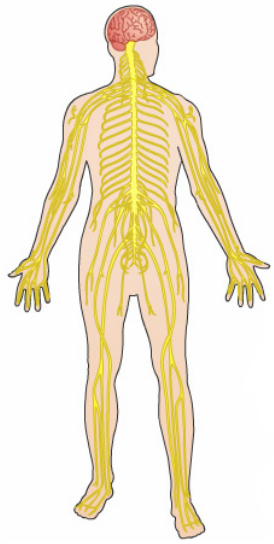
Neurofibromatosis

Introduction

Neurofibromatosis is a genetic disorder that affects the ways cells grow in your nervous system. It causes tumors to grow on nerves. There is no cure for neurofibromatosis.



Nervous System

Most people with neurofibromatosis only have mild symptoms. But, in rare cases, the tumors may become cancerous. Severe symptoms of neurofibromatosis may require surgery.

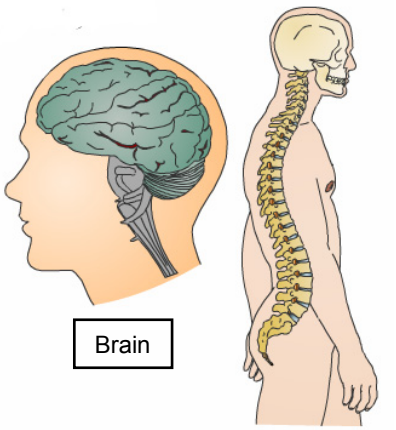
This reference summary explains neurofibromatosis. It discusses symptoms, complications and treatment of the disorder.

Neurofibromatosis

Neurofibromatosis is a genetic disorder. This means the disorder is caused by changes in a person's genes. Genes tell the body to make certain substances.

- Neurofibromatosis changes the way cells grow in your nervous system. It causes tumors to form on nerve tissue. These tumors may develop anywhere in your nervous system, including the:
- Brain.
- Spinal cord.
- Peripheral nerves.

The brain is the control center of the body. It controls the five senses, as well as the ability to move, think and speak.



Brain

Spine

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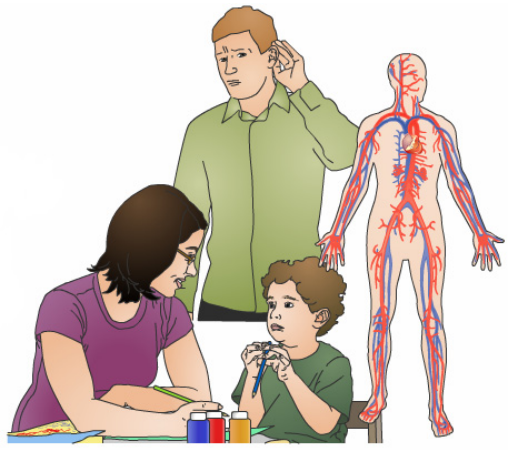
A network of nerves carries messages back and forth between the brain and the rest of the body. This includes the spinal cord. The spinal cord is protected by the vertebrae, or bones of the spine. Peripheral nerves are the nerves outside the brain and spinal cord. They bring signals from the eyes, ears, face, arms, legs, muscles and the rest of the body to the brain.

There are three types of neurofibromatosis. They include:

- Type 1, or NF1. This type usually starts in childhood.
- Type 2, or NF2. This type usually starts during the teen years.
- Schwannomatosis. This type affects people in early adulthood.



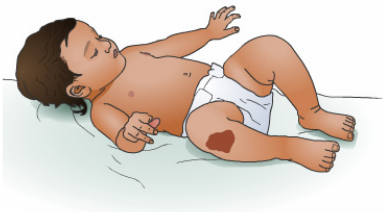
Symptoms

People with neurofibromatosis often have only mild symptoms. Neurofibromatosis may cause:

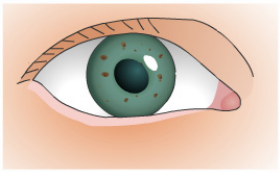
- Hearing loss.
- Learning problems.
- Heart and blood vessel problems.

Severe neurofibromatosis may cause nerve compression. This can lead to deafness, strokes, chronic pain or loss of vision.

The symptoms of neurofibromatosis are different depending on which type you have. They can also vary from person to person. Symptoms of NF1 begin in childhood. They include:



- Flat, light brown spots on the skin, known as café au lait spots. Café au lait spots are very common. They are harmless. The spots are usually present at birth or appear during the first years of life.
- Freckles in the armpits or groin area.
- Soft bumps on or under the skin, called neurofibromas. Neurofibromas are benign, or noncancerous, tumors. They can be located anywhere in the body. Most develop in the skin or under the skin. But they can also grow inside the body. If a neurofibroma affects more than one nerve, it is known as a plexiform neurofibroma.



- Tiny bumps on the iris of the eye, called Lisch nodules.

NF1 may also cause:

- Below average height.
- Bone deformities, such as scoliosis or bowed legs.
- Larger than average head size.
- Learning disabilities, such as problems with language or ADHD.

ADHD stands for Attention Deficit Hyperactivity Disorder. Symptoms include getting distracted easily, fidgeting or squirming a lot and impatience.

NF2 is much less common than NF1. Signs and symptoms of NF2 are usually caused by tumors at the base of the brain on the nerves that go to the ears. These tumors are called acoustic neuromas or vestibular schwannomas.

Acoustic neuromas are benign tumors. But they can affect the nerves they grow on. These nerves carry sound and balance signals to the brain.

Symptoms of NF2 often begin in the late teen and early adult years. They include:

- Gradual hearing loss.
- Poor balance.
- Ringing in the ears.

Sometimes, NF2 can cause tumors to grow in other nerves of the body. This can cause:

- Facial droop.
- Numbness and weakness in the arms or legs.
- Pain.
- Vision problems.

Causes

Neurofibromatosis is caused by genetic abnormalities. The genes that cause abnormalities may be inherited, meaning that they are passed down from family member to family member. But they may happen spontaneously.

Last reviewed: 12/24/2018

Neurofibromatosis happens when certain genes that make proteins to regulate cell growth do not work properly. The gene does not make enough of the protein. This allows cells to grow in an uncontrolled way.

The biggest risk factor for neurofibromatosis is a family history of the disorder. About half of NF1 and NF2 cases are inherited. About 15% of Schwannomatosis cases are inherited.

Diagnosis

Neurofibromatosis is usually diagnosed in childhood or early adulthood. To diagnose the condition, your health care provider will perform a physical exam. He or she will ask questions about your family and medical history.

Genetic tests can diagnose NF1, NF2 and schwannomatosis. Your DNA will be sent to a lab to be examined. Genetic tests include:

- A blood sample.
- A cheek swab.

Prenatal genetic tests, such as amniocentesis, can confirm neurofibromatosis in a developing baby. Amniocentesis is a genetic test that examines the amniotic fluid. Amniotic fluid surrounds and protects the baby in the womb.

Imaging tests, such as x-rays, CT scans and MRIs, may be used identify:

- Bone abnormalities.
- Tumors in the brain or spinal cord.
- Small tumors throughout the body.

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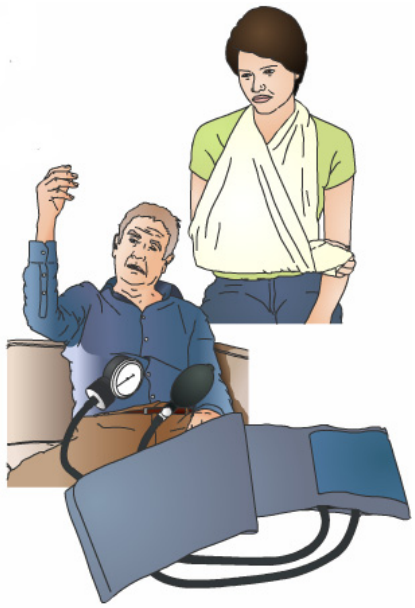
Complications

Complications of neurofibromatosis usually happen when tumor growth harms nerve tissue or puts pressure on internal organs.



Complications of NF1 include:

- Bone fractures or weak bones.
- Cancer.
- Epilepsy and stroke.
- High blood pressure.
- Hydrocephalus, the buildup of excess fluid in the brain.

Epilepsy is a group of disorders that cause problems in the normal functioning of the brain. These problems can produce seizures, unusual body movements, a loss of consciousness or changes in consciousness, mental problems or problems with the senses.

The tumors caused by neurofibromatosis are usually benign. But in some cases these tumors become malignant, or cancerous.

People with NF1 have a higher risk of other forms of cancer, such as:

- Brain tumors.
- Breast cancer.
- Leukemia.
- Soft tissue cancers. Soft tissue cancers begin in the muscles, fat, fibrous tissue or blood vessels in the body.

Complications of NF2 include:

- Brain and spinal tumors that require surgery.
- Facial nerve damage.
- Partial or total deafness.
- Skin lesions.

Last reviewed: 12/24/2018

Treatment

Neurofibromatosis treatment focuses on healthy growth, easing symptoms and managing complications. There is no cure.
If you have schwannomatosis, medications can help control your pain.

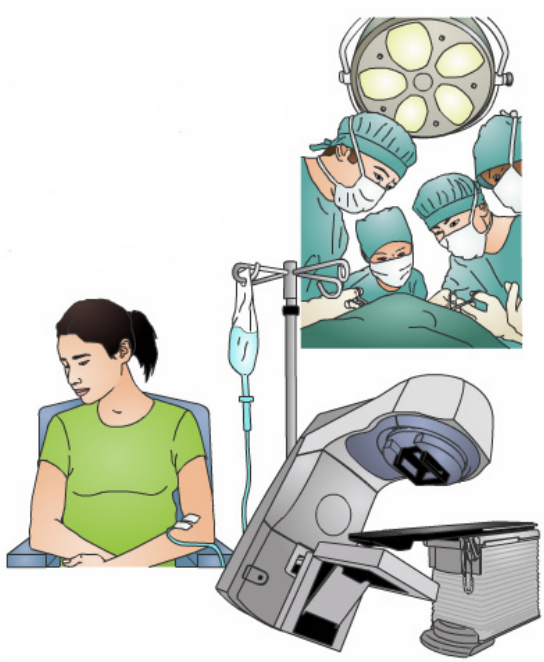
Neurofibromatosis can cause large tumors, tumors that cause pain or tumors that press on a nerve or on the brain. Surgery may help relieve symptoms caused by these types of tumors.

If you have NF2, your health care provider may recommend surgery or stereotactic radiosurgery.

If you have NF2 and hearing loss, hearing aids and implants may help improve your hearing.

Malignant tumors and other cancers associated with neurofibromatosis are treated with standard cancer therapies. These include:

- Surgery.
- Chemotherapy. Chemotherapy is a cancer treatment that uses drugs to kill cancer cells.
- Radiation therapy. Radiation therapy is a cancer treatment that uses high-energy x-rays or other types of radiation to kill cancer cells or keep them from growing.



If you have a child with NF1, monitor your child for:

- New neurofibromas or changes in existing ones.
- High blood pressure.
- Normal physical and learning development.
- Skeletal changes or problems.
- Vision problems.

Caring for a child with NF1 can be difficult. You may find it helpful to join a support group for parents.

Last reviewed: 12/24/2018

Summary

Neurofibromatosis is a genetic disorder that affects the ways cells grow in your nervous system. It causes tumors to grow on nerves. There is no cure for neurofibromatosis. The symptoms of neurofibromatosis are different depending on which type you have. They can also vary from person to person. Most people with neurofibromatosis only have mild symptoms. But, in rare cases, the tumors may become cancerous. Severe symptoms of neurofibromatosis may require surgery.

Neurofibromatosis is caused by genetic abnormalities. The genes that cause abnormalities may be inherited, meaning that they are passed down from family member to family member. But they may happen spontaneously.

Neurofibromatosis treatment focuses on treating symptomatic tumors with surgery or radiation therapy. It also focuses on managing possible complications.

Last reviewed: 12/24/2018