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. 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 pseudoarthrosis), 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.

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]. 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].

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

![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](image)

![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](image)
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].

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 growth of neural tissue and characterized by involvement of skin, peripheral nerves, subcutaneous tissue, eyes, and skeletal system.
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. 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. 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. 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

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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., 1983 | 37 years / female | Intraarticular NF | Posterolateral | CR | 1 | OR+hip spica | 1 year | Painless, stable, mobile hip |
| Phillips and McMaster, 1987 | 18 years / female | Local extra articular NF | Superior | No Rx | - | - | - | Painless, limited abduction |
| Haga et al., 1994 | 8 years / male | Intraarticular NF | Superolateral | No Rx | - | - | 6 years | Died at 14 years |
| Haga et al., 1994 | 2 years / female | Intraarticular NF | Superolateral | No Rx | - | - | 2 years | Had contralateral hip D/L at 4 years |
| Odent et al., 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., 2007 | 30 years / female | Neuropathic+ local NF | Anterior | OR+osteotomy (acetabulum in+femoral) | - | - | 5 years | Stable, mobile hip |
| Lampasi et al., 2008 | 28 years / male | Neuropathic (spinal NF) | Posterolateral | Girdle stone arthroplasty | - | - | 3 years | Pain free and walking unaided |
| Galbraith et al., 2011 | 18 years / female | Local extra articular NF | Superior | CR+skeletal traction | 1 | CR | 12 years | Walking unaided |
| Tangsataporn et al., 2012 | 39 years / male | Local extra articular NF | Lateral | THR | - | - | 1 year | Painless, stable, mobile hip |
| Current study, 2014 | 16 years / male | Intra+extra articular NF | Anterior | OR+arthrodesis | - | - | 1 year | Walking unaided, painless, stable hip |

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

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Figure 7: Clinical photographs showing functional outcome at 1-year followup
the trochanteric distalisation.\textsuperscript{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.\textsuperscript{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.\textsuperscript{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.

\textbf{References}

1. Fienman NL, Yakovac WC. Neurofibromatosis in childhood. J Pediatr 1970;76:339-46.
2. National Institutes of Health Consensus Development Conference Statement: Neurofibromatosis. Bethesda, Md, USA, July 13-15, 1987. Neurofibromatosis 1988;1:172.
3. Galbraith JG, Butler JS, Harty JA. Recurrent spontaneous hip dislocation in a patient with neurofibromatosis type 1: A case report. J Med Case Rep 2011;5:106.
4. Nakasone S, Norimatsu H, Hamasaki N, Kinjo S, Kinjo Y, Ibaraki K, \textit{et al}. A case report of recurrent dislocation of the hip joint with neurofibromatosis. Orthop Surg Traumatol 1989;38:511-14.
5. Lachiewicz PF, Salvati EA, Hely D, Ghelman B. Pathological dislocation of the hip in neurofibromatosis. A case report. J Bone Joint Surg Am 1983;454:414-5.
6. Phillips JE, McMaster MJ. Pathological dislocation of the hip in neurofibromatosis. J R Coll Surg Edinb 1987;32:180-2.
7. Odent T, Ranger P, Aarabi M, Hamdy RC, Fassier F. Total hip arthroplasty in a patient with neurofibromatosis type 1 and recurrent spontaneous hip dislocation. Can J Surg 2004;47:219-20.
8. Guilleminet M, Creyssel J, de Mourgues G, Fischer L. Von Recklinghausen’s neurofibromatosis. Congenital hypertrophy of the lower limb in childhood and spontaneous luxation of the homolateral hip in adult age. Presse Med 1970;78:1269-71.
9. Lampasi M, Greggi T, Sudanese A. Pathological dislocation of the hip in neurofibromatosis: A case report. Chir Organi Mov 2008;91:163-6.
10. Haga N, Nakamura S, Taniguchi K, Iwaya T. Pathologic dislocation of the hip in von Recklinghausen’s disease: A report of two cases. J Pediatr Orthop 1994;14:674-6.
11. Endo H, Mitani S, Sugihara S, Kuroda T, Nakahara S, Ozaki T. Nontraumatic subluxation of the hip after spine surgery for scoliosis in a patient with von Recklinghausen’s disease. J Orthop Sci 2007;12:510-4.
12. Lucet L, Elayoubi L, Defives T, Mejjad O, Le Loet X, Cambon-Michot C, \textit{et al}. Anterior pathologic dislocation of the hip in adulthood complicating Von Recklinghausen neurofibromatosis. Rev Rhum Ed Fr 1993;60:79-80.
13. Kuroda M, Nakase H, Yasui N, Ochi T, Takahashi Y, Hirabayashi S. Nontraumatic dislocation of the hip in von Recklinghausen’s disease: A case report. Clin Orthop Surg 1999;1151:1151-4.
14. Tangsataporn S, Shakib A, Kuzyk PR, Backstein DJ, Gross AE, Safir OA. Secondary Hip Osteoarthritis due to Neurofibroma Treated with Total Hip Replacement. Case Rep Orthop 2012;2012:173921.
15. Hope DG, Mulvihill JJ. Malignancy in neurofibromatosis. Adv Neurol 1981;29:33-56.
16. Feldman DS, Jordan C, Fonseca L. Orthopaedic manifestations of neurofibromatosis type 1. J Am Acad Orthop Surg 2010;18:346-57.

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