

NEUROFIBROMATOSIS OF AN ISOLATED FOOT: A CASE REPORT

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ABSTRACT

A neurofibroma is a benign nerve sheath tumor usually involved with the peripheral nervous system. Neurofibromas are commonly associated with neurofibromatosis type 1, including being an inclusion criteria, but are not always associated with the disease. Neurofibromas are uncommon in the foot and ankle and account for less than 10% of all soft tissue tumors of the foot and ankle.^[1] Peripheral nerve tumors are most common in the face, neck, and flexor surfaces. Neurofibromas are characterized by a combination of cells of the nerve sheath including: Schwann cells, peri-neural cells, and fibroblasts.^[2] We report herein a rare case of a neurofibroma present in the left foot and ankle region of a 20 year old male.

KEYWORDS: Neurofibroma, Schwann cells, neurofibromatosis.

INTRODUCTION

Neurofibromas are among the most common and debilitating complications of neurofibromatosis type 1 (NF1). They account for substantial morbidity, including disfigurement, functional impairment, and may even be life threatening. Plexiform neurofibromas are also subject to transformation into malignant peripheral nerve sheath tumor. The current mainstay of treatment of plexiform neurofibromas and of malignant peripheral nerve sheath tumors is surgical resection.^[3] Neurofibromas arise from a combination of neural cells including Schwann cells, peri-neural cells and fibroblasts. Neurofibromas often present painless but can cause debilitating pain and motor sensory dysfunction. Neurofibromatosis I, also known as Von Recklinghausen disease, is an autosomal dominant condition which is clinically characterized in part by pigmented skin lesions known as café-au-lait spots, benign cutaneous and subcutaneous tumors known as neurofibromas, distinctive bone lesions, and focal malformations of the iris. It is the most common single gene disorder in humans and results from the defective protein neurofibromin, which is thought to act as a tumor suppressor.^[4] Neurofibromas are most commonly associated with NF1.

CASE REPORT

A 20 year old male presented to open patient door (OPD) with complaint of swelling, pain and disfigurement of left foot and ankle since 15 years of age (Figure 1,2). The swelling was smaller in size initially which progressed gradually. The pain was fluctuating in severity, did not correlate with specific activities, and was only partially

relieved with medication. On local examination, there was multilobulated swelling over dorsum of foot and ankle region. The skin over the swelling was intact but had mild protrusion, as the mass was fixed to the deep tissue. It did not pulsate or fluctuate. There was no bruit on auscultation. This lesion was initially painless, but gradually become painful and tender. The patient also experienced persistent numbness over the swelling in the later 6 months. Other physical examinations revealed mild weakness of the left gastrocnemius and decreased Achilles reflex, with normal strength of the right tibialis anterior, extensor hallucis longus and extensor digitorum longus. The patient had no past history of malignancy or family history of neurofibromatosis. There was no previous trauma, surgical procedure or skin disease at the site. Ultrasonography revealed an ill-defined, solid and hypoechoic mass (7.5 cm × 6.8 cm × 10.1 cm). No blood flow was noted in the lesion during color doppler survey. Magnetic resonance imaging demonstrated a well defined multilobulated soft tissue mass lesion approximate size 7.5 cm × 6.8 cm × 10.1 cm in anterolateral aspect of left foot and ankle which is isointense on T1 and heterogeneously hyperintense on T2/PD images showing extensive post contrast enhancements and on the medial aspect there is a fibrotic area (T1 and T2 hypointense) of approx. size 2.2 cm × 2.4 cm × 4.7cm and fibrotic bands within the lesion causing displacement and encasement of tibialis anterior, extensor hallucis longus and extensor digitorum longus and abutting the cortex of talus, navicular and medial cuneiform and shows extensive post contrast enhancement. Diagnosis of neurofibromatosis was confirmed by histopathological analysis. Patient was

counseled about surgical management of deformity and referred to tertiary care centre.



Figure 1: Multilobulated swelling over anterior aspect of foot and ankle region.



Figure 2: Multilobulated swelling over anterior aspect of foot and ankle region.

DISCUSSION

Neurofibromatosis is a genetic disorder that primarily affects cell growth of neural tissue. The prevalence ranges from 1 in 2,500 to 1 in 7,800 in the population.^[5-6] It can cause tumors to grow on the nerves at any location and at any time.^[7] Neurofibromatosis displays wide variability in clinical presentation. There are at least two distinctive forms: NF-1 and NF-2. NF-1 shows no predilection for race or sex.^[8] Inheritance is autosomal dominant, although 50% of patients do not have a family history and correspond to sporadic mutations. Foot schwannoma is a rare benign tumor. This tumor is reported throughout the body, with the incidence for pedal involvement shown to be 2.93%, based on a literature reviewed.^[9] There have also been reports of multiple schwannomas without other stigma indicating NF 1 or NF 2.^[10] This type of disease did not match the characteristics of NF 1, in which there are such as multiple neural tumors dispersed anywhere on or in the body, café au lait spots, and Lisch nodules as seen in index patient.

During the physical and radiographic examination no bony changes were observed in the present case. Bone involvement is common in patients with neurofibromatosis (51-71% of cases).^[11] The treatment of neurofibromatosis is little more than symptomatic because the disease is progressive. There is no specific treatment.^[12] Therapy is designed to prevent or manage complications. According to the literature, clinical follow-up should be conducted and surgery performed when the lesions compromise the patient's function or cosmetic appearance.^[12-13] The risks, possible complications and benefits of a surgical strategy must be considered before intervening. Regardless of whether or not previous surgical procedures have been performed malignization affects 5% of cases. When malignization occurs, the prognosis is unfavorable.^[11] The patients with neurofibromatosis must always be followed up clinically to evaluate the course of the disease.

CONCLUSION

Neurofibromas can be a harmful soft tissue mass in the body. Although neurofibromas are benign they do have a small incidence to transform into malignancy in the body. Neurofibromas can also cause crippling pain and affect the body's function. Early diagnosis can help prevent nerve damage or deformity.

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