International Journal of Health Sciences and Research
www.ijhsr.org ISSN: 2249-9571

Case Report

Musculoskeletal Manifestations in Patients with Marfan Syndrome with Unusual Presentation: A Case Report

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Received: 03/04//2014 *Revised: 24/04/2014* *Accepted: 05/05/2014*

ABSTRACT

Marfan syndrome is rare condition and an autosomal dominant systemic disorder that affects connective tissue with an estimated prevalence of about 10 cases per 100,000 births and characterized with clinical manifestations of pectus carinatum and pectus excavatum thoracolumbar scoliosis ,positive wrist sign, positive thumb sign, joint hypermobility , pes planovalgus deformity and protrusio acetabuli .Though the clinical presentation is usually typical, conventional radiological and biochemical investigation helps in confirming the diagnosis. We present a rare case of Marfans syndrome with most of the skeletal manifestation and radiological features as pictorial assay with an unusual presentation of bilateral gluteal pain.

Key words: Protrusion acetabuli, dural ectasia, bilateral gluteal pain, Marfans

INTRODUCTION

Marfans syndrome is a rare condition with normal intelligence and skeletal manifestation of pectus carinatum, thoracolumbar scoliosis, joint hypermobility, positive wrist and thumb sign. Foot being in pes planovalgus deformity and protrusio acetabuli was seen on a pelvic radiogragh. Estimated prevalence of this syndrome is about 10 cases per 100,000 births highly lethal and an average age at death is 30 – 40 years with no geographic and sex predilection and occurs by autosomal dominant inheritance.

CASE REPORT

A 12 year old female child presented with bilateral gluteal pain and difficulty in walking since 15 days .she was apparently normal until then. She was admitted in the pediatric ward to rule out potts spine and her perinatal history was uneventful. General physical examination reveals patient is poorly built and nourished with normal intelligence her facial features were malar hypoplasia dental crowding retrognathia (fig 1) along with following skeletal manifestations pectus carinatum barrel shaped chest (fig 2), hypermobility of the metacarpophalangeal joint with positive thumb sign (fig 3) positive wrist sign (fig 4), arachnodactyly (fig 5) fixed flexion

deformity of the elbow joint (fig 6) , thoracolumbar scoliosis , pes planovalgus deformity and hallux valgus (fig 7) .Her skeletal survey shows posterior wedging of the lower lumbar vertebras and thoracolumbar scoliosis and barrel shaped chest.(fig 8) X ray pelvis shows protrusio acetabuli more on the right side.(fig 9)

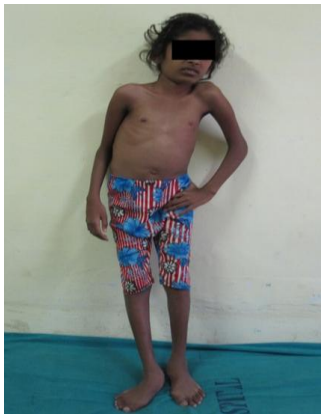
Fig 1 thin built and poorly nourished.

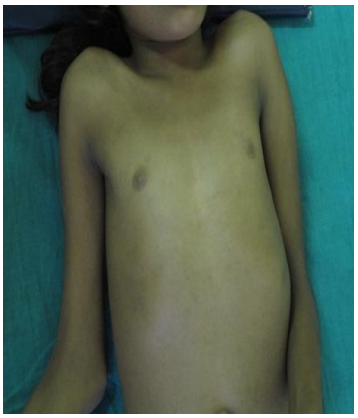
Fig 2 pectus carinatum barrel shaped chest.


Fig 3 positive thumb sign

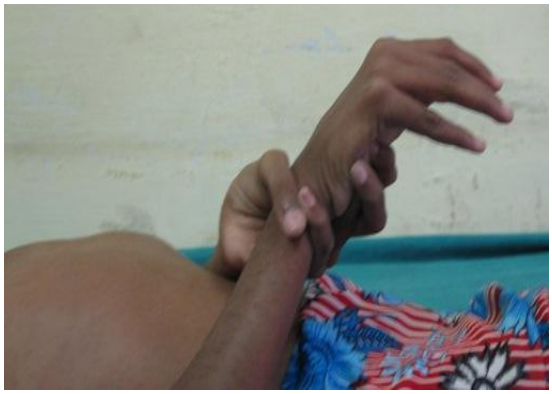
Fig 4 wrist sign.

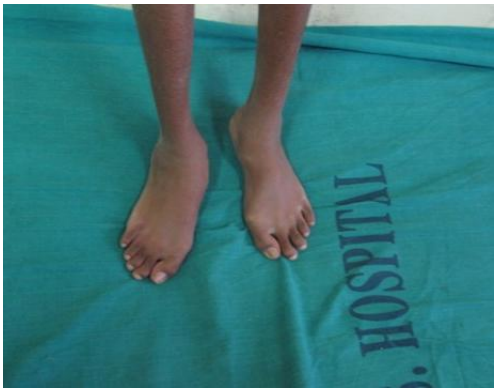

Fig 7 Pes planovalgus deformity and hallux valgus.

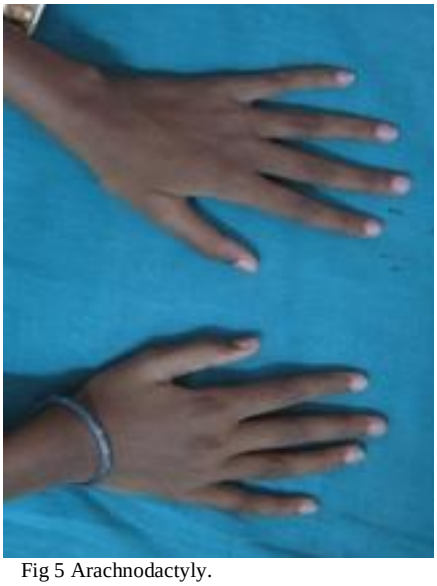
Fig 5 Arachnodactyly.


Fig 6 fixed flexion deformity of the elbow.

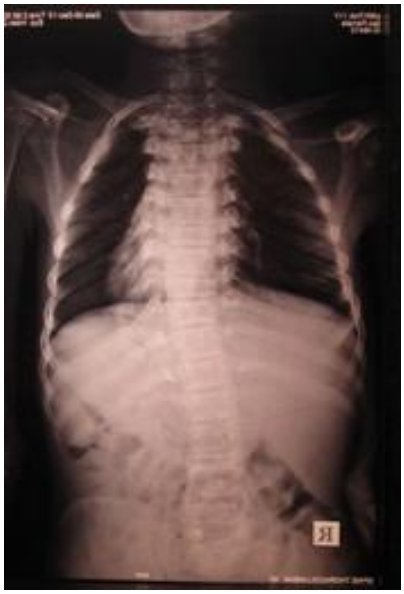

Fig 8 Thoracolumbar scoliosis

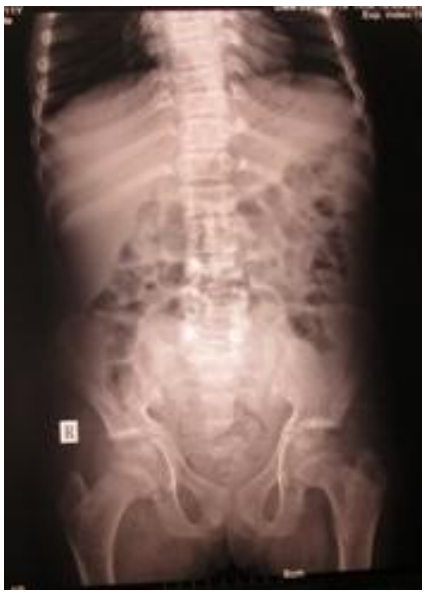

Fig 9 Protrusio acetabuli.

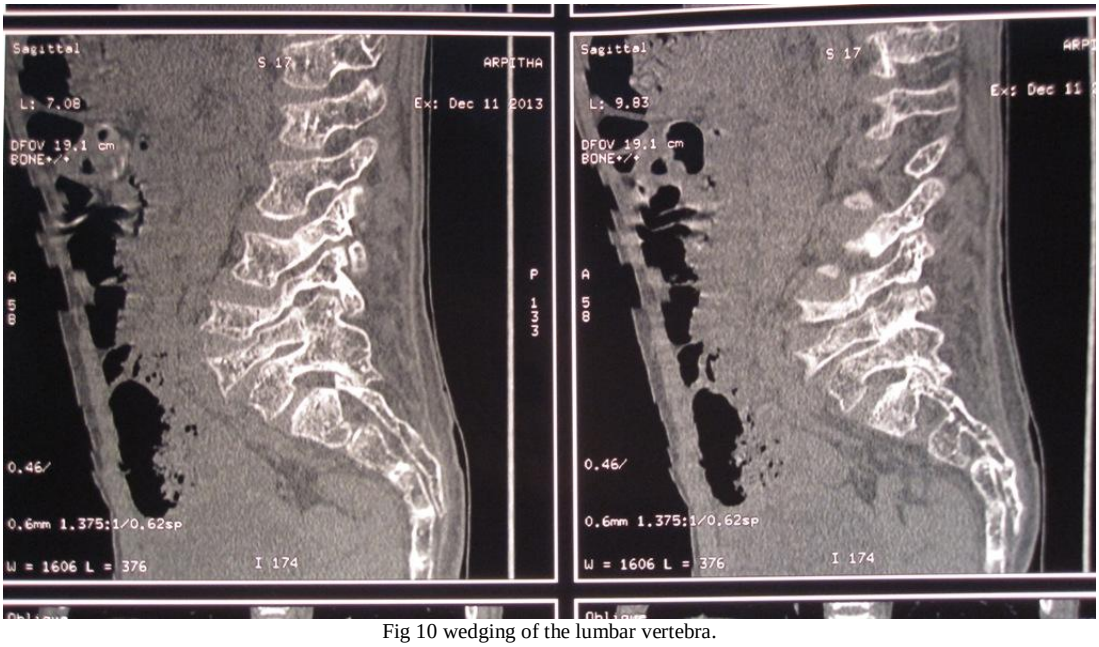

Fig 10 wedging of the lumbar vertebra.

MRI of the spine revealed posterior wedge compression of the L3 and L4 lumbar vertebras with elongated bilateral pedicles of L3 L4 L5 and S1 vertebra and spodylolysis at L4 and L5 level with sacralization of L5 vertebra .(fig 10)Large irregular extradural multiloculated cystic lesion displacing the conus with dural ectasia at the lumbar region was noted.(fig 11) With the above skeletal manifestations and radiological features marfans syndrome was suspected as per the Ghents criteria with bilateral gluteal pain being an unusual presentation.

DIFFERENTIAL DIAGNOSIS

Beals syndrome otherwise called as Congenital contractural arachnodactyly is an autosomal dominant disorder is less severe than Marfan syndrome with the differentiating features of marfanoid

habitus, arachnodactyly, crumpled ears, camptodactyly of the fingers and adducted thumbs, mild contractures of the elbows, knees, and hips, and mild calf muscle hypoplasia. [1] Shprintzene Goldberg syndrome differentiating feature are Craniosynostosis, mental retardation Loeyse Dietz syndrome usually presents with easy bruising, thin and velvety skin , bifid uvula cleft palate, hypertelorism, diffuse aortic and arterial aneurysms, craniosynostosis, clubfoot, cervical spine instability. Weille Marchesani syndrome classical features would be microspherophakia, brachydactyly, joint stiffness. Ehlers Danlos syndrome present with characterstic facies, dystrophic scars translucent elastic skin, arterial aneurysm and severe valvular insufficiency

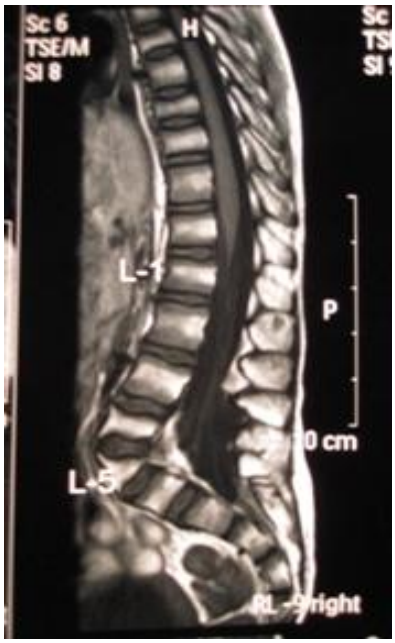

Fig 11 Dural thinning and ectasia.

DISCUSSION

Marfan syndrome is a rare disorder with the skeletal manifestation that may develop and progress during childhood. Scoliosis is a common skeletal manifestation affects 50- 60% of the marfan patients leading to marked deformity pain and restricted mobility as the age progresses. [2,3] Marfan scoliosis is found in a similar proportion in both sexes. Back pain is more common in patients with Marfan syndrome who also suffer from scoliosis than in other scoliosis patients. [4] Significant curve progression is more prevalent in Marfan scoliosis. [5] Forty-five percent of the patients had acetabular protrusion, with one-half of these being unilateral and one-half bilateral. Scoliosis was associated with acetabular protrusion in 90% of cases. [6] Scoliosis present in Marfans syndrome tends to progress faster and resistant to bracing when compared to idiopathic scoliosis. [7,8] Back pain gluteal pain is common in patients with dural ectasia which is present in 69% of the patients affected with Marfans syndrome which can be seen on MRI imaging of the spine .Dural ectasia is seen almost always in the lumbar region. [9] Joint hypermobility arthralgia, myalgia is common, affecting 85% of children under 18. Patients with the features of Marfan's syndrome (even without major diagnostic criteria) have a high incidence of progressive scoliosis, protrusio acetabuli, and various foot deformities. [10] Diagnosis is usually made by defining the involvement of multiple systems. As per the Ghents nosology the musculoskeletal criteria require at least 4 of the following features categorized under the major criteria

1. Pectus carinatum
2. Pectus excavatum requiring surgery
3. ULSR < 0.86 or span:height > 1.05
4. Wrist and thumb signs
5. Scoliosis > 20^0 or spondylolisthesis
6. Reduced extension of the elbows < 170^0
7. Pes planus
8. Protrusio acetabulae

Or 1 major feature with 2 of the following:

1. Pectus excavatum
2. Joint hypermobility
3. High arched palate
4. Dental Crowding

5. Characteristic face. [11]

Many patients are diagnosed before the age of ten where as few patients with four criteria develop at a later stage of their life. [12] Diagnosis is challenging as it requires definition of diverse clinical manifestation and input from other specialists. . Failure to make a diagnosis or making an inappropriate diagnosis of Marfans syndrome has social, lifestyle and medical consequences for the individual as well as the family.

CONCLUSION

In summary, the musculoskeletal manifestations of Marfan syndrome are diverse and include the entire skeleton in various degrees. Keeping in mind an unusual presentation of bilateral gluteal pain with above skeletal manifestation diagnosis of Marfans syndrome can be suspected. Treatment options vary according to the severity of symptoms presented by patients and are directed at the symptomatology like in our case Taylor Knight brace was applied to prevent the progression of the scoliotic deformity .

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How to cite this article: Vijay C, Ravishankar R, Adarsh T et. al. Musculoskeletal manifestations in patients with marfan syndrome with unusual presentation: a case report. Int J Health Sci Res. 2014;4(6):249-253.

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