A CASE REPORT OF RARE PRESENTATION OF MARFAN’S SYNDROME
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ABSTRACT: Marfan syndrome is a heritable condition that affects the connective tissues of Human body. It can affect any body systems including the skeleton, eyes, heart and blood vessels, nervous system, skin, and lungs. Estimates indicate that approximately 1 in 3,000 to 5,000 individuals have Marfan syndrome and most individuals with Marfan’s syndrome have another affected family member which are due to new genetic mutations. We present to you an interesting rare case which reported to our hospital few months back with features of Marfan’s syndrome. Without cardiac or respiratory involvement and associated Mutism and Hypogonadism. In the family no other member was affected.

KEYWORDS: Marfan’s syndrome, Mutism, Hypogonadism, Arachnodactyly, sinus of Valsalva, Prognathism.

INTRODUCTION: Marfan syndrome is named after Antoine Marfan, the French pediatrician who first described the condition in 1896. Marfan syndrome is a multisystem connective tissue disorder usually associated with mutation in fibrillin, and occasionally with mutation in TGFBR1 or 2. The minimal birth incidence is 1 in 9800.¹ About 27% of cases arise from new mutation. Marfan syndrome is a variable autosomal dominant disorder with characteristic cardiovascular, eye and skeletal features. Progressive aortic dilatation, usually maximal at the sinus of Valsalva, associated with aortic valve incompetence leads to aortic dissection or rupture and is the principal cause of mortality, but mitral valve prolapsed with incompetence may be significant, and lens dislocation, myopia, and arthritis associated with chronic joint laxity can cause substantial morbidity.²

The diagnosis is commonly considered in a young person with a tall, thin body habitus, long limbs, arachnodactyly, pectus deformities, and sometimes scoliosis. Some clinical findings such as a high arched palate with dental crowding, skin striae, recurrent hernia or recurrent pneumothorax may increase suspicion. Family history may be helpful, but around 27% of cases arise from new mutation.³

To make the diagnosis of Marfan’s syndrome more consistent and of more prognostic value, the Berlin diagnostic criteria of 1988 were revised and the clinical features codified as the Ghent nosology in 1996.⁴ Ectopialentis (subluxation of lens) is a hallmark feature of Marfan’s syndrome which is present in approximately 60% to 80% of patients⁵ and was also noticed in the present cases. Ectopialentis is usually bilateral, symmetrical, and upward. The diagnosis can be made by looking for iridodonesis (tremor of iris), phacodonesis (abnormal movement of lens), and a deep anterior chamber in the non-dilated eye.⁵
CASE REPORT: A tall 23 year old man presented with sudden onset of blurred vision, mucopurulent discharge and watering from the right eye to the ophthalmology outpatient. The patient being planned for surgery was sent to medical OPD for fitness. On general examination: Height: 163 cms. Arm span: 171 cms. His vital signs showed pulse: 76/ min regular, low volume, B.P: 90/60 mm of Hg in right upper arm and respiratory rate: 18/min abdomino thoracic type with temperature normal. On detailed CNS examination the patient was found to have microcephaly, arachnodactyly, marfansbuild, mutism, prognathism, distal muscle wasting (Hallus valgus left side), lumbar lardosis, protruded abdomen, no secondary sexual characters and left undescended testis.

No deafness and bilateral immature cataract was found. The examination of other systems Cardiac, respiratory and abdomen revealed no abnormality. Interestingly, apart from absence of secondary sexual characters, one of his testis on left side did not descend. On investigations chest x ray did not suggest any pulmonary or cardiac pathology. ECG shows prominent R in V1 which a sign of right axis deviation. Routine blood and biochemical analysis were normal.
Fig. 2: Dry scaly skin with long and tapering first toe

Fig. 3: Long spidery fingers (Arachnodactyly)

Fig. 4: Head and neck examination revealed a convex profile with long and narrow face

Fig. 5: Prognathism

Fig. 6: Undecended testis
The thumb sign (Steinberg's sign) is elicited by asking the patient to flex the thumb as far as possible and then close the fingers over it. A positive thumb sign is where part of the thumb is visible beyond the ulnar border of the hand, caused by a combination of hypermobility of the thumb as well as a thumb which is longer than usual.

The wrist sign (Walker's sign) is elicited by asking the patient to curl the thumb and fingers of one hand around the other wrist. A positive wrist sign is where the little finger and the thumb overlap, caused by a combination of thin wrists and long fingers.
**DISCUSSION:** Most patients who have Marfan’s syndrome are usually diagnosed incidentally when they present for a routine physical examination. Marfan’s syndrome primarily involves the skeletal, ocular, and cardiovascular systems. Typically, patients with Marfan’s syndrome present with tall stature, ectopic lens, aortic root dilatation, and a positive family history. Less frequently, the diagnosis is made when a patient presents with complications of the syndrome, such as aortic dissection, or with involvement of the pulmonary, skin/integument, or nervous systems. Presentation of the disease varies greatly, even among family members. Some persons with Marfan’s syndrome experience only mild effects, whereas others have severe problems. Fibrillin-1 mutation causes some Marfan-like disorders with a better prognosis. Recently, mutations in the transforming growth factor b-receptor 2 (TGFBR2) gene on chromosome 3 and in the TGFBR1 gene on chromosome 9 were found in some families with apparent Marfan syndrome. These “Marfan’s syndrome type 2” families seem less likely to have ectopic lens. Skeletal manifestations are the cardinal signs of Marfan’s syndrome and usually gain the attention of a physician. The most common features include tall stature with the lower segment of the body greater than the upper segment and long, slender limbs, or dolichostenomelia; thin body habitus with increased arm span-to-height ratio; long, slender fingers, or arachnodactyly; deformities of the chest, such as pectus carinatum or pectus excavatum, scoliosis and highly arched palate with crowded teeth and dental malocclusion. Other less common manifestations include hypermobility of joints, flat foot (pes planus), reduced extension of elbows (<170°), and elongated face (dolichocephalia). Most of the features were seen in the present two cases except hypermobility of joints and flat foot (pes planus). Patients should be examined for arachnodactyly; positive wrist or Walker’s sign (the distal phalange of the first and fifth fingers of the hand overlap when wrapped around the opposite wrist); and positive thumb or Steinberg sign (the thumb projects beyond the ulnar border while completely opposed within the clenched hand which were positive in both the cases).

Cardiovascular manifestations are the most serious complications and determine the prognosis and survival in Marfan syndrome. Abnormalities include aortic root dilatation, aortic regurgitation, aortic dissection, and aortic aneurysm, which most commonly involves the ascending aorta but can involve the descending aorta. Mild aortic annulus and root dilatation with trivial mitral regurgitation, tricuspid regurgitation, and aortic regurgitation was noticed in the first patient.

Ectopic lens (subluxation of lens) is a hallmark feature of Marfan syndrome which is present in approximately 60% to 80% of patients and was also noticed in the present cases. Ectopic lens is usually bilateral, symmetrical, and upward. The diagnosis can be made by looking for iridodonesis (tremor of iris), phacodonesis (abnormal movement of lens), and a deep anterior chamber in the nondilated eye. Striae may occur over the shoulders and buttocks as noticed in the first case. Pulmonary manifestations include spontaneous pneumothorax and apical blebs. Marked dilatation of the dural sac may be seen frequently in computed tomography or magnetic resonance imaging scans, but the condition is usually asymptomatic.

The diagnosis can be established by a comprehensive clinical evaluation and diagnostic criteria have been established. The Ghent criteria are based upon family/genetic history, involvement of organ systems (primarily skeletal, cardiovascular, and ocular), and whether the...
clinical sign is major or minor. Major criteria are specific for Marfan syndrome and are rarely present in the general population. According to these criteria, Marfan syndrome in a patient with unequivocal family history is diagnosed when there is major involvement in one organ system (skeletal, cardiovascular, or ocular) and involvement of a second organ system. If the patient has no first-degree relative who is unequivocally affected by Marfan syndrome, the patient must have major criteria in at least two different organ systems and involvement of a third (skeletal, cardiovascular, and ocular) to be diagnosed with Marfan’s syndrome.

CONCLUSION: Marfan’s syndrome is an inheritable connective tissue disorder and is rare as compared to acquired connective tissue disorders. Marfan’s syndrome is one of the most common single gene defects with a prevalence of around 1 in 9800 population. It is characterized by diverse clinical manifestations. Genetic testing is nonspecific, and the diagnosis is based on clinical criteria. Despite the morbidity and mortality associated with Marfan’s syndrome, appropriate medical and surgical management can improve and extend the lives of many patients, and advancing research holds the promise of further improvements in the future.

REFERENCES:
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