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Case Report

Giant Pigmented Neurofibroma - An Unusual Variant of A Usual Tumor

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ABSTRACT

Neurofibromas (NFs) are benign nerve sheath tumors which are relatively common. Pigmented neurofibroma is a rare variant of neurofibroma showing melanin production and constitute less than 1% of all cases. The clinical diagnosis is difficult to establish requiring the histopathological examination to differentiate between the melanotic neurofibroma and other pigmented tumours. We report an unusual case of giant pigmented neurofibroma without striking clinical pigmentation with stigmata of neurofibromatosis.
Key words: Neurofibroma , Pigmented neurofibroma , Melanotic neurofibroma

INTRODUCTION

Neurofibromas (NFs) are benign nerve sheath tumors of the peripheral nervous system which were first described by Smith in 1849 and later by Von Recklinghausen in 1882. They are relatively common, slowly growing, and painless tumors. [1] They are usually found in individuals with neurofibromatosis type 1 (NF1), a genetically inherited disease and arise from schwann cells that exhibit inactivation of the NF1 gene that codes for the protein neurofibromin. The majority of NFs are solitary lesions that occur in the dermis or subcutis. Multisystemic involvement is common and an increased incidence of malignancies, osseous defects and congenital dislocations, oral pathology, endocrine disorders, autonomic involvement, GI tract involvement, hypertension, and vascular anomalies may be present. [1]

Pigmented neurofibroma (PNF) is a unique pathologic subtype of neurofibroma containing melanin producing cells. The pigment is not usually appreciated on gross examination and requires histologic examination. The pigmented cells which are dendritic or epithelioid in shape are dispersed throughout the tumour and express both S100 protein and melanin markers. [2,3] The presence of melanin producing cells in PNF can be explained on the basis of origin of both, the melanocytes and schwann cells from multipotent neural crest cells. Although melanogenic potential of schwann cells is suggested and proved in the literature, very few reports provide

ultrastructural confirmation. We document the clinicopathological and ultrastructural findings of giant pigmented neurofibroma and discuss the differential diagnosis.

CASE REPORT

A 28 years old male presented to us with a large swelling in left gluteal region & multiple small swellings all over the body since childhood. He had noticed a small, slightly elevated nodule over left gluteal region in childhood and the tumour had gradually increased in size. There was difficulty in walking since childhood and recent onset of dragging pain. There was no history of fever, loss of weight, hearing impairment or tinnitus. There was no family history of similar disease.

General physical examination revealed multiple cutaneous swellings all over the body and multiple Cafe au lait spots (Figure 1). Local examination revealed a huge pendulous swelling around 32x16x10 cms, involving the left gluteal region and extending up to the calf with sparing the anal orifice (Figure 2). No pigmentation or ulceration was seen. The swelling was soft in consistency. In view of presence of stigmata of neurofibromatosis, patient was diagnosed clinically to have giant neurofibroma and was advised surgery.

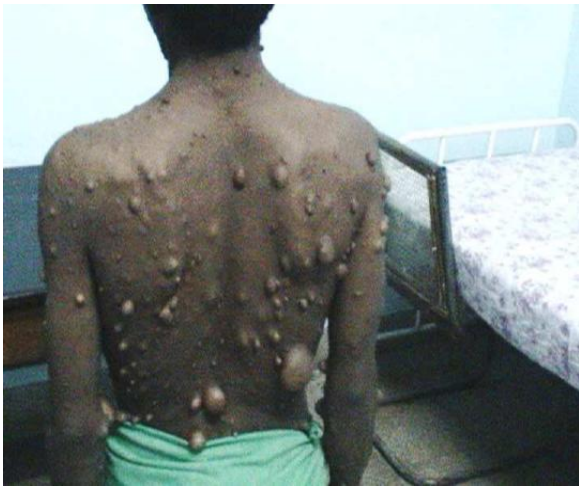

Figure 1 Patient with multiple neurofibromas

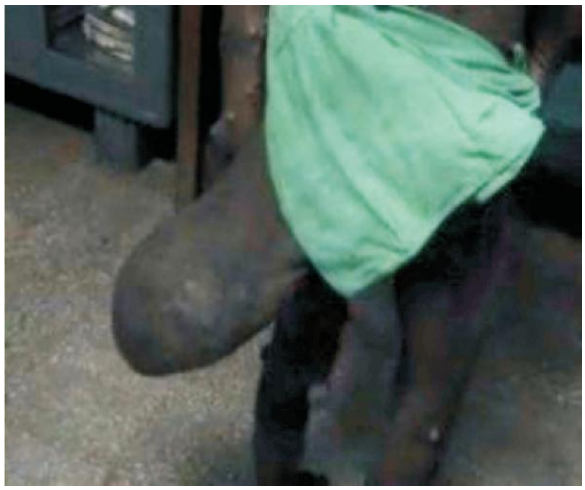

Figure 2 Large pendulous swelling of left gluteal region extending up to the calf

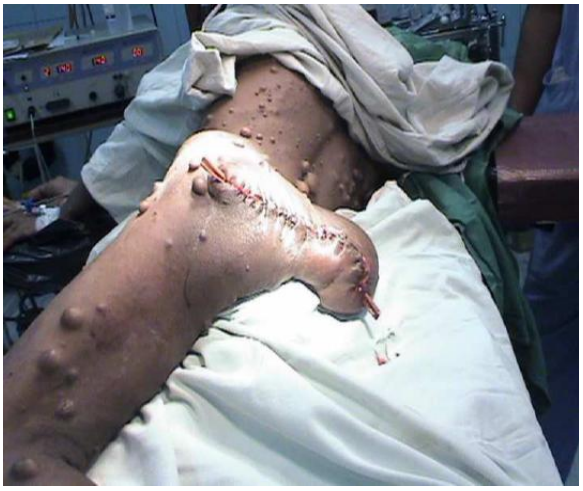

Figure 3 Intraoperative photograph after excision of tumor

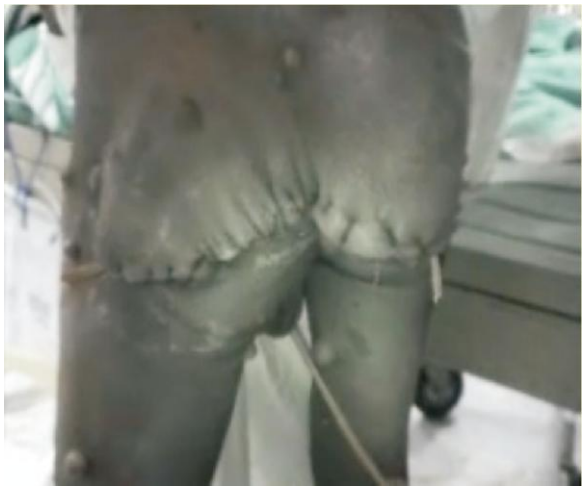

Figure 4 Postoperative photograph

Patient underwent excision of tumor under spinal anaesthesia. The weight of excised tumor was 20 kilograms. On gross examination the resected specimen measured 34x14x13 cms and was covered with thick nonpigmented skin. The cut surface revealed an ill demarcated, solid, grayish white to tan,glistening, soft to rubbery tumour involving the dermis and subcutis. There was haemorrhage but no evidence of necrosis (Figure 3 and 4).

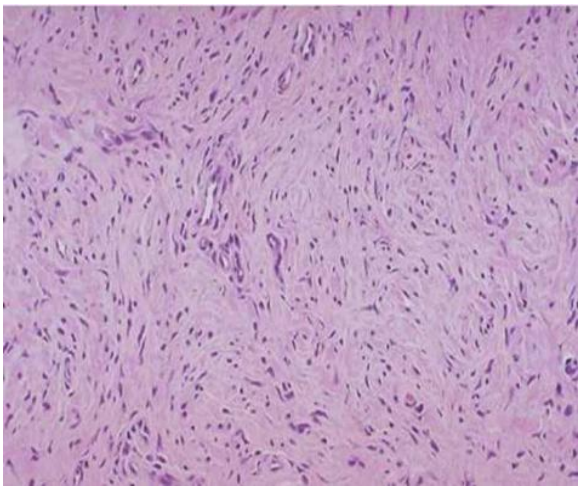

Figure 5 Histopathology showing pigmented neurofibroma

On microscopic examination the tumour had characteristic appearance of diffuse neurofibroma with uniform matrix of fine fibrillary collagen and short fusiform to rounded schwann cells embedded in it. The peculiar feature was the presence of spindled, dendritic and epithelioid pigmented cells in clusters throughout the tumour. The pigment was granular and brownish black (Figure 5). Tumour infiltration into the subcutaneous fat was observed. On immunohistochemistry, spindle cells were positive for S-100 protein, Melan-A, and CD34, but negative for HMA-45 and melanin producing cells were S-100 (+), Melan-A (+), HMB45 (+), but CD34 (-). Histopathologically the tumor was diagnosed as giant diffuse pigmented neurofibroma of left gluteal region. Patient has been on regular follow up for the past 17 months without any complaints.

DISCUSSION

Pigmented neurofibroma also named melanotic neurofibroma are rare variants usually showing only faint macroscopically obvious pigmentation and accounting for less than 1% of all neurofibromas. They are more frequent in dark skinned populations with an age range of 2-71 years and usually involve head and neck, buttock, lower leg and thigh. [2,3] Because of the diffuse pattern of growth these lesions may recur, but malignant transformation or metastasis has not been known.

Macroscopically pigmented neurofibromas involve dermis and subcutis and can vary between 1 to 50 cm in size with a glistening, gray tan cut surface, ill-defined margins and rubbery consistency. Pigmentation within the tumour varies from absent to striking blackish flecks. [2-4] The resected tumour in our case had above features without macroscopic pigmentation.

Microscopically pigmented neurofibromas have a typical histological appearance of a neurofibroma with scattered melanin laden cells. Melanin producing cells tend to be located in the deep dermis and subcutis, accompanying neurofibroma components in the more superficial dermis. [3] According to Motoi et al this unique distribution pattern of melanin producing cells in pigmented neurofibromas can be a major useful tool for distinguishing this tumour from other pigmented tumours. [3] Immunohistochemical studies demonstrate that the non pigmented spindle cells are positive for S-100 protein, Melan-A, and CD34, but negative for HMA-45. But the melanin producing cells are S-100 (+), MITF (+), Melan-A (+) and HMB45 (+/-), but CD34 (-). [2]

The differential diagnosis of pigmented neurofibroma includes pigmented dermatofibrosarcoma protuberans, cellular blue nevus and melanotic schwannoma. Pigmented dermatofibrosarcoma protuberans (DFSP) also called storiform neurofibroma or Bednar tumor is an uncommon tumour with a predilection for the truncal region which is not usual for pigmented neurofibroma.The uniform fibroblastic cells, repetitive storiform pattern, strong positivity for CD 34 and lack of S-100 protein immunoreactivity usually make this distinction apparent. [5,6]

Cellular blue nevus presents typically in young adults in the lumbosacral and buttock region. The tumour has dumbbell like configuration and exhibits a more solid, organoid or nested growth than pigmented neurofibroma. It typically lacks prominent proliferation of schwann cells, abnormal nerve trunks, fibrillary collagenous matrix and Wagner Meissner bodies. [7] Melanotic schwannoma can be differentiated from pigmented neurofibroma by the presence of cellular areas alternating with hypocellular areas, cells with peculiar syncytial quality, presence of psammoma bodies, large and epithelioid pigmented cells and are S-100 (+), MITF (-), Melan-A (-), tyrosinase (-) and HMB45 (-). [5,6]

CONCLUSION

Pigmented neurofibroma is a unique subtype of neurofibroma which is rare and contains melanin producing cells. Most occur in patients with neurofibromatosis and are of diffuse type, although some have features of both diffuse and plexiform types. Because of the diffuse pattern of growth these lesions may recur, but malignant transformation or metastasis has not been known. The histomorphological features with unique pattern of melanogenesis, ultrastructural findings and immunohistochemistry will enable us to diagnose this entity and differentiate it from other pigmented tumours of skin.

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