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Thoracolumbar Paraspinal Heterotopic Ossification (H.O)

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Authors' contributions

This work was carried out in collaboration between all authors. Author ME designed the study. Author MHJ performed the statistical analysis, wrote the protocol and wrote the first draft of the manuscript, managed the analyses of the study. Author RI managed the literature searches. All authors read and approved the final manuscript.

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Case Study

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ABSTRACT

A rare disorder in the skeletal system is the fibrodysplasia ossificans progressive (FOP) that is a sever, disabling disease, autosomal dominant, occasionally sporadic, ectopic ossifying condition and involvement of the muscles together with malformation the great toe.
We report a case of the thoracolumbar heterotopic (H.O) in the 16 years Old Iranian female student. The patient presented with about a history of the back pain and restriction of the lumbar motion especially in the flexion and bilateral short hallux valgus.
This is the first report of an atruamatic thoracolumbar paraspinal H.O in the our center. We don't the awareness from fibrodysplasia ossificans progressiva.
The understanding and awareness of the FOP is essential for the physicians according to the extra-skeletal heterotropic bone formation and the congenital hallux.

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Keywords: Fibrodysplasia ossificans progressive; heterotopic ossification; malform great toe.

1. INTRODUCTION

The fibrodysplasia ossificans progressiva (FOP) is a rare genetic inflammatory disorder of the connective tissue. The term Fibrodysplasia ossificans progressive is preferred to the myositis ossificans because the ectopic osteogenesis occurs in the connective tissue within the muscles, faciae, ligaments, tendons and the joint capsules, rather than in the muscle fibers themselves [1,2]. It is a condition with prevalence of 1 case in 2 million. The hallmarks of the disease are heterotropic calcification of the skeletal muscles and the abnormalities of the bilateral big toes which may be the only abnormality present at birth [3]. The abnormality of big toes is the characteristic and helps to distinguish from the other bone and the muscle problems [4]. The most patients are misdiagnosed early in the life before the appearance of heterotopic ossification and undergo the diagnostic procedures that can cause the lifelong disability [4,5]. The most patients with the FOP are misdiagnosed during the childhood as having the sarcomatus or the aggressive fibromatosis and undergo unnecessary and harmful the operation. The nearly 90% of the FOP are misdiagnosed in the worldwide and the 67% undergo the dangerous and unnecessary procedures that lead to permanent the disability in more than of fifty percent the all patients [5]. The few physicians are the awareness of the classic features of the FOP or the clinical significance of the congenital malformation of the great toes. The genetic tests for the FOP are available recently before the appearance of heterotopic ossification for the avoidance of unneeded surgery or treatment. The genetic cause of the FOP lies in the (Activin Receptor IA) ACVR1 gene, which encodes a type 1 BMP the trans-membrane receptor. The mutation in this gene causes inherited and sporadic FOP [6,7].

2. CASEREPORT

Following institutional review board and ethical approval, this study was conducted in St Zahra Hospital Isfahan University of Medical Sciences.

We report a case of the thoracolumbar heterotopic (H.O) in a 16 years Old Iranian female student.

The patient presented with about the history of back pain and the restriction of lumbar motion especially in the flexion and the bilateral short hallux valgus.

In the past medical history; she has not the history of family and consumption drug and trauma and disease. But she has the mild mental retardation.

She had the progressive pain in her lower back without the neurologic symptom or the radiculopathy. The pain was increased with the physical activity and the change of position.

We could palpe the multiple, small lamps (the tumor like lesions) over the her back that was firm, immobile on examination (Fig. 1).

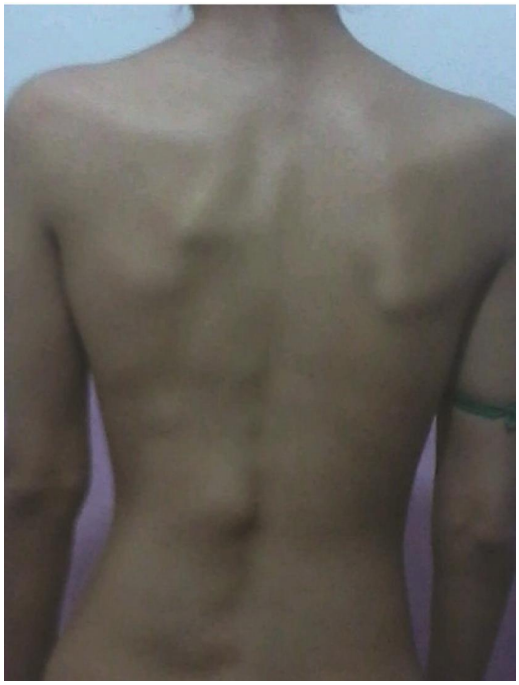


Fig. 1. Multiple rigid mass in back and periscapular region

These masses were painful in the some area and located only within the left thoracolumbar muscles.

The gait analysis was normal but she had the little flexible deviation to the left side. The great toes were the short and monophalangic in the valgus deformity (Fig. 2).

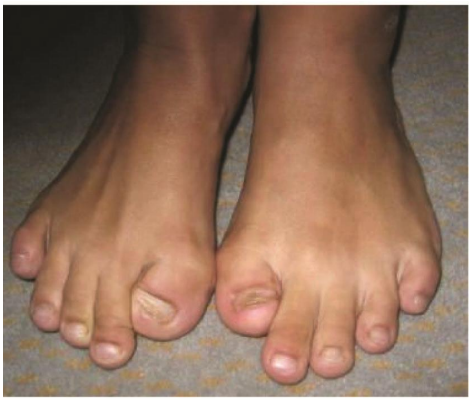


Fig. 2. Malformed great toes with hallux valgus

The muscles force and sense and deep tendon reflexs were the normal.

The many lab tests such as blood profile; kidney and liver function tests, thyroid and parathyroid functional tests, calcium and phosphorus and alkaline phosphates and serum vitD3 level were normal.

We had taken multiple imaging studies such as the thoracolumbar radiography that demonstrated an extensive ossified mass along the paraspinal muscles extend to subcutaneous tissue from T3 vertebral level to L5 vertebral level (Fig. 3).

In tri-dimensional CT scan was reported a plate-like and oval shaped ossification areas seen in the left paraspinal muscles from T10 vertebral level to_L5 vertebral level (Fig. 4).

In magnetic resonance imaging (T1-weighted - T2-weighted) was defined the elongated para spinal mass that had the characteristic finding of the mature hypertrophic ossification (H.O) with a well defined peripheral rim of the hypo intense T1W and T2W signal (Fig. 5).

In the Whole body scan with 20 mci 99TC-MDP was shown several hot spots radionuclide uptake in the left site of the thoracolumbar area (Fig. 6). The rest of skeletal was intact.

This is the first atruamatic the thoracolumbar paraspinal report of hypertrophic ossification in this center. We decided to the excisional biopsy for the improving lumbar the pain and motion.

We wasn't aware about the fibrodysplasia ossificans progressiva in this center to now. In the surgery time we removed all the mature bone with a few millimeter of the soft tissue margin and send to the pathologic center (Fig. 7). After the several days we was informed from the pathologic report "Heterotopic endochodral" ossification without the malignancy. Therefore we understood that the correct diagnosis of the patient is a rare disease with the name of fibrodysplasia ossificans progressive. The patient was treated with corticosteroid, indometacine (NSAID) and celexib (cox-inhibitor) in the post operation.

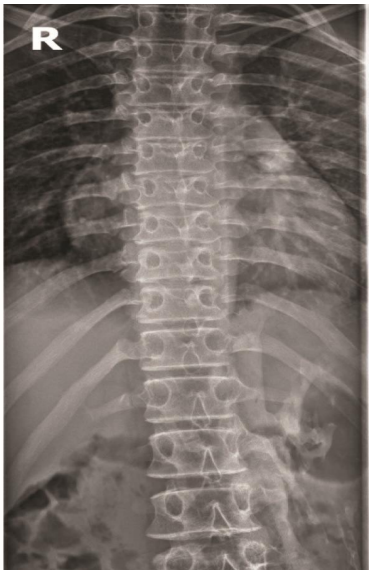
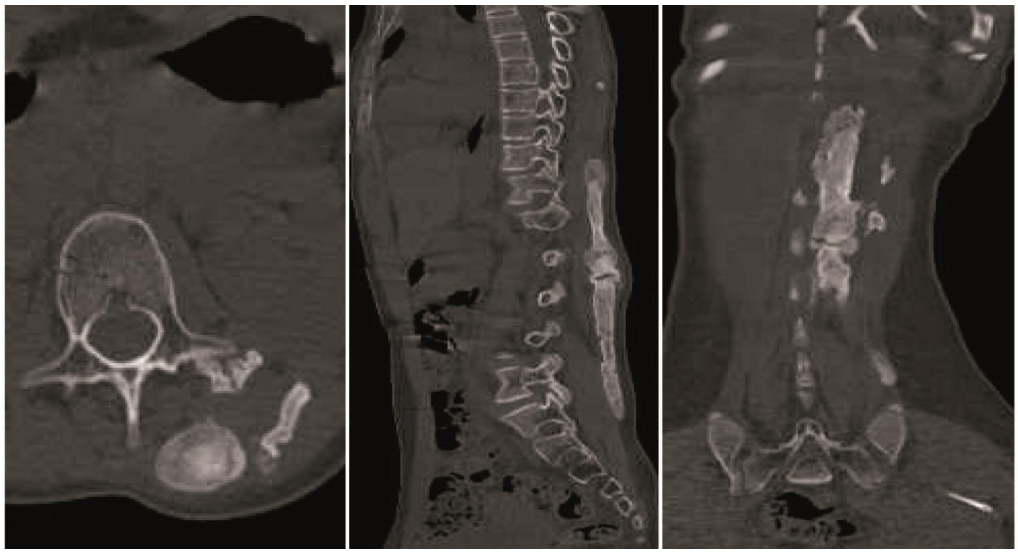


Fig. 3. Anterior – posterior thoracolumbar X-Ray shows paravertebral ossification

3. DISCUSSION

The Gay patin was reported the first case of FOP in the 1692, who presented a case of the young patient that "become hard as wood" [6]. Then the Munchmeyer applied a term of the myositis ossificans progressive in the 1867. However, it turned out inappropriate, because the primary changes affect the connective tissue. The etiology of disease is unknown although the familial cases were reported with the autosomal dominant pattern and which is occasionally the result of spontaneous de novo mutation [7,8]. The initial presentation, which is usually in the first two decades of the life involves painful and the hard soft tissue swellings over the affected muscles, ligaments, tendons and so, lead to ossification. It usually occurred from the birth to the age of 16 years (mean age 4.6 yrs), following the spontaneous or the trauma-induced "flare-ups" [8].



Axial **Sagittal** **Coronal**

Fig. 4. CT- Scan Indicates bony mass in paravertebral muscles
Axial, sagittal and coronal slices in CT scan

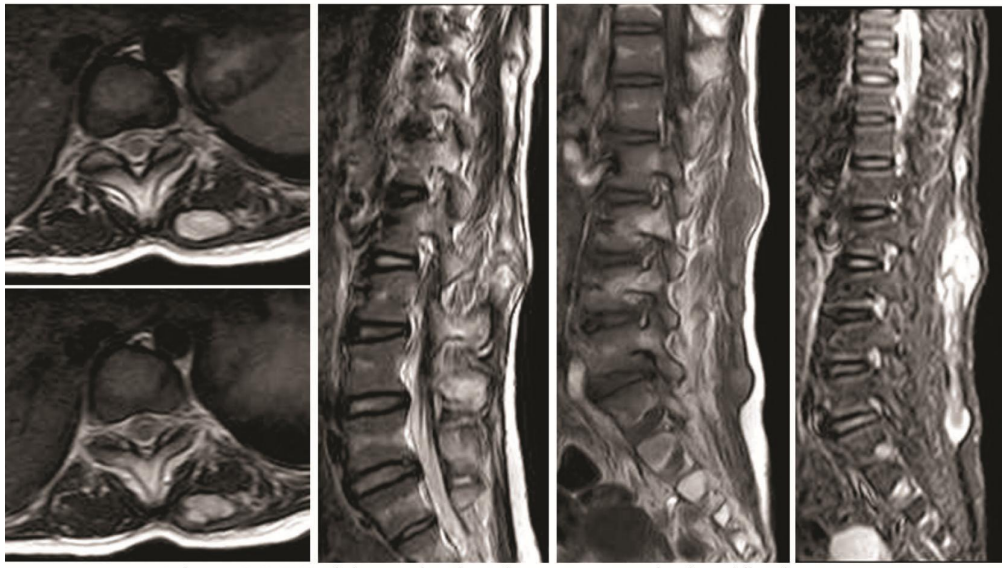


Fig. 5. MRI of thoracolumbar shows paravertebral ossification
Axial T2W, T2W left parasagittal, T1W left parasagittal and STIR respectively (left to right)

As early as possible, the FOP should be diagnosed and non- invasively based upon the clinical and radiological findings. The mainstay of the diagnosis is bilateral great toe anomaly present from the birth, reported in the 79% to 100% of the patients. The most characteristic deformity is the microdactyly of the both hallux due to the single phalanx in the valgus position. The most commen anomalies are the short first metacarpus and the fifth finger with clinodactyly [9].

The ectopic ossification occurs the lifelong with the records of the initial appearance at the mean age of three or five years the almost all patients. There is well defined pattern for the heterotopic

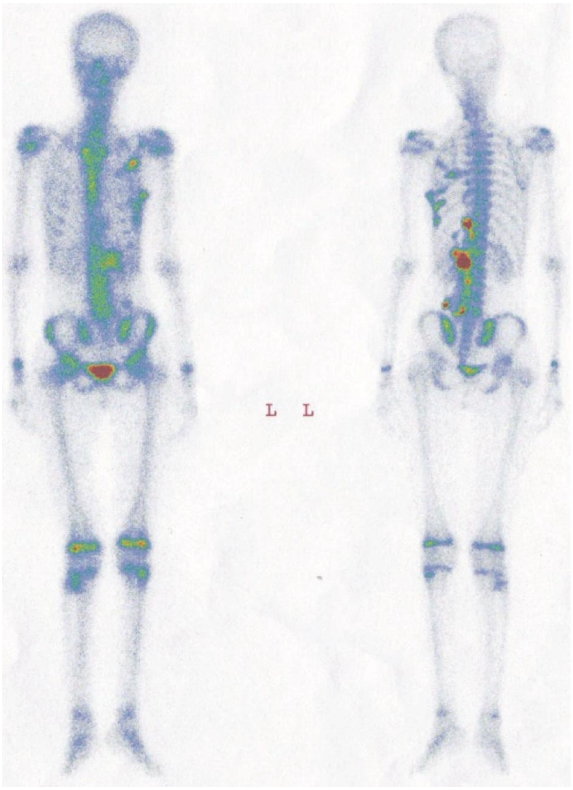


Fig. 6. Increase uptake in 99TC-MDP whole body scan

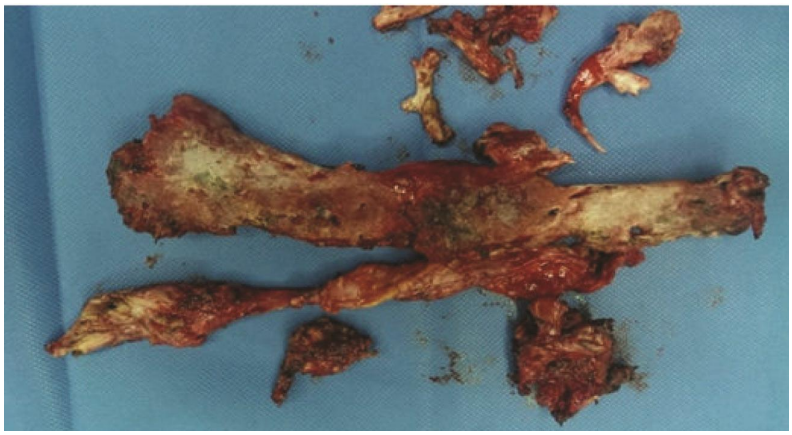


Fig. 7. Excisional biopsy

ossification, first in the axial body (the paraspinal muscles) and then the shoulders and hips are affected more than the distal limbs. Where heterotopic ossification is less obvious, radiographic findings may assist in the diagnosis. The Scoliosis is a common finding and is often the result of the asymmetric heteropic bones formation [10].

The diagnosis of the FOP is clinical and radiological findings and based on the presence of three major criteria: 1) congenital malformation of the great toes 2) progressive heterotopic endochondral ossification 3) progression of the disease in well-defined anatomical and temporal patterns. The ossification appears proximally before distally, axially before appendiculary,

cranially before caudally, and dorsally before venterally.

The differential diagnosis includes other the genetic illnesses such as the progressive osseous heteroplasia (POH), Albright hereditary osteodystrophy (AHO), osteoma cutis, ankylosing spondylitis (AO), still's disease and klippel-feil-syndrome. The FOP must be differentiated from the inflammatory osseous tumor, and the aggressive fibromatosis [11].

The routine laboratory tests including calcemia and phosphatemia are usually the normal. The radiographic examination may be help in the diagnosis minor osseous formation. The Bone scan with 99-TC-MDP may show early the heterotopic ossification and the assessment of disease extension [10,11].

There is no effective known treatment for the FOP. The drug treatment was published by the Kaplan et al guidelines and currently on the new guidelines published by the same authors that use of corticosteroid in the acute phase and using a combination of leukotriene inhibitor and a Cox-inhibitor [11]. The other drugs such as NSAIDs (indometacine and brofen …) and the calcium chelator (etidronate) bisphosphonates (aldronate), single dose radiotherapy have been tried by the some physicians. The physiotherapy and rehabilitation also beneficial and advisable. The diagnosis is based on the history and the clinical involvement. The biopsy and the surgical excision may be avoided because excision is futile occasioally, because surgical trauma predictably leads to the stimulation of new and more robust heterotopic ossification at the operative site [12]. Now days, the discovery of the FOP gene establishes a milestone in the understanding and the diagnosis of the disease and for the genetic counseling in the suspicious cases and minimizing trauma and painful flare ups [11,12].

4. CONCLUSION

In Conclusion, The understanding and awareness of the FOP is the essential for the physicians especially for pediatricians and orthopaedic surgeons. Although all patients with malformed great toes will not have FOP, the consideration of FOP must be part of the differential diagnosis. Once swelling of soft tissue occurs, the clinical diagnosis is certain and genetic testing that can be used to prevent iatrogenic harm.

CONSENT

As per international standard or university standard written patient consent has been collected and preserved by the authors.

ETHICAL APPROVAL

As per international standard or university standard, written approval of Ethics committee has been collected and preserved by the author(s).

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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