

## Von Recklinghausen Disease (Neurofibromatosis Type 1) and Lisch Nodules: Clinical Imaging of the Body and Retina in a 66-Year-Old Patient

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### IMAGE DESCRIPTION

In this 66-year-old patient, clinical examination reveals multiple cutaneous neurofibromas scattered across the trunk and limbs, typical of von Recklinghausen's disease. Also noted are large café-au-lait spots and axillary lentiginosities. Ophthalmic examination shows Lisch nodules in the iris (pigmented melanocytic hamartomas), visible on slit-lamp examination. Retinal imaging (OCT and angiography) may reveal astrocytic hamartomas or cotton-wool spots, although these are often asymptomatic. No optic glioma is observed in this elderly patient.

### Mode of Transmission

NF1 is transmitted in an autosomal dominant pattern. The NF1 gene (17q11.2) encodes neurofibromin. The patient has an affected child and belongs to a sibship of three individuals (at least one other sibling is likely a carrier and should be evaluated).

### Abnormalities to Look For

Given this presentation, one must systematically search for: spinal neurofibromas with compression (spinal MRI), central nervous system tumors (optic gliomas, astrocytomas), vascular abnormalities (arterial stenosis), scoliosis, pseudoarthrosis, pheochromocytoma, and possible cognitive impairments.

### Proposed Treatment

There is no curative treatment. Management is multidisciplinary: surgical removal of bothersome or suspicious neurofibromas (risk of malignant schwannoma), annual ophthalmologic follow-up, and monitoring with brain and spinal MRI. In the affected child, monitoring for learning disabilities is recommended. Genetic counseling is offered to the siblings.



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