

peer reviewed CASE REPORT

## Neurofibroma of the tibia and fibula: a case report

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### Abstract

This case report describes and illustrates neurofibromatosis Type 1 with a giant neurofibroma of the tibia and fibula, diagnosed in a patient in his late 20s. The clinical history, investigations, results, etiology, epidemiology, radiographic appearance and treatment of this case are also discussed.

### Keywords

Von Recklinghausen's Disease, rounded nodules, café au lait, tumours, autosomal dominant.

### Case report

The patient in this case report had fractured his left tibia as a child and had undergone successful surgical repair at a tertiary hospital. Three years prior to the current examination he had been seen at an academic hospital due to a dog bite on his left calf which had become swollen. On examination he was well, yet walked with a slight limp. His left calf and ankle were extensively swollen, painful and hard on palpitation. There was a dark discolouration/pigmentation extending from the posterior portion of his left calf down to the ankle.

He was referred to a radiology department for plain film x-rays of his left tibia and fibula. The anteroposterior and lateral plain film radiographs demonstrated a marked deformity in both bones. There was degenerative and extensive changes in the ankle joint as well as severe soft tissue swelling (Figure 1). The main lesion was located in the mid-shaft of the tibial metaphysis. The lesion had an expansile and well corticated 'benign looking' appearance. No pathological fracture or abnormal soft tissue calcifications were noted (Figure 2). Features of this lesion were in keeping with unusual osseous involvement of the tibia by neurofibromatosis Type 1. A histology test was ordered as a sarcoma was suspected. The histology results confirmed a diagnosis of neurofibromatosis Type 1, with a giant neurofibroma in the left calf.

### Discussion

Neurofibromatosis Type 1 (NF1), formerly known as Von Recklinghausen's Disease,

is one of the most common inherited disorders affecting 1 in 3000-4000 individuals. A neurofibroma is a benign tumour that arises within the nerve sheath of Schwann cells. Specific sites of origin may occur as single or multiple neurofibromata that arise in the same patient. These tumours produce rounded nodules, giving a loculated appearance within both superficial and deep nerves. This neoplasm is associated with areas of skin pigmentations, referred to as café au lait patches. In some sites the tumours may be associated with a mixture of bone destruction as well as reactive new bone formation as a result of pressure on neighbouring bony structures<sup>[1]</sup>.

Typical features of NF1 include: neurofibroma tumours, pigmented café-au-lait patches on the skin, osseous malformations (as can be seen in this case), learning disorders and a predilection to selected malignancies, such as malignant peripheral nerve sheath tumours, for example optic gliomas and meningiomas<sup>[2]</sup>.

Two or more of the following criteria are required for diagnosis of NF1: six or more café-au-lait spots over 5 mm in pre-pubertal individuals and over 15 mm in post-pubertal individuals, two or more neurofibromas

of any type or one plexiform neurofibroma, freckling in the axilla or groin, optic glioma, two or more Lisch nodules, presence of a distinct osseous lesion, sphenoid wing dysplasia or thinning of a long bone with or without pseudoarthrosis, and lastly a first degree relative who meets the above criteria for NF1<sup>[3]</sup>. NF1 is one of the most common single gene syndromes and is characterized by a range of distinguishing, but variable, manifestations such as orthopedic manifestations of congenital pseudoarthrosis (non-union) of the tibia, scoliosis as well as other skeletal defects including sphenoid wing dysplasia and rib pencilling, have been



Figure 1: Anteroposterior view of the left lower leg showing main lesion involving tibia and fibula (thin arrow). Also note thinning of the fibula (thick arrow).



**Figure 2:** Lateral view of the tibia and fibula showing expansile nature of lesion (see arrows). The ankle joint is also markedly deformed.

predominantly associated with NF1. Approximately half of the patients with NF1 have some form of quantifiable skeletal abnormality<sup>[4]</sup>.

Being an autosomal dominant disorder, it has an almost even division between spontaneous and inherited mutations. Penetrance of NF1, in other words the regularity or frequency in which the gene produces its effect, approaches 100% by age 20 and if the patient has the mutation then certain manifestations will be exhibited, although expressivity is highly inconsistent, even among family members with the same mutation. An individual with mild clinical findings may have a child with a more severe phenotype, or vice versa<sup>[5]</sup>.

The patient in this case report only suffered dermatologic and orthopedic manifestations with café-au-lait spots, tibial bowing as well as thinning of the bones of the lower leg. Many of the clinical manifestations of NF1 are dependent upon the patient's age. Café-au-lait spots, plexiform neurofibromas, and tibial dysplasias are typically recognized within the first year of life, whereas optic pathway gliomas and axillary freckling may not be apparent until 3-5 years of age<sup>[6]</sup>.

NF1 occurs in all ethnic and racial groups and affects both genders equally. Morbidity, due to complications of NF1, occurs mainly in adults but occasionally in children. In addition to neurofibromas, individuals affected with NF1 may exhibit a multitude of other clinical manifesta-

tions that affect different organ systems, with neurological, dermatologic, ophthalmologic, and orthopedic impairments being the most common<sup>[3]</sup>.

Adults with NF1 should be monitored for neurological complications resulting from neurofibroma growth, as well as for signs of malignant transformation of a plexiform neurofibroma. The patient's age should be considered, as an appreciation of the age-dependent nature of many of the features of NF1, will aid the physician in proper management and treatment of patients with NF1. Cortical thinning of the long bones or dysplastic

bony changes can lead to repeated pathological fractures with incomplete healing, and may result in the appearance of a false joint, or pseudoarthrosis<sup>[6]</sup>. The patient described in this case report had cortical thinning and dysplastic bony changes but no pathological fractures or pseudoarthrosis.

A dermatologist has a primary role in recognizing and differentiating NF1 from other conditions based on a careful skin examination, making appropriate referrals once the diagnosis is made, and managing symptomatic or disfiguring cutaneous neurofibromas. Currently, neurofibromas are only subjected to surgical removal. Symptomatic (i.e. painful and bleeding) neurofibromas are most commonly removed but, depending on the community, this may not be handled by dermatology<sup>[4]</sup>.

Shortly before writing up of this case report, the patient had undergone an above knee amputation (AKA) of the left tibia. The patient was successfully fitted with a prosthesis and was discharged once well enough to return home.

### Conclusion

NF1 may manifest with a variety of signs and symptoms which, although specific to NF1, may be experienced with other disorders as well. This case report further highlights the important role plain film imaging has in the diagnosis and management of patients, given the vast technological advances currently taking place

within radiography. Plain film imaging is thought to remain the mainstay for diagnosis of skeletal abnormalities even more so in the primary and rural health care setting.

### References

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