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School Delay In A Child Revealing Marfan Crystalline Ectopia In A Family

Auteurs:

Salma Hassina, Robbana L, Krichene MA,Hasnaoui I, Tebbay N,hazil Z, Bekkar B,Bardi C,Sinnate A,Akkanour Y,Serghini L,Abdallah EL, Berraho A

Ophthalmology B, Specialty Hospital Rabat Morocco

Summary:

Introduction: Marfan syndrome, a rare genetic disorder, presents a variety of symptoms, including visual disturbances associated with crystalline ectopia. Early diagnosis and multidisciplinary follow-up are essential to improve patients' quality of life and prolong their life expectancy.

Observation: We report the case of a family with marfan ectopia of the crystalline lens, discovered when their young child was behind in school.

Conclusion The presence of a hereditary factor in the family reinforces the diagnosis. Surgery, notably phacophagy with anterior vitrectomy, is often necessary to treat severe ocular complications.

Key words: ectopy, marfan,retardation,lens

Introduction:

Marfan syndrome (MS) is an autosomal dominant disorder, a rare pathology resulting in multisystem disorders that mainly affect soft connective tissues (1). In children, all systems can be affected, but the symptoms that lead to the diagnosis of SM are most often skeletal (large stature associated with scoliosis and/or thoracic deformity) or ocular (2).

Materials and methods:

In our article, we report the case of a small child referred during a school screening caravan for educational delay due to visual disorders. A thorough examination revealed ectopia in the father, sister and grandmother.

Observation :

Case 1: This is a 6-year-old child with a second-degree consanguinity with a sibling of two. The child was referred for visual disorders. The ophthalmological examination revealed a visual acuity at near finger count bilaterally, which could not be improved with correction. The child also had a strong myopia of -11D and -14D. The adnexal examination was normal, but the anterior segment examination revealed a superotemporal subluxation on the right and a ruptured zonule on the left in the superonasal region. All zonules appeared to be ruptured (figure 1). The fundus was normal except for chorioretinal atrophy.

The ocular pressure was normal in both eyes at 12mmHg. The complete general examination, as well as the pediatric and cardiological examinations, were normal.

Further questioning was carried out with the mother, who reported that the sister, father, and paternal grandmother had visual problems. They were called in for a complete ophthalmological examination :

The sister's examination showed that her visual acuity was 4 meters bilaterally, and it could not be improved with correction. She had high myopia with a prescription of -13D in one eye and -12D in the other. The examination of the front part of her eyes revealed that her lenses were partially dislocated upwards, with a torn structure called the zonule. There was also a formation called a palisade at the 9 o'clock position in the inner part of the right eye, along with diffuse chorioretinal atrophy in both eyes. The pressure inside her eyes was normal at 13 mm Hg.

The cardiological examination found a heart murmur caused by a problem with the mitral valve. The results of the biological tests were normal, and the X-rays of the chest and spine showed no abnormalities. The cardiac ultrasound revealed that there was leakage of the mitral valve, which was treated with beta-blockers.

Both children were found to have mental retardation.

In Case 3, the father's visual acuity with finger-counting at a distance of 1 meter was 2/10 in his right eye with a correction of +11, and 2/10 in his left eye with a correction of +13. The father had no lenses in either eye, and the examination of the back part of his eyes showed that his lenses were dislocated in both eyes. The pressure inside his eyes was 23 mm Hg in the right eye and 21 mm Hg in the left eye. An ultrasound scan confirmed that the lenses were dislocated in both eyes, and it also revealed that the retina was flat and the axial length of the eyes was 25 mm.

Case 4: the grandmother's acuity was 3/10 with a correction of +14 on the right and +12 on the left. Eye pressure was 20 mm Hg on the right and 21 mm Hg on the left. Both eyes were aphakic. The fundus was difficult to perform due to the patient not dilating. An ultrasound scan showed bilateral lens dislocation and cataract. The retina was flat with an axial length of 26.5 mm (figure 4).

The father and grandmother had a long Marfan morphotype, and the clinical examination of the other devices was normal. Biological examinations were normal, as were chest, spine, and limb X-rays. Cardiac ultrasound was without abnormality.

The patients underwent surgery: the two children underwent lens extraction with anterior vitrectomy of both eyes, with a 1-month interval between the two eyes. The father and grandmother underwent complete posterior vitrectomy followed by laser banding.

Discussion:

Marfan syndrome (MDS) is a rare disorder that affects both sexes equally. The first case was reported in 1896 by Marfan himself. It is caused by mutations in the gene that codes for fibrillin-1. This genetic disease can affect various parts of the body, including the heart and blood vessels, lungs, bones, joints, eyes, and skin. One common manifestation is the weakening of the aorta or an increased risk of heart problems (3).

The clinical expression of the syndrome primarily involves musculoskeletal damage. This, along with ocular and cardiac involvement, is a major diagnostic criterion (4).

According to several studies conducted by different authors, the diagnosis of MDS in the absence of a hereditary factor is based on major criteria, which include the involvement of two different systems and the involvement of a third organ. In the presence of a hereditary factor, which is also a major criterion, the involvement of another system is sufficient for diagnosis. In our case, the hereditary factor was present (5).

Among the numerous ocular anomalies observed in patients with Marfan syndrome, lens ectopia is the most common, affecting 50-80% of individuals, and is typically bilateral. The severity of lens ectopia can vary from slight displacement visible only when the pupil is dilated to significant subluxation, which places the lens equator in the pupillary axis and causes monocular diplopia (6).

Minor ocular criteria include an abnormally flat cornea, increased axial globe length, and hypoplastic iris or hypoplastic ciliary muscle leading to anisocoria. The major ocular criterion is crystalline ectopia. Since 2010, crystalline ectopia has been considered a fundamental clinical feature of Marfan syndrome, facilitating its differentiation and improving accuracy in diagnosis (7) (8).

When the ectopia is uncomplicated and does not cause significant refractive errors, optical correction is the preferred treatment. Surgery is recommended for ectopia cases where visual acuity is less than 3/10, according to some authors (9) (10). Phacophagy combined with anterior vitrectomy through the pars plana remains the most commonly performed procedure (78%). This is believed to reduce the risk of postoperative retinal detachment by decreasing traction forces on the vitreous base. After phacoemulsification, some authors have suggested optical correction using a posterior chamber, scleral-fixed intraocular lens (11).

Conclusion:

Marfan syndrome, like most syndromes, cannot be cured. However, some of its symptoms can be treated. Early detection and ongoing monitoring of children suspected of having the syndrome can improve their quality of life and significantly increase life expectancy. A multidisciplinary approach is crucial for providing optimal care to patients and their families.

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Figures :

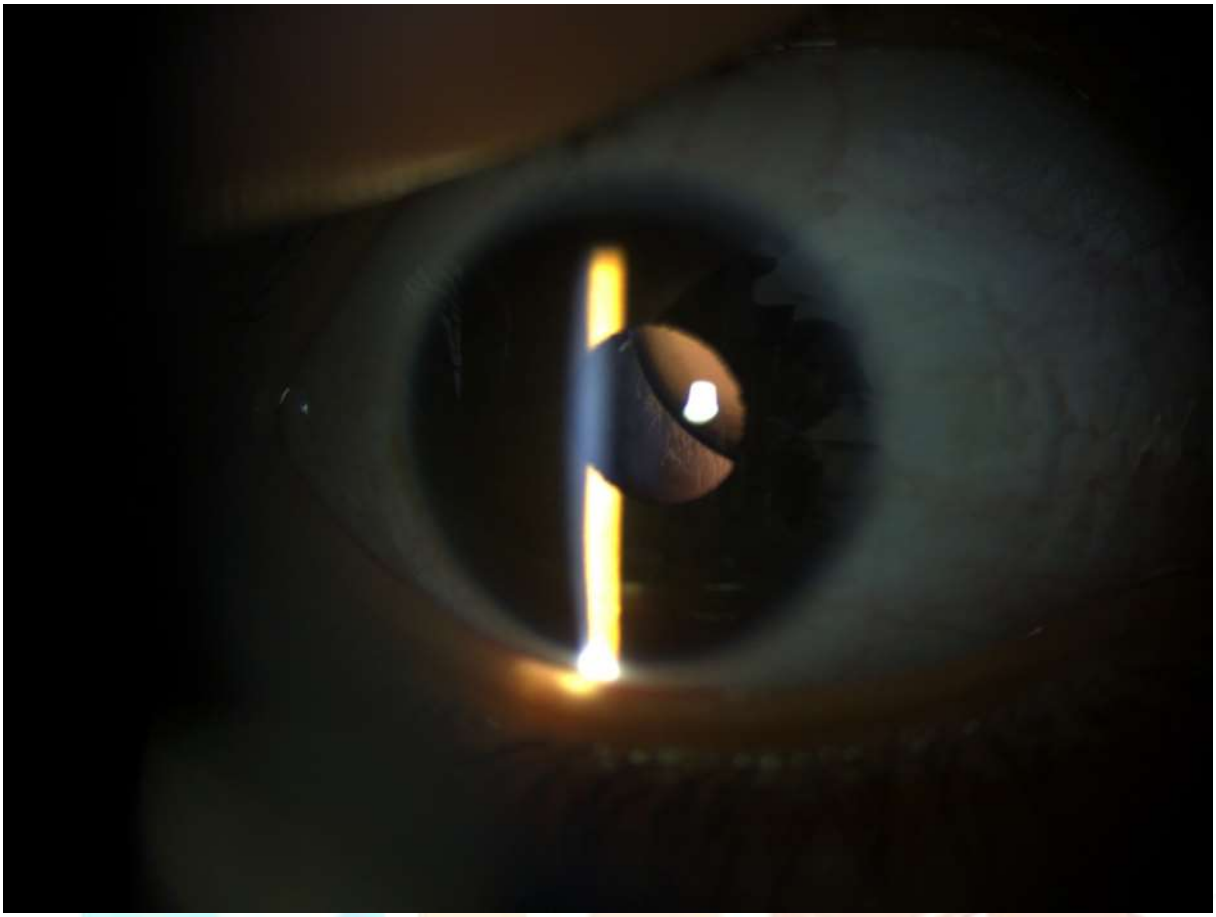

Figure 1 A: right superotemporal subluxation with ruptured zonule

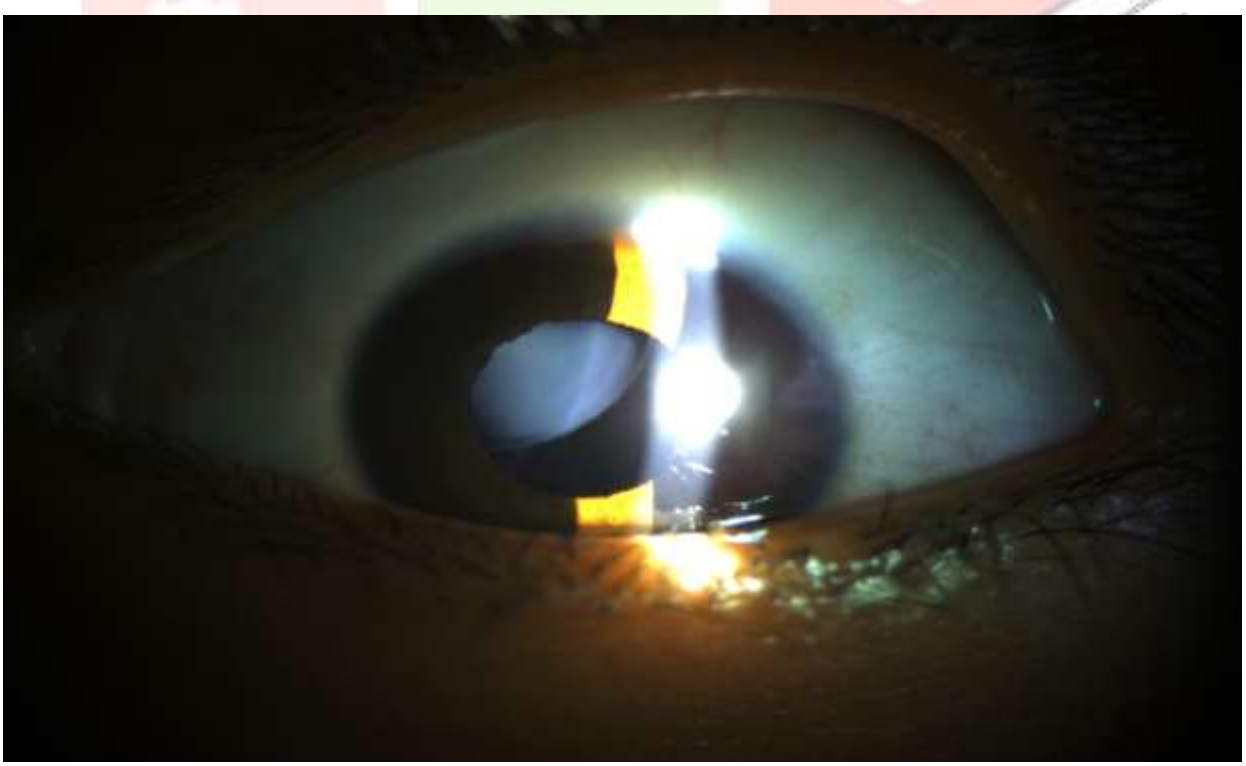

Figure 1 B: supero nasal subluxation all zonules appeared ruptured

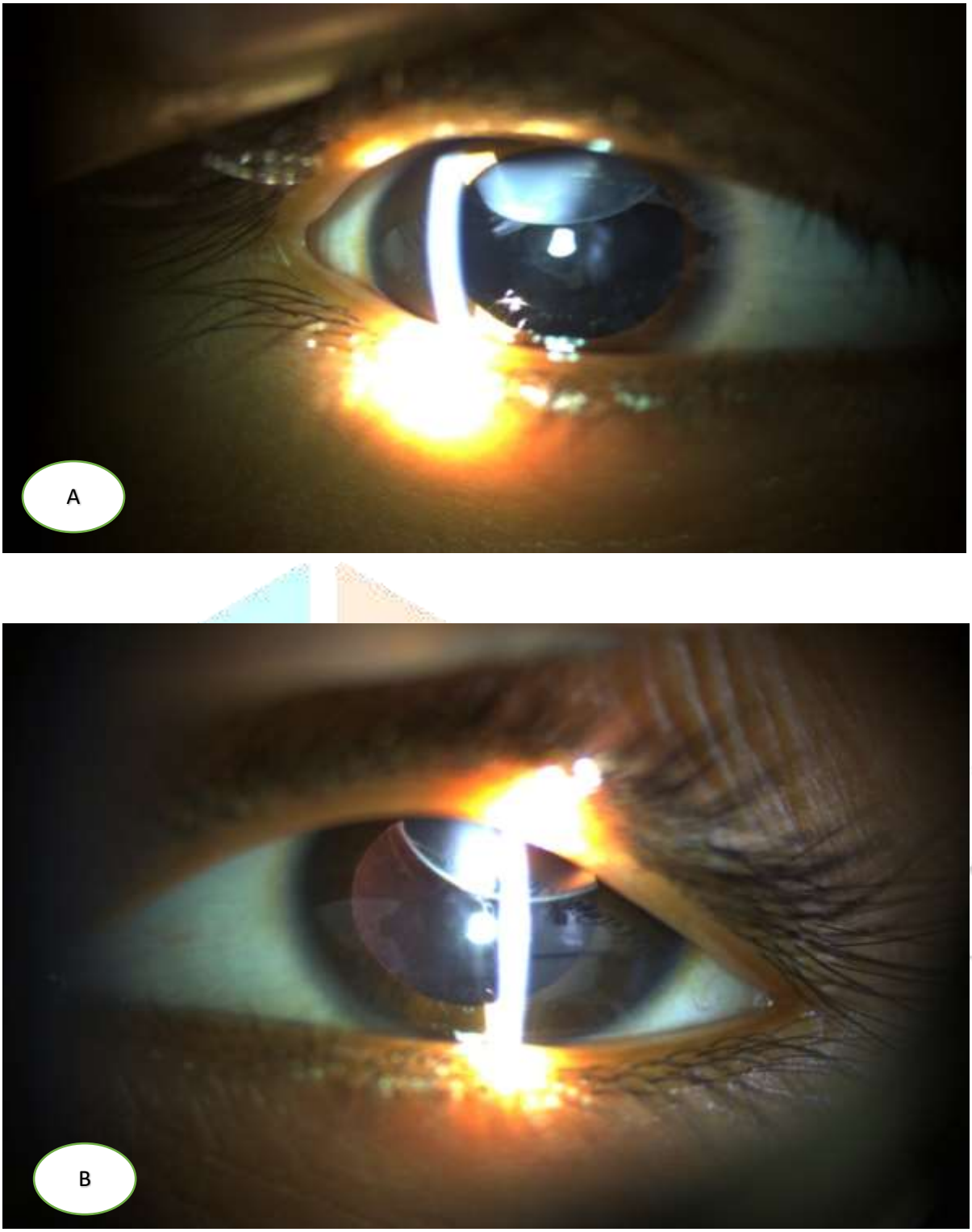

Figure 2 A and B: bilateral superior subluxation

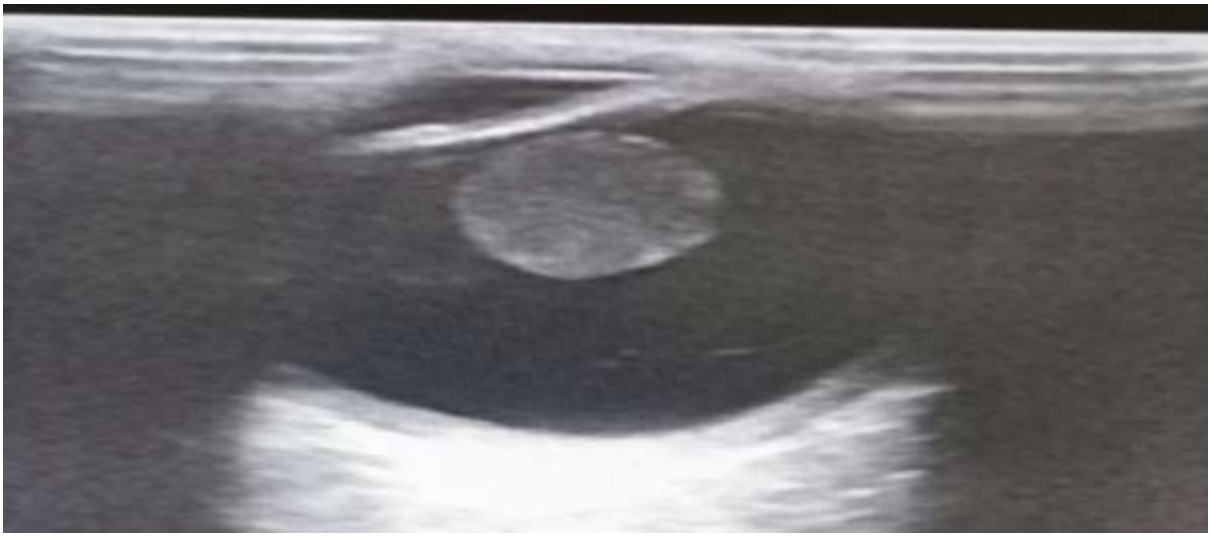

Figure 3: Lens dislocation in the father

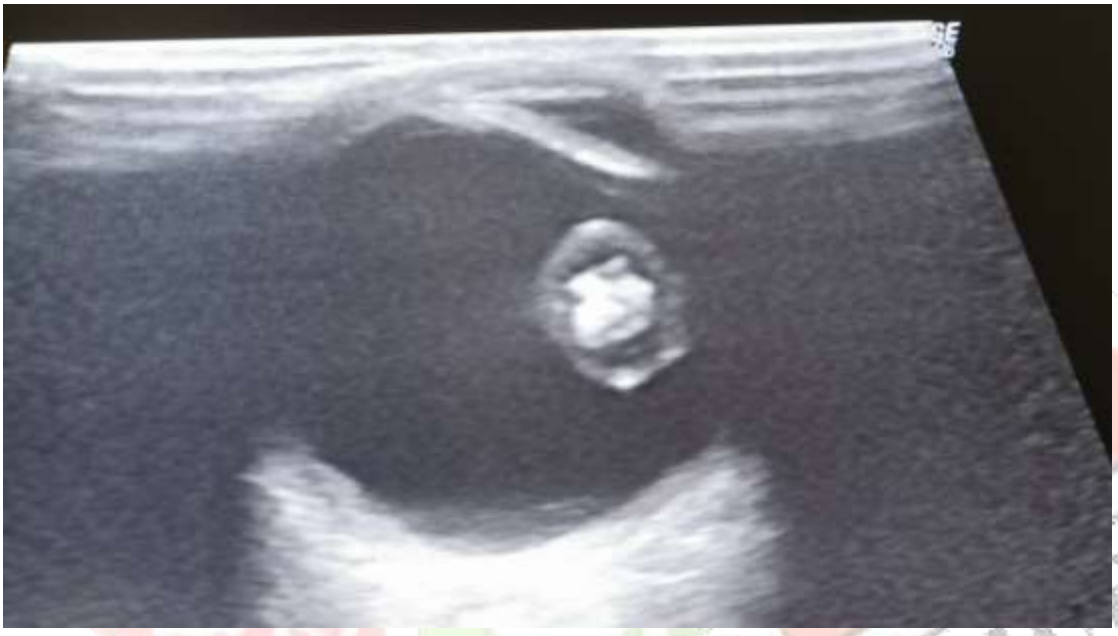

Figure 4: Cataract lens dislocation in the grandmother