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Case Report

Marfan's Syndrome with Acute Inferior Wall ST-segment Elevation Myocardial Infarction in A Combined Family of 13 Cases

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Abstract

Marfan's syndrome (MFS) is a rare autosomal dominant connective tissue disorder, often associated with genetic mutations and most patients have a family history of the disease. Its clinical manifestations are mainly skeletal, ocular and cardiovascular pathologies. We report a case of a 34-year-old male diagnosed with Marfan's syndrome at the age of 22 and presenting with ocular symptoms. Following an episode of sudden chest pain, he was admitted to the hospital with an acute inferior wall ST-segment elevation myocardial infarction, and was diagnosed with a mid-right coronary artery occlusion by coronary angiography. He underwent coronary artery balloon dilatation to restore grade 3 flow, and a stent was implantated 4 months later. In a subsequent epidemiologic survey of 26 family members from 4 generations, 13 members were diagnosed with Marfan's syndrome, all of them presented with ocular signs. After more than 3 years of follow-up of the entire family, there is currently no one with coarctation of the aorta and no sudden death. Since some patients with Marfan's syndrome can present with acute chest pain complicated by aortic coarctation, if the coarctation involves the aortic root it can lead to severe stenosis or occlusion of the coronary arteries, resulting in electrocardiograms showing typical changes for a ST-segment elevation myocardial infarction. Considering the complexity and life-threatening nature of Marfan's syndrome, it is important consider the various etiologies of chest pain in patients with Marfan's syndrome.

Keywords

Marfan's syndrome; myocardial infarction; balloon dilatation; eye signs; lens replacement

Introduction

Marfan's syndrome (MFS), is an autosomal dominant connective tissue disorder that has been linked to mutations in the gene encoding protofibrillar protein 1 (FBN1) [1], which is located on the long arm of the human chromosome 15 [2]. The incidence is 1/5000 to 1/10,000 [3]. The diagnosis is based on the 2010 revised version of the Ghent criteria [4], which places greater emphasis on the importance of aortic root dilatation and lens ectasia. A systematic rating scale is used to assess characteristic symptoms in patients with MFS, with a total score of 20 points, and a score of 7 or more having diagnostic value. The risk of the disease in children is 50% if one parent has the disease, and most patients have a family history of the disease [5]. It is a multisystem disease, with signs and symptoms in the eyes, bones, and cardiovascular system as the primary clinical manifestations [2,6–9]. The main manifestations are aortic root dilatation, aortic root aneurysm, acute aortic coarctation, long bone overgrowth, lens dislocation and high myopia. Clinically, there are two types: those with all three signs are called complete, those with two are called incomplete. In MFS, 80% of patients present with cardiovascular complications, followed by skeletal and ocular abnormalities [10]. The leading cause of death is rupture of aortic coarctation aneurysms [11]. The CARE checklist was used when writing this casereport (**Supplementary Fig. 1**).

Case Report

We report the case of a 34-year-old male with a 12 year history of MFS, with skeletal signs and ocular signs of the incomplete type. In 2010, he underwent lens replacement in the left eye and in May 2020, he underwent lens replacement in the right eye. The patient was admitted to the hospital on 30 November 2020 for "persistent anterior precordial pain for 1 hour", after drinking alcohol, which persisted without relief, accompanied by sweating, nausea, and vomiting. He had no radiating pain in the back of the shoulders, palpitations, and dizziness. An electrocardiogram (ECG) showed sinus bradycardia, second-degree type I atrioventricular block, and ST-segment elevation in leads II, III, and aVF (Fig. 1). He had no history of hypertension and diabetes, but a history of smoking 20 cigarettes per day for 10 years. Admission examination showed a blood pressure of 130/80 mmHg, height: 176 cm, weight: 63 kg, clear breath sounds and no rales. The heart rate was 74 beats/minute and regular without murmurs. The admission diagnosis was coronary atherosclerotic heart disease, acute inferior ST-

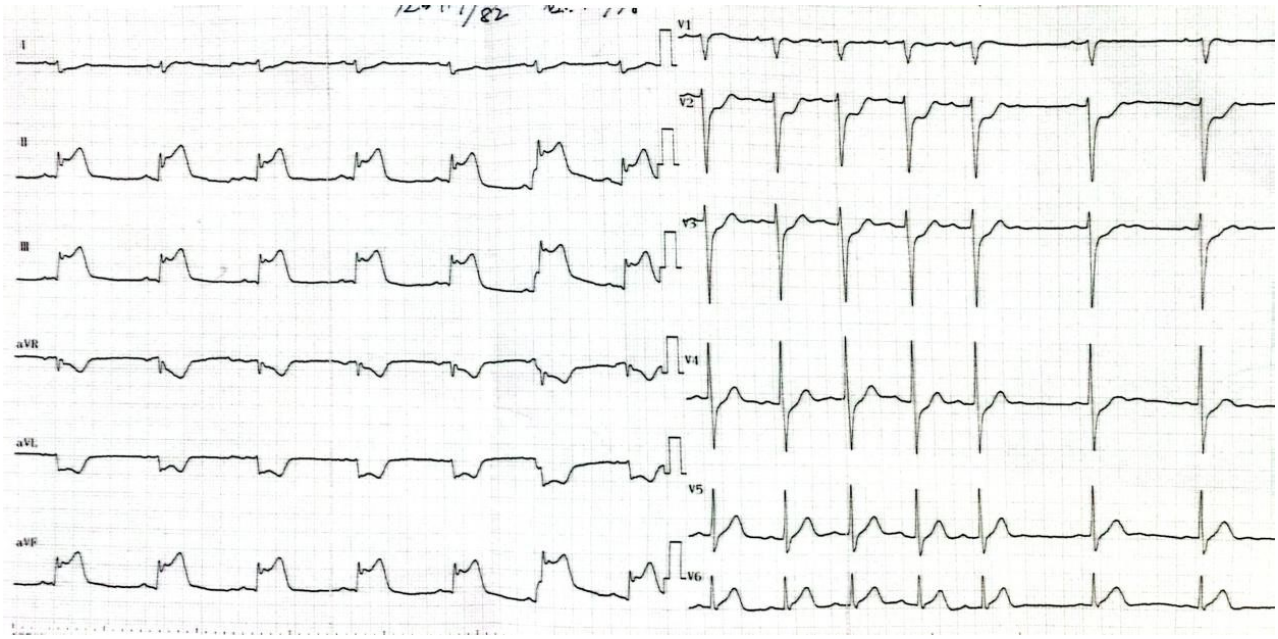

Fig. 1. Electrocardiogram on admission to the emergency department.

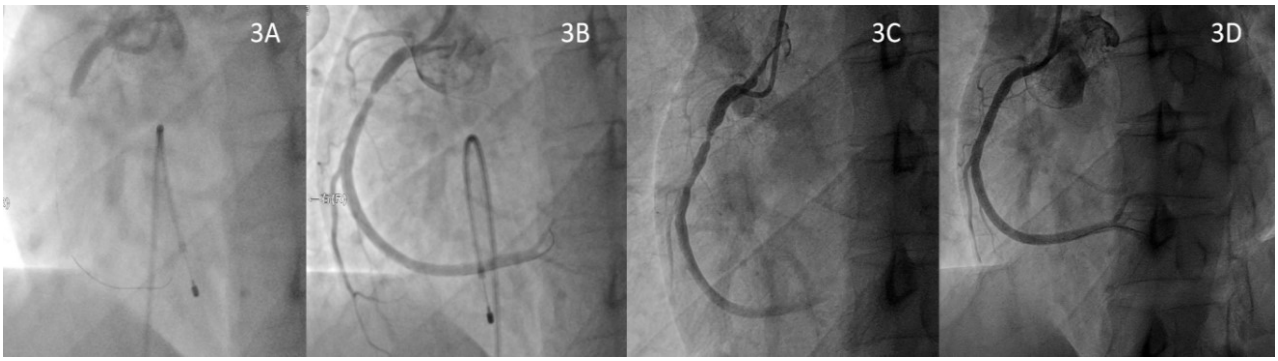

Fig. 2. Coronary Angiography Intervention. (A) Preoperative right coronary artery; (B) Postoperative right coronary artery; (C) 4 months after first right coronary artery procedure; (D) Poststenting right coronary artery.

segment elevation myocardial infarction, cardiac function class I (killip classification). Because of the previous history of MFS, a Computer Tomography scan was performed which no showed no evidence of a coarctation or aortic dissection and no pulmonary embolus. Emergency coronary angiography showed occlusion of the mid right coronary artery. After percutaneous transluminal coronary angioplasty (PTCA), a thrombus was detected in the mid-portion of the coronary artery, and a dark-red thrombus of about 3 mm thick and 5 mm long was extracted, resulting in Thrombolysis In Myocardial Infarction grade 3 blood flow, with a residual 80% stenosis. (Fig. 2A–C) Stent implantation was not performed. An echocardiogram showed normal size of all cardiac chambers, reduced motion of the basal segment of the left ventricular inferior posterior wall, no valve abnormalities, no dilatation of the ascending aorta and the aortic sinus, and a Left Ventricular Ejection Fractions of 57%.

Subsequently, the family line was investigated for the prevalence of MFS, the following items were investigated: current age, age at diagnosis, height/weight, extended finger distance between arms/height, presence of lens ectasia, history of lens surgery, and cardiac ultrasound for the presence of aortic root dilatation or aortic sinusoidal aneurysm. The epidemiologic investigation of the family line is shown in (Table 1), the family genealogical chart is shown in (Fig. 3): 13 of the 26 members of the family had MFS in 4 generations. The eldest grandson of his great-uncle (IV1), who was diagnosed with bilateral subluxation of the lens due to MFS at the age of 9 years old in 2008, was treated surgically. Ocular disorders that had plagued several members of the family for many years were confirmed to be the result of lens ectasia due to MFS, a total of 13 individuals were diagnosed, 10 of whom underwent successive lens replacements.

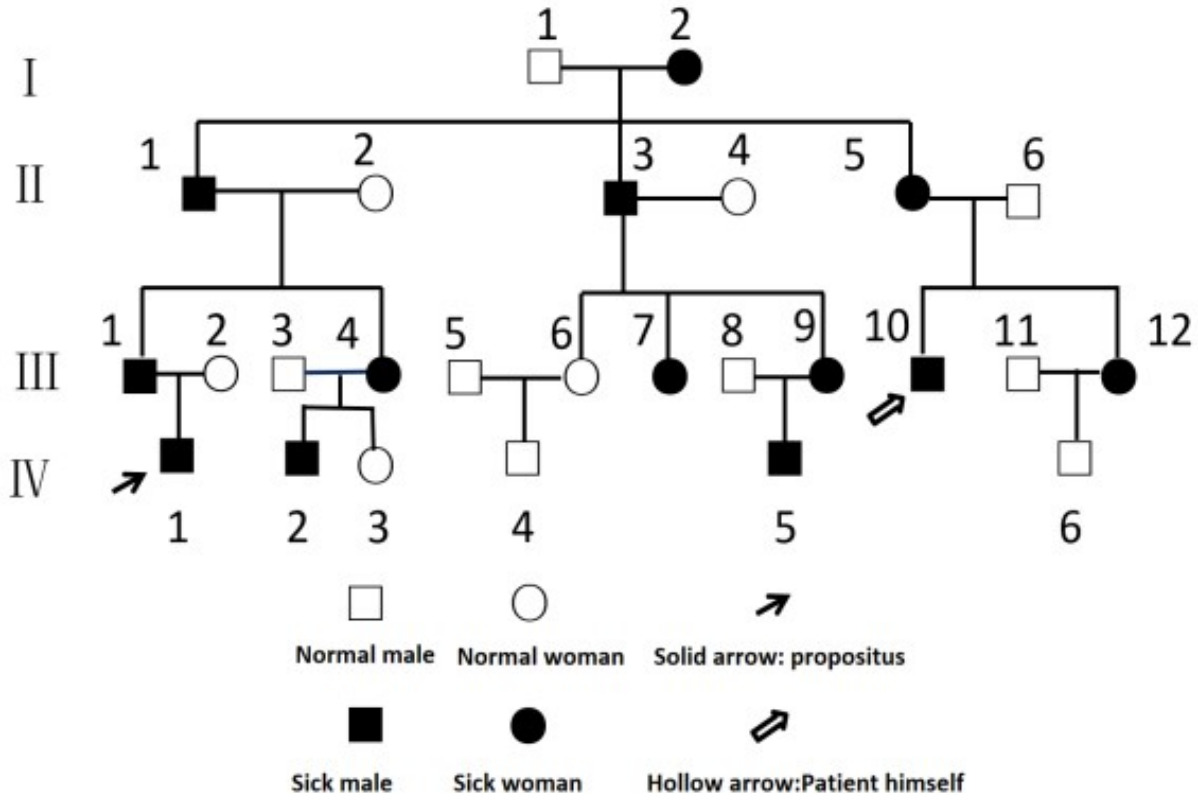

Fig. 3. Family Marfan's syndrome Survey.

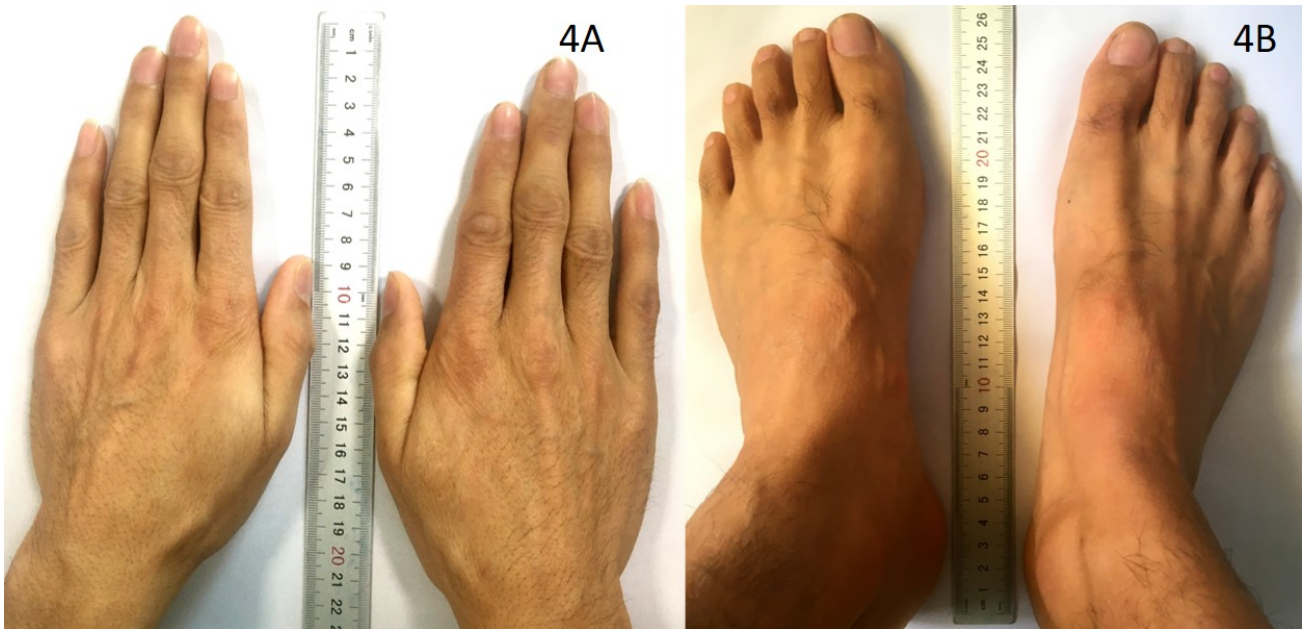

Fig. 4. Patient's limbs. (A) Hands; (B) Feet.

Discussion

MFS is a multi-system disease, with symptoms or signs in the eyes, bones and cardiovascular system as the first manifestation, among which the cardiovascular system is the most seriously affected, manifesting as elastic lamina fibers dysplasia in the large arteries, abnormal dilatation of the aorta, aortic aneurysmal changes and aortic valve insufficiency, which can lead to aortic coarctation. Lens ectasia

Table 1. Epidemiological survey of the family lineage.

Member No.	Sex	Age (years)	Age of diagnosis (years)	Height/Weight (cm/kg)	Arm extension/height	Lens ectopia	History of lens surgery	Heart echocardiography
I2	women	81	69	166/56	1.00	+	–	–
II1	man	62	50	175/60	0.96	+	–	–
II3	man	60	48	176/64	0.98	+	–	–
II5	women	56	44	167/67	1.01	+	+	–
III1	man	39	27	176/65	0.98	+	+	–
III4	women	37	25	166/60	1.02	+	+	–
III7	women	37	25	165/58	0.99	+	+	–
III9	women	36	24	171/60	0.97	+	+	–
III10	man	34	22	176/63	1.05	+	+	–
III12	women	31	19	165/53	1.02	+	+	–
IV1	man	21	9	177/68	1.02	+	+	–
IV2	man	20	8	174/70	0.99	+	+	–
IV5	man	18	6	178/72	1.03	+	+	–

is present in approximately 50–80% of patients with MFS, and lens ectasia is also the only diagnostic criterion for its ocular involvement [12]. Lens ectasia often leads to binocular refractive error, amblyopia, high myopia, corneal astigmatism, iris hypoplasia or ciliary muscle hypoplasia leading to pupil reduction, cataracts, glaucoma, retinal detachment and even blindness [13,14].

We report the case of MFS in which the patient had ocular signs as the first manifestation. He now presented with sudden chest pain, which was confirmed by enhanced CT and cardiac ultrasound to be free of cardiovascular system lesions such as aortic dilatation and valvular insufficiency, aneurysmal dilatation of the aortic root, and aneurysm of the aortic coarctation. He was found to have an acute occlusion of the right coronary artery resulting in an acute inferior wall ST-segment elevation myocardial infarction. Wu Y and Wu YZ [15] have reported the death of a 25-year-old female patient with Marfan's syndrome after an acute inferior and posterior wall myocardial infarction due to coronary artery involvement by an aneurysm of the aortic root. Xu *et al*. [16] reported a 30-year-old male MFS patient with an acute anterior and high lateral wall ST-segment elevation myocardial infarction due to an ascending aortic coarctation involving the coronary arteries and with malignant arrhythmias such as ventricular tachycardia and ventricular fibrillation, requiring surgical treatment. Zhang *et al*. [17] reported a 28-year-old male diagnosed with an acute inferior wall ST-segment elevation myocardial infarction (STEMI) due to sudden onset of acute chest pain, which was ultimately determined to be caused by a type A aortic dissection due to Marfan's syndrome involving the right coronary artery. He underwent a Bentall procedure, and is stable 2-years following surgery.

Surgery should be the first choice in the treatment of acute inferior wall myocardial infarction due to MFS combined with an ascending aortic coarctation, aortic root tear, and involvement of the right coronary artery. In our patient, we restored TIMI grade 3 flow after PTCA at the site of the mid right coronary artery occlusion. Current guidelines do not clarify whether patients with MFS combined with an acute myocardial infarction are candidates for emergency stenting, and no cases have been reported. Therefore, we did not perform immediate stent implantation but suggested that coronary angiography be repeated after several months, and then stent implantation be performed if the residual stenosis was still present, or if angina. A coronary angiogram after 4 months showed a residual stenosis of 80%, so a stent was implantated (Fig. 2D), The patient has been followed for more than 3 years without any cardiovascular events or recurrent as angina.

Epidemiologic investigation showed that 13 members of the patient's family in four generations had MFS, none of them had cardiovascular system involvement, and none of had aortic coarctation or sudden death after more than 3 years of follow-up. All family the members did undergo relevant genetic monitoring for MFS. Skeletal system abnormalities in this patient were characterized by tall and thin stature, slender limbs, little subcutaneous fat, long head (32 cm), zygomatic bone convexity (cheek bone projection), high palate, flat chest, long and slender metacarpals, phalanges and phalanges, and long and slender fingers and toes in the form of spider fingers (toes) (Fig. 4). In both hands: the thumb was 6 cm, the index finger was 8.5 cm, the middle finger was 10 cm, the ring finger was 9 cm, and the pinky finger was 7 cm. Arms' span (185 cm) was larger than the height (176 cm). The clinical characteristics of MFS in the eye are often characterized by lens subluxation, in addition to refractive error, amblyopia, myopia, and glaucoma, and are treated by cataract or dislocated lens removal combined with Intraocular lens implantation. In this family line, there were 26 individuals in 4 generations, 13 persons were affected (7 males and 6 females), with a prevalence of 50%, and there were persons in each generation with a high rate of exophthalmos, all of whom presented with oc-

ular signs of subluxation of the lens and detachment of the retina. Three patients had both lens subluxation and retinal detachment, the patient himself (III10), the patient's sister (III12), and the patient's second uncle (II3), while the remaining 10 showed bilateral lens subluxation, all of the remaining 12 patients in the family, except for I2, II5, and III1, underwent ophthalmic surgical treatment.

At present, there is no specific treatment for MFS. The condition of MFS can change dynamically over time, so it is necessary to carry out regular physical examinations [18], echocardiography, and Computer Tomography\Magnetic Resonance Imaging to monitor cardiovascular involvement, such as the diameter of the aortic root and ascending aorta, the opening and closing of the valves, chest pain, shortness of breath, palpitations, and other symptoms, and seek medical attention once these symptoms occur. A ruptured aneurysm, aortic coarctation, and severe cardiac valvular insufficiency should be treated with surgery to reduce the risk of sudden death. This case suggests that patients with MFS presenting with chest pain should not only be considered for a ruptured aortic coarctation aneurysm, but for an acute myocardial infarction caused by coronary artery atherosclerosis.

Availability of Data and Materials

The authors confirm that the data supporting the findings of this study are available within the article.

Author Contributions

ZZS contributed to the design of this work. QW contributed to the interpretation of data. ZHC analyzed the data. ZJM and YDY contributed to acquisition, analysis, interpretation of the data and image. All authors contributed to editorial changes in the manuscript. All authors read and approved the final manuscript. All authors agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

Ethics Approval and Consent to Participate

This study was conducted in accordance with the Declaration of Helsinki. Written informed consent and ethical approval was obtained from the patient for the publication of any recognizable images or data contained in this article. This study has been approved by the Third People's Hospital of Datong, ethics approval number (2024) Scientific Research Paper Luncheon No. (07).

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Conflict of Interest

The authors declare that the research was conducted in the absence of any commercial or financial relationships that could be construed as a potential conflict of interest.

Supplementary Material

Supplementary material associated with this article can be found, in the online version, at https://doi.org/10.59958/hsf.7833.

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