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# Non- Dissecting Large Aortic Arch Aneurysm Leading to Chronic Aortic Insufficiency Presenting as Acute Heart Failure Revealing Marfan Syndrome

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**ABSTRACT**

**Introduction:** Marfan syndrome is an autosomal dominant disorder of connective tissue which has many clinical symptoms and whose prognosis depends on associated cardiovascular complications, dominated by progressive aortic root dilation, which may lead to aortic dissection, rupture or aortic regurgitation. Prevention of these life threatening complications is of major importance.

**Case presentation**: We report here a case of a 42-year-old man, with a history of two retinal detachments in 2015 and 2018, who presented with severe aortic valve regurgitation and severe heart failure secondary to significant aortic root dilatation revealing Marfan syndrome. Unfortunately, before referring the patient to the surgeon, the patient ended up dying from an unrecovered cardiac arrest.

**Conclusion:** It is very important to recognize Marfan syndrome on time and the preventive actions should be undertaken in order to avoid life-threatening consequences of this disorder. Echocardiography and especially the angioscanner represent the main tools used for the diagnosis of gravity and the follow-up of this condition.

## INTRODUCTION

Marfan syndrome is an autosomal dominant pathology of the connective tissue, with primary involvement of musculoskeletal, cardiovascular, and ocular systems. The most serious complication in patients with Marfan syndrome presents progressive aortic root dilation that may lead to aortic dissection, rupture or aortic regurgitation, which used to be the main cause of death in this patient category prior to the era of successful preventive therapies (1).

Thus, we propose a recent and complex case of Marfan syndrome and a review of the literature of this rare pathology highlighting the cardiovascular, orthopedic, and ophthalmological manifestations.

## CASE REPORT

The following report presents the case of a 42-year-old man, presented to the emergency for a five- day history of new-onset dyspnea at rest, orthopnea, and productive cough with green phlegm. For the previous six months he had presented exercise dyspnea and had marked limitation in activity due to symptoms (NYHA classII-->III).

His past medical history was a pulmonary tuberculosis treated in 2000 and two retinal detachments in 2015 and 2018 that were never investigated due to the patient being lost to follow- up. both parents were alive and well with no known cardiac or other medical, also their brothers, sisters, aunts, uncles, cousins, or grandparents.

He was 184 cm tall and weighed 68 kg. Had elongated arms,

arachnodactyly, ligamentary hyperlaxit and serious chest wall deformities, which included thoracic scoliosis of the vertebral column and moderate pectus excavatum and Flatfeet. On clinical examination, the vital signs were: apyrexia at 37˚C, respiratory rate of 28 cycles/min, BP: 106/44 mmHg, heart rate, 110bpm, SPO2: 94% on room air. The rest of the examination showed a regular heart sounds with a soft high-pitched early diastolic murmur best heard at the right upper sternal border with an irradiation all along the left edge of the sternum, of intensity 4/6th, B2 is decreased and mitral systolic murmur of intensity 3/6$^{th}$. The pleuropulmonary examination revealed crackles at the pulmonary bases. An electrocardiogram (ECG) showed normal sinus rhythm, normal PR, evidence of left atrial enlargement, and left ventricular hypertrophy. The echocardiogram showed severe aortic valve regurgitation secondary to significant aortic root dilatation (89 mm diameter at the Valsalva sinus), moderate mitral regurgutation and left ventricular dilatation, with anormal systolic function (Figure 2). Contrast CT of the chest demonstrated severe enlargement of aortic root measuring 76 mm, cardiomegaly and osseous deformities previously mentioned (Figure 3).

In accordance with Ghent criteria, patient was diagnosed as having Marfan syndrome, complicated by severe aortic root aneurysm and aortic insufficiency with severe heart failure. Unfortunately, before referring the patient to the surgeon, the patient ended up dying from an unrecovered cardiac arrest.

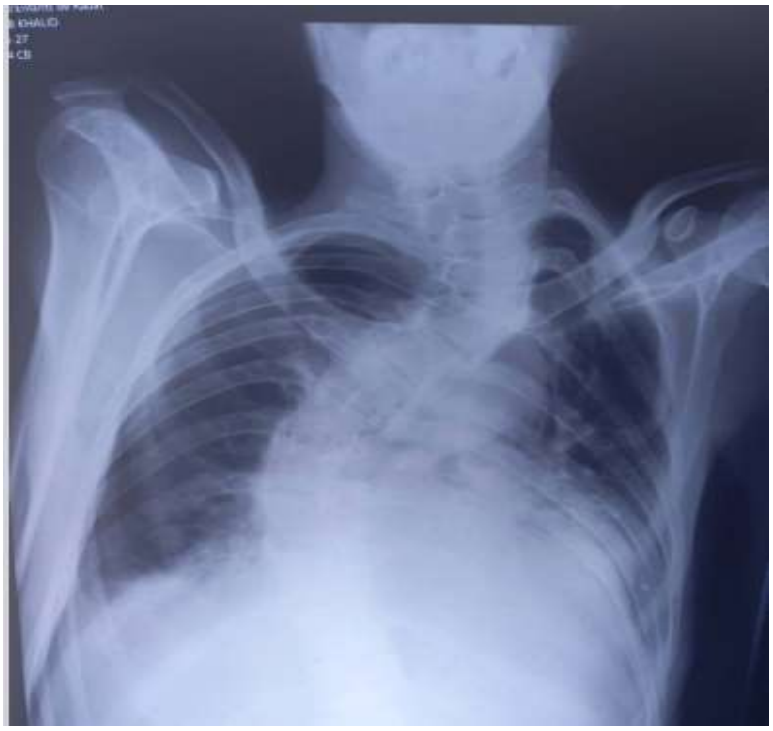

**Figure 1 :** Frontal chest radiograph: disruption of the anatomical configuration of the heart; cardiomegaly, kyphoscoliosis.

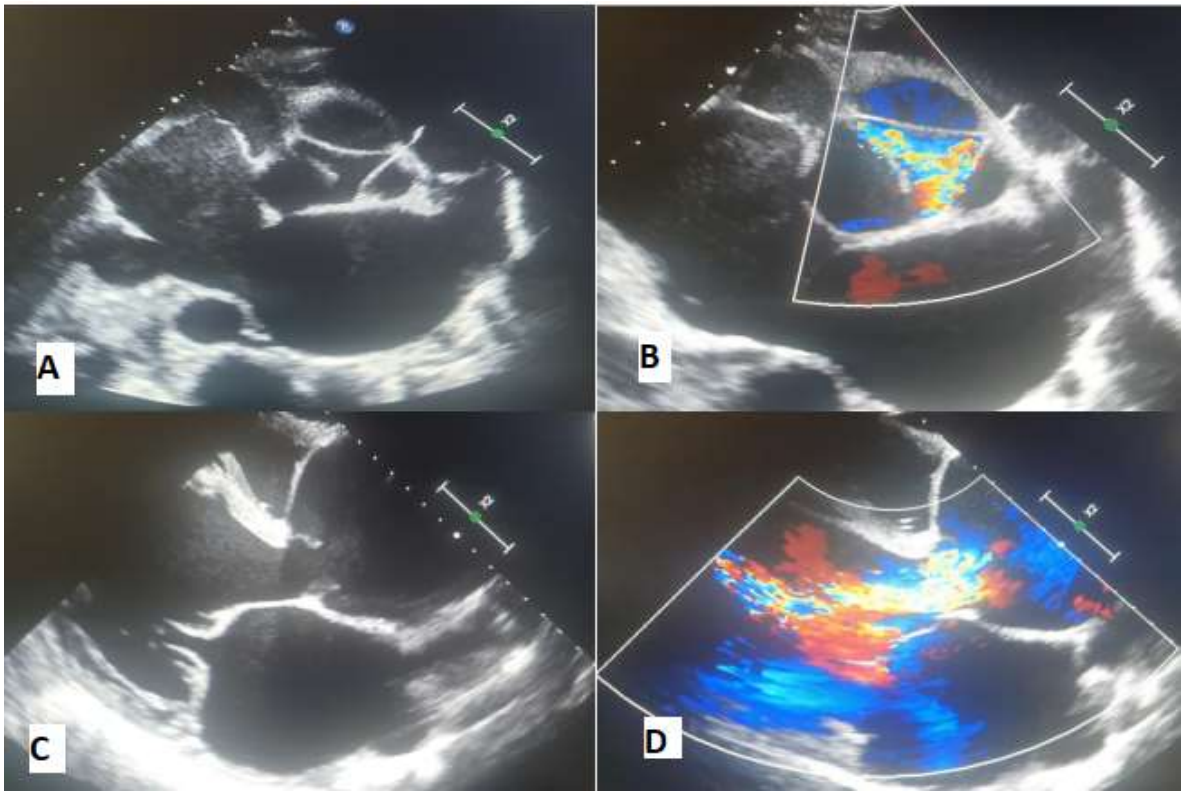


**Figure 2 :** Transthoracic echocardiogram showing 89 cm ascending aortic aneurysm without dissection at the level of sinus of valsalva (C) and severe aortic regurgitation (A,B,D)

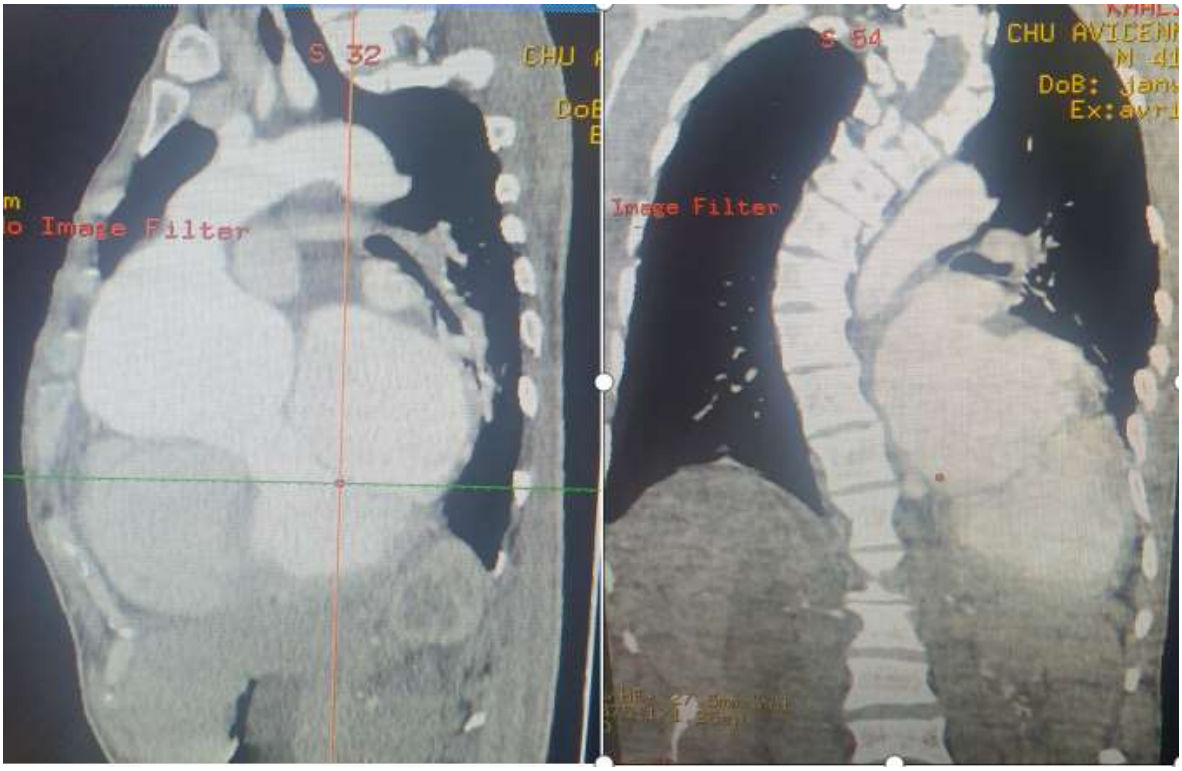


**Figure 3 :** Aortic root dilatation seen in cardiac computed tomography (A), scoliosis (B)

**DISCUSSION**

Marfan syndrome is the most common in herited multi-systemic disorder of connective tissue, with a reported incidence of 2-3 per 10000 population, without gender, racial,

or ethnic predilection. The gene responsible is carried on a non-sex chromosome (Chromosome 15). Which explains why this condition affects both sexes without distinction. The responsible gene carries a deleterious mutation, which is why a parent of a kid with Marfan syndrome has an estimated 50% risk of being affected by the syndrome. Cases have been described in which the parents are free of the syndrome and a mutation has occurred spontaneously in the egg or sperm after its formation (2,3). This phenomenon, which occurs in about one in 10,000 births, still represents one third to one quarter of Marfan individuals (3).

The Marfan syndrome is characterized by an alteration in the production of fibrillin due to a mutation in the FBN1 gene. Fibrillin 1 allows the organization of elastin fibers constituting the extracellular matrix of connective tissue, whose role is to ensure the support of organs. Therefore a mutated fibrillin 1 weakens the connective tissue of the body, whether tendinous, ligamentous, cartilaginous, vascular or heart valves (4,5). Other mutations have been identified, notably the mutation of the TGFBR2 and TGFBR1 gene in patients with Marfan syndrome (6,7).

The circumstances of discovery are diverse, because this syndrome is rich in clinical signs and the clinical manifestations are not of the same intensity in all carriers, the characteristics of Marfan depend on the patient's age when the disease is first diagnosed, some neonatal forms remain very disabling while other forms appear at an advanced age of the patient's life (8,9). Our patient was 42 years old, male, with no family history of cardiovascular disease.

The cardiovascular features were first outlined by McKusick in 1955 (10). This Cardiovascular signs are the main threat to the vital prognosis of patients, as the weakening of the aortic wall with dilatation of the root of the aorta is often complicated by aortic dissection, which darkens the vital prognosis of patients. Dilatation of the ascending aortic is part of the criteria major cardiovascular of Marfan syndrome (11). The Aortic regurgitation is a common valvulopathy associated with thoracic aortic aneurysms that is generally described as an early diastolic decrescendo murmur (12). However, there are peripheral signs including Musset's sign (systolic head bobbing), Mueller's sign (systolic pulsation of uvula), Quincke's sign (capillary pulsation), and Corrigan's sign (forceful bounding pulse with rapid upstroke followed by a collapse), the latter two were observed in our case (13), Although not part of a routine physical examination, clinicians should look for these signs if they have high suspicion for thoracic aortic aneurysm in the setting of non- specific symptoms with new-onset diastolic murmurs. Cardiomyopathy is another manifestation that can be seen with aortic aneurysms when aortic regurgitation is involved. The patient was found to have severe reduction in LVEF, In this case, the patients develop non-specific symptoms including chest pain, shortness of breath, lightheadedness, dizziness, palpitations, and lower extremity swelling. Although ruling out underlying coronary artery disease in patients presenting with acute onset cardiomyopathy is a diagnostic priority, valvular heart disease including aortic regurgitation should be evaluated. Accordingly, echocardiography examination with special emphasis on evaluation of ascending aorta and heart valves is mandatory in patients with Marfan syndrome.

The study of coronary artery anatomy is traditionally performed through invasive angiography. However, this canal so be safely done with cardiac CT, especially in patients with marfan syndrom (14, 15) taking advantage of the high negative predictive value of this exam (15); this is one indication for which cardiac CT can replace invasive angiography (16).

Management and treatment of the aorta in Marfan syndrome patients should be based on regular imaging of the aorta to evaluate its size and to determine the eventual dilation and its progression, administration of Beta adrenergic receptor antagonist therapy and prophylactic aortic repair when aorta reaches a size that may threaten dissection or aortic regurgitation. In the pre open-heart surgery era, the average life expectancy of patients with Marfan syndrome was 45 years (17), whereas in our time, owing to preventive measures, it has extended up to 70 years (18). We can state without a doubt that our patient was diagnosed with advanced Marfan syndrome with serious cardiovascular and osteoarticular complications, which darkens his vital prognosis.

## CONCLUSION

Marfan syndrome is a genetic disease of connective tissue that can manifest itselfby cardiovascular, pulmonary, orthopaedic, ophthalmological and cutaneous signs. It is very important to recognize Marfan syndrome on time and the preventive actions should be undertaken in order to avoid life-threatening consequences of this disorder. Echocardiography and especially the angioscanner represent the main tools used for the diagnosis of gravity and the follow-up of this condition.

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