POLYOSTOTIC FIBROUS DYSPLASIA

Adil Qayyum, Ruqqayia Adil, Faisal Basheer, Jawad Jalil*

Combined Military Hospital Abbottabad, Pakistan, *Combined Military Hospital Multan, Pakistan

ABSTRACT

Fibrous dysplasia is a non inherited skeletal disorder in which bone-forming cells fail to mature and produce too much fibrous or connective tissue. We report a case of 3 years old female with limping gait and limb length discrepancy. X-ray lower limb showed lucent expansile lesions in metaphyseal regions of right femur & tibia. Skeletal survey showed unilateral monomelic similar like lesions involving right lower limb and right iliac bone, right humerus and radius. On the basis of X-ray and biopsy findings, diagnosis of polyostotic fibrous dysplasia of right upper and lower limb was made. She was referred to Rehabilitation department for management of her limb length shortening and bone deformities.

Keywords: Fibrous dysplasia, Limb length shortening, Polyostotic.

INTRODUCTION

Fibrous dysplasia also known as Lichtenstein – Jaffe disease, is a non inherited skeletal disorder in which bone-forming cells fail to mature and produce too much fibrous or connective tissue. There is abnormal differentiation of osteoblasts, which leads to replacement of normal marrow and cancellous bone by immature woven bone with fibrous stroma[1]. Areas of healthy bone are replaced with this fibrous tissue.

The importance of early diagnosis is that if not detected earlier, the disease may progress causing deformities of limbs, skull & face. Early detection can prevent loss of vision from orbital involvement or malignant transformation of the lesion.

The defect occurs at some point after conception, most likely early in fetal development. Monostotic fibrous dysplasia, characterized by involvement of only one bone, is considerably more prevalent than the polyostotic form. Males and females are thought to be affected evenly, although recent research has shown a slight female preponderance. Any bone may be affected, the long bone, skull, and ribs most often[2]. In monostotic fibrous dysplasia, ribs and proximal femoral site accounts up to 28% and 23% respectively.

CASE REPORT

My patient 3 years old female reported in children OPD at CMH Abbottabad, with vague complaints of limping gait for last 3 to 4 months. No other clinical complaints were present. On examination, the child's right lower limb was 2 cm shorter than the left side. X-ray lower limb was advised which showed multiple, lucent, expansile lesions in metaphyseal regions of right femur & tibia with surrounding sclerosis and internal ground glass haze. Provisional Diagnosis of Fibrous dysplasia was made and skeletal survey was done. Unilateral monomelic lesions are noted, involving right lower limb and right iliac bone along with similar like lesions in right humerus and radius (fig-1). No other bone was involved.

Her serum alkaline phosphatase was also raised. Bone biopsy was done for further confirmation of diagnosis, which showed small nonmineralized trabeculae of woven bone in bland cellular and collagenous matrix in the lesions. More radiolucent lesions were composed of predominantly fibrous elements, whereas

Correspondence: Dr Adil Qayyum, MCPS, FCPS, Classified Radiologist, Radiology Department CMH Abbottabad, Pakistan.
Email:qayyum.adil@gmail.com
Received: 11 August; revised received: 17 Sep 2014; accepted: 30 Sep 2014

more radiopaque lesions contained greater proportion of woven bone. On the basis of X-ray and biopsy findings, diagnosis of polyostotic fibrous dysplasia of right upper and lower limb was made. Detailed history was taken to rule out any history of acne, precocious puberty, pigmented cutaneous lesions and endocrine abnormalities to rule out McCune Albright syndrome. She was referred to Rehabilitation department for further management of her limb length shortening, to improve joint mobility and to correct bone deformities.

DISCUSSION

Fibrous dysplasia in itself is not a rare disorder; it is reported to represent 5% to 7% of benign bone tumors. It is primarily a developmental abnormality of the bone-forming mesenchyme in which fibrous tissue gradually expands and replaces the bone. It is believed to be a non-neoplastic hamartomatous developmental lesion of bone, of unknown origin. Fibrous dysplasia is a sporadic condition that results from a postzygotic mutation in the GNAS1 (guanine nucleotide binding protein, α – stimulating activity polypeptide1) gene. In most cases, the radiographic characteristics of polyostotic Fibrous Dysplasia and the clinical information are sufficient to allow the practitioner to make a diagnosis without a biopsy.

Males and females are thought to be affected evenly, although recent research has shown a slight female preponderance. Our patient was also a female patient. There is wide range of presentation between 10 and 70 years of age, with 75% of patients presenting before the age of 30 years. Mean age of polyostotic fibrous dysplasia is 8 years. Our patient was 3 years old.

It is important to have an earlier diagnosis of fibrous dysplasia to prevent the further complications and identify patients who will benefit from non surgical or surgical treatment. The disease can be diagnosed & managed earlier before progressing towards deformities of limbs, skull & face. Early detection can prevent loss of vision from orbital involvement or benign / malignant transformation of the lesion. Follow-up is important in fibrous dysplasia to prevent deformities as a result of the disease and check for recurrence. In 50 percent of cases, fibrous dysplasia will re-occur.

In our case, patient presented with limping gait and fibrous dysplasia was incidentally discovered. International studies also showed the condition is often an incidental finding and is usually painless. Children usually present with leg pain, limp and pathological fracture.

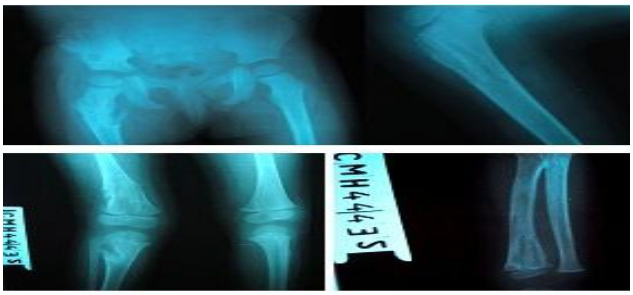

Figure-1. Unilateral monomelic polyostotic fibrous dysplasia.

Alternatively it may present due to bony expansion or remodeling[3]. Morbidity may arise from compression and displacement of adjacent structures. This is particularly true in craniofacial fibrous dysplasia, where the content of the orbit or cranial nerves may be compressed. The distribution of bones in polyostotic fibrous dysplasia is often unilateral and monomelic. Femur, which is the commonest being 91 % involved. The other common sites are tibia 81%, pelvis 78%, skull & facial bones 50%, foot, ribs, upper extremities, lumbar spine, clavicle and cervical spine[4].

The radiographic features on various modalities are quite diagnostic. The lesion has typical ground-glass opacities which may be completely lucent (cystic) or sclerotic with well circumscribed lesions on plain radiographs. Extremities like femur is a common site with classical radiographic features[5]. Our patient also presented with similar lesions in upper & lower limbs. It may also lead to bowing deformities,

shepherd's crook deformity of femoral neck, discrepant limb length, looser zones and premature fusion of growth plates leading to short limb/stature. CT scan also confirms ground-glass opacities with well-defined borders, expansion of bone, with intact overlying bone and endosteal scalloping. MRI is not particularly useful in differentiating fibrous dysplasia from other entities as there is marked variability in the appearance of the bone lesions, and they can often resemble tumour or more aggressive lesions. T1W sequence show heterogeneous signal, usually intermediate. T2W with heterogeneous signal, usually low, but may have regions of higher signal and T1W post contrast images may have heterogeneous contrast enhancement[6]. Nuclear scan demonstrates increased tracer uptake on Tc99 bone scans (lesions remain metabolically active into adulthood).

Fibrous dysplasia might be monostotic or polyostotic or involve large area of the skull. The lesions of fibrous dysplasia appears in three distinctive clinical patterns. The most severe form of FD is McCune-Albright syndrome, which is more commonly found in females and is associated with short stature due to premature closure of the epiphyses and with endocrine abnormalities and pigmented cutaneous lesions. Another severe form is Mazabraud syndrome. It is characterized by the association of polyostotic fibrous dysplasia of the bones with solitary tumours of large muscle groups, occurring predominantly in the lower limbs, and myxomas[7].

Pathological fractures are the most common complication of this entity as bone affected by fibrous dysplasia is weaker than normal and thus susceptible to fractures. Sarcomatous de-differentiation (osteosarcoma, fibrosarcoma, malignant fibrous histiocytoma or rarely chondrosarcoma) is occasionally seen (less than 1%) and is more common in the polyostotic form[8].

The differential diagnosis of fibrous dysplasia includes lesions like Paget's disease, Non ossifying fibroma, Simple bone cyst, Enchondroma, Adamantinoma, Aneurysmal bone cyst, Osteofibrous dysplasia, Diffuse sclerosing osteomyelitis and Giant cell tumour. The main factor that guide the approach are the patients age, location of the lesion, symptoms along with classical radiographic appearance.

Treatment may include reduction in risk of complication such as rickets or fractures and medications to strengthen bones. Medication known as bisphosphonates have been shown to reduce pain associated with the disease. Physiotherapy is done to improve joint mobility and surgery to correct bone deformities. Radiotherapy is contra-indicated not only because the tumor is radioresistant but also because of the probable increase of the capacity for the dysplasia sarcomatous transformation. Usually the prognosis is good although complications occur more frequently among young patients or those with polyostotic forms of the disorder.

CONFLICT OF INTEREST

This study has no conflict of interest to declare by any author.

REFERENCES

1. Bhattacharyyal, Islam N, Cohen D. Diagnostic discussion. Fibrous dysplasia of bone. Todays FDA 2014; 26(1):47-50
2. MacDonald-Jankowski D. Fibrous dysplasia: a systematic review. Dentomaxillofac Radiol 2009: 38(4):196-215
3. V Nitvasri, PS Haris, T Bose, A Balan. Fibrous dysplasia- a 13-year retrospective radiographic analysis in a south Indian population. Dentomaxillofac Radiol 2011; 40(5):282-9
4. Thomsen MD, Rejnmark L. Clinical and radiological observations in a case series of 26 patients with fibrous dysplasia. Calcif Tissue Int 2014; 94(4):384-95
5. Mrabet D, Rekik S, Sahli H, Ben AM, Meddeb N, Sellami S. An extensive hemimelic polyostotic fibrous dysplasia: a case report. Rheumatol Int 2012; 32(4):1075-8
6. Park SK, Lee IS, Choi JY, Cho KH, Suh KJ, Lee WJ, at al. CT and MRI of fibrous dysplasia of the spine. Br J Radiol. 2012; 85(1015):996-1001
7. Faivre L, Nivelon-Chevalier A, Kottler ML, Robinet C, Khau van Kien P, Lorcerie B, et al. Mazabraud syndrome in two patients: clinical overlap with McCune-Albright syndrome. Am J Med Genet 2001; 99:132-6
8. Garg MK, Bhardwaj R, Gupta S, Mann N, Kharb S, Pandit A. Sarcomatous transformation (Leiomyosarcoma) in polyostotic fibrous dysplasia. Indian J Endocrinol Metab 2013;17(6):1120-3