Case Report

Pathological fracture dislocation of the acetabulum in a patient with neurofibromatosis-1

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Abstract

Skeletal neurofibromatosis (NF) commonly manifests as scoliosis and tibial dysplasias. NF affecting the pelvic girdle is extremely rare. Pathological fracture of the acetabulum leading to anterior hip dislocation in a patient with NF-1 has never been reported in the literature. The paper presents the clinical symptomatology, the course of management and the successful outcome of such a rare case of NF-1. Histopathological and immunohistochemistry studies showing abundant spindle cells, which are S-100 positive and of neural origin are the classical hallmarks of neurofibromatous lesions. Tumor resection and iliofemoral arthrodesis can be considered as a valid option in young patients with pathological fracture dislocation of the acetabulum.

Key words: Acetabulum, neurofibromatosis, pathological fracture
MeSH terms: Acetabulum, neurofibromatosis, fracture, bone, pathological

Introduction

Neurofibromatosis type-1 (NF-1) is a multisystem, autosomal dominant disorder of peripheral nerves affecting nearly 1/3000 individuals worldwide.[1] It was first described by a German pathologist, Friedrich Daniel von Recklinghausen. Inherited or spontaneous mutation of the neurofibromin gene located on chromosome 17 is responsible for this diverse disorder. Common skeletal manifestations of NF-1 include spinal deformities, congenital tibial dysplasia (congenital bowing and pseudarthrosis), sphenoidal dysplasia and cystic lesions in bones. Pathological fracture of the acetabulum with anterior dislocation of hip secondary to osseous involvement of the acetabulum, femoral head, and pubic rami has never been documented in a case of NF-1. Appropriate patient consent was obtained.

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Access this article online	
Quick Response Code:	Website: www.ijoonline.com
	DOI: 10.4103/0019-5413.173524

Case Report

A 16-year-old boy presented with the complaints of pain in the left hip associated with the inability to bear weight following a trivial fall. On examination, the affected limb was 1.5 cm short, abducted and externally rotated. Joint line was tender and attempted movements were painful. On general examination, patient had 8 café au lait spots over the body, bilateral axillary freckles and multiple palpable neurofibromas in the subcutaneous tissues of forearm, thighs and back [Figure 1]. Patient met three out of seven criteria described for the diagnosis of NF-1 [Table 1].[2] Plain radiograph and computed tomography scan of pelvis revealed an ill-defined lytic lesion causing pathological fracture - dislocation of the left hip [Figure 2]. Magnetic resonance imaging (MRI) showed additional soft tissue involvement and joint effusion [Figure 3]. MRI picture was in favor of a giant cell tumor. Ultrasound guided fine-needle aspiration cytology showed scanty cellularity with round to oval cells having minimal pleomorphism; hyperchromatic nucleus and moderate cytoplasm with spindle cells and osteoblasts. These features were suggestive of a sarcomatous lesion.

A wide local excision followed by arthrodesis of the joint was planned. Considering the extent of bony and soft tissue involvement, we used a modification of the ilioinguinal and iliofemoral approach to have a wide exposure. We used the conventional ilioinguinal incision and combined it with femoral part of the iliofemoral incision [Figure 4a]. On exposing the pelvis, anatomy was distorted. The deformed femur head and acetabulum with deficient pubic rami were visualized [Figure 4b]. There was extensive soft tissue

involvement adjacent to the acetabulum and lower part of the ilium. Femur osteotomy at the level of lesser trochanter was done. The entire acetabulum with 2-3 cm clear margin of the ilium was resected along with the abnormal soft tissue. Iliofemoral arthrodesis was done using a 14 hole stainless steel dynamic compression plate [Figures 4c-d, and 5].

Histopathology revealed dense collagenous tissue cores with spindle cells having blunt nuclei with minimal atypia and no mitosis or necrosis [Figure 6a]. On immunohistochemistry, cells were S-100 positive and of neural origin [Figure 6b]. These findings were consistent with neurofibroma. The postoperative period was uneventful. Partial weight bearing was allowed at 6 weeks and full weight bearing at 10 weeks. At 1-year followup, the patient was comfortable, pain free, able to ambulate unassisted, stand on one limb, sit and climb stairs without any difficulty [Figure 7].

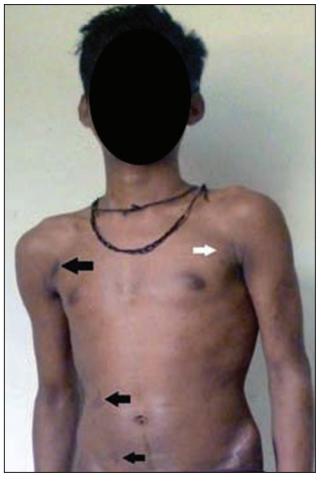

Figure 1: Clinical photograph showing skin lesions - café au lait spots (black arrows) and axillary freckling (white arrow)

Discussion

Type 1 neurofibromatosis or Von Recklinghausen disease, is a multisystem disorder that primarily affects the cell

Table 1: Criteria for diagnosis of NF-1 (at least 2 or more features)

Characteristic features
More than six cafe au lait spots, at least 15 mm in greatest diameter in adults and 5 mm in children
Two or more neurofibromas of any type or one plexiform neurofibroma
Freckling in the axillae or inguinal regions (Crowe sign)
Optic glioma
Two or more Lisch nodules (iris hamartomas)
A distinctive bone lesion, such as sphenoid dysplasia or thinning of the cortex of a long bone, with or without pseudarthrosis
A first-degree relative (parent, sibling or offspring) with NF-1 by the above criteria

NF=Neurofibromatosis

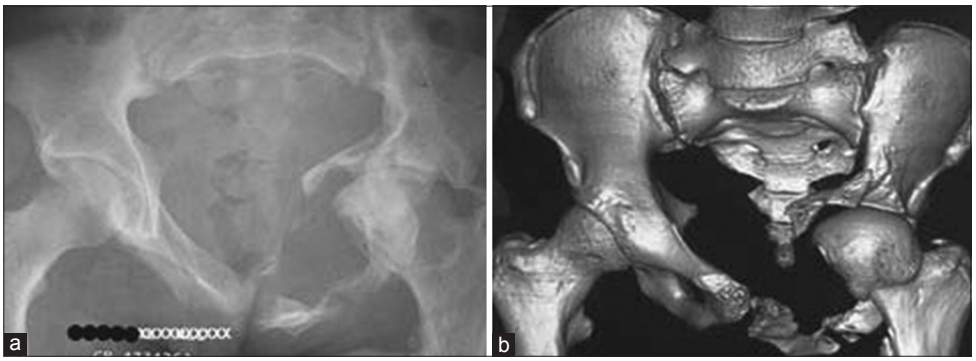

Figure 2: Preoperative radiograph anteroposterior view (a) and computed tomography scan (b) of pelvis showing an ill-defined lytic lesion destroying anterior column of acetabulum, pubic rami and part of the femoral head

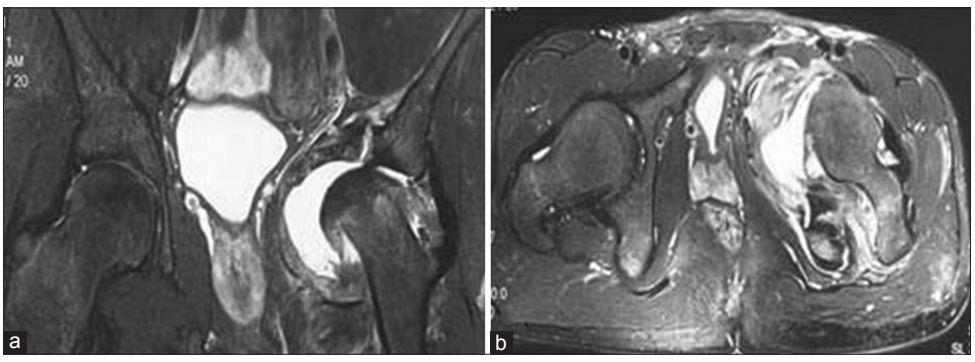

Figure 3: Coronal (a) and axial (b) sections of magnetic resonance imaging of pelvis showing expansile lytic lesion of acetabulum and pubic rami with soft tissue involvement and joint effusion

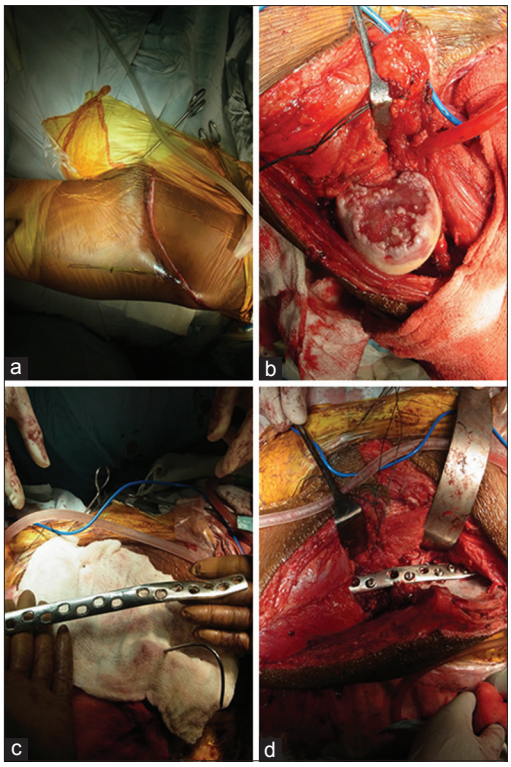

Figure 4: Intraoperative photographs showing (a) Skin incision (b) deformed femoral head (c) 14 hole DCP (d) iliofemoral arthrodesis

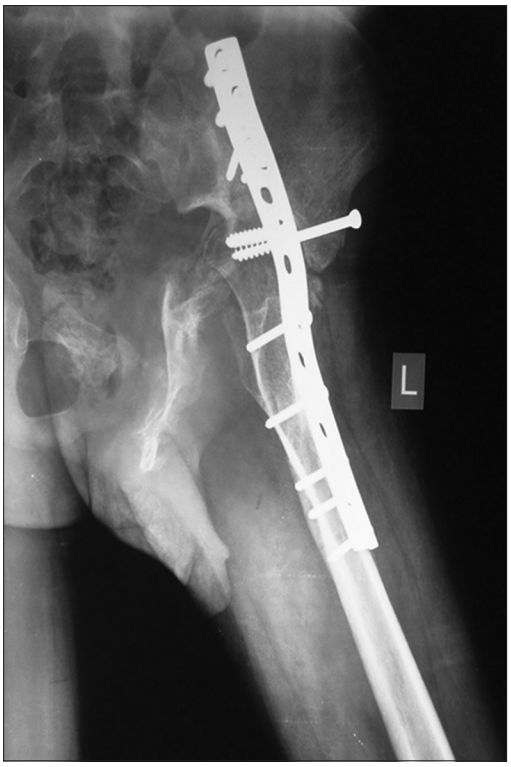

Figure 5: Postoperative radiograph after tumor resection and iliofemoral arthrodesis

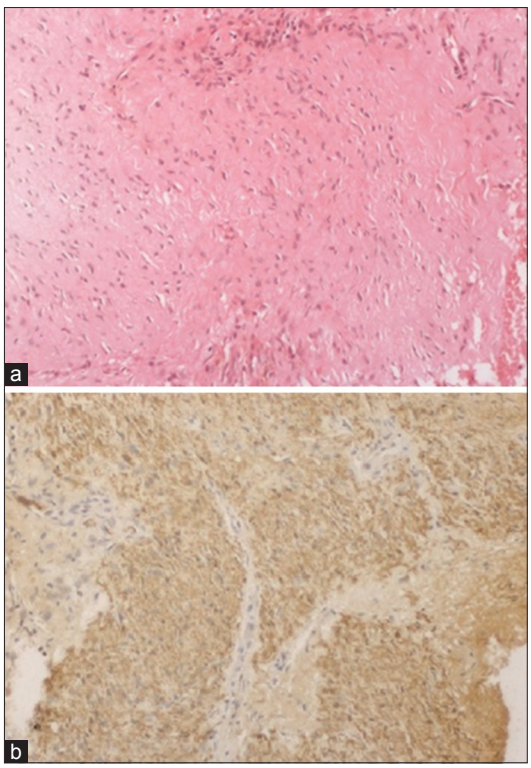

Figure 6: (a) Histopathological photomicrograph showing dense collagenous tissue cores and spindle cells with blunt nuclei. (b) Immunohistochemistry showing neural marker S-100 positivity

growth of neural tissue and characterized by involvement of skin, peripheral nerves, subcutaneous tissue, eyes, and skeletal system. The orthopedic manifestations of NF-1 are listed in Table 2. Although involvement of the musculoskeletal system is common, there have been only a few cases of subluxation/dislocation of hip in patients with NF [Table 3].[3-14] On reviewing the literature, the etiology of hip instability leading to pathological subluxation/dislocation in patients with NF-1 can be classified as local and remote. Most of the cases are secondary to

Table 2: Orthopedic manifestations of NF-1

Salient features
Common
Kyphoscoliosis
Congenital bowing/pseudoarthrosis of tibia/ulna
Unilateral segmental limb hypertrophy or local gigantism
Plexiform neurofibromas
Neurogenic tumors and soft tissue sarcomas
Metabolic bone disease (osteopenia)
Uncommon
Short stature
Spondylolisthesis
Bone cysts in the pelvis and tarsal bones
Sub-periosteal bone proliferation
Defects and thinning of bones
Atlantoaxial dislocation
Hypoplasia of ilium
Charcot's neuropathic arthropathy
Tendon ruptures
Pathological hip subluxation/dislocation

NF=Neurofibromatosis

Figure 7: Clinical photographs showing functional outcome at 1-year followup

local (intra and peri-articular) neurofibromas, which can result in mass effect, bony erosions (ilium, acetabulum, and femoral neck), acetabular dysplasia, narrowing of the femoral neck, coxa valga, increased femoral neck offset, capsuloligamentous laxity, and synovial membrane proliferation.[3-5,8,10,11,13] Remote causes of hip instability include intra spinal neurofibromas/schwannomas leading to motor deficit (hip abductor weakness) or sensory deficit (charcot's neuropathic arthropathy), limb length discrepancies secondary to hemi-hypertrophy of lower limb and abnormal biomechanical alteration in the spinopelvic alignment due to scoliosis.[6,7,9-11] Endo *et al*. described anterior subluxation of hip secondary to decreased femoral head coverage resulting from decreased lumbosacral lordosis and posterior pelvic inclination following scoliosis correction.[11] Until date, there has been no case of NF-1 reported in the literature with pathological fracture of the acetabulum with anterior dislocation of hip attributable to a neurofibroma involving the acetabulum, pubic rami and femoral head.

The various treatment options described for pathological hip dislocations in NF-1 include closed reduction, open reduction, shelf operation with fascia lata tenorraphy, rotational acetabular osteotomy with femoral varus osteotomy, girdle stone resection, total hip replacement with

Table 3: Comprehensive literature review on published cases of hip dislocation/subluxation in NF-1

Study, year	Age/sex	Type of lesion	Direction of dislocation/ subluxation	Initial Rx	Number of redislocations	Subsequent Rx	Followup	Final outcome
Lachiewicz *et al.*,[5] 1983	37 years/ female	Intraarticular NF	Posterosuperior	CR	1	OR+hip spica	1 year	Painless, stable, mobile hip
Phillips and McMaster,[6] 1987	18 years/ female	Local extra articular NF	Superior	No Rx	-	-	-	Painless, limited abduction
Haga *et al.*,[10] 1994	8 years/ male	Intraarticular NF	Superolateral	No Rx	-	-	6 years	Died at 14 years
Haga *et al.*,[10] 1994	2 years/ female	Intraarticular NF	Superolateral	No Rx	-	-	2 years	Had contralateral hip D/L at 4 years
Odent *et al.*,[7] 2004	26 years/ female	neuropathic (abductor weakness)	Posterolateral	CR	4	OR+shelf procedure followed by THR followed by cup revision	11.5 years	Stable hip
Endo *et al.*,[11] 2007	30 years/ female	Neuropathic+ local NF	Anterior	OR+osteotomy (acetabulum in+femoral)	-	-	5 years	Stable, mobile hip
Lampasi *et al.*,[9] 2008	28 years/ male	Neuropathic (spinal NF)	Posterosuperior	Girdle stone arthroplasty	-	-	3 years	Pain free and walking unaided
Galbraith *et al.*,[3] 2011	18 years/ female	Local extra articular NF	Superior	CR+skeletal traction	1	CR	12 years	Walking unaided
Tangsataporn *et al.*,[14] 2012	39 years/ male	Local extra articular NF	Lateral	THR	-	-	1 years	Painless, stable, mobile hip
Current study, 2014	16 years/ male	Intra+extra articular NF	Anterior	OR+arthrodesis	-	-	1 years	Walking unaided, painless, stable hip

CR=Closed reduction , OR: Open reduction, THR=Total hip replacement, NF=Neurofibromatosis

the trochanteric distalisation.[7,9,11,14] The rate of re dislocation is very high in most of the cases, subsequently requiring a secondary surgical procedure for stabilization. Since only a handful of cases have been described in the literature, it is difficult to comment upon the best line of management. In our case, arthrodesis was the best possible option since the bone stock after tumor resection was so inadequate that none of the above mentioned procedures could be tried. Moreover, the fear of redislocation, which might necessitate repeated surgeries, was negated. The age of the patient also favored arthrodesis. The surgical approach was modified since there was extensive bony and soft tissue involvement. A combination of ilioinguinal and iliofemoral approach was employed to have a better exposure of the hip joint.

The neurofibromatous tumors associated with NF-1 are usually benign; however, there is a 2-5% chance of malignant transformation, especially with plexiform neurofibromas.[15] Plexiform neurofibromas are diffuse, poorly defined nerve sheath tumors arising from multiple nerve fascicles and surrounding tissues. They are more prone for hemorrhage, dysfunction, pain, disfigurement, and malignant transformation.[16] There was no clinical or radiological evidence of recurrence or malignant transformation in our patient at 1-year followup. Overall, the patient had a good clinical and functional outcome.

This case sheds light on the unusual manifestation of a familiar genetic disorder. The importance of general systemic examination cannot be underestimated. Subtle clinical signs such as skin patches, axillary freckling, and subcutaneous neurofibromas can be easily missed. A thorough clinicoradiological evaluation and accurate histopathological examination helps in clinching the diagnosis. Orthopedic surgeons must be aware about the various management options available and tailor them as per the needs of their patient. Iliofemoral arthrodesis offered a good functional outcome with improved quality of life in our case.

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How to cite this article: Saibaba B, Sen RK, Sharma M, Nahar U. Pathological fracture dislocation of the acetabulum in a patient with neurofibromatosis-1. Indian J Orthop 2016;50:103-7.

Source of Support: Nil, **Conflict of Interest:** None.