Plexiform Neurofibromatosis

A 10-year-old boy presented with a gradually progressive swelling over his right arm for last 7 years (*Fig. 1*). Examination revealed a soft, diffuse swelling measuring 10 × 12 cm over posterior aspect of right arm. The lesion was non-tender and freely mobile over the underlying tissues. In addition, he had multiple hyperpigmented macules with serrated margins over the trunk (cafe *au lait* macules) and multiple freckle like macules (axillary freckles) in both axillae. He also had multiple soft nodules in the skin (mollusca fibrosa) which were widely dispersed over trunk and limbs. An ophthalmological examination revealed multiple pigmented iris hamartomas (Lisch nodules) in both eyes. A clinical diagnosis of Neurofibomatosis type I with plexiform neurofibroma of right hand was made. A biopsy from lesion on right hand showed a whorled proliferation of spindle shaped cells consistent with neurofibroma.

Neurofibromatosis is a genodermatosis of neuroectodermal origin characterized by multiple cutaneous turnours (mollusca fibrosa), pigmented '*cafe au lait*' macules, axillary freckles, lisch nodules in iris and variable involvement of central nervous system. The genetic defect is localised to chromosome 17 and is transmitted in an autosomal dominant pattern. Plexiform neurofibroma presents as a diffuse and elongated swelling along the course of a nerve trunk/plexus, These tend to infiltrate into deeper structures like fascia, muscles and bone. There is a localized or segmental hypertrophy of underlying soft tissue resulting in a gross deformity of the involved part. The incidence of malignant transformation into neurofibrosarcoma is upto 5% of cases. Surgical excision is the treatment of choice,

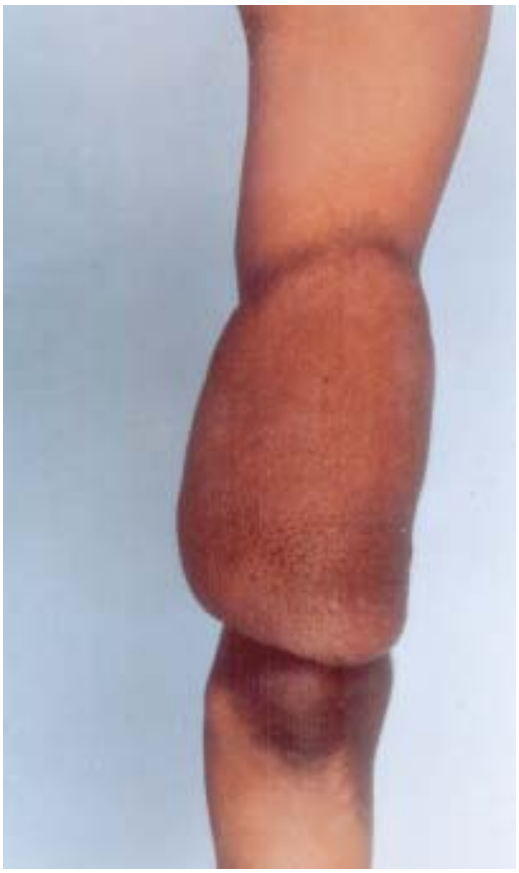

Fig. 1. Plexiform neurofibroma on right elbow.

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