

## Neurofibromatosis type 1

### What is neurofibromatosis type 1?

Neurofibromatosis (nur-oh-fie-**broe**-mah-**toe**-sis) type 1 (also called NF1) is a condition that causes skin changes as well as tumors along the nerves in the body. The tumors are **usually not cancer**.

NF1 affects one in every 2,500 babies. About 120,000 people in the United States have it. NF1 affects all races and both sexes equally.

### What causes NF1?

NF1 is a genetic condition. This means it is caused by a mutation (change) in the NF1 gene. It is present at birth, and nothing can prevent it. See the education sheet, "Genetic conditions."

About half of all people with NF1 have inherited the gene that causes it from a parent. The other half have it because of a new change in the gene. There is a blood test to identify genetic changes that can cause NF1. See the education sheet, "Genetic conditions."

When a person with NF1 has a child, there is a 50% chance that the baby will receive the NF1 gene.

### How is it diagnosed?

To diagnose NF1, the doctor will do a physical exam. **Two** of the following must be found in order to decide that someone has NF1:

- at least 6 café-au-lait spots (brown birthmarks). These are often seen on the skin in young children, and may get larger with age.
- freckles in the armpit or groin.
- two or more small neurofibromas - lumps on or under the skin. They may appear just before puberty.
- at least one plexiform neurofibroma - larger, deeper tumors around the nerves. Sometimes they cannot be seen or felt. They usually don't cause a problem, but a few may develop into cancer.
- two or more iris Lisch nodules - spots on the colored part of the eye.
- optic pathway tumor or optic glioma - tumor or thickening of the optic nerve in about 10 to 15% of affected children.
- tibial dysplasia - curved lower leg bone.
- sphenoid dysplasia - abnormally shaped bone around the eye.
- first degree relative (mother, father, bother, sister, son, or daughter) who has NF1.

Some of the signs of NF1 may not show up until later in life. This means that NF1 may not be diagnosed until later in life, especially if no one else in the family has it.

## Can other problems happen?

The following problems are more common in NF1 and need to be watched for and treated if necessary:

- learning disability - about a 50 % chance
- attention deficit disorder (ADD/ADHD)
- larger head size than average
- shorter than average
- early puberty
- high blood pressure
- scoliosis (curved spine)
- headaches

## What is the treatment?

There is no way to predict what problems a person with NF1 will have, or how serious those problems will be. The features of NF1 can be very different among people in the same family. Most people with NF1 do not have many of the health problems listed in this sheet, and live long, healthy lives.

There is no cure. Treatment depends on the problems the person has.

Café-au-lait spots and freckles are not a problem and need no treatment.

Children and adults should have their blood pressure checked regularly. High blood pressure may be caused by these rare, but treatable conditions:

- narrowing of the artery to the kidney (renal artery stenosis)
- tumor of the adrenal gland (pheochromocytoma)

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Other treatments may include:



Problem	Treatment
ADHD	Medicine and non-medicine therapies
learning disability	special education
scoliosis (curved spine) curved lower leg	early care by an orthopaedist (a doctor who specializes in bones)
neurofibromas	if painful or irritating, they can be removed
plexiform neurofibroma	watch carefully; if too large, or becomes cancer, surgery or chemotherapy may be needed
optic pathway tumor	regular eye exams; rarely need chemotherapy

## How should I care for my child?

All people with NF1 should be seen yearly by a doctor or nurse practitioner familiar with the condition, as well as an ophthalmologist (eye doctor). Referrals to other specialists may be needed.

## When should I call the clinic?

- any new and persistent pain
- concerns about vision or headaches
- learning or behavior concerns

## Questions?

This sheet is not specific to your child, but provides general information. If you have any questions, please call your clinic.

For more information about neurofibromatosis, visit:

- The Children's Tumor Foundation  
[www.CTF.org](http://www.CTF.org)
- Neurofibromatosis Inc., Minnesota  
[www.nfinc-mn.org](http://www.nfinc-mn.org)