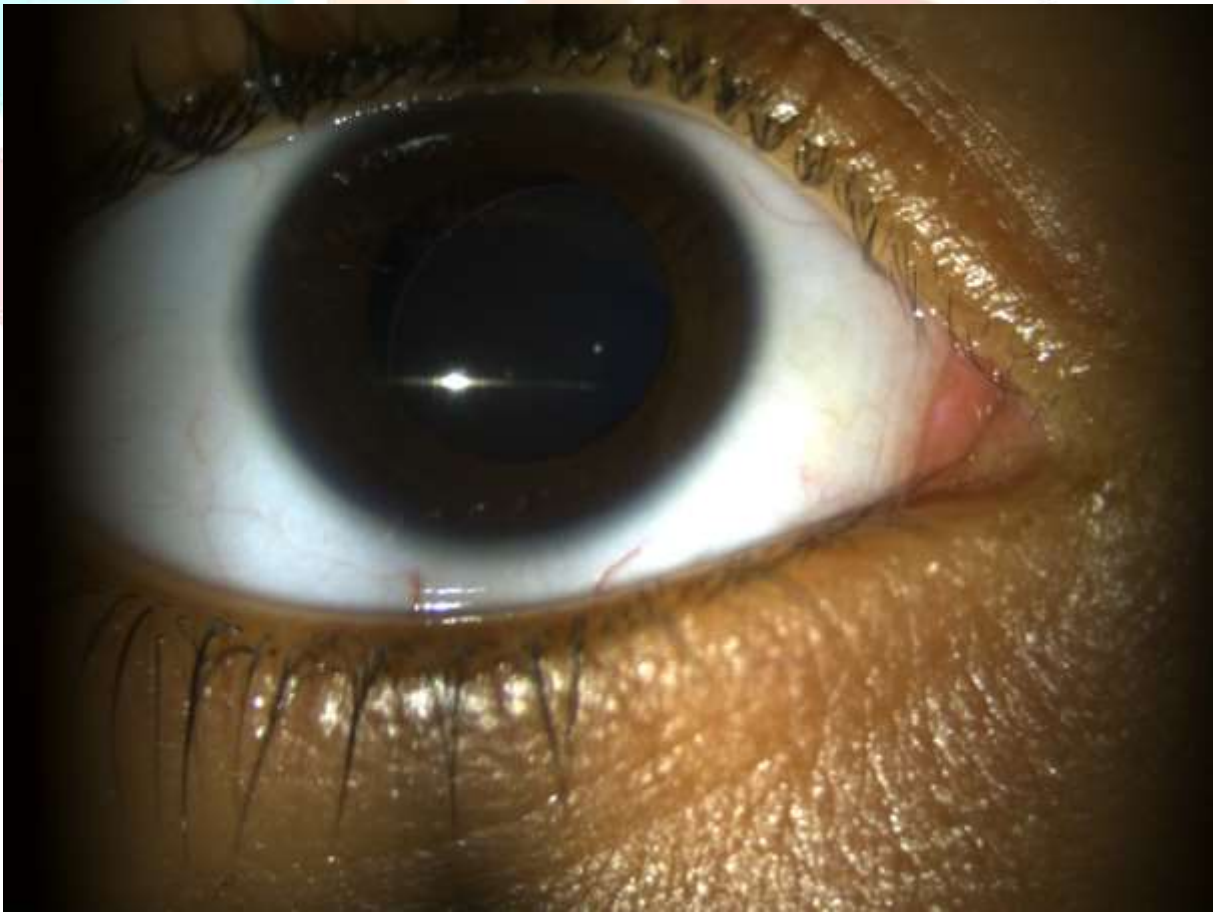

Figure 1 : ectopia of the lens sup inferonasal

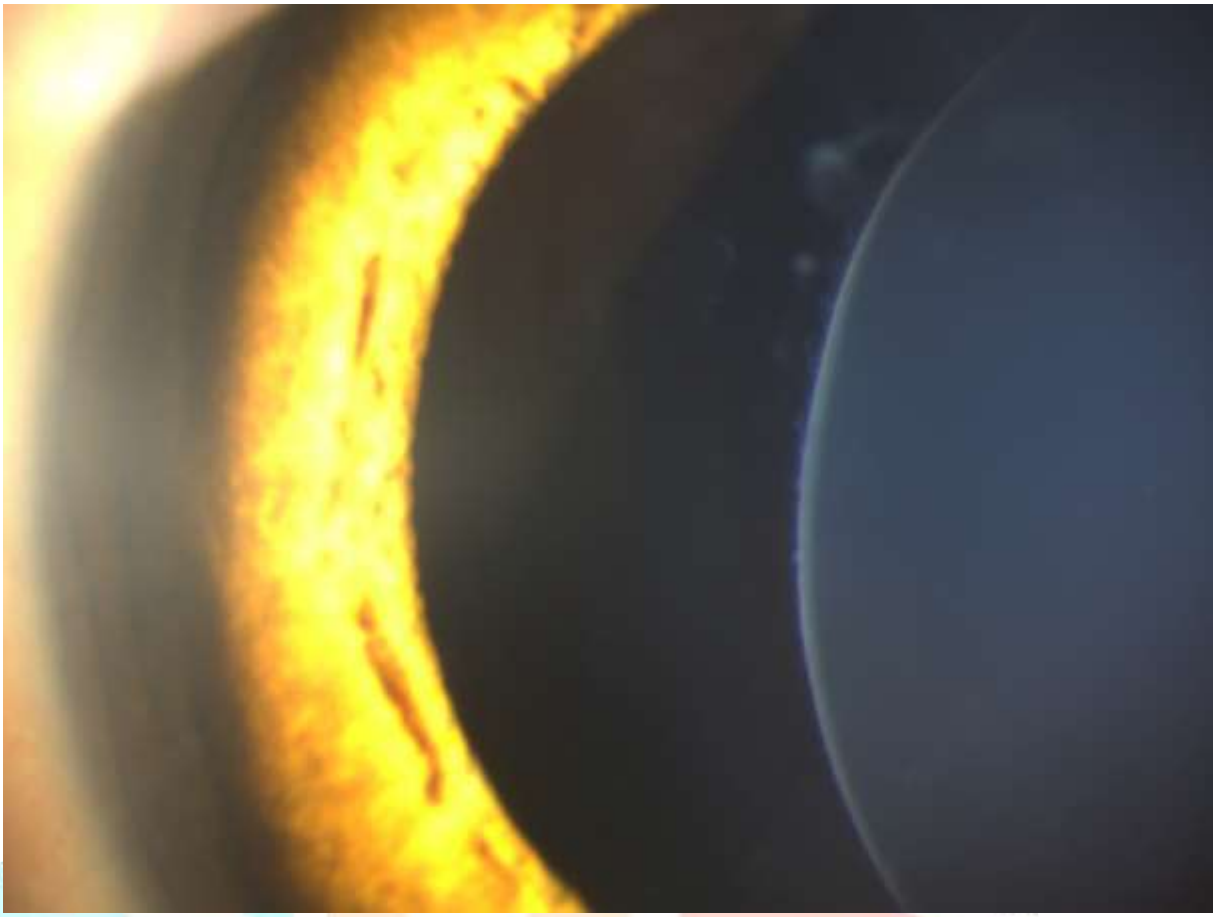

Figure 2 : ectopia of the lens with zonular rupture

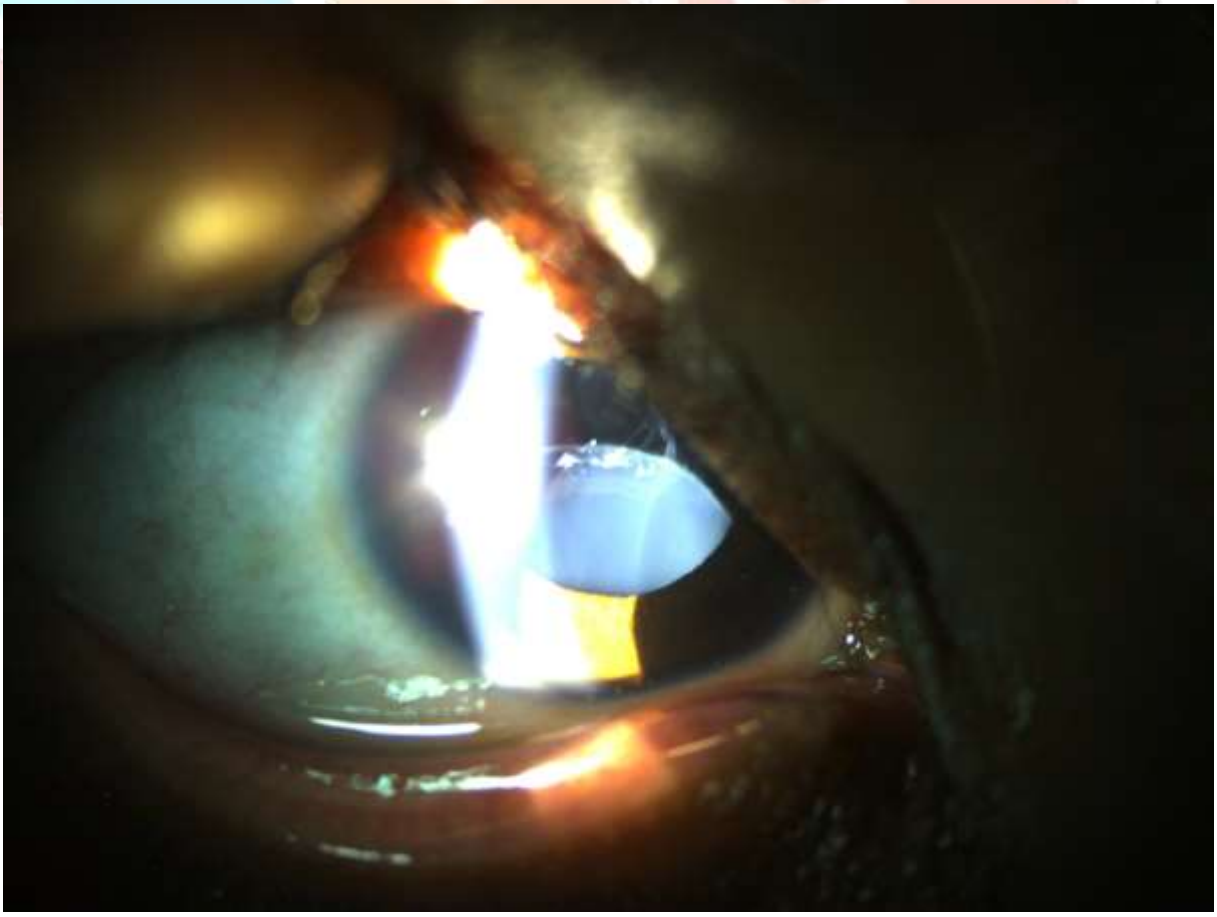

Figure 3 : subtotal subluxation on marfan

Discussion

Our study indicates that 65% of patients were from consanguineous marriages. A study by Bittles (1)showed that consanguinity increases the prevalence of rare genetic diseases, particularly in populations where consanguineous marriages are common. Al-Hussain et al. (2) also reported a high incidence of genetic disorders in inbred populations, including Marfan syndrome and homocystinuria.

The age at diagnosis in your study (2 to 25 years, mean age 12 years) is similar to other studies. For example, according to Faivre et al. (3) patients with Marfan syndrome are often diagnosed at a young age (mean 15 years), with similar symptoms of reduced visual acuity and lens dislocation. In the Brenton et al. (4) homocystinuria study, patients are often diagnosed in childhood or adolescence with symptoms of lens dislocation and visual problems.

The main symptoms observed in your study are decreased visual acuity and poor visual behavior, with a progressive decline in 80% of cases. Similar results are reported in studies of Marfan syndrome and homocystinuria, where progressive visual impairment is a common feature. The study by Bhandari et al. observed severely reduced visual acuity in patients with lens dislocation, similar to your findings(5).

Miksch et al. reported high success rates for retinal detachment surgery in patients with genetic eye disorders. All our patients benefited from phacophagia and anterior vitrectomy(6).

Traboulsi has demonstrated that surgical procedures such as anterior vitrectomy and the use of aphakic spectacles are effective in improving visual acuity in patients with crystalline ectopia(7). In terms of amblyopia treatment, a study by Birch et al. showed significant improvements in visual acuity in children treated early for amblyopia linked to genetic disorders(8).

CONCLUSION

cristalline ectopia Early diagnosis and intervention, particularly in populations with high rates of consanguinity, are essential to improve visual outcomes. Surgical interventions such as phacophagia and anterior vitrectomy, combined with the use of aphakic spectacles and treatment of amblyopia, have shown promising results in terms of improving visual acuity and managing complications.

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