

Frequently Asked Questions About NF



NF is a group of genetic conditions that cause tumors to grow on nerves throughout the body.



What is NF?

NF refers to a group of genetic conditions that cause tumors to grow on nerves throughout the body. The types of NF include neurofibromatosis type 1 (NF1), and all types of schwannomatosis (SWN), including *NF2*-related schwannomatosis (*NF2*-SWN). Some type of NF occurs in approximately **1 in every 2,000 births**.

What are the types of neurofibromatosis?

Neurofibromatosis 1 (NF1): This is the most common type of NF and occurs in about **1 in every 2,500** births. NF1 is characterized by six or more café-au-lait (light brown) spots and neurofibromas (small benign tumors) on or under the skin. About half of the individuals with NF1 also have learning disabilities. Some patients with NF1 develop softening and curving of bones and curvature of the spine (scoliosis). Occasionally, tumors may develop in the brain, on cranial (brain) nerves, or on the spinal cord. While NF tumors are generally benign (not cancerous), they may cause health problems by pressing on nearby body tissues. Sometimes a benign tumor may become malignant (cancerous), but 90 percent of people with NF1 will never develop a malignant tumor. NF1 is usually diagnosed in childhood.

Schwannomatosis (SWN): Schwannomatosis is an umbrella term for a group of genetic conditions in which tumors called schwannomas grow on the nerves. Schwannomas are benign, meaning they are not cancer, however, sometimes they press on nerves, blood vessels, or nearby organs, which can cause pain or other symptoms. The types of schwannomatosis are named by the genetic information used to diagnose the condition and include *NF2*-related schwannomatosis (see below), *SMARCB1*-related schwannomatosis, *LZTR1*-related schwannomatosis, 22q-related schwannomatosis, and schwannomatosis NOS (not otherwise specified) or NEC (not elsewhere classified). Some type of schwannomatosis occurs in **1 in every 20,000** births. Excluding *NF2*-related schwannomatosis, approximately **1 in 70,000** individuals are born with schwannomatosis and are usually diagnosed in adulthood.

***NF2*-related schwannomatosis (*NF2*-SWN):**

This type of NF occurs in about **1 in 25,000** people. *NF2*-SWN is usually diagnosed with the onset of hearing loss or tinnitus (ringing in the ears), which is the result of benign tumors that form on the vestibular nerve in the brain. The hallmark of *NF2*-SWN is the appearance of bilateral vestibular schwannomas, benign tumors on both sides of the vestibular nerve. People with *NF2*-SWN may also develop schwannomas in other parts of the body or may develop other types of benign brain or spinal tumors. Many people with *NF2*-SWN may develop cataracts and other kinds of eye abnormalities, although these are usually treatable. ***NF2*-related schwannomatosis was formerly called neurofibromatosis type 2,** but was reclassified under the umbrella term schwannomatosis in 2022. *NF2*-SWN is usually diagnosed in young adulthood.





Is NF1 the same as Elephant Man's disease?

No. NF is not Elephant Man's disease. For many years, scientists believed that Joseph Merrick, the so-called "Elephant Man," had NF1. However, in 1986, it was established that he had Proteus syndrome, an extremely rare condition.

Can one type of NF turn into another?

No. The types of NF are all distinct conditions.

Will NF affect someone's life expectancy?

Most people with NF will have a normal life expectancy. Some patients develop more serious complications that may shorten their lives.

Is there any way to predict how severe a person's NF will be?

It depends. The type and severity of the symptoms of NF1 vary from person to person, and cannot be predicted in advance. Even when the condition is inherited, a child's symptoms may be different from those of the affected parent. When *NF2-SWN* is inherited, however, a child's symptoms tend to be similar to those of the affected parent. When *NF2-SWN* occurs spontaneously, the type of genetic variant can predict to a certain extent what the course of the disease will be. It is not possible to predict the course of schwannomatosis at this time.

How does someone develop NF?

Half of the people who develop NF1 or NF2-SWN inherit it from a parent. The others develop it by chance, as the result of a spontaneous change in a specific gene in an egg or sperm cell. Every person affected by NF1 or NF2-SWN has a 50 percent chance of passing the condition on to their offspring. Schwannomatosis is less well understood, but the majority of cases appear to occur by chance, not because they are inherited.

Is there a blood test for NF?

A blood test is available for all forms of NF. This test determines if someone has one of the gene changes responsible for these conditions. These tests are not done routinely because a clinical diagnosis (based on observable signs of NF) is considered reliable in most cases. Sometimes genetic testing may be used to confirm a clinical diagnosis. The decision of whether or not to pursue genetic testing is a personal one. It is important to speak with a qualified healthcare professional about the appropriate risks, benefits, and limitations of testing.



Is there a cure?

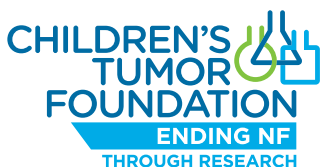
There is no cure at this time for NF, however, the US Food and Drug Administration (FDA) approved the first-ever treatment for NF in 2020, Koselugo (selumetinib), for inoperable plexiform neurofibromas. Clinicians and scientists are optimistic that with funding for more research, more treatment options to improve the lives of the millions of people living with NF will be attainable.

What should someone do if they think they have NF?

Only a trained healthcare professional familiar with the condition can make a diagnosis of NF. If you think you or your child has NF, contact a doctor for an evaluation. You can find a list of NF specialty clinics across the country by contacting the Children's Tumor Foundation at 800-323-7938 or by visiting **ctf.org/doctor**.

What other resources are available?

The Children's Tumor Foundation (CTF) offers many opportunities to help you manage your NF, learn more, feel supported, and know that you are not alone. Founded in 1978, CTF began as the first grassroots organization dedicated solely to the goal of finding treatments for NF. Today, the CTF is a highly recognized global nonprofit foundation, the leading force in the fight to end NF, and a model for other innovative research endeavors. Learn more by visiting our website at **ctf.org**.



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