Neurofibromatosis [Von Recklinghausen's disease]

Neurofibromatosis is clinically divided into Type I and Type II. Type I affects peripheral nerves and will be discussed in this section. Type II is the central type consisting of acoustic neuroma and has nothing to do with peripheral neurofibromatosis. The Type I disease is a familial dysplasia, inherited as an autosomal dominant trait, with an incidence of about one in every 3,000 births. The condition becomes clinically manifest in the first few years of life with the presence of small café-au-lait spots that increase in number and size over a time. Unlike the café-au-lait spots seen in fibrous dysplasia, those in neurofibromatosis have a smooth edge, sometimes referred to as the "Coast of California". If a patient has more than six lesions with smooth-edged café-au-lait spots greater than 1-2 cm in diameter, the diagnosis of neurofibromatosis can be made. Later in life, the patient will develop numerous cutaneous neurofibromas that are referred to as fibroma molluscum and have the appearance of small, pedunculated lipomas. The most pathognomic feature of neurofibromatosis is the large, plexiform neurofibromata associated with the larger nerves that can involve an entire extremity and be associated with loose, hyperpigmented skin that produces an elephant-like, gross distortion of the skin anatomy referred to as elephantiasis neuromatosa or elephant man syndrome. Skeletal deformation can be associated with neurofibromatosis, including scoliosis (which can be quite angular resulting in paresis), spinal meningioceles, scalloping of vertebral bodies, and pseudarthrosis of the distal tibia. There can also be associated hypertrophy or localized gigantism in the hand or foot. Unlike the solitary neurofibroma, patients with neurofibromatosis run a 10 per cent chance of developing a malignant neurofibrosarcoma that usually occurs during adult life and carries an extremely poor prognosis for survival.