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Case Report

Giant pedunculated neurofibroma of the thigh with apical ulceration in a patient with neurofibromatosis type 1: a case report and review of operative management

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ABSTRACT

Neurofibromatosis type 1 (NF1) is an autosomal dominant neurocutaneous disorder arising from mutations in the NF1 tumour-suppressor gene on chromosome 17q11.2. It is characterised by café-au-lait macules, axillary freckling, Lisch nodules, and the development of multiple cutaneous and plexiform neurofibromas. Giant solitary pedunculated neurofibromas are a rare and functionally disabling manifestation, particularly when complicated by secondary ulceration, which carries a risk of infection and raises concern regarding malignant transformation. A 56-year-old woman with NF1 presented to the outpatient surgical clinic with a large, pedunculated soft-tissue mass over the upper lateral left thigh that had progressively enlarged over many years and had recently developed an ulcer at its apex with intermittent serous discharge. Following thorough pre-operative workup-including magnetic resonance imaging (MRI) of the thigh, ultrasound-guided fine-needle aspiration cytology (FNAC), and cardiorespiratory assessment-the patient underwent elective surgical excision under general anaesthesia. Histopathological examination confirmed a benign peripheral nerve sheath tumour consistent with neurofibroma. Surgical excision remains the definitive treatment for large, symptomatic or ulcerated neurofibromas. Careful pre-operative imaging and meticulous dissection are essential to achieve complete resection whilst avoiding neurovascular injury. Lifelong multidisciplinary surveillance is mandatory given the systemic nature of NF1 and the risk of malignant peripheral nerve sheath tumour (MPNST) transformation.

Keywords: Neurofibromatosis type 1, NF1, Neurofibroma, Peripheral nerve sheath tumour, Surgical excision, Soft-tissue tumour, Thigh

INTRODUCTION

Neurofibromatosis type 1 (NF1), historically termed von Recklinghausen's disease after the German pathologist Friedrich Daniel von Recklinghausen who first delineated the condition as a single nosological entity in 1882, is one of the most prevalent hereditary tumour-predisposition syndromes in humans, affecting approximately 1 in 2500-3000 live births irrespective of sex, ethnicity, or geographic region.[1-3] The condition follows an autosomal dominant pattern of inheritance with complete penetrance and highly variable expressivity; however, up to 50% of cases arise as de novo germline mutations in the NF1 gene.[1,3]

The NF1 gene encodes neurofibromin, a Ras-GTPase activating protein that functions as a tumour suppressor by accelerating the hydrolysis of active Ras-GTP to

inactive Ras-GDP.[3,5] Loss-of-function mutations lead to constitutive Ras pathway activation and disinhibited cellular proliferation within neural crest-derived cell lineages including Schwann cells, perineurial cells, endoneurial fibroblasts, and mast cells-the principal constituents of neurofibroma stroma.[5,6]

The diagnostic criteria for NF1, established by the National Institutes of Health (NIH) Consensus Conference and subsequently refined, require the presence of two or more of the following: (i) six or more café-au-lait macules exceeding 5 mm in prepubertal or 15 mm in postpubertal individuals; (ii) two or more neurofibromas of any subtype or one plexiform neurofibroma; (iii) axillary or inguinal freckling (Crowe's sign); (iv) optic pathway glioma; (v) two or more Lisch nodules (iris hamartomas); (vi) a distinctive osseous lesion such as sphenoid wing dysplasia or tibial pseudarthrosis; or (vii) a first-degree relative with the NF1.[4]

Cutaneous and subcutaneous neurofibromas are the hallmark lesions of NF1, typically appearing in the second decade of life and increasing in number and size with advancing age, particularly during pregnancy and the perimenopausal period.[1,3,4] Although individually benign and non-life-threatening, they may become disfiguring, pruritic, painful, or mechanically obstructive.[10] Giant pedunculated variants-less commonly reported in the literature pose additional challenges including recurrent trauma, secondary ulceration, superimposed infection, and the need to exclude MPNST, which carries a lifetime risk of approximately 8-13% in NF1 patients and represents the leading cause of disease-related mortality.[7,11,12] We report a case of a giant pedunculated neurofibroma of the left thigh with apical ulceration in a woman with NF1, detailing the pre-operative workup, operative technique, and histopathological findings.

CASE REPORT

A 56-year-old woman was referred to the outpatient general surgery clinic with a longstanding history of multiple cutaneous lumps distributed over the trunk and extremities. The dominant complaint was a large, progressively enlarging pedunculated mass located over the upper lateral aspect of the left thigh, which had been present for several years but had recently developed an ulcer at its apex with intermittent serous discharge. The patient reported generalised fatigue and disturbed sleep, likely attributable to discomfort from the mass. There was no history of acute haemorrhage, rapid size increase raising concern for MPNST transformation, fever, or constitutional symptoms. Her past medical and surgical histories were unremarkable. There was no documented family history of neurofibromatosis, suggesting a possible de novo mutation, although formal genetic counselling had not been undertaken previously.

Physical examination

The patient was alert, orientated, and haemodynamically stable. Vital signs on the day of assessment were: blood pressure 120/70 mmHg, heart rate 82-86 beats per minute (regular rhythm), peripheral oxygen saturation 97-99% on room air, temperature 36.6°C (97.8°F), and respiratory rate 21 breaths per minute. Cardiovascular examination revealed dual heart sounds with no added murmurs. Respiratory assessment demonstrated bilateral vesicular breath sounds with no adventitious sounds. Neurological assessment of all four limbs revealed full power (MRC grade 5/5) with intact sensory and motor function. Abdominal examination was unremarkable aside from the presence of numerous small sessile neurofibromas scattered across the abdominal wall.

Local examination of the left thigh demonstrated a large, soft, pedunculated mass with a broad pedicle arising from the upper lateral thigh region. The lesion was non-tender, compressible, and did not exhibit a positive transillumination sign. The skin overlying the bulk of the mass was intact, with multiple small satellite neurofibromas visible across the thigh. The apex of the pedunculated mass bore a well-demarcated ulcer with a fibrinous base and no active bleeding, consistent with chronic pressure-related ischaemic change (Figure 1).

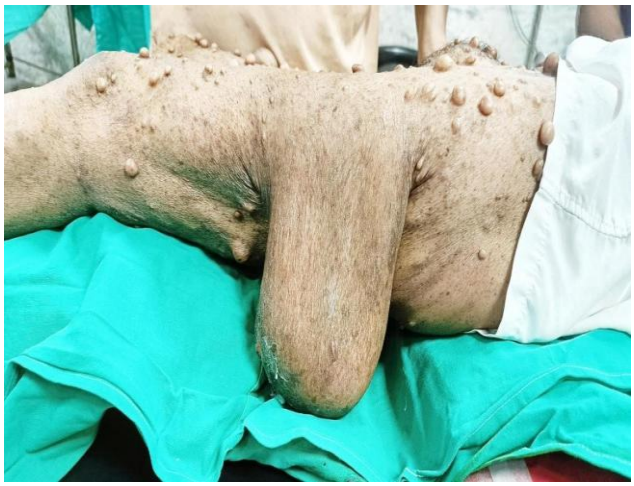

Figure 1: Clinical photograph demonstrating multiple cutaneous neurofibromas.

*Distributed across the left thigh with the dominant giant pedunculated neurofibroma visible in the right of frame. Note the diffuse cutaneous involvement characteristic of NF1.

Investigations

A structured pre-operative workup was undertaken. Haematological and biochemical investigations-including full blood count, renal and hepatic function panels, coagulation screen (PT-INR), and viral serology (HIV 1 and 2 antigen/antibody, hepatitis B surface antigen, and hepatitis C antibody)-were all within normal reference ranges with the exception of haemoglobin,

which was mildly reduced at 10.2 g/dl, consistent with the chronic anaemia of systemic disease seen in longstanding NF1. Urinalysis and urine culture were unremarkable. Electrocardiography and two-dimensional echocardiography demonstrated sinus rhythm with the normal biventricular function as well as no valvular pathology.

Ultrasound-guided FNAC of the dominant thigh mass yielded a haemorrhagic aspirate; cytological smears revealed blood and loosely cohesive stromal fragments without atypical cytomorphology, raising the differential of a vascular or mesenchymal lesion and warranting formal histopathological characterisation. Sonographic measurement of the lesion recorded dimensions of 53×37×39 mm with a calculated volume of approximately 40 ml.

MRI of the left thigh demonstrated a large, elongated soft-tissue mass in the posterolateral compartment, superficial to the deep fascia, with no intra-articular extension and no evidence of invasion into the adjacent intermuscular septa, neurovascular structures, or bone. Signal characteristics were consistent with a benign peripheral nerve sheath tumour with no features to suggest malignant transformation (target sign on T2-weighted sequences; Figure 3).[8]

HRCT of the thorax, performed as part of the cardiorespiratory fitness assessment, incidentally revealed bilateral fine fibrotic changes in the lower lobes with associated bilateral emphysematous changes. These findings did not preclude general anaesthesia but were noted for ongoing respiratory surveillance.

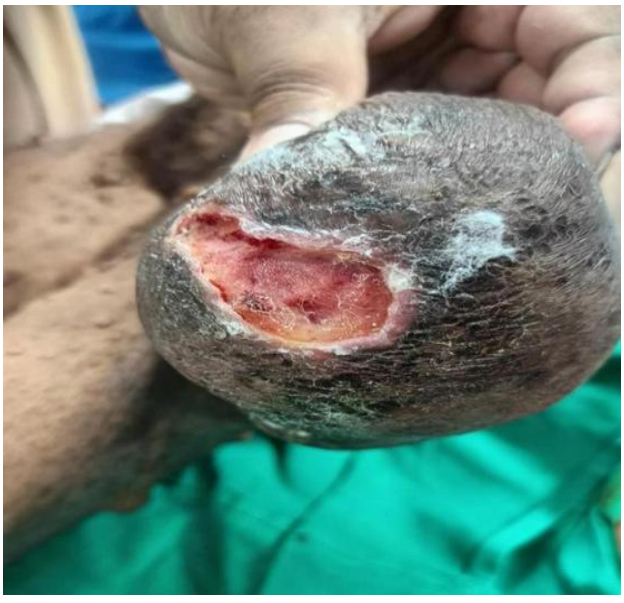

Figure 2: Close-up view of the apical surface of the pedunculated neurofibroma.

*Illustrating the ulcerated zone with a fibrinous slough and surrounding indurated skin. No active haemorrhage was present at the time of examination.

Figure 3: Pre-operative axial and coronal MRI images of the left thigh.

*The posterolateral soft-tissue mass (arrow) is well-circumscribed and confined to the subcutaneous compartment without deep fascial breach or neurovascular encasement.

Operative management

Following written informed consent for the procedure, anaesthesia, blood transfusion, and acknowledgement of high-risk features (mild anaemia, incidental pulmonary findings), the patient was admitted electively for surgical excision. She was positioned supine on the operating table and general anaesthesia was induced following standard pre-operative antiseptic skin preparation (ASS) and antiseptic surgical draping (ASD). An elliptical transverse skin incision was fashioned circumferentially around the pedicle of the mass, and gradual, progressive, anatomical dissection was performed using monopolar electrocautery, taking care to identify and preserve underlying neurovascular structures. The mass was dissected free from the underlying tissue in its entirety without capsular breach, achieving macroscopically clear margins. Meticulous haemostasis was secured, and wound closure was performed in layers using 2-0 monofilament non-absorbable sutures to the skin.

Histopathological findings

The excised specimen was submitted in formalin to the department of histopathology. Macroscopic examination described a pedunculated soft-tissue mass with a grey-white, whorled cut surface. Microscopic sections demonstrated a hypocellular to moderately cellular spindle-cell neoplasm with wavy, buckled nuclei, collagenous stroma, myxoid change, and interspersed mast cells-features pathognomonic of neurofibroma. There was no evidence of nuclear pleomorphism, increased mitotic activity, necrosis, or features suggesting transformation to MPNST. The final histopathological

diagnosis was benign peripheral nerve sheath tumour consistent with neurofibroma.

The post-operative course was uneventful. The patient was discharged on the second post-operative day with wound care instructions and appropriate analgesia. Follow-up at four weeks demonstrated satisfactory wound healing with no evidence of local recurrence. She was referred to a clinical genetics service for formal NF1 counselling and long-term surveillance planning.

DISCUSSION

NF1 is the most prevalent form of neurofibromatosis, accounting for approximately 90% of all cases of the condition. Riccardi and Eichner recognised four principal clinical subtypes: (i) peripheral neurofibromatosis (NF1); (ii) central or bilateral acoustic neurofibromatosis (NF2); (iii) segmental or mosaic neurofibromatosis; and (iv) cutaneous fibromatosis.[1] The incidence of NF1 is well established at approximately 1 in 2500-3000 live births, with no predilection for sex, ethnicity, or geography.[1-3]

Neurofibromas are mixed-cell tumours composed of neoplastic Schwann cells, perineurial-like cells, endoneurial fibroblasts, and a variable admixture of mast cells within a collagen-rich, frequently myxoid extracellular matrix. Three principal growth patterns are recognised: cutaneous neurofibromas, subcutaneous neurofibromas, and plexiform neurofibromas-the last carrying a 10-15% lifetime risk of MPNST transformation.[6,7] The histopathological features in the present case-wavy buckled nuclei, collagenous and myxoid stroma, and interspersed mast cells-are entirely consistent with the published morphological criteria for benign neurofibroma as described by Woodruff and Carroll and are distinct from the nuclear pleomorphism, hypercellularity, and elevated mitotic indices that characterise MPNST.[5,6]

Giant pedunculated neurofibromas are a recognised but uncommon complication of NF1. Fewer than 100 cases of truly giant neurofibromas, arbitrarily defined as exceeding 5 cm in greatest dimension, have been reported in the indexed literature.[11,13] The present lesion, measuring 53×37×39 mm on ultrasound with a calculated volume of approximately 40 ml, falls within the lower range of published giant neurofibroma dimensions. By contrast, Batista et al reported a pendulous thigh neurofibroma measuring in excess of 15 cm in a patient with NF1, requiring staged debulking.[11] Similarly, Kransdorf demonstrated that soft-tissue tumours of the thigh in the benign peripheral nerve sheath category rarely exceed 8 cm in mean diameter in large referral series.[13] The secondary apical ulceration observed in our patient is likewise described in prior case reports of pendulous lesions, arising from chronic pressure-induced ischaemia in a gravity-dependent mass, and mandates prompt surgical intervention to prevent secondary infection and progressive tissue loss.[11]

The role of pre-operative MRI in the evaluation of large neurofibromas cannot be overstated. In the present case, MRI delineated the anatomical extent of the lesion, confirmed the absence of deep fascial invasion and intra-articular extension, and demonstrated no high-risk features of malignant transformation such as peripheral enhancement, perilesional oedema, or infiltrative margins. The identification of the 'target sign'-central T2 hypointensity surrounded by peripheral hyperintensity-strongly favours benignity and is present in approximately 70% of neurofibromas on T2-weighted sequences.[8] Bhargava et al specifically validated this MRI sign in a paediatric cohort, demonstrating a sensitivity of 70% and specificity of 92% for distinguishing benign from MPNSTs.[8] Mautner et al further demonstrated that whole-body MRI is invaluable for documenting total tumour burden in NF1, allowing objective monitoring of lesion progression and identification of candidates requiring intervention-a strategy directly applicable to ongoing surveillance in the present patient.[14] FNAC yielded a haemorrhagic specimen in our case, a recognised pitfall in hypervascular neurofibromas; formal histopathological examination of the resected specimen therefore remained indispensable for definitive diagnosis, consistent with published recommendations that pre-operative cytology alone is insufficient for peripheral nerve sheath tumour characterisation.[6,8]

Regarding surgical technique, the elliptical pedicle-based excision employed in this case is well-suited to pedunculated lesions and is consistent with the approach most widely reported in the literature for large cutaneous neurofibromas.[11,16] Lin et al described a comparable elliptical incision technique for a giant pendulous neurofibroma of the lower extremity, achieving primary wound closure without tension grafting, analogous to the outcome in the present case.[16] Complete en bloc resection, as achieved here, is universally accepted as the principal determinant of local disease control. Needle et al in their single-institution series of plexiform neurofibromas, demonstrated that incomplete resection was the strongest predictor of local recurrence and symptomatic progression, underscoring the importance of macroscopically clear margins.[9] The use of monopolar electrocautery facilitates haemostasis in these highly vascular lesions, though care must be taken not to transmit heat to adjacent neural structures-a principle consistently emphasised in neurofibroma excision literature.[16] Adjuncts such as laser ablation and radiofrequency destruction have been reported for smaller lesions in patients with a high lesion burden, but surgical excision remains the gold standard for large, symptomatic, or ulcerated tumours, as endorsed by current NF1 management guidelines.[4,17]

The lifetime risk of MPNST in NF1 patients is estimated at 8-13%, representing the leading cause of NF1-related mortality.[7,12] Evans et al reported a cumulative MPNST incidence of 8% by age 50 in a large UK NF1 cohort, with the highest risk observed in patients with a high

internal plexiform tumour burden.[7] Ducatman et al in their landmark clinicopathological analysis of 120 MPNSTs, identified NF1 as the underlying predisposition in 50% of cases, and noted that high-grade transformation was associated with rapid symptom progression and deep anatomical location-neither of which was present in our patient.[12] The absence of nuclear pleomorphism, elevated mitotic activity, and tumour necrosis on histopathological examination in the present case is therefore reassuring, though lifelong multidisciplinary surveillance for MPNST transformation remains mandatory, consistent with current NF1 guidelines.[4,15]

The incidental finding of bilateral pulmonary fibrosis and emphysema on HRCT in the present patient is a pertinent reminder that NF1 is a multisystem disorder. Pulmonary manifestations of NF1-including interstitial lung disease, bullous emphysema, and pulmonary hypertension-have been described in the literature and are thought to arise from neurofibromin loss in pulmonary interstitial mesenchymal cells.[13,15] Zamora et al characterised NF1-associated lung disease in a case series and literature review, noting bilateral lower-lobe fibrosis and emphysema as the most common pattern, mirroring the HRCT findings in our patient.[13] Ferner comprehensively reviewed the systemic manifestations of NF1 and emphasised that pulmonary complications, though less prevalent than cutaneous or neurological features, may significantly impact anaesthetic risk and perioperative planning-a consideration that was directly relevant in the pre-operative assessment of our patient, in whom incidental fibrotic and emphysematous changes necessitated cardiorespiratory specialist review before elective surgery.[15]

Beyond the immediate operative considerations, present case highlights the broader impact of NF1 on quality of life. Valeyrie-Allanore et al demonstrated, in a cross-sectional study of 128 NF1 patients, that health-related quality of life was significantly impaired compared with age-matched controls, with cosmetic disfigurement, pain, and sleep disturbance among the most frequently reported domains of impairment-all of which were directly experienced by the present patient secondary to giant pedunculated lesion.[10] These findings underscore the importance of not only surgical intervention but also holistic multidisciplinary management, including psychological support, pain management, and genetic counselling, for patients with NF1.[17]

CONCLUSION

We present a case of a giant, pedunculated, ulcerated cutaneous neurofibroma arising in the context of NF1, successfully managed by elective surgical excision with an uneventful recovery. This case underscores several important clinical principles: the necessity of comprehensive pre-operative imaging to characterise the lesion and exclude malignant transformation; the value of formal multidisciplinary input encompassing surgical, anaesthetic, and cardiorespiratory specialists; the efficacy of meticulous anatomical dissection in achieving complete resection; and the imperative of histopathological confirmation even when pre-operative imaging is reassuring. Patients with NF1 require lifelong surveillance given the systemic nature of the condition and the non-negligible risk of MPNST. Genetic counselling and cascade screening of first-degree relatives should be offered to all affected individuals.

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