

CASE REPORT

Intra-articular hip neurofibromas in von Recklinghausen's disease

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Abstract

Neurofibromatosis is a genetic disorder with osteo-articular manifestations. The intra-articular location of neurofibroma is scarce.

KEYWORDS

hip, intra-articular neurofibroma, neurofibromatosis

1 | INTRODUCTION

Neurofibromatosis type I (NF 1) is a disease of neural crest origin, affecting ectodermal and mesodermal tissues. It is a common autosomal dominant disorder known to have associated skeletal manifestations. The most involved bones are the sphenoid wing, vertebrae, tibia, and fibula. However, intra-articular neurofibroma is a rare presentation in NF 1. We herein report a case of a 24-year-old woman with known NF 1 complaining of paroxysmal right hip pain the last 2 years. Pelvic computed tomography scan revealed iliac bone scalloping and intra-articular neurofibromas. This case illustrates a rare complication in NF 1 consisting of hip arthritis without subluxation or dislocation.

Neurofibromatosis type I (NF 1), also named von Recklinghausen's disease, is a genetic disorder caused by a mutation of the NF 1 gene.¹ NF 1 can result either from an autosomal dominant lineage or from a spontaneous

NF 1 gene mutation. The most frequent characteristics include café au-lait macules, axillary freckling, iris Lisch nodules, multiple neurofibromas, and bone defects.²

Neurofibromas cause accelerated bone and soft tissue growth.³ These lead to several skeletal manifestations considered as 'dysplasias'.² Preferentially, it appears within the long bone and vertebrae. The reasons behind such a striking predisposition are unclear. Intra-articular neurofibroma is an exceedingly rare location in NF 1, and only a few cases had been reported.⁴⁻⁹ In this article, we report an exceptional case with hip joint and iliac bone involvement. Written informed consent was obtained from the patient for the publication of this case report.

2 | CASE DESCRIPTION

A 24-year-old female has complained of mechanical and paroxysmal pain in the right hip for 24 months. She had a

known familial history of NF 1. Her father had a clinical feature of NF 1.

She has a Trendelenburg gait with a leg discrepancy.

Physical examination revealed café-au-lait spots (Figure 1). The right hip was limited, especially in abduction motion.

Neurological examination was within normal limits. Laboratory tests were slightly normal.

The groin X-ray showed scalloping of the right iliac wing and the great trochanter (Figure 2).

Pelvis computed tomography scan (CT scan) pointed out a coxo-femoral space narrowing associated with neurofibromas in the coxo-femoral joint (Figure 3).

We referred our patient to surgery to excise the intra-articular neurofibromas.

3 | DISCUSSION

Neurofibromatosis type I was first described by the German pathologist Frederick von Recklinghausen in 1882.¹⁰ It represents one of the most common autosomal dominant disorders, occurring in 1 in 3000 birth worldwide.^{11,12} NF 1 is secondary to hamartomatous cellular abnormalities affecting ectodermal and mesodermal tissues. The neural crest is a transitory embryonic structure whose cells form neuronal neural supportive, pigmentary, endocrine, and other tissues.

In classical NF 1, skeletal defects result from abnormalities of the neural ectoderm and mesoderm.¹³ The characteristic sites of NF 1-associated osseous manifestations are the long bones (usually the tibia and the fibula), vertebrae,



FIGURE 1 Clinical exam detecting multiple café-au-lait spots

and sphenoid wing. While focal bony lesions may cause profound clinical consequences, a minority of people with NF 1 are symptomatic.¹⁴ Scoliosis, congenital tibia pseudarthrosis, metabolic bone disorders, and disturbed growth patterns are all known orthopedic manifestations of NF 1.¹⁴

We herein report a rare case of intra-articular neurofibromas revealed by hip pain and limping in a 24-year-old woman with NF 1. Although several associations between NF 1 and the hip location, intra-articular neurofibromas were a rare occurrence. These associations were due to intraosseous cystic lesions, periosteal bone proliferation, coxa valga, and increased femoral offset. Only a few described cases had a dislocation hip with or without intra-articular neurofibromas.^{5,7,8,12,15–22}



FIGURE 2 Pelvic radiographic showing a scalloping in the right great trochanter and the ischial branch (white arrow)



FIGURE 3 Sagittal CT revealing the intra-articular neurofibromas (white arrow) and the scalloping in the great trochanter

A joint penetration of extra-articular neurofibromas provoked secondary arthritis, bony erosion, and even deformity.^{6,17,19}

The computed tomography tool was helpful in bone dysplasia demonstration. However, pelvic magnetic resonance imaging (MRI) would have been more accurate in assessing neurofibroma extension and eliminating malign transformation.

Treatment may be conservative or surgical. For the current case, we preferred the second alternative to avoid joint dislocation. Obviously, the intra-articular growth of neurofibromas may distant the joint capsule and lead to hip dislocation.

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None.

CONFLICT OF INTEREST

None declared.

AUTHOR CONTRIBUTIONS

HF, MM, and KM analyzed and interpreted the patient data. DB and TW were major contributors in writing the manuscript, and all the authors read, revised, and approved the final manuscript.

CONSENT

Written informed consent was obtained from the patient for the publication of this case report.

DATA AVAILABILITY STATEMENT

The data that support the findings of this study are available from the corresponding author upon reasonable request.

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