

Segmental neurofibromatosisPawar S¹, Wankhade V², Singh R P³, Kodale P⁴

Figure 1 : Nodules on dorsum of right hand



Figure 2 : Multiple soft to firm nodules
On the Right Forearm

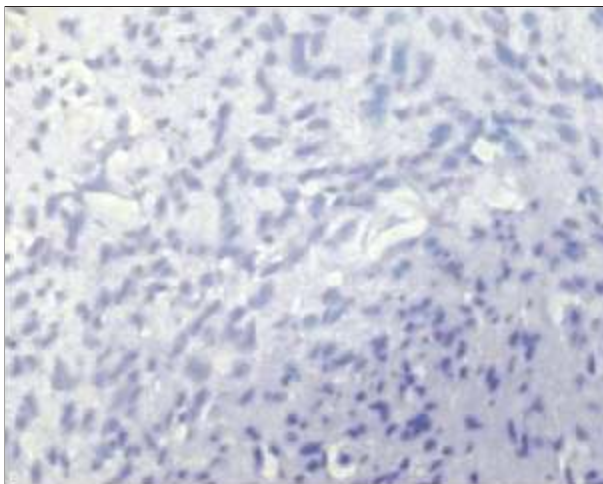


Figure 3 : Interlacing spindle shaped cells
arranged in myxoid matrix H&E40 X

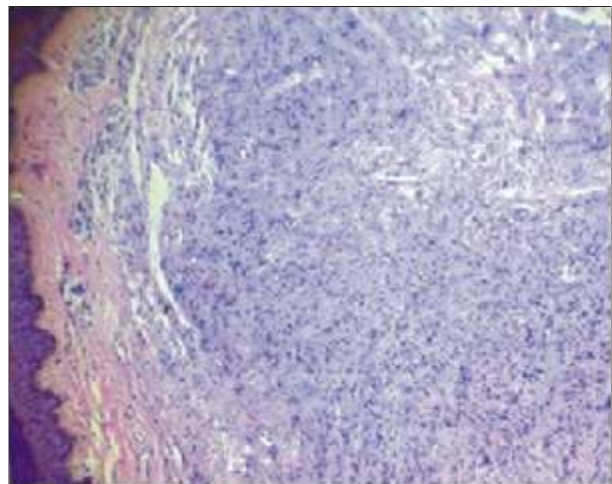


Figure 4 : Encapsulated Tumor in dermis
with normal epidermis H & E 10X

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A 50 year old female presented to department of dermatology with multiple asymptomatic nodules on the right forearm which had gradually evolved over a period of 10 yrs. There was no family history of similar lesions. Physical examination showed multiple, skin colored, and painless, firm nodules of variable size in a dermatomal pattern on the extensor aspect of the right arm extending from the dorsum of

hand to cubital fossa (**Fig.1&2**). Cafe-au-lait macules, axillary freckling, skeletal deformities were absent. On slit lamp examination no lisch nodules were found. Histopathological examination of one of the nodules revealed a non encapsulated tumor of the dermis, with a normal overlying epidermis (**Fig. 3**). The tumor consisted of interlacing bundles of spindle cells in myxoid matrix (**Fig. 4**). On the basis of clinicopathological correlation a diagnosis of segmental neurofibromatosis was reached.

Segmental neurofibromatosis (SNF) is a rare subtype of neurofibromatosis (Nf1) with its prevalence estimated between 0.0014%-0.002%.¹ This localized form of neurofibromatosis was first described by Gammel in 1931.² Riccardi classified SNF as type Vof neurofibromatosis type 1. It is defined as café-au-lait macules or neurofibromas in a single unilateral segment of the body, with no crossing of the midline, no family history and no systemic involvement. Females are affected twice as often as males with bimodal peaks of onset at 10-30 yrs and 50-70 yrs.³ Cervical and thoracic regions are commonly involved and the disease is unilateral in majority. ¹³SNF of the face and the extremities is uncommon. It can rarely be bilateral, multidermatomal and hereditary³, The most common manifestation of SNF is neurofibromas, and less frequently, café-au-lait spots and axillary freckling. Pigmentary changes and plexiform

neurofibromas appear in childhood, whereas neurofibromas usually develop during adolescence.⁴ Roth et al. subdivided SN into four variants: true segmental, localized with deep involvement, hereditary and bilateral.⁵ The regionally restricted distribution of skin lesions in segmental neurofibromatosis and the lack of antecedent family history of the affected person whose offspring can show full blown NFI have suggested that SNF results from a post zygotic NFI gene mutation.⁶

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