MARFAN’S SYNDROME: CASE REPORT AND LITERATURE REVIEW

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SUMMARY

Marfan’s syndrome is a rare genetic disease, autosomal dominant. The most affected organs are eyes (myopia, subluxation of the lens); skeleton (hyperlaxity, arachnodactyly, scoliosis, dolichostenomelia) and cardiovascular system (aortic pathology). The severity of this disease is related to its cardiovascular damage. We proposed to carry out a review of the literature from the first case reported in the ophthalmology department of the University Hospital of Brazzaville.

INTRODUCTION

Marfan’s syndrome is a rare genetic disease, with an autosomal dominant transmission. It is due to connective tissue injury, in relation to an anomaly of fibrillin, which is a component of elastin (1-5). This disease affects all organs of the body, with very variable degrees of clinical manifestations. The most affected organs are eyes, the cardiovascular system and the skeleton (1-4). We proposed to carry out a review of the literature from the first case reported in the ophthalmology department of the University Hospital of Brazzaville.

CASE REPORT

An eight year-old-girl was seen for bilateral visual blur. At the age of three she was already suffering from myopia, -5 diopters (D). As soon as she was educated, a new pair of glasses was prescribed to her every nine months due to a rapid progression of this myopia, which reached the value of -12 D when she was seven years old. Furthermore, parents noted the exceptional flexibility of the joints of their child, as well as its large size compared to other children of the same age in the family.

On admission, the review noted, on both sides, high myopia -18D, an inferonasal subluxation of the lens (Figure 1). The fundus was normal.
In general terms, we note dadolichostenomelia, thinness (Size = 1.40 m, weight = 25 kg for a Body Mass Index of 12.75), anarachnodactyly (Figure 2), thoracodorsal scoliosis (Figure 3), a large upper limbs. Cardiovascular examination was normal.

Surgery (lensectomy) was carried out on both sides. The post-operative period was simple. A month after this intervention, visual acuity was 10/10 on both sides with a correction of -1D.

The most recent cardiovascular evaluation was normal.

**LITERATURE REVIEW**

Marfan’s syndrome is associated with a mutation in the fibrillin-1 gene (FBN1), sometimes appearing from the birth with obvious signs and rapid progression of the disease. The gene is located on the chromosome 15 (5). It was described nearly 1000 different mutations in this gene causing more or less severe disease (6).

There also seems to increase the activity of TGF-β (Transforming Growth Factor), whose blood test shows high levels in case of Marfan’s syndrome, which presents a potential interest as a test diagnosis (7, 8). A mutation in the gene TGFBR2 coding for the receptor of the TGF-β, gives a related syndrome, sometimes called << Marfan syndrome type 2 >>, the evolution and prognosis is close to the classic syndrome (9, 10). Marfan’s syndrome is a rare genetic disease, its prevalence is 1/5000(11). However, in a third of cases are spontaneous mutations, not inherited (12).
Circulating cardiovascular complications. The ophthalmologist must never forget the need for cardiovascular monitoring for life of his patient, the risk of complications although this is not proven. beta-blockers cushion the impact of systolic flow on the weakened media of the aorta. They reduce the progression of aortic dilatation and may reduce the risk of complications. Original rhythm of ventricular disorders can cause sudden death. Beta-blockers at a high level are not recommended.

**CONCLUSION**

Once the diagnosis of Marfan's syndrome posed, the ophthalmologist must never forget the need for cardiovascular monitoring for life of his patient, because the mortality of this disease is related to its cardiovascular complications.


