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Case Report

Lens Dislocation in a 9-Year-Old Child with Marfan Syndrome

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Abstract

Introduction: Marfan syndrome is a rare genetic disorder linked to the mutation in the FBN1 gene, which codes for fibrillin. It is transmitted in the autosomal dominant mode. Its prevalence is estimated at 1/5000 individuals. Cardiovascular, skeletal and ophthalmic disorders are the most frequent

Clinical Observation: We report the case of a 9-year-old male child with uneventful family history brought by his parents for blurred vision. The visual acuity without correction was "count fingers at 2 meters". Slit lamp examination showed bilateral posterior dislocation of the lens in the vitreous, the Intra-ocular pressure (IOP) was 12 mmHg in both eyes, the fundus was normal. The general examination found a tall child measuring 1.48 m of height for 26 kg. He had slender limbs showing the sign of the thumb (Steinberg) and the sign of the wrist along with a moderate pectus excavatum. Cardiac ultrasound showed dilation of the ascending aorta. According to Gant's criteria (Ghent), we concluded to the diagnosis of Marfan syndrome.

Discussion: The diagnosis of Marfan syndrome is difficult to establish. At first, it is mainly clinical with the association of major and minor signs. Genetic research is possible as well as skin biopsy.

Lens ectopia is the most common ophthalmologic manifestation in Marfan syndrome. According to Gant's criteria, we concluded to the diagnosis of Marfan syndrome.

Conclusion: Marfan syndrome is oftentimes discovered during an ophthalmic consultation. The management of its various disorders and the follow-up imply a frank collaboration between many practitioners.

Keywords: Dislocation of the Lens; Marfan; Child

Introduction

Marfan syndrome is a genetic disorder linked to the mutation of the FBN1 gene, which codes for fibrillin on chromosome 15, sometimes to TGFBR1 or 2 [1-3]. It is transmitted in the autosomal dominant mode. Its prevalence is estimated at 1/5000 individuals [4]. It was first described by Antoine - Bernard Marfan in a young girl with unusual musculoskeletal characteristics in 1896, while Bürger first described ophthalmological characteristics in 1914 [5,6]. Multisystemic disorder, cardiovascular, skeletal and ophthalmological disorders are the most frequent [3,7]. The diagnosis of Maran's syndrome is difficult to establish. At first, it is mainly clinical with the association of major and minor signs [8,9]. Genetic research is possible as well as a skin biopsy. Histological studies have shown an abnormal expression of fibrillin in the lens capsule and the ciliary epithelial surface at the attachment of the zonules [10]. Ectopia of the lens is the most common ocular manifestation in Marfan syndrome. It is found in 50 to 80% of affected eyes and around 41% are initially seen for ocular pathology and diagnosed by an ophthalmologist [4,11,12]. Rare pathology, we report the case of a child with bilateral posterior dislocation of the lens.

Clinical Observation

He was a 9-year-old child with no known family history brought in our office by his parents for blurred vision. The visual acuity without correction was "count fingers at 2 meters". Slit lamp examination showed bilateral posterior dislocation of the lens in the vitreous (Figure 1), IOP was 12 mmHg in both eyes, the fundus was normal. The general examination shows a tall child measuring 1.48 m for 26 kg, slender limbs showing signs of the thumb (Steinberg's sign) and the wrist, a moderate pectus excavatum (Figure 2). Cardiac ultrasound showed dilation of the ascending aorta (Figure 2). According to Gant's criteria (Ghent), we made the diagnosis of Marfan syndrome.

Figure 1: Bilateral lens dislocation in the vitreous.

Figure 2: Skeletal features: slender limbs (a), sign of the thumb (b), sign of the wrist (c).

Discussion

Lens ectopia is the most common ophthalmologic manifestation in Marfan syndrome, 50 to 80% [4,11,12]. Although the disease is first described in a young girl [5], Charif-Chefchaouni., *et al*. reported a male predominance in a series of 20 cases with an average age of 14 years whose extremes range from 4 to 41 years [11]. A male predominance of 53% is reported by Mary J Roman., *et al* [13]. However, Kumar A., *et al*. reported a female predominance in a series of 4 cases including 3 women [14]. Laffargue F., *et al*. reported a series of 17 cases aged less than 10 years [15]. The ophthalmic complications of Marfan are among others, posterior dislocation of the clear or opacified lens, rhegmatogenic detachment of the retina, secondary glaucoma [11,14,16]. In our series the fundus was normal as in that of Omolasse., *et al*. [4] in a 12-year-old girl. Cardiovascular and skeletal damage are also common. Dilation of the ascending aorta is a major sign of cardiac involvement [7] which can be complicated by dissection and aortic rupture, aortic or mitral insufficiency leading to cardiac insufficiency [2,15]. The skeletal manifestations reported by several authors are marked, among other things, by the large size of the subjects, ligament hyperlaxity, scoliosis, signs of the thumb and wrist, pectus carinatum and pectus excavatum, flat feet [2-8,10-15]. Scoliosis and deformation of the rib cage in the keel or funnel can lead to a restrictive disabling syndrome, thus the frequency of apical bubbles and spontaneous pneumothorax is increased [2]. Pneumothorax affects 5 to 11% of patients [6]. In our case, there was pectus excavatum without lung damage.

Conclusion

A multi-system disorder, Marfan syndrome is most often discovered during an ophthalmological consultation. The management of the various disorders and the follow-up imply a frank collaboration between ophthalmologists, cardiologists, orthopedists, pulmonologists and many others.

Conflict of Interest

No conflict of interest.

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