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A Case Presentation of Marfan Phenotype

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ABSTRACT
INTRODUCTION: It is an autosomal dominant connective tissue disorder affecting bones, joints, ligaments.
INCIDENCE:1/10000- No ethnic or gender prediliction; male:female=1:1
GENETICS: Mutation in fibrillin-1(FBN-1) gene on chromosome 15(15q21), 30%sporadic mutation.
CLINICAL FEATURES:skeletal abnormalities:
DOLICHOSTENOMELIA- the patient is tall, the lower segment is more than upper segment by atleast 5cm(long narrow limbs). the arm span is more than height of patient by atleast 5cm(arm span: height>1.05)
ARACHNODACTYLY- Long thin fingers and toes
Skull- dolichocephalus
High arched palate with crowded teeth.
Chest and spine-pectus excavatum, pectus carinatum, straight back syndrome,kyphosis, scoliosis.
Ligamentous laxity and joint hypermobility

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I. Introduction:

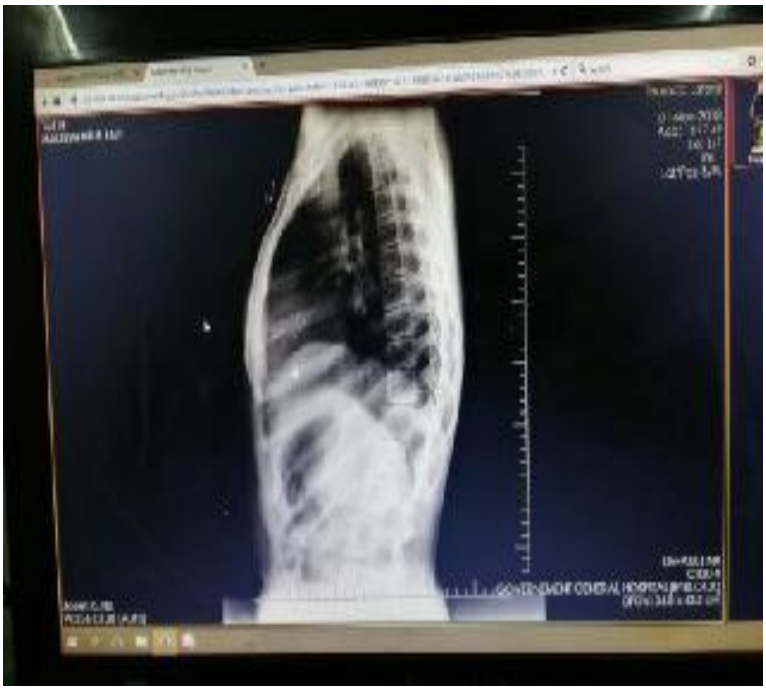
Antoine Marfan was the first to describe Marfans syndrome in 1896 in a five and half year old girl. It is an autosomal dominant inherited disorder that affects many system of body like skeletal, ocular andcardiovascular systems etc. It has no gender pedilection with equal distribution in both males and females. The prevalence of the disease is 1 in 10,000 livebirths .it mostly affects skeleton,lungs,eyes, heart and aorta. The skeletal abnormalities include dolichostenomelia i.e, the lower segment is more than upper segment by atleast 5cm. The arm span is more than height by atleast 5cm. Arm span :height>1.05. there is arachnodactyly i.e., long fingers and toes, the cranial abnormalities include dolichocephalus. Joint hypermobility and ligament laxity is common in marfans syndrome.the chest and spine abnormalities include pectus excavatum, pectus carinatum, kyphosis, scoliosis, straight back syndrome. Here I am presenting a case of marfans disease with only skeletal abnormalities without any involvement of other systems.

II. Case Presentation:

A 14years old female patient came along with her mother to the opd with chief complaint of pain in left hip since 4 months. There was no history of any trauma or fever. There is history of second degree consanguity.On general examination the patient had tall stature with arm span of 182cm exceeding her height of 172cm, therefore armspan to height ratio is about 1.05. Other systemic examinations was normal

On examination of left hip there were no swellings, anterior joint line tenderness was present. Straight leg raising test is 30 degrees. Flexion at hip joint is 90 degree and further flexion is painful and limited, extension, abduction, adduction, external rotation movements are normal. Sensory and motor examination are normal. No regional lymphadenopathy was noted. The patient was evaluated radiologically and there were no significant findins. The blood chemistry and serology were within normal limits except for elevated serum ALP of 227IU/lt.

Provisional diagnosis of marphan syndrome was done and was symptomatically treated for hip pain with analegics, immobilisation and rest with skin traction



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III. Discussion:

DIFFERENTIAL DIAGNOSIS:

Differential diagnosis include homocystinuria,familial arachnodactaly,Ehler Danlos syndrome and MENIIb. Serum methionine test must be done to rule out homocystinuria. Molecular techniques may be useful in differentiating between them.

MANAGEMENT:

Ligamentous laxity and bone overgrowth leads to several problems and need o be managed by an orthopaedician. Surgical stabilisation of spine may be needed. Pectus excavatum may also need surgical intervention. Protrusio acetabulae may be associated with pain and functional limitation of movements, surgical intervention may not be needed in them. Pes planus is associated wih internal rotation of ankle, muscle cramps, leg fatigue. Parents and doctors should be sensitive to cosmetic issues inherent in marfans syndrome. Regular eye checkups may be needed regularly to identify refractive error or amblyopia. Patients with marfan syndrome need frequent assessment of aortic root. Regular ECGs need to be taken atleast at yearly intervals.

IV. Conclusion:

Marfans syndrome is most commo ninherited connective tissue disorder with various clinical manifestations. In this case the patient exhibits only skeletomuscular features of marfans syndrome like arachnodactyly, dolichostenomelia. The patient was treated for her left side hip pain with analgesics, rest with skin traction.

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