Neurofibromatosis Type 1

Neurofibromatosis type I, previously called von Recklinghausen disease, is an autosomal-dominant disorder characterized by a malfunction of the neurofibromin 1 gene on chromosome 17. Patients typically present with neurofibromas, optic gliomas, Lisch nodules, hamartomas and cafe au lait spots. The neurofibromas in patients with neurofibromatosis type I have a higher propensity to undergo malignant degeneration at an increased rate as compared to the general population. The course of disease is extremely variable with some patients displaying minimal symptoms, while others develop progressive disease with spinal deformities, such as scoliosis and disfiguring skin lesions. Patients with neurofibromatosis type I also have a higher incidence of pheochromocytomas than the general population.



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Pathophysiology

Autosomal Dominant
Domino
This disease is inherited in an autosomal- dominant fashion.

Chromosome 17
Sign Labeled 17 with Chromosome
Neurofibromatosis type I is characterized by malfunction on the neurofibromin 1 gene on chromosome 17.

Signs and Symptoms

Neurofibromas
Neurofibromas on Body
A neurofibroma is a lesion or enlargement of the peripheral nervous system thought to derive from Schwann cells. They can arise sporadically or in association with neurofibromatosis type I and can infiltrate and encase major structures in the body.

Optic Glioma
Glowing Eye
A glioma is a tumor type that originates from glial cells in the CNS. An optic glioma is a glioma that affects the optic nerve and is one of the manifestations of neurofibromatosis type 1.

Lisch Nodules
Fish Knobs
Lisch nodules are pigmented nodules that project from the surface of the iris. While they commonly do not affect vision, they can be useful in diagnosing neurofibromatosis type I.

Hamartomas
Hammerhead-shark
Hamartomas are focal malformations in tissue that resemble a neoplasm, but they are benign and grow at the same rate as surrounding tissues. Lisch nodules are commonly described as hamartomatous, and are even referred to as iris hamartomas.

Cafe Au Lait Spots
Coffee-cow
Cafe au lait translates from French to “coffee with milk.” These hyperpigmented macules are commonly considered to be a type of pigmented birthmark. However, having six or more café au lait spots >5 mm in diameter before puberty, or >15 mm after puberty, is a major diagnostic feature of neurofibromatosis type I.

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Scoliosis

Curved-spine and Skull

Scoliosis, which is an abnormal, sideways curvature of the spine, is a common manifestation of neurofibromatosis type I.

Pheochromocytoma

Phiat-chrome

A pheochromocytoma is an adrenal gland tumor that secretes the catecholamines epinephrine and norepinephrine. It can lead to severe, even life-threatening hypertensive episodes with associated symptoms. Patients with neurofibromatosis type I have a higher incidence of pheochromocytomas than the normal population.

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