

Fact Sheet:

Neurofibromatosis Type 2



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Neurofibromatosis Type 2 (NF2), also called Bilateral Acoustic Neurofibromatosis, is very different from NF1 and a much rarer genetic disorder affecting about 1 in 40,000 births. Alterations in the NF2 gene cause Neurofibromatosis Type 2. Normally, the NF2 gene produces a protein that inhibits cell division particularly in the specialized cells that insulate the nerve cells of the brain and spinal cord (Schwann cells). The altered gene produces an abnormal protein that cannot properly control cell division, leading to tumour development.

Onset is unique to each individual with NF2. Some get their first symptoms during late teenage years or in their early 20's. A few people develop symptoms in childhood and some do not have problems until their 40's.

NF2 is characterized by benign tumours that grow on the cranial and spinal nerves. There are few visible signs of NF2; however, there are symptoms that can indicate the presence of the disorder. NF2 often causes slow-growing, benign tumours to grow along the eighth cranial nerve, which leads from the brain to the inner ear. These tumours are called acoustic neuromas or vestibular schwannomas. The most common early symptoms are hearing loss, ringing in the ears (tinnitus), and loss of balance caused by tumours growing on the nerve from the ear to the brain. If tumours are growing in other parts of the brain, signs and symptoms vary according to location and can include seizures, changes in vision or sensation, and fluid build up in the brain.

Although tumours on the eighth cranial nerve are most common, persons with NF2 can also develop tumours on other nerves also. These tumours are called schwannomas because they arise from the Schwann cells. Schwann cells support and protect nerve cells and provide nerves with the insulation they require to conduct information. The symptoms of a schwannoma will depend on its location in the body. The dumbbell-shaped spinal cord schwannoma is a particularly troublesome, yet common, tumour that forms in people with NF2. As the tumour grows along the spinal cord, it can cause numbness, tingling, pain, and weakness in various parts of the body. If left untreated, the tumours may even cause paralysis.

In addition to schwannomas, persons with NF2 occasionally develop other sorts of tumours which grow on the coverings of the brain and spinal cord. These tumours (called ependymomas and meningiomas) may cause many different kinds of neurological symptoms depending on their location. As with schwannomas, a physician may detect signs of a tumour on a detailed neurological examination before a patient can detect symptoms in everyday life.

A person with NF2 can also develop cataracts at a relatively early age but these cataracts may be asymptomatic. Usually, cataracts are found in people over 50, but in patients with NF2, "juvenile cataracts" can form much sooner. Cataracts are a clouding of the eye's lens and on occasion make distance vision difficult or cause problems with glare.

How is NF2 Diagnosed?

NF2 is generally a clinical diagnosis. Although genetic tests are available for NF2, the detection rate is only between 65% and 70%.

The most useful imaging modality for NF2 is magnetic resonance imaging (MRI). It is the mainstay for diagnosis and screening for central nervous system tumours and spinal cord tumours that commonly occur with this disorder. Typically, NF2 patients will have an annual MRI study of the head, but screening for spinal cord tumours may be done less frequently. Some doctors may recommend MRIs of the spine only when symptoms occur.

How do people inherit NF2?

NF2 is inherited in an autosomal dominant pattern which means only one copy of the altered gene is necessary to cause the disorder. In about half of all cases, an affected person has one affected parent. The other half result from new alterations which means that neither parent is affected. All individuals have a 50% chance of passing the disorder on to their children. NF2 affects both genders equally and is located on the 22 chromosome.

What treatments are available for NF2?

Presently, the only treatments available for the tumours of NF2 are surgery and radiation therapy. Most individuals with NF2 require at least one operation during their lifetime. Since these tumours lie on nerves and or near the brain and spinal cord, their surgical removal is not without risk. Surgery in small and delicate places may cause further injury to nerves and further neurological problems. For these reasons an attempt should be made to postpone surgery until the risk of further damage by the tumour itself out weights the risk of possible surgical damage. When surgery is no longer an option for a particular person because of their medical problems or the size or location of a tumour, radiation therapy may be considered. As with any surgery, radiation therapy has both risks and benefits which must be carefully considered.

There are two very common tests that may be helpful in defining its nature and progression. An MRI (magnetic resonance imaging) scan and audiometry (a hearing test). MRI scans are used to visualize the anatomy of the body. They are most commonly taken of the brain, but may also be used to “see” the spine or nerves in the arms and legs.

Emotional impacts often accompany a diagnosis of NF2 for the individual, as well as family and other loved ones. Anxiety and fear can be alleviated with responsible guidance and support of appropriate medical professionals and the utilization of peer support and community resources. Community resources are a vital link to possible assistance in meeting the needs of an individual. Keep open dialogue with your doctor, become educated about NF2, and reach out to your community. Help is available.

MRI (Magnetic Resonance Imaging)

To scan the patient has to lie very still on a small bed which slides into a donut-shaped machine. Magnets are activated around the patient. At some point the patient may be injected with a dye that enhances the appearance of some parts of the brain. No x-irradiation is used. Although MRI scans can produce very detailed structural information (what your body looks like), they cannot show functional information (how well your body is working). Audiometry will show how well the hearing portion of the eighth cranial nerve is working.

Audiometry

To undergo audiometry a person puts on earphones in a soundproof room. Sounds of different intensity and frequency are given to each ear and the ability to detect them is monitored. Information from the audiometry augments the structural information from an MRI.