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Marfan Syndrome with Atypical Pneumonia: A Case Report

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ABSTRACT

Marfan syndrome (MFS) is an autosomal dominant systemic disorder of the connective tissue. Children impacted by the MFS carry the mutation in one of their two alleles of the gene that codes the connective tissue protein fibrillin-1 (FBN-1). MFS influences their effects upon most organs and tissues, in particular the skeleton, lungs, eyes, heart, and the large blood vessel that distributes blood from the heart to the reminder of the physique. A case report of MFS with atypical pneumonia has been reported with positive Steinberg sign, thumb sign and wrist sign. Existence of long legs with the positive Walker Murdoch sign. There were no complications such as an aortic aneurysm, ocular lens luxation, or mitral valve prolapse were remarked. Since the patient was lacking such complications based on MFS, he was treated only for atypical pneumonia.

Keywords: Marfan Syndrome, Fibrillin-1, Thoracic Aortic Aneurysm or Dissection, Ectopia Lentis

INTRODUCTION

An autosomal dominant hereditary connective tissue disorder (CTD) is Marfan syndrome. primarily brought on by mutations in the FBN1 gene, which codes for fibrillin-1 (FBN-1), a structural element of the extracellular matrix (ECM) and a regulator of the bioavailability of transforming growth factor β (TGF-β)[1].

MFS is a rare pleiotropic disease (one in 5000) that is defined by two genetic criteria (existence of a pathogenic mutation in the FBN1 gene in the presence of TAAD or EL) and three clinical criteria (thoracic aortic aneurysm and/or dissection [TAAD], Ectopia Lentis [EL], and systemic features [SFs, multi systemic manifestations] with score ≥7). The diagnosis can only be made based on the clinical features because there is currently no confirmatory biochemical test available[1].

A number of bodily systems can exhibit signs of MFS, but the skeletal system, the cardiovascular system, the ocular system, the pulmonary system, and the nervous system—the fibrous membrane that surrounds the brain and spinal cord—are the most common[2].

Numerous anatomical characteristics associated with MFS, including tall stature, scoliosis, chest wall deformities, and mitral valve prolapse, are also rather common in the general population.

Atypical pneumonia or primary atypical pneumonia is a mild acute respiratory disease characterized by gradual onset occurrence of constitutional symptoms as well as symptoms localized to the respiratory tract.[3]

CASE REPORT

A 16year old male patient developmentally normal secondary order born to non-

congenious married couple brought with c/o cough and cold since 4 weeks, insidious in onset, low grade, not associated with expectoration from 1st 3 weeks. Aggravated since last week. Cough has associated with expectoration. Expectorate is white in color, more in the morning, not associated with chest pain. c/o fever since2 days, low grade intermittent type, not relieves on taking medication. H/o admission in the past I/v/o viral fever. H/o septoplasty done at 7 year of age I/v/o DNS.

The medical history revealed that he has MFS with atypical pneumonia. General examination disclosed an elongated face, malar hypoplasia. Head to toe examination shows that he is tall and thin built, the cheek bone (malar or zygomatic) is smaller or absent and Dolichocephaly is also observed. In ENT examination, it shows a narrow and high arched palate and his oral cavity is having crowded teeth.

Chest is bilateral, symmetrical and pectus excavatum is noticed with a curved spine, scoliosis and the abdominal findings are normal.

Presence of long arms and fingers (upper limb) with positive Steinberg sign, thumb sign and wrist sign. Presence of long legs with a positive Walker Murdoch sign.

On examination, vitals such as SpO_2, Pulse rate, Respiratory rate and Blood pressure was normal and the systemic examination was also normal. In the laboratory investigation, Total count-12800$mm^3$and C reactive protein levels-108.0mg/dl are increased and other parameters such as Haemoglobin, Platelet, Red blood cells, Hematocrit, Urea, Creatinine, Na+, K+, Cl-, mean corpuscular volume, mean corpuscular haemoglobin, mean corpuscular haemoglobin concentration are found normal.

Echocardiography reports shows no coarctation of aorta and shows situs solitus, levocardia, normal chamber, normal Pulmonary artery pressure, intact septae and good biventricular function. He was referred to ophthalmology department for further findings, and there were no ocular complaints. Dilated Fundoscopy was done and found both eyes with normal fundus and other ophthalmological interventions are not found.

The patient was provided with treatment including Tab. Aziwan 500 mg, once daily (at night), Tab. PCT 500mg 4 times daily, Tab Pantop 40mg once daily and syrup Coughwar 5ml three times daily and while discharging he was advised with Tab Aziwan 500mg once daily for 16 days, syrup Inkrizz 5 ml twice daily for 2 months and Tab PCT 500mg SOS and advised for further review.

DISCUSSION

A disorder of the connective tissue known as Marfan syndrome (MFS) is inherited autosomally dominantly and is caused by mutations in the fibrillin-1 (FBN1) gene. MFS is a multiorgan-involved, severe, chronic, and potentially fatal illness for which there is no effective treatment. When an index patient meets the major criteria in two systems and meets the Ghent criteria for involvement in a third system, a clinical diagnosis known as MFS is made. The clinical manifestations include prolapse of the mitral valve and aortic aneurysm, lung emphysema, ocular lens luxation, and long bone growth. For Marfan patients, the primary cause of morbidity and death is aortic disease, which progresses to aneurysmal dilatation and dissection[4].

Patients diagnosed with MFS have been observed to have respiratory disorders, including distal airspace enlargement, which has been historically referred to as emphysema and frequently leads to spontaneous lung rupture. The development of the distal alveolar septum is impaired in the early postnatal period, which is indicative of lung abnormalities.

It is difficult to diagnose MFS, although international standards have been put forth. Worldwide adoption of the Ghent criteria from 1996 occurred (table.1); however, in 2010, new diagnostic criteria for MFS were published, which placed greater emphasis on aortic root aneurysm and Ectopia Lentis[5].

Treatment options for various kinds of cardiovascular issues include medications. Among the medications are beta-blockers. β-blockers assist in lowering blood pressure and heartbeat force. They can lower the risk of aortic dissection, which is the tearing of the aorta between its layers, and they may help prevent or delay aortic dilatation, or an enlarged aorta[6].

In many cases, beta-blockers are the first-choice medication for patients with Marfan syndrome. ARBs, or angiotensin receptor blockers, according to recent studies, the ARB medication losartan may stop aortic growth. A clinical trial comparing this drug to beta-blockers in individuals with Marfan syndrome is currently in progress. Inhibitors of the angiotensin-converting enzyme (ACE-I) also lessen aortic stress and aid in blood pressure regulation. If someone does not respond well to β-blockers or angiotensin receptor blockers, they might be prescribed. braces to prevent the deterioration of spinal curvatures. The majority of Marfan syndrome patients with cardiac issues benefit from scheduled surgery. Before there is a life-threatening condition, such as an aortic dissection, surgery is done[7].

People with Marfan syndrome benefit physically and psychologically from exercise. Consume a diet high in fruits, vegetables, whole grains, and other nutrients and well-balanced. Steer clear of weight lifting, competitive sports, and some leisure activities. While some parents may not require counseling, it is important for parents to acknowledge and manage their initial emotions upon discovering that their child has Marfan syndrome[8].

TABLE I. Diagnostic Criteria for MFS in Adults According to the 2010 Ghent Nosology.[9]

In the absence of a family history of MFS:
1. Aortic root Z-score ≥2 AND ectopia lentis
2. Aortic root Z-score ≥2 AND an FBN1 mutation
3. Aortic root Z-score ≥2 AND a systemic score* ≥7 points
4. Ectopia lentis AND an FBN1 mutation with known aortic pathology In the presence of a family history of MFS (as defined above):
1. Ectopia lentis
2. Systemic score* ≥7
3. Aortic root Z-score ≥2
*Points for systemic score
Wrist AND thumb sign = 3 (wrist OR thumb sign =1)
Pectus carinatum deformity= 2 (pectus excavatum or chest asymmetry =1)
Hindfoot deformity = 2 (plain pes planus =1)
Dural ectasia =2
Protrusio acetabula = 2
Reduced upper segment/lower segment ratio AND increased arm/height AND no severe scoliosis = 1
Scoliosis or thoracolumbar kyphosis = 1
Reduced elbow extension = 1
Facial features (3/5) = 1 (dolichocephaply, enophthalmos, downslanting palpebral fissures, malar hypoplasia, retrognathia)
Skin striae = 1
Myopia >3 diopters = 1
Mitral valve prolapse =1

CONCLUSION

In this case the patient exhibits the features such as an elongated face, malar hypoplasia and presence of long arms and fingers (upper limb) with positive Steinberg sign, thumb sign and wrist sign. The patient was solely treated for atypical pneumonia, as there were no complications observed, including aortic aneurysm, ocular lens luxation, and mitral valve prolapse.

Abbreviations

Sl. no	Abbreviation	Expansion
1	MFS	Marfan syndrome
2	CTD	Connective Tissue Disorder
3	FBN-1	Fibrillin-1
4	ECM	Extracellular Matrix
5	TGF-β	Transforming Growth Factor-β
6	TAAD	Thoracic Aortic Aneurysm
7	EL	Ectopia Lentis
8	ENT	Ear, Nose and Throat
9	ARBs	Angiotensin Receptor Blockers
10	ACE-I	Angiotensin Converting Enzyme Inhibitor

Declaration by Authors

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