Solitary asymptomatic keloid- like mass

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CLINICAL FINDINGS

A 30-year-old male patient presented with asymptomatic, small, skin-colored, raised mass over the right thumb. The lesion was first noticed at two years of age and appeared as a small, flesh-colored papule. It started to increase in size gradually until the present state, taking the appearance of a keloidal mass. The lesion was initially painless, but became more painful by the age of 21. There was no associated itching or discharge from the lesion. The patient did not complain of any systemic illness and there was no family history of similar condition.

Clinical examination revealed a 2 × 2 cm solitary, flesh-colored, indurated mobile keloidal nodule located overlying the right proximal interphalangeal joint. Nails and mucous membranes were not affected and showed no significant abnormalities. Routine laboratory investigations including CBC, blood sugar, hepatic and renal profile revealed no abnormal findings. X-ray of both hands showed no attachment of the lesion to underlying bones (Fig. 1,2).

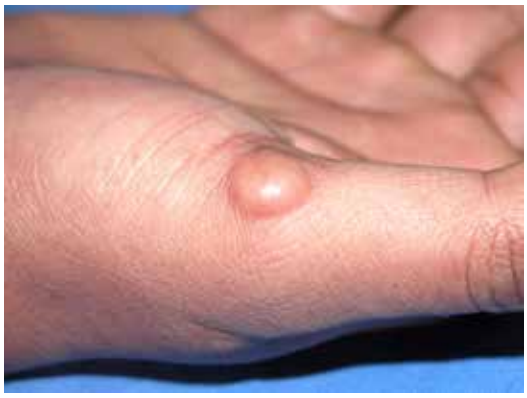

Fig. 1 Skin colored, firm, mobile mass 2×2cm over the proximal interphalangeal joint of Rt thumb.

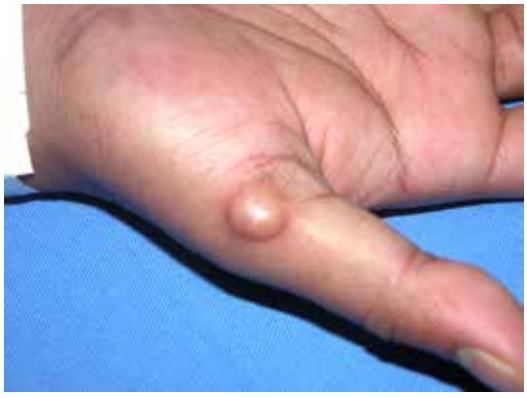

Fig. 2 Mass overlying the right proximal interphalangeal joint.

What is your clinical differential diagnosis?

1. Keloid
2. Giant tumor of tendon sheath
3. Ganglion
4. Adnexal tumor

Total excisional biopsy was performed for removal of the tumor and histological analysis revealed well defined, unencapsulated dermal mass formed of wavy spindle shaped cells and mast cells in homogenous eosinophilic stroma which was consistent with solitary neurofibroma (Fig. 3,4).

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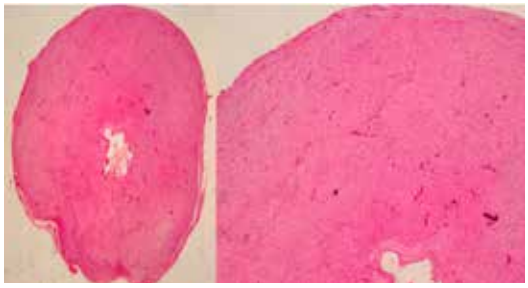

Fig. 3 Well defined, un-encapsulated dermal mass formed of spindle shaped cells.

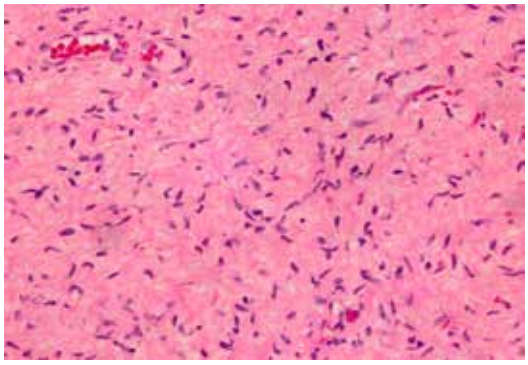

Fig. 4 Wavy spindle shaped cells and mast cells in homogenous eosinophilic stroma.

DIAGNOSIS

Solitary Neurofibroma

DISCUSSION

There are three types of peripheral neural sheath tumors (PNST) including schwannoma, neurofibroma, and neurogenic sarcomas.[1] Schwannomas and neurofibromas, as benign PNSTs, account for 10% of benign soft-tissue tumors.[2]

Neurofibromas can develop in any peripheral encapsulated nerve of the body, which can be invaded by the neurofibroma and can be divided into: (1) Solitary or isolated neurofibromas, which more commonly originate from cutaneous nerves; (2) diffused, most likely to arise from the nerves in the subcutaneous tissues of the head and neck; (3) plexiform, which are diffused masses with tortuous expansion along the branches of the parent nerve.[2]

The pathogenesis of solitary neurofibroma not associated with NF-I is poorly understood. Storlazzi et al. using G-banding and fluorescence *in situ* hybridization analysis showed that somatic inactivation of NF-1 gene located on chromosome 17 through chromosomal translocation leads to increased and abnormal production of neurofibromin which regulates ras-mediated cell growth pathway leading to increased levels of activating proteins p21ras and p13 which causes cellular proliferation of Schwann cells associated with neurofibroma.[3]

The differential diagnosis for soft tissue tumor of the digit is extensive and includes both benign and malignant entities. The most common benign soft tissue tumors of the digits are ganglion cysts, giant cell tumors, fibromas, glomus tumors, sarcoma, and malignant fibrous histiocytoma have high rates of local recurrence and metastasis, and thus may require amputation for adequate treatment.[4]

Given this extensive differential diagnosis, imaging is an important step in the pre-operative evaluation of the patient. In addition to regular X-rays, suggested imaging studies for the finger tumors include ultrasound and MRI to assess extent of the lesion, involvement of surrounding structures and to narrow the differential diagnosis.[5] Ultrasound is relatively inexpensive and easily accessible, but technically difficult and conveys less information. MRI offers superior imaging with greater anatomic detail and a higher likelihood of accurate diagnosis, but is more expensive. Most soft tissue tumors will have similar clinical presentations and therefore all soft tissue tumors of the digit must be biopsied prior to complete resection due to malignant potential.[5]

Both schwannomas and neurofibromas are derived from a neoplastic Schwann cell. Histologi-

cally, neurofibromas feature neoplastic Schwann cells, perineural-like cells, and fibroblasts in a mucocollagenous matrix. These are histologically distinct from schwannomas, which present as spindle-shaped Schwann cells embedded in Antoni A and B patterns of compact, elongated cells and loose, less cellular regions, respectively. Both entities stain positive for S-100, as this stain is specific to neural crest derived tissues. Neurofibromas stain positive for CD34, an intercellular adhesion protein and cell surface glycoprotein with a poorly understood function, and stain negative for EMA, a cell surface mucin glycoprotein expressed by most glandular and ductal epithelial cells and some hematopoietic cells. In contrast, schwannomas stain positive for EMA and negative for CD34.[6]

After histopathological diagnosis is established, symptomatic benign lesions should be completely resected with care taken not to disrupt nearby structures, in particular the digital nerves. Involved tissue should be resected as closely as possible to the tissue bed.[7] Malignant lesions will likely require the care of a multidisciplinary team with assessment of metastasis and consideration for the role of radiation therapy or chemoradiotherapy.[8]

Regarding the location and size of tumor, the symptoms are variable. Discoloration of skin accompanied with disfiguration in the cutaneous form of NF is expected. Whereas, deeper masses can cause neurological symptoms and destruction as a result of compression of the neighboring organs. Neurofibromas are often asymptomatic, but can cause debilitating pain and motor sensory dysfunction. They are usually benign and grow slowly, but sometimes degenerate to become malignant.[9]

Diagnostic difficulties and delays are common due to the lack of characteristic features of these lesions and in some cases due to unexpected sites. Neurofibromas on fingers are rare and reports in literature are scant. In our case, diagnosis of neurofibroma was not considered clinically due to the unusual site of the tumor. But in retrospect, the lesion was typical solitary neurofibroma.[10]

Histologically, neurofibromas in all form of neurofibromatosis can be unencapsulated, poorly circumscribed tumors that can incorporate all aspects of the nerve including the surrounding Schwann cells and the axons within.[11]

In this case, clinical examination showed small, firm keloid-like mass overlying the proximal interphalangeal joint of the right thumb. Histopathological examinations revealed well defined, unencapsulated dermal mass formed of wavy spindle shaped cells and mast cells in homogenous eosinophilic stroma which was consisting with solitary neurofibroma.

The patient was treated by complete excision of the tumor which was in accordance with the treatment protocol for solitary neurofibroma.[12] No signs of recurrence or NF-1 were noted over a follow-up period of 9 months.

Solitary neurofibromas may become malignant, although it is extremely rare. Malignant transformation is usually seen with multiple neurofibromas and those associated with Von Recklinghausen disease or MEN-III syndrome.[13]

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