SUNTEXT REVIEWS

SunText Review of Case Reports & Images
ISSN: 2766-4589

Open Access
Case Report
Volume 3:3

Acute Aortic Dissection on Ascending Aortic Aneurysm in a Patient with Marfan Disease: A Case Repot

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Received date: 19 September 2022; **Accepted date:** 28 September 2022; **Published date:** 05 October 2022

Citation: Redha L (2022). Acute Aortic Dissection on Ascending Aortic Aneurysm in a Patient with Marfan Disease: A Case Repot. SunText Rev Case Rep Image 3(3): 154.

DOI: https://doi.org/10.51737/2766-4589.2022.054

Abstract

Marfan syndrome is a rare genetic disorder. It is characterized by the involvement of one or more organs and can in particular cause skeletal disorders (tall stature, scoliosis), ophthalmological (ectopia of the lens), cardiac (dilation of the aorta).

Keywords: Kyphoscoliosis; Arachnodactyly; Adolichesteomelia

Introduction

Marfan syndrome is a rare genetic disorder. It is characterized by the involvement of one or more organs and can in particular cause skeletal disorders (tall stature, scoliosis), ophthalmological (ectopia of the lens), cardiac (dilation of the aorta). The prevalence of Marfan syndrome is estimated at one person in 5000 [1,2]. The aim of this work is to report a case of type an aortic dissection in a patient with Marfan's disease with significant thoracic and skeletal deformity.

Observation

We report the observation of a young woman with a family history of Marfan (mother had a minor form and the two brothers had a major form) presented to cardiological emergencies for tearing retrosternal chest pain evolving for a week. Clinically, the patient was very thin with marfanoid appearance presenting kyphoscoliosis, arachnodactyly, adolichesteomelia, positive thumb and wrist sign, high myopia with bilateral dislocation of the lens. Her brachial blood pressure is 150/90 mmHg on the right and 145/90 mmHg on the left. Cardiopulmonary auscultation is normal. The blood work shows a normal haematological examination and ionogram. The chest X-ray showed dorsal scoliosis with significant chest deformity. ECG was in regular sinus rhythm with fine QRS. Echocardiography showed aortic dissection on onion bulb dilation of the aortic sinuses with grade II aortic insufficiency and correct LV function with EF at 70%, LV not dilated at 45/27 mm and PAPS at 35 mm Hg. Thoracic CT angiography showed type A aortic dissection on aneurysmal dilation of the sinuses of Valsalva 74.5/67 mm with extension of the aortic dissection over the entire height of the thoraco-abdominal aorta and on the proximal portions of the iliac arteries. The decision of the medico-surgical staff was to reject this patient given the high operative risk linked to the importance of the thoracic malformations and the deterioration of her general condition (Figure 1).

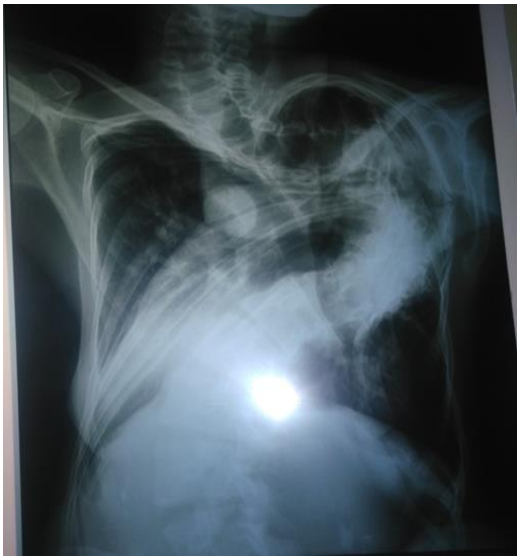

Figure 1: *Chest x-ray showed huge sternal, spinal and costal deformity.*

Discussion

Belonging to the group of rare diseases, Marfan syndrome currently affects a population of approximately 1/5000 individuals, i.e. 12,000 patients in France. It is a disease of the supporting tissue that affects different systems and whose vital prognosis is determined by aortic damage. The functional prognosis depends on ophthalmological and rheumatologically involvement. Marfan's syndrome is an autosomal dominant disease, as is the case of our patient, which generally results from a mutation of the Fibrillin type 1 gene. It can more rarely be a mutation of the gene coding for a TGF-beta receptor. Borderline forms are frequent and sometimes pose difficult nosological problems. The clinical signs appear during life at a variable age. The suspicion of a Marfan syndrome in a patient can be the domain of the general practitioner, in front of the association of skeletal signs, a family history and one of the warning signs (Ghent criteria) as it is the case of our patient. It is also the domain of the cardiologist and cardiac surgeon in the face of aortic dilation or aortic dissection and/or mitral valve prolapse in childhood. The diagnosis is based on a set of clinical, Para clinical and sometimes evolutionary arguments, which lead to the diagnosis, according to the Ghent criteria [3-5]. Echocardiography should be performed to look for aortic dilatation (taking into account Roman's standard curves), bicuspid aortic valve and mitral valve prolapse. The genetic study in molecular biology is proposed: To confirm the diagnosis in the event of an incomplete phenotype (Ghent diagnostic criteria), to propose a basic treatment in order to prevent complications and treat existing symptoms. The initial prescription for treatment with Beta-blocker limiting aortic dilation must be initiated by a cardiologist [6-9]. The treatment is multidisciplinary. Aortic monitoring, as a preventive measure in the event of significant dilation of the aorta. The diameter from which the surgery is performed depends on its absolute value (50 mm as a rule), but also on its evolution, the history family [10]. The goal of the surgery is to replace the initial part of the aorta, which is particularly fragile. This can be done by keeping the patient's aortic valve (ascending aortic plasty) or by combining it with aortic valve replacement, usually by a mechanical valve (modified Bentall procedure). The preservation of the valve is technically more delicate and must therefore be carried out by a surgeon who has experience. The risk is the appearance of an aortic leak, which may require further intervention. The presence of a mechanical valve requires lifelong anticoagulant therapy. In case of dissection of the ascending aorta, emergency surgery is necessary in all cases. Surgery is possibly considered in a second step in case of dissection of the descending aorta with dilation of the latter. Cardio-vascular mitral surgery by valvular plasty in the event of significant mitral leakage. It was impossible to operate on this patient with altered general condition and enormous thoracic deformity type dorsal scoliosis and deep sternal depression although she suffered from a type an aortic dissection on aneurysmal dilation of the sinuses of Valsalva at 74.5/67 mm. After cardiovascular surgery, treatment with beta-blocker should be maintained and prevention of endocarditis should be systematic. Follow-up makes it possible to assess the evolution, in particular cardiological and ophthalmological, and to propose surgery at the best time. Cardiac and aortic ultrasound at least annually, more often if rapid change in aortic diameter or if an indication for surgery is discussed (Figure 2).

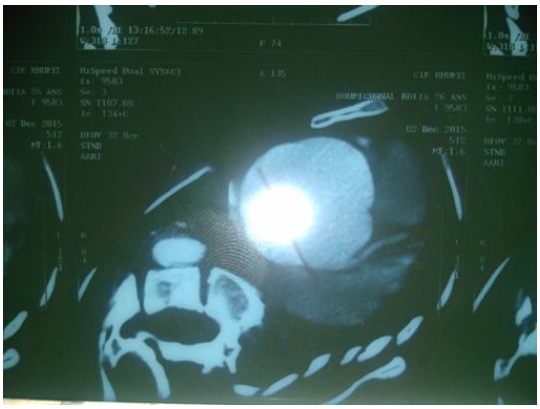

Figure 2: *Thoracic CT angiography showed aortic dissection on onion bulb dilation of the aortic sinuses with thoracic deformity.*

Scanner synchronized with ECG or MRI of the aorta depending on availability:

-If aortic dissection is suspected.

-To confirm the measurement of a dubious aortic diameter on the ultrasound. In all cases, on an annual basis in the event of descending aortic dissection, at least every 5 years in the absence of aortic dissection.

Conclusion

The announcement of the diagnosis is an integral part of the overall care process. The diagnosis is sometimes difficult, especially in children. Molecular research is not the solution. Management is multidisciplinary and continuous. It requires cardiac monitoring: vital, paediatric monitoring: functional and psychological monitoring: like any genetic disease. The key elements of surveillance are the root of the aorta and the mitral valve, the eye and the search for musculoskeletal complications. Aortic complications are prevented by limiting physical and sporting activity (isometric sports, endurance sports, contact sports, combat sports, competition).

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