

Understanding NEUROFIBROMATOSIS

Guide to Symptoms, Causes, and Support

What is Neurofibromatosis?

Neurofibromatosis refers to a group of genetic conditions involving the development of tumors that may affect the brain, spinal cord, and the nerves that send signals between the brain and spinal cord and all other parts of the body. Most tumors are non-cancerous, although some may become cancerous.

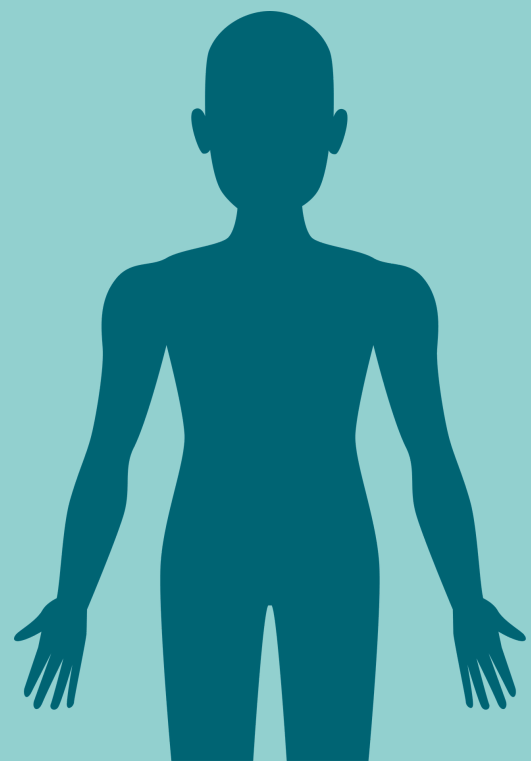
There are three main types: NF1, NF2-related schwannomatosis, and Schwannomatosis.

Causes and Inheritance

NF is an autosomal dominant condition. It means that an individual only needs one mutated copy of the gene from one parent to develop the disorder. There is a 50% probability of a parent passing the gene to their offspring.

Approximately 50% of diagnosed cases occur in individuals with no family history of the disorder. This results from a spontaneous mutation in the sperm or egg cell prior to conception.

It's not completely known why tumors develop in people with NF. However, tumor development is partly due to genetic changes that affect proteins responsible for controlling the growth of nervous system cells. Without the normal function of these proteins, cell growth increases, leading to the formation of tumors.



Common Signs and Symptoms

Café-au-lait Macules are flat, pigmented skin patches that are light brown in appearance. The presence of six or more of these spots is often a primary diagnostic indicator for NF1.

Unlike standard sun-induced freckles, NF-related freckling typically appears in skin folds, such as the armpit or groin regions.

Neurofibromas are small, soft tumors that develop on or just beneath the surface of the skin. They generally increase in number as an individual ages.

In cases of NF2, tumors often grow on the cranial nerves responsible for hearing and balance, leading to gradual hearing loss, ringing, or chronic dizziness.

Treatment and Support

There is no cure for NF, but treatments are available to help manage symptoms and address other conditions that may develop. Many people with NF won't require lengthy treatment for any single symptom during their lives. However, they should get regular screenings to detect signs and symptoms, such as routine eye and physical exams. They should be seen regularly by a specialist, even if they don't have any symptoms.

Medications

Surgery

Cancer treatment

Therapeutic support