# Neurofibromatosis 1: Diagnosis and Management

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**ABSTRACT**

Von Recklinghausen or neurofibromatosis type 1 is an autosomal dominant condition that results in skin changes and noncancerous tumors along the nerves in the body. It is blind in regards to ethnicity, race, or sex. With a prevalence rate of 1 in 3,000 to 1 in 3,500 individuals, it is likely that the nurse practitioner will encounter an individual with neurofibromatosis sometime in his/her career. The aim of this article is to give a brief synopsis of the etiology, clinical manifestations, and symptomatic treatment as well as guidance on monitoring, when to refer, health promotion, and teaching of the neurofibromatosis type 1 patient.

**Keywords:** assessment, complications, neurofibromatosis, treatment



There are 2 main types of neurofibromatosis (NF), with NF type 1 (NF1) being the most common and accounting for about 90% of all cases.[1] NF1, also known as Von Recklinghausen disease, is a condition that results in skin changes and noncancerous tumors along the nerves in the body.[2] NF1 is blind in regards to ethnicity, race, or sex.[3] It is an autosomal dominant disorder caused by a single-gene mutation or deletion.[3] One half of all NF1 cases are spontaneous mutations, with the other half occurring as a result of inheritance.[4] More than 100,000 individuals or about 1 in 3,000 to 3,500 in the United States are affected.[5]

NF2 occurs less frequently and is usually diagnosed in early adulthood. It affects approximately 1 in 50,000 individuals.[6] Although the occurrence is less than NF1, because of the complications of NF2, individuals with this type have a shortened life expectancy and often experience blindness and deafness.[6] This article focuses on NF1.

## ETIOLOGY

The NF1 gene is located within the long arm of chromosome 17. The exact role of the NF1 protein product neurofibromin is not fully understood. It is known that neurofibromin plays an important role in the regulation of the activity of Ras proteins. One role of these proteins is to relay instructions to the cells to promote cell division and growth.[7] In normal individuals, neurofibromin helps inactivate Ras proteins. In individuals with NF1, neurofibromin is not present in sufficient quantities to inhibit cell growth properly. This results in the formation of neurofibromas along the nerves. These growths are generally benign but have the ability to become malignant.[8]

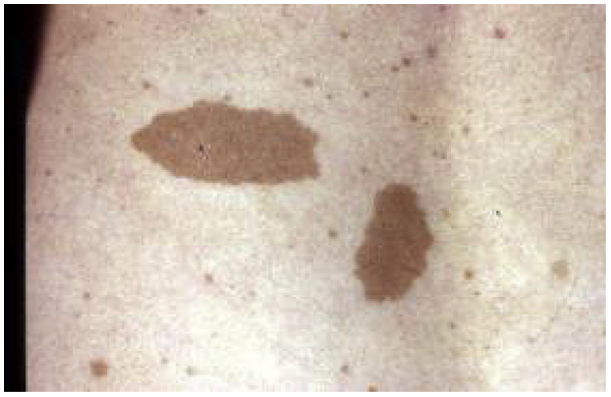
## DIAGNOSIS

Early diagnosis is challenging because of its extremely variable characteristics. Some individuals may be mildly affected showing minimal signs, whereas others are severely afflicted.[3] Typically, NF1 is diagnosed through clinical assessment including a thorough history and physical examination. These assessment findings are compared with standardized diagnostic criteria outlined by The National Institutes of Health (NIH).

The NIH developed diagnostic criteria for NF1 based on clinical features that occur commonly.[9] The diagnosis is made if an individual presents with 2 or more of the following features:

1. Six or more café au lait macules > 5 mm in prepubertal individuals and > 15 mm in diameter in adults (Figures 1 and 2)[3]

**Figure 1.** Dark cafe au lait spots.



2. Two or more neurofibromas of any type; there are 4 types: cutaneous, subcutaneous, modular plexiform, and diffuse plexiform (Figure 3)[3]
3. Freckling in the axillary or inguinal regions (not generally apparent at birth but begin to appear around 4-5 years of age)[3]
4. Optic glioma visual pathway tumors most often presenting as grade I pilocytic astrocytomas
   a. Generally grow between the ages of 15 months to 7 years of age and then become dormant[3]
5. Two or more Lisch nodules: hyperpigmented spots on the iris (present in 90% of all individuals with NF1) (Figure 4)[3]
6. Abnormal development of the spine (scoliosis), the temple (sphenoid) bone, or the tibia[6]
7. A first-degree relative (parent, sibling, or offspring) with NF1[6]

**Figure 2.** Light cafe au lait spots.

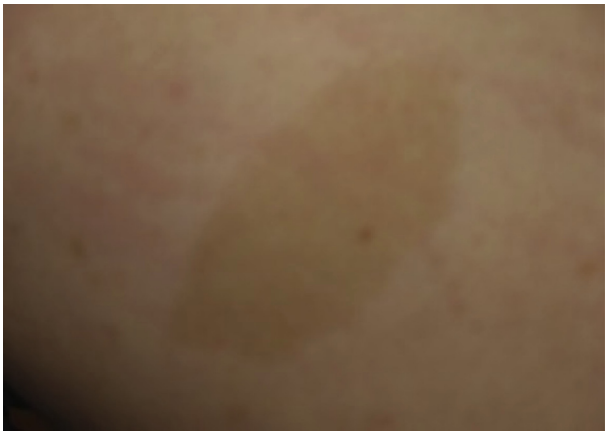
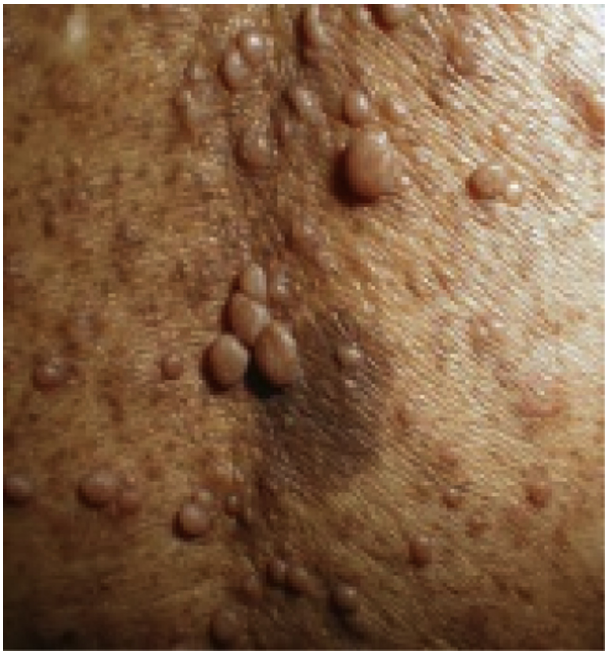


**Figure 3.** Neurofibromas.



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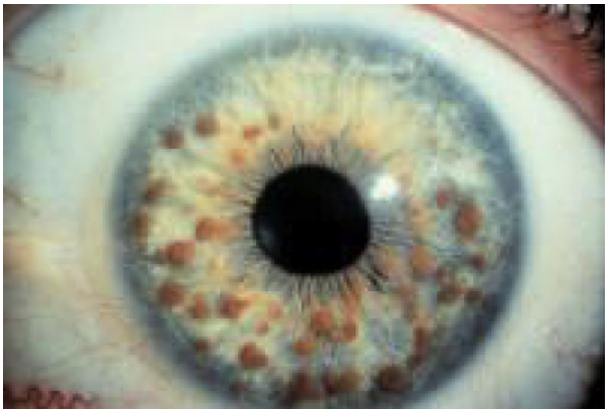
Genetic testing is available; however, it is not indicated in most cases. Because of the cost of the test and the accuracy of diagnosis through clinical assessment, it is not a necessary component of diagnosis. Genetic testing may be useful in cases in

**Figure 4.** Lisch nodules.



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