Neurofibromatosis type 1 (NF1)

**Record Status**
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**Authors’ objectives**
Neurofibromatosis type 1 (NF1), also known as von Recklinghausen disease, is a heritable disorder characterized by altered skin pigmentation and an increased risk for benign and malignant neurological tumors. Inherited in an autosomal dominant manner, NF1 is found in individuals of all ethnic backgrounds and races, with an average prevalence of 1 in 3500 individuals. NF1 is caused by variants in the NF1 gene, a large gene consisting of 60 exons and spanning approximately 350 kilobases on chromosome 17 at band q11.2. Approximately 50% of NF1 patients have an affected parent, while the remaining patients have the condition as the result of a de novo variant involving the NF1 gene. NF1 encodes the neurofibromin protein, a negative regulator of Ras guanosine triphosphatases (GTPases). Deleterious gene variants, the majority of which result in premature protein truncation, are often unique and distributed throughout the NF1 gene. Whole-gene deletions account for approximately 5% of NF1 cases and are typically associated with a more severe phenotype. The penetrance of NF1 gene variants is age dependent but essentially complete by the age of 20, although the condition is characterized by exceptional phenotypic variability. The most common features of NF1 include café au lait macules (CALMs; flat skin lesions of increased pigmentation), axillary (underarm) or inguinal freckling, cutaneous or subcutaneous neurofibromas (benign tumors of the peripheral nerve sheath), plexiform neurofibromas (tumors that grow along the length of the peripheral nerve), Lisch nodules (benign iris hamartomas), and intellectual disabilities. In addition, NF1 patients may develop optic pathway gliomas and malignant peripheral nerve sheath tumors. They are also at an increased risk for other types of tumors, such as brain tumors and pheochromocytomas (catecholamine-secreting tumors of the adrenal gland). Additional features of NF1 include vascular problems, hypertension, congenital heart disease, scoliosis, bone dysplasia (particularly of the tibia and sphenoid bone), seizures, short stature, and macrocephaly.

The diagnosis of NF1 is based on specific clinical criteria established by the National Institutes of Health (NIH) Consensus Development Conference in 1987, which state that a diagnosis of NF1 can be made in any individual fulfilling at least two of seven specified features (one of which is a positive family history of NF1 in a first-degree relative). Current management involves the treatment of specific disease symptoms, such as benign or malignant tumors, hypertension, and intellectual disabilities.

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