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Magnetic resonance imaging of elephantiasis neuromatosa

Abstract We present the case of a 43-year-old man with neurofibromatosis type 1 who developed elephantiasis neuromatosa of his left leg. The gross limb enlargement was extremely disfiguring, and resulted in such severe disability that he was only able to walk a very short distance using crutches. Previous debulking procedures had resulted in massive blood loss, and prior to attempting further surgical intervention MRI studies were requested. Taking advantage of the excellent tissue characterisation and multiplanar imaging capabilities of MRI, we were able to assess the extent of soft tissue and osseous involvement. The use of recently developed MR angiographic sequences enabled us to non-invasively provide detailed images to assess the relationship of the lesions to the major vessels, as well as the vascular supply and angiographic features of the lesions themselves. This article describes our MRI-based findings, which precluded debulking surgery in this unusual manifestation of neurofibromatosis.

Key words Neurofibromatosis type 1 · Plexiform neurofibroma · Elephantiasis neuromatosa · MRI · MR angiography

Introduction

Neurofibromatosis type 1, or von Recklinghausen disease, is an autosomal dominant condition characterised by cutaneous lesions, skeletal abnormalities, and multiple nerve sheath tumours [1–4]. The peripheral nervous system is selectively involved with relative sparing of the visceral and central nervous system. Peripheral nerve tumours in neurofibromatosis 1 involve neural supporting tissues, with the two most common benign forms being neurofibromas and schwannomas, which may be either solitary or multiple. Cutaneous neurofibromas develop as well-circumscribed, sessile or pedunculated tumours. Benign plexiform neurofibromata are a hallmark of neurofibromatosis 1, and result from tumour-like overgrowth of fibromyxoid tissue from the nerve sheath. Plexiform neurofibromata can occasionally lead to gigantism, with massive hypertrophy of the skin, soft tissues and underlying skeleton of an extremity, known as elephantiasis neuromatosa.

We present the case of a 43-year-old man with neurofibromatosis type 1 who presented with gross elephantiasis neuromatosa of his left leg. This was severely disabling and he was only able to walk short distances with crutches. Previous surgical attempts to debulk the lesion had resulted in massive blood loss. Prior to further surgery we undertook MRI scanning, including dynamic contrast-enhanced magnetic resonance angiographic sequences (MRA). This enabled us to evaluate the soft tissue and osseous abnormalities, as well as providing detailed assessment of vascular involvement.

Case report

A 43-year-old man with neurofibromatosis type 1 presented to the plastic surgeons in 1997 for debulking of a large plexiform neurofibroma involving his left leg. This had been present for many years, but was gradually increasing in size. It had reached such vast proportions that he was unable to continue work as a window-cleaner (Fig. 1).
A debulking procedure in 1973 had resulted in marked blood loss. A further debulking procedure was attempted in 1993, and at this time the neurofibromatous tissue was found to be extensively infiltrating quadriceps and the adductor muscles, precluding excision. A large mass of tissue was removed, with more than 3 l blood-loss. Histology of the excised specimen showed neurofibromatous tissue with no evidence of malignancy.

In 1997 he represented, severely disabled by the further increase in size of his left leg. On examination he had extensive cutaneous neurofibromata over his trunk with café au lait spots. The left leg was grossly enlarged, particularly in the thigh. The skin hung down in loose folds over the knee itself. Large dilated superficial veins were seen in the thigh. Radiographs of his pelvis, femurs and knees demonstrated hyperplasia of the left femur, with an irregular bone contour and disorganised trabecular pattern (Fig. ). There was mild medial subluxation of the left tibia on the femur, and marked lateral dislocation of the patella.

Surgical exploration was again undertaken. Despite the removal of more than 1 kg of tissue, this only represented a small fraction of the lesion, and further resection was not possible without risking life-threatening bleeding. Histology again confirmed neurofibromatous tissue with no evidence of malignancy.

In view of the risks of major haemorrhage, prior to further attempts at surgical debulking, an MRI study coupled with MR angiography, was requested to ascertain the extent of soft tissue, osseous and vascular involvement. T1, T2 and STIR images were obtained in coronal and axial planes. Dynamic