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Neurofibromatosis.

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Perspective

Rather than being a single disease, neurofibromatosis encompasses three distinct diseases in which tumours form in the brain, spinal cord, and the peripheral nerves that carry messages between the brain, spinal cord, and the rest of your body.

- Type 1 Neurofibromatosis (NF1), formerly known as von Recklinghausen illness
- Type 2 Neurofibromatosis (NF2)
- A disease known as Schwannomatosis (SWN)

The majority of tumours are benign (noncancerous), but a small percentage of them go on to develop into cancer (malignant). Tumors in these patients are not fully understood, but it appears that gene alterations that play important roles in inhibiting the development of nervous system cells are a contributing factor. Because of these mutations, the genes responsible for controlling the capacity of cells to operate normally-NF1, NF2, SMARCB1, and LZTR1-cannot make normal proteins. Tumors arise when these proteins stop performing their usual functions in the body. These tumors are generally non-cancaerous.

Neurofibromatosis affects men and women of all races and ethnicities equally. Certain oncogenes have undergone genetic mutation as a result of the problem. These can be passed down through genetics, or they might emerge on their own throughout a person's formative years. The three distinct NF subtypes are the consequence of various mutations. However, the nerve cells that support the neurons are what cause neurofibromatosis. Neurofibromas (tumours of the peripheral nerves) are more prevalent in NF1 than in NF2 or schwannomatosis, which are tumours of Schwann cells. Symptoms, physical examination, medical imaging, and a biopsy are all used in the diagnosis process. Rarely will genetic testing be used to bolster a diagnosis.

As Neurofibromatosis has three types, each caused by a unique chromosomal abnormality.

An alteration in the NF1 gene on chromosome 17 causes the condition known as NF1. Cancer of the nervous system type 2 (NF2) is caused by an alteration in the tumor-suppressor gene NF2. The numerous mutations on chromosome 22 that lead to Schwannomatosis are the cause of the disease.

Neurofibromatosis is an autosomal dominant disease, meaning that only one copy of the faulty gene is required for the condition to manifest itself. If one of the parents has neurofibromatosis, the child has a 50% risk of getting it as well. Children with NF1 inherited from parents with various degrees of the illness will have milder forms of the disorder than those who do not. Neurofibromatosis can come in a variety of forms, including:

Nervofibromatosis type I, in which nerve tissue develops tumours (neurofibromas) that may be harmless, but which might cause significant injury by squeezing nerves and other tissues.

Type II Neurofibromatosis is characterised by the development of bilateral acoustic neuromas (tumours of the vestibulocochlear nerve or Cranial Nerve 8 (CN VIII) also known as schwannoma), which commonly results in hearing loss. Schwannomosis is a painful condition in which schwannomas grow on the spinal and limb nerves.

Early-life neurofibromatosis type 1 can lead to learning and behavioural issues; around 60% of children with NF1 have some trouble in school, according to research. Several warning signs that the person may be in danger:

- Six or more light brown dots on the skin (referred to as "café au lait spots")
- At least two neurofibromas found.
- At least two new growths on the iris of eye.
- Abnormal spine growth (scoliosis)

Type 2 neurofibromatosis patients might have skin symptoms similar to type 1 patients, although this isn't always the case. Auditory loss is the most prevalent sign and symptoms of NF2. Tumor pressure on the auditory nerve causes the hearing loss. The same amount of pressure can also produce headaches, dizziness, and nausea.

Localized pain is the most common schwannomatosis symptom. There is a lot of discomfort because of the tumours that are close by that are increasing the pressure on the tissues and nerves.

Approximately 1 in 3,500 Americans has NF1, about 1 in 25,000 has NF2, and about 1 in 40,000 has schwannomatosis, according to the CDC. In all three cases, both sexes are equally at risk. Symptoms of NF1 might appear as early as birth or develop before the age of 10. Despite the fact that the disease deteriorates over time, most NF1 patients live normal lives. Symptoms of NF2 may not appear until adolescence or early adulthood. NF2 raises the chance of premature mortality. Symptoms of Schwannomatosis usually appear in early childhood and might get worse with time. Those with schwannomatosis might expect a normal lifespan. Clinical characteristics alone may not be able to tell someone with NF2 from someone with SWN. It's possible that genetic testing is required to accurately diagnose people with symptoms of these diseases who don't have a family history or bilateral vestibular schwannomas (those that occur on both sides of the body). Some instances call for genetic testing, such as prenatal diagnosis or if a clinical diagnosis is ambiguous.

If the patient has symptoms that point to schwannomas on the

peripheral nerves, then more extensive brain and spinal-cord imaging using MRI is required.

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