



NEUROFIBROMATOSIS

<http://www.aocd.org>

What is Neurofibromatosis?

Neurofibromatosis is an autosomal dominant inherited disorder that affects approximately 1 out of 3,000 people. There are two types of Neurofibromatosis. The most common is Neurofibromatosis type 1, where 85% of the affected individuals present with skin manifestation of brown freckling spots (café-au-lait macules) on the trunk, freckling in the armpit, brown dome shape lesions (neurofibromas) on the body, and brown pigmentations in the eyes (Lisch nodules) which are present in one fourth of patients under six years old and 94% in adults. Neurofibromatosis type 2 affects hearing with a tumor called acoustic neuroma and skin manifestation is minimal.

How is Neurofibromatosis diagnosis?

If there are six or more brown freckling lesions greater than 5mm in diameter in prepubescent age or six or more lesions greater than 15mm in diameter in adults, then there is presumptive diagnosis of Neurofibromatosis. Thus, the patient should visit a dermatologist for further evaluation if they have these skin findings.

What are some systemic manifestations of Neurofibromatosis?

Patients with Neurofibromatosis may have signs and symptoms of constipation, headache, mentally challenged, crooked spine, enlarge head, malignancies, premature or delayed puberty, seizures, speech impediment, short stature, and tumors in the brain.

Who should get genetic counseling?

Offsprings of patients with Neurofibromatosis have a 50% chance of inheriting this autosomal dominant disease. Thus, all family members and relatives should be screen for the brown pigmentations in the eyes (Lisch nodules), brown freckling spots (café-au-lait macules) on the body, and brown dome shape lesions (neurofibromas).

What are the support system and management for Neurofibromatosis?

There are many academic centers in the U.S. that have specialists that provide the management of Neurofibromatosis in a multi-disciplinary approach. Patients can search the **Children's Tumor Foundation** website to find the clinic closest to their location. Patients should be monitored closely to detect malignancies. Skin tumors can be surgically removed. Lastly, magnetic resonance imaging can be used for screening family members.

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