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Abstract title: Early Cutaneous Signs of Neurofibromatosis Type 1

Goal/Purpose: To provide nurse practitioners with information of early recognition of neurofibromatosis type 1 (NF 1) signs and symptoms.

Objective: Nurse practitioners will be able to recognize early signs of neurofibromatosis type 1.

Abstract

A 19-year old female presented with freckling in the axillary region. She was diagnosed with neurofibromatosis type 1 (NF 1), also known as von Recklinghausen disease, a disorder with multiple malformations and tumors involving the skin, nervous system, and skeleton. NF 1 is an autosomal dominant disorder that occurs with an incidence of approximately 1 in 3000. NF 1 affects males and females equally as well as all races and ethnic groups. It is very important for health care providers to recognize early manifestations of this disorder that can lead to learning disability, developmental delay, dysmorphic facial features, high blood pressure, short stature, scoliosis and optic gliomas. The diagnostic criteria are based on specific clinical features that include two or more of the following: six or more café-au-lait macules, two or more neurofibromas of any type, freckling in the axillary or inguinal regions, optic gliomas, two or more Lisch nodules, thinning of the long bone cortex, a first-degree relative with NF 1.

There is no one overall treatment or cure for NF 1. Individual manifestations are treated symptomatically. Screening includes: assessment of skin, eye examinations, blood pressure evaluation, assessment of growth and development, and imaging. Patients and families are encouraged to seek counseling and information on the inheritance of the disorder, prognosis, and psychosocial adjustment.