Plexiform neurofibromatosis

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A 34-year-old female presented with large pedunculated lesion over right shoulder associated with cafe au lait macules and neurofibromas since childhood [Figure 1]. The lesion over the right shoulder gradually increased over the time and was painless. The lesion extended over right arm and breast region without any posterior extension over scapular area. Radiograph of right shoulder did not show any bony involvement. Histopathology revealed normal epidermis with well-circumscribed collections of spindle cells in the dermis [Figure 2].

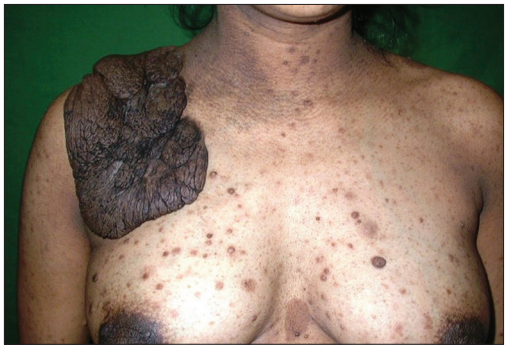

Figure 1: Giant brown pedunculated lesion over right shoulder

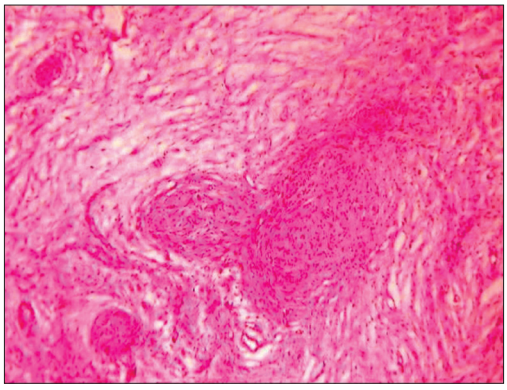

Figure 2: Hematoxylin and eosin-stain showed well-circumscribed collections of spindle cells in the dermis (×100)

Neurofibromatosis type 1 is a benign autosomal dominant genetic disorder with complete penetration. It is caused due to mutation of a gene present on chromosome 17q11.2 that encodes protein neurofibromin which is negative regulator of RAS proto oncogene signal transduction pathway. It affects approximately 1 in 3 000 individuals.[1]

Plexiform neurofibroma is a least common form of neurofibromatosis which occurs in 5% and shows rapid proliferation of the neural tissue in the subcutaneous fat. It occurs in either solitary nodular or diffuse form which is also known as elephantiasis neurofibromatosis. They have risk of development of malignancy which ranges from 5% to 28%. The risk of malignancy increases with the duration of disease as well as according to size of lesion. The commonest malignancy in plexiform neurofibromatosis is Malignant Peripheral Nerve Sheath Tumors (MPNST). Among patients with MPNST, the chance of recurrence of malignancy is up to 47%, with metastasis occurring in 20% of cases.[2]

The commonest site of Plexiform neurofibromatosis is along the trigeminal nerve distribution. Positron Emission Tomography scan may be used to exclude malignancy along with Magnetic Resonance Imaging to define such tumors.[3]

Surgical excision along with genetic counseling remains the treatment of choice, but patients with plexiform neurofibroma have to keep follow up on regular basis as they have risk of development of malignancy or recurrence.[3]

REFERENCES

1. Easton DF, Ponder MA, Huson SM, Ponder BA. An analysis of variation in expression of neurofibromatosis (NF) type I (NF I): Evidence for modifying genes. Am J Hum Genet 1993;53:305-13.
2. McCarron KF, Goldblum JR. Plexiform neurofibroma

with and without associated malignant peripheral nerve sheath tumor: A clinicopathologic and immunohistochemical analysis of 54 cases. Mod Pathol 1998;11:612-7.
3. Sehgal VN, Srivastava G, Aggarwal AK, Oberai R. Plexiform neurofibromas in neurofibromatosis type 1. Int J Dermatol 2009;48:971-4.

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