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Neurofibromatosis: Symptoms, Diagnosis and Treatment

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Commentary

Neurofibromatosis is a nervous system genetic condition. Tumors develop on the nerve tissues. Neurofibromatosis problems primarily influence nerve cell tissue growth and development. Neurofibromatosis type 1 (NF1) and neurofibromatosis type 2 (NF2) are the two diseases (NF2). The most frequent kind of neurofibromatosis is NF1. Schwannomatosis, a third and more uncommon variety of neurofibromatosis, was recently discovered, but physicians still don't know much about it [1-5].

Recklinghausen disease, Von Recklinghausen disease, Von Recklinghausen's phakomatosis, Von Recklinghausen's neurofibromatosis, neurofibromatosis (multiple), neurofibromatosis-pheochromocytoma-duodenal carcinoid syndrome, or peripheral neurofibromatosis are all terms used to describe NF1. Multiple café au lait spots (tan or light brown patches of skin) and neurofibromas (soft, squishy growths) appear on or beneath the skin. It can also result in enlarged or misshapen bones, as well as spine curvature (scoliosis). Tumors can form in the brain, on the cranial nerves, or in the spinal cord. Learning problems affect 50 percent to 75 percent of patients with NF1.

Bilateral acoustic neurofibromatosis, vestibular schwannoma neurofibromatosis, and central neurofibromatosis are all names for NF2. Multiple tumours on the cranial and spinal nerves define this disease, which is far less prevalent than NF1. The initial signs of NF2 include tumours that damage both auditory nerves, as well as hearing loss that begins in the teens or early 20s. A person with NF has an average life expectancy of around 8 years fewer than the general population. Individuals with neurofibromatosis type 1 have a higher lifetime risk of both benign and malignant malignancies (NF1).

Neurofibromatosis symptoms

- Several café au lait places (typically 6 or more)
- Several freckles in the armpit or groyne
- Lisch nodules are little growths in the iris (coloured part) of the eye that normally do not impede vision.
- Neurofibromas are benign tumours that develop on or beneath the skin, and occasionally even deep into the body. These tumours are benign (harmless). They can, however, become malignant or cancerous in rare situations.
- Deformities of the bones, such as a twisted spine (scoliosis) or bent legs

Neurofibromatosis causes

Neurofibromatosis is a condition that is frequently hereditary (passed on by family members through your genes). However, roughly half of those newly diagnosed with the illness have no family history of it. This is because a sudden mutation (change) in your DNA might trigger it. You can transmit the mutated gene on to future generations after this transformation occurs. Neurofibromatosis is caused by a variety of genetic mutations, including:

Tumors that develop along the optic nerve and cause vision issues

- **Neurofibromatosis 1 (NF1):** The NF1 gene on chromosome 17 produces neurofibromin, a protein that regulates cell development. This gene mutation results in a lack of neurofibromin and unregulated cell proliferation.
- **Neurofibromatosis 2 (NF2):** The NF2 gene on chromosome 22 produces the protein merlin, also known as schwannomin. Tumors are suppressed by it. The loss of merlin and unregulated cell proliferation are caused by changes to this gene.
- **Schwannomatosis:** This kind of neurofibromatosis is connected to mutations in the two known genes linked to schwannomatosis, SMARCB1 and LZTR1, which both inhibit tumours.

Neurofibromatosis diagnosis

Neurofibromatosis is diagnosed using a number of tests, including:

- Physical examination
- Medical history
- Family history
- X-rays
- Computerized tomography (CT) scans
- Magnetic resonance imaging (MRI)
- Biopsy of neurofibromas
- Eye tests
- Tests for particular symptoms, such as hearing or balance tests
- Genetic testing

Neurofibromatosis treatment

There's no cure for neurofibromatosis. Treatments focus on controlling symptoms. There's no standard treatment for NF, and many symptoms, such as café au lait spots, do not need treatment. When treatment is necessary, options may include:

- Surgery to remove problem growths or tumors
- Treatment that includes chemotherapy or radiation if a tumor has turned malignant, or cancerous
- Surgery for bone problems, like scoliosis
- Therapy (including physical therapy, counseling, or support groups)
- Cataract removal surgery
- Aggressive treatment of pain linked to the condition
- Stereotactic radiosurgery
- Auditory brainstem and cochlear implants

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