How does someone develop neurofibromatosis?
Half of the people who develop NF1 or NF2 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 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 three forms of NF. This test determines if someone has the gene responsible for these disorders. 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 genetic health care 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 actionable treatment options to improve the lives of the 2.5 million people living with NF will be attainable.

What should someone do if they think they have NF?
Only a trained health-care professional familiar with the condition can make a clinical 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 the CTF website: www.ctf.org.

What other resources are available?
The Children’s Tumor Foundation, a nonprofit organization dedicated to finding effective treatments for NF through support of medical research, is the oldest and largest national foundation for NF in the United States. You can find information on NF, support groups, NF specialists, research news, educational events, family events, and many other ways to support the NF community by visiting our website: www.ctf.org.
What is neurofibromatosis (NF)?

Neurofibromatosis (NF) is a term for three genetic disorders, NF1, NF2, and schwannomatosis. NF can cause tumors to grow on nerves throughout the body.

What are the types of neurofibromatosis?

Three types of neurofibromatosis exist:

**Neurofibromatosis 1 (NF1):** This is the most common type of neurofibromatosis. NF1 occurs in about 1 in 3,000 people. 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 people with NF1 also have learning challenges. 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 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.

**Neurofibromatosis 2 (NF2):** This type of neurofibromatosis occurs in about 1 in 25,000 people. NF2 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 is the appearance of bilateral vestibular schwannomas, benign tumors on both sides of the vestibular nerve. People with NF2 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 may develop cataracts and other kinds of eye abnormalities, although these are usually treatable. NF2 is usually diagnosed in young adulthood.

**Schwannomatosis:** This type of neurofibromatosis affects about 1 in 40,000, or even fewer, people. Individuals with schwannomatosis may develop benign tumors, called schwannomas, on nerves anywhere in the body. These tumors may cause pain that may be hard to manage. Schwannomatosis is usually diagnosed in adulthood.

Is neurofibromatosis the same as Elephant Man’s disease?

No. Neurofibromatosis 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 three types of NF are all distinct disorders.

Will NF affect someone’s life expectancy?

Most people with NF will have a near 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 is inherited, however, a child’s symptoms tend to be similar to those of the affected parent. When NF2 occurs spontaneously, the type of genetic mutation can predict to a certain extent what the course of the disorder will be. It is not possible to predict the course of schwannomatosis at this time.