Case Report

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Surgery Section

A Rare Case of Diffuse Neurofibroma Mimicking as Hairy Naevus or Vascular Malformation: A Surgeon's Perspective

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ABSTRACT

Nevi are areas of melanocytic hyperplasia or neoplasia. These collections can be found in the epidermis, partially in the dermis, or completely within the dermis. They commonly develop in childhood and young adulthood and sometimes spontaneously regress. Exposure to Ultraviolet (UV) radiation is associated with increased density of these lesions. In contrast, neurofibromas are benign proliferations made up of all nerve elements and arise as fleshy and non tender, sessile, or pedunculated masses on the skin. They can arise sporadically or in association with type 1 Neurofibromatosis (NF), and in these cases, are associated with café-au-lait spots and Lisch nodules. In the present report, authors present a case diagnosed clinically as a giant naevus on the dorsum of the right hand in an adult patient (26-year-old female) with no other associated secondary swellings elsewhere in the body. However, further radiological investigations revealed a case of vascular malformation. Postoperatively, histopathological examination showed features of diffuse neurofibroma. The present case report enables the surgeon to consider various rare differential diagnosis for swelling in the extremities, even when it is not reflected in the clinical or radiological findings. Thus, a detailed history, physical examination, and various diagnostic tests, keeping in mind all possible differential diagnosis, should be performed.

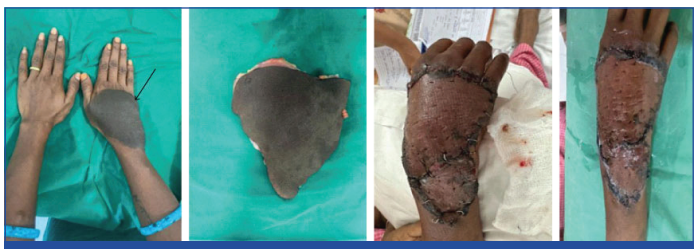
Keywords: Melanocytic hyperplasia, Nevi, Type 1 neurofibromatosis, Ultraviolet radiation

CASE REPORT

A 26-year-old female presented with a swelling on the dorsum of her right hand that had been present for five years. The swelling, measuring approximately 11×5 cm, had gradually increased in size and was accompanied by skin darkening and hair [Table/Fig-1]. Upon inspection, the swelling was observed to be flat, solitary, irregular in shape, approximately 10×8 cm, non pulsatile, with hyperpigmentation on the dorsum of the right hand. On palpation, it was soft, non tender with no local rise in temperature, compressible, with a smooth surface, variable consistency, fixity to the underlying tissues, and the presence of hair over it. A clinical diagnosis of a hairy naevus was made based on the examination findings. Complete blood count, kidney function, and coagulation profile all showed results within the normal range.

On a colour Doppler of the right upper limb, a large (approximately 10×11 cm), hypoechoic soft-tissue density lesion was noted in the palmar aspect of the hand in the subcutaneous plane, with a maximum depth of 20 mm. Few hyperechoic linear striations were noted within. Multiple vascular feeders to the lesion were noted, predominantly showing venous flow on colour Doppler (PSV-3-4 cm/s), likely suggestive of slow flow vascular malformation.

Following the clinical presentation and imaging results, the patient underwent surgical intervention. The swelling was completely excised without any harm to the underlying structures, and split-thickness skin grafting was performed [Table/Fig-2,3].



[Table/Fig-1]: Swelling over the dorsum of right hand. **[Table/Fig-2]:** Excised specimen. **[Table/Fig-3]:** Postoperative wound. **[Table/Fig-4]:** Follow-up image of the wound (one month follow-up). (Images from left to right).

Histopathological study showed melanophages and a tumour, predominantly showing diffuse growth with focal plexiform areas. The tumour is composed of elongated cells with scanty cytoplasm and elongated wavy nuclei, infiltrating the subcutaneous tissue. Neurofibroma-diffuses type with S-100 positive. Immunohistochemistry (IHC) confirmed the diagnosis of neurofibroma. Staple removal was performed on postoperative day 10, with the surgical site being healthy and no signs of infection noted. The patient was discharged on postoperative day 12. She was advised to follow-up after one month, during which the graft site was found to be healthy, and no signs of recurrence were seen [Table/Fig-4].

DISCUSSION

Neurofibroma is a benign, heterogeneous peripheral nerve sheath tumour arising from the connective tissue of the peripheral nerve sheath, especially the endoneurium, and accounts for approximately 5% of all benign soft-tissue tumours in large surgical series [1]. Three types of neurofibromas are described: localised, plexiform, and diffuse. Diffuse neurofibroma is a less common subtype of neurofibroma that has received little attention in the imaging literature. It has been reported to occur most commonly among children and young adults, typically involving the skin and subcutaneous tissues of the head and neck [1].

Siddiqui S et al., reported a case of a 16-year-old girl who presented with a gradually progressive swelling around the right ankle and heel, which was initially diagnosed as a case of a vascular malformation. However, it subsequently turned out to be a diffuse neurofibroma [2].

Spinelli C et al., reported a case of a two-year-old boy diagnosed with Neurofibromatosis Type 1 (NF1) , who presented with a palpable swelling on the left nuchal region showing ultrasonographical characteristics of a venolymphatic malformation. However, it was later histologically reported to be a superficial plexiform neurofibroma [3].

A case study by Miraglia E et al., found that 45.3% of people with Neurofibromatosis Type 1 (NF1) had a familial history, 90%

of patients exhibited axillary and inguinal freckling, 78.1% had neurofibromas, and 96.5% of patients had Cafe Au Lait Macules (CALM) [4]. Other skin conditions rarely present were vitiligo, Becker's naevus, melanoma, poliosis, lipoma, psoriasis, spilus naevus, juvenile xanthogranuloma, and naevus anaemicus [4]. A light brown patch of varying sizes and locations known as Naevus Spilus (NS) is distinguished by numerous, unevenly shaped, smaller, darker brown macules that are overlaid. Rarely has the relationship between NF and NS been documented. The release of growth factors by fibroblasts, such as nerve growth factor and stem cell factor, may account for this correlation [4,5].

Imaging findings can be observed using ultrasound and Magnetic Resonance Imaging (MRI). It can be seen as one or more regular-shaped, clear-boundary hypoechoic solid masses on ultrasound [6,7]. There is no sign of blood flow, and the internal echo is homogeneous. It displays a low signal on T1-weighted imaging and a high signal on T2-weighted imaging on plain MRI [8,9]. The T2 signal is significantly enhanced to varying degrees, with small tumours displaying a uniform signal and larger tumours showing uneven signals. In the present case report, ultrasound showed a large (approximately 10×11 cm), hypoechoic soft-tissue density lesion in the palmar aspect of the hand in the subcutaneous plane, with a maximum depth of 20 mm. Few hyperechoic linear striations were noted within. Multiple vascular feeders to the lesion were noted, predominantly showing venous flow on colour Doppler (PSV- 3-4 cm/s), likely suggestive of slow flow vascular malformation.

The definitive diagnosis is made through histopathological examination, which is considered the gold standard. Pathologically, the tumour tissue exhibits slightly clear boundaries and appears as strip or bead-shaped grayish-white tissue. It has a tough consistency and is connected to the nerve bundle proximally and distally, following the direction of the nerve. Microscopically, it is composed of Schwann cells, fibroblasts, perineurial cells, and mast cells [6,10]. According to immunohistochemical analysis, the S-100 protein is positively stained, and the Ki-67 Proliferation Index (PI) is typically between 0% and 5% [7,11].

Surgical excision is the ultimate treatment; however, imaging plays a major role in determining the size, site, and shape of the tumour, as well as its relationship with surrounding tissues, and for guiding the formulation of a surgical strategy.

Diffuse neurofibromas share morphological features with localised cutaneous neurofibromas, but they display a distinct growth pattern. These tumours extensively infiltrate the dermis and subcutaneous connective tissue, ensnaring fat and appendage structures, resulting in a plaque-like appearance.

The NF1 is linked to a significant percentage of malignant peripheral nerve sheath tumours (around 50%). However, only a tiny percentage of NF1 patients-roughly 5%-develop malignant peripheral nerve sheath tumours. Malignant peripheral nerve sheath tumours typically affect the main nerve trunks and manifest as a soft-tissue mass along with neurologic signs and discomfort [4]. These signs suggest a poor prognosis and higher mortality. Regular follow-up is essential to detect any indications of malignancy, thereby suggesting a need to consider it as one of the differential diagnosis in hand masses for early detection and timely management.

CONCLUSION(S)

The present case is a unique case discussing a rare presentation of an unusually large Naevus over the dorsum of the right hand, which turned out to be a diffuse neurofibroma in an adult. Due to the hand's various tissues and predisposition for a variety of tumours, it is imperative to consider diffuse neurofibroma as a probable cause of hand masses. Diagnosing hand tumours before surgery can be challenging. Overall, a comprehensive approach involving thorough examination, medical history, and imaging studies is necessary for accurate diagnosis and surgical planning to ensure complete tumour removal and prevent recurrence.

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