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Case Report

Prevention of pathological fracture in fibrous dysplasia of proximal femur in a 24-year-old female a rare presentation - a case report

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ABSTRACT

Fibrous dysplasia (FD) was historically mentioned in the medical literature in 1938 by Dr. Lichtenstein and in 1942 by Dr. Lichtenstein and Jaffe. The term Jaffe-Lichtenstein syndrome is sometimes used synonymously with monostotic FD or to denote cases of polyostotic FD with café au lait spots, but no endocrine dysfunction. Our case is a 24-year-old female, housewife presented to our hospital with a complaints of right hip pain and backache with difficulty in walking of 6 months duration. She had dull aching pain which increased in intensity with the course of time, there was no radiation, but was aggravated by movements. She was managed with dynamic hip screw along with curettage of the lesion and bone graft, after 9 months of follow up, she had no pain over her hip and there was calcification over the previous lesion, our patient had a very favourable outcome.

Keywords: Fibrous dysplasia of proximal femur, Dynamic hip screw, Bone graft

INTRODUCTION

Fibrous dysplasia (FD) was historically mentioned in the medical literature in 1938 by Dr. Lichtenstein and in 1942 by Dr. Lichtenstein and Jaffe. The term Jaffe-Lichtenstein syndrome is sometimes used synonymously with monostotic FD or to denote cases of polyostotic FD with café au lait spots, but no endocrine dysfunction.[1,2]

FD is a rare benign bone lesion resulting from congenital dysplasia of bone, in which osteoblasts cells abnormally differentiate leading to woven immature bone formation by replacement of normal cancellous bone cells. This is originated by missense gene mutation at chromosome 20q13.[2,3]

FD may involve one bone (monostotic) or more than one bone (polyostotic). It can involve any bone in human body, but the long bone, skull, and ribs are common. The incidence rate of FD is difficult to estimate. The prevalence of FD has been estimated almost 1/30, 000.[4] Both genders are equally affected. It is usually an incidental imaging finding. It is a slowly progressive disease which can involve any bone in human body, but the long bone, skull, and ribs most affected.[5] FD localized to long bones is easy to diagnose considering adequate clinical and radiological information and typically through microscopic findings. This report is a rare case which has been reported only twice earlier. The case is about the monostotic FD of right hip bone.

CASE REPORT

A 24-year-old female, housewife came to our hospital with a complaints of right hip pain and backache with difficulty in walking of 6 months duration. She had dull aching pain which increased in intensity with the course of time, there was no radiation, but was aggravated by movements. There was no history of other systemic illness. On examination tenderness was elicited in right posterior iliac

bone area, distal peripheral neurovascular status was normal (Figure 1). On X-ray of pelvis (AP-view) there was well defined lytic lesion with ground glass appearance over greater trochanteric region and intramedullary, expansible lesion with a smooth sclerotic margin over left proximal femur neck area, neck-shaft angle measured on the AP radiograph, defined as the angle between the axis of the femoral neck and the anatomical axis of the femur, there was no varus or valgus deformity of femur.

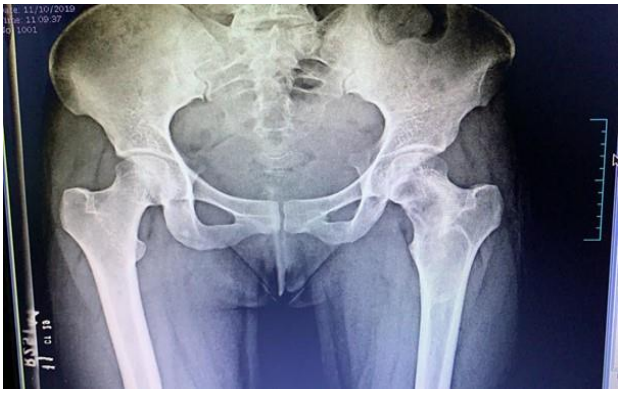

Figure 1: Radiograph of pelvis showing the lesion.

Magnetic resonance imaging (MRI) of both hip joints was done which showed altered signal in the head, neck, and proximal shaft of left femur suspicious of an indolent lesion like FD, cortical thinning, and focal breach of the cortex of medial aspect of neck of femur suggestive of old pathological fracture with no evidence of soft tissue, oedema, or collection in the region (Figure 2).

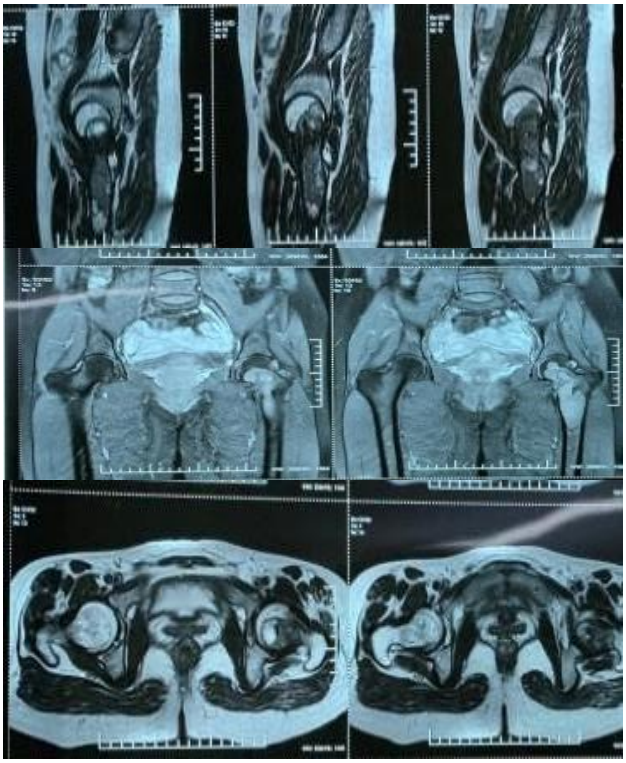

Figure 2: Magnetic resonance images.

Surgery

Patient was planned for open reduction and internal fixation with dynamic hip screw (DHS), under all aseptic precautions and in spinal and epidural anaesthesia, lateral approach for left hip was taken. The neck of femur was identified under image intensifier and from the lytic region, sclerotic tissue was curated and tissue was sent for histopathology examination, after curettage tricortical bone graft was harvested from iliac crest and graft was transferred into the defect which was left after curettage, then for prevention of any subsequent pathological fracture, a four hole long barrel DHS plate with 135 angulation was used along with derotation screw, wound closure was done and post operatively patient was advised nil weight bearing walk for 4 weeks along with oral weekly bisphosphonates for 6 months and calcium therapy, partial weight bearing was started at 4 weeks post-operative period, then full weight bearing was done. Physiotherapy with bisphosphonates were continued for 6 months (Figure 3 and 4).

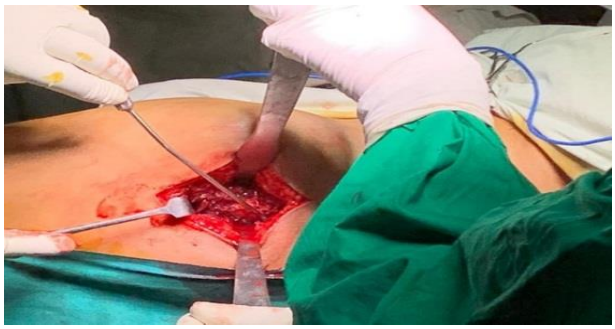

Figure 3: Surgical approach.

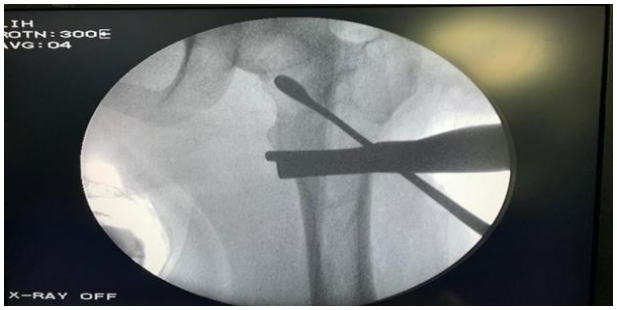

Figure 4: Identification of the lesion under C-arm.

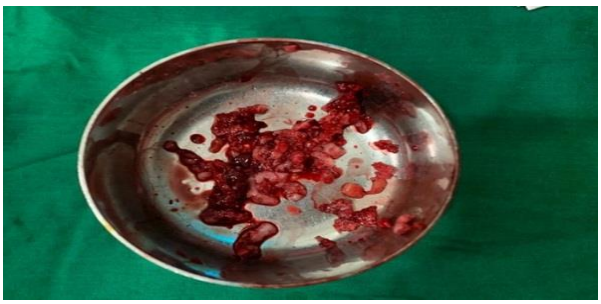

Figure 5: Tissue removed from the site of the lesion.

DISCUSSION

FD is a considerably rare bone disorder. Bones affected by this disorder are replaced by abnormal scar-like (fibrous) connective tissue. This abnormal fibrous tissue weakens the bone, making it fragile and susceptible to fracture. Pain is the most common noticed complaint in the affected areas.[6] As children grow, affected bone may become misshaped (dysplastic). It may only affect solitary bone (monostotic disease), or the disorder can have large spread, affecting multiple bones throughout the body (polyostotic disease). The severity of the disorder shows variations in intensity of involvement from one person to another. Any part of the skeleton can be affected, but the long bones of the legs, the bones of the face and skull (craniofacial area), and the ribs are most often affected. Diagnosis in children or young adults is made clinically and radiologically, but mild cases may go undiagnosed until adulthood.[7] In some cases, FD may not require treatment and it may show spontaneous resolution; in most other cases, certain medications and surgical intervention may be recommended.

Symptoms associated with FD mostly depend upon the specific bones involved in the body as any part of the skeleton can be affected, but the long bones of the arms and legs, the bones of the face and skull (craniofacial area), and the ribs are most often affected. Most of the times monostotic FD presents as a painless swelling on the ribs. FD affecting the spine can cause abnormal curvature of the spine (scoliosis).[8] When the long bones of the legs are affected, this can lead to frequent pathological fractures with trivial trauma due to weight bearing when walking or standing. In addition, the long bones can eventually become bowed.

The underlying cause of FD is incompletely understood. Many researchers believe that the disorder is caused by a mutation in a gene called GNAS1.[9,10] The gene mutation occurs after fertilization of the embryo and is therefore not inherited, so passage of affected genes to future generations is also not possible. Individuals affected with FD have some cells with a normal copy of this gene and some cells with the affected gene (mosaic pattern). The variability of symptoms is attributed to the ratio of healthy cells to abnormal cells.

The GNAS1 gene encodes for a protein known as a G-protein. In this mutation a gain-of-function is seen in the GNAS1 gene resulting in the overproduction of this G-protein. Which leads in the overproduction of a molecule known as cyclic adenosine monophosphate (cAMP), which is involved in the differentiation of osteoblasts in bone. Osteoblasts are bone-forming cells that form new bone. The human skeleton is constantly dynamic. So, it is believed that the disease involves increased bone turnover. Bone turnover being a normal process in which bone is produced and resorbed in equilibrium which is maintained by interaction between osteoclasts and osteoblasts. The interaction is a complex process that involves multiple factors. Improper differentiation of osteoblasts due to mutation of the GNAS1 gene is believed to contribute to the development of FD.[10,11] The activity of the osteoclasts in removing bone probably allows skeletal progenitor cells including immature osteoblasts and fibrous tissue to have more space to grow and multiply.

When the cells from different system are involved, such as endocrine system or skin cells are involved in addition to osteoblasts, it results into McCune Albright syndrome.[11]

In our case along with prevention of future possibility of pathological fractures and removal of the affected tissue and inducing osteoblastic activity with tri-cortical bone graft harvested from iliac crest was the main purpose of surgery.

The DHS plate has several advantages in the prevention of pathological fracture of the femoral neck in our patient as it provides stronger angular stability than other implants, allowing it to withstand greater forces which are generated in the hip region while walking and weight bearing. It functions as a scaffold with prevents moulding of the weaker dysplastic bone. In addition, FD destroys normal bone matrix and replaces it with fibro-osseous tissue, which lacks anchoring strength, so a dynamic plate provides necessary compression and maintenance of the normal architecture of the joint. And lastly it does incorporate a dynamic compression mechanism, allowing it to be utilized in large bone voids filled with bone graft to minimize collapse.[12] Curettage weakens the bone more and may lead to pathological fracture so a tricortical or a cancellable bone grafting of void is necessary as the cancellous grafts are rapidly reabsorbed and replaced by fibrous tissues the cortical autologous grafts survive longer than cancellous bone grafts, and allogeneic cortical bone grafts are replaced by a host bone at the slowest rate. Oral bisphosphonates (BPs) have been used to treat the disease the FD and to inhibit osteoclastic remodulation of osteoid. It reduces the likelihood of a new fracture while simultaneously demonstrating radiological improvement such as progressive ossification of the lesion, cortical thickness, and reduction in lesion size.[13] Our study has a limitation of sample size, which is mainly due to the rarity of the disease.

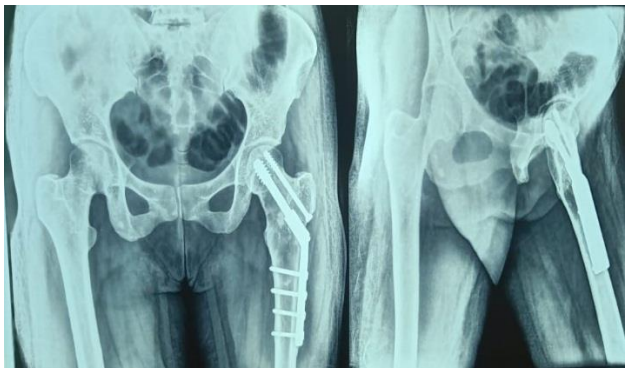

Figure 6: Radiographs done at 9 months of recovery.

Figure 7: Clinical recovery of the patient.

CONCLUSION

Even though numerous treatment modalities for FD have been reported in the literature, all possible efforts should be made by the treating surgeon to reduce complications such as implant failure, recurrence of lesion, and prevention of pathological fractures. Based on the optimal radiological and clinical outcome of our case, as the patient was able to squat, walk freely without any pain, so we conclude that the DHS with bone graft can be employed for internal fixation as a means for prevention of a pathological fracture in monostotic femoral neck lesion without any long-term complications (Figure 6 and 7).

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