Peripheral Nerve Disorders: Chapter 53. Neurofibromatosis type 1 (NF1): diagnosis and management (Handbook of Clinical Neurology)

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Neurofibromatosis 1 (NF1) is an inherited neurocutaneous disease that has a major impact on the nervous system, eye, skin, and bone. Individuals with NF1 have a predisposition to benign and malignant tumor formation and the hallmark lesion is the neurofibroma, a benign peripheral nerve sheath tumor. The gene for NF1 was cloned on chromosome 17q11.2 and neurofibromin, the NF1 protein, controls cell growth and proliferation by regulating the proto-oncogene Ras and cyclic adenosine monophosphate (AMP). Advances in molecular biology and mouse models of disease have enhanced our understanding of the pathogenesis of NF1 complications and facilitated targeted therapy. Progress has been made in developing robust clinical and radiological outcome measures and clinical trials are underway for children with learning difficulties and for individuals with symptomatic plexiform neurofibromas.

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