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Plexiform neurofibroma

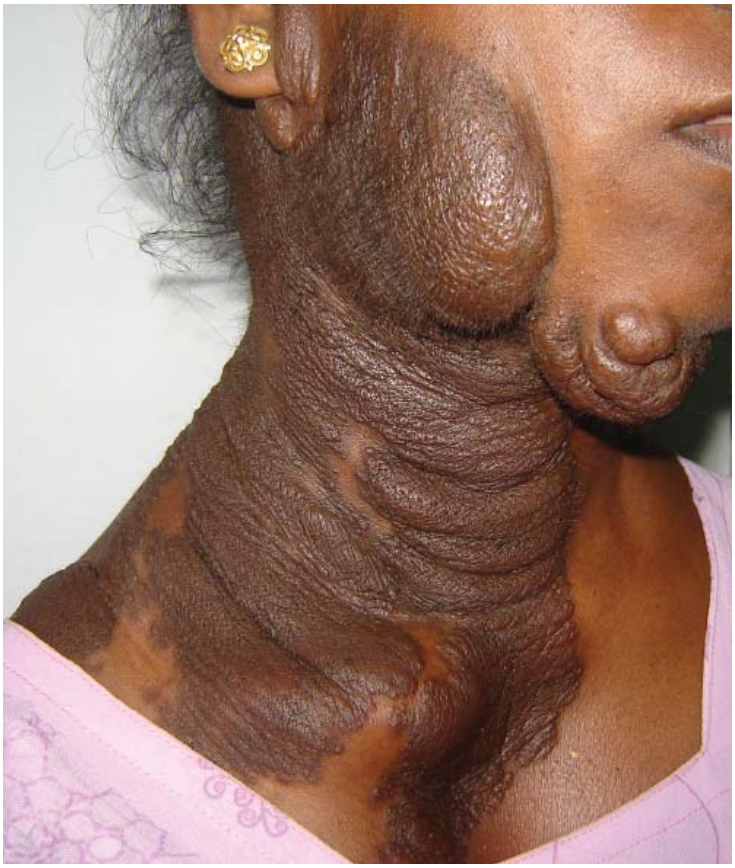

FIGURE 1. Diffuse plexiform neurofibroma.

A 19-year-old woman presented with diffuse swelling on her neck and face that had been growing gradually over the past 12 years. There was no family history of similar swelling.

Examination revealed a diffuse, hyperpigmented, nodular swelling on the right side of her neck and extending upward to the lower part of the face and the scalp (**Figure 1**). The rest of the physical examination was normal.

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A diagnosis of plexiform neurofibroma was made based on the typical clinical presentation. The patient had come primarily because of cosmetic concerns but refused surgery. She was offered genetic counseling and was scheduled for regular follow-up.

Plexiform neurofibroma is mostly associated with autosomal dominant neurofibromatosis type 1, characterized by cutaneous findings such as café-au-lait spots, axillary freckling, skeletal dysplasias (usually of the tibia, fibula, ribs, and vertebrae), Lisch nodules in the eye (iris hamartomas), and neural tumors. Rarely, it may also be seen in germline p16 mutation-positive heritable melanoma and Cowden syndrome.

Diffuse plexiform neurofibroma of the face and neck rarely appears after the age of 1, and rarely develops on other parts of the body after adolescence. In contrast, deep nodular plexiform neurofibroma often originates from spinal nerve roots and usually becomes symptomatic in adulthood.

Plexiform neurofibroma has an 8% to 12% chance of changing into a malignant peripheral nerve-sheath tumor. Continuous pain in the tumor, rapid tumor growth, hardening of the tumor, or weakness or numbness in an arm or leg with a plexiform neurofibroma suggests malignant transformation.

The diagnosis is clinical, and the management involves a multidisciplinary team including a physician, geneticist, neurologist, surgeon, and ophthalmologist. Genetic counseling should be offered to patients for assessment of family risk, to look for other possible conditions such as Cowden syndrome, for marital and family planning, and whenever the diagnosis is unclear. ■

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