

# Neurofibromatosis – A Rare Genetic Disorder

About one-third of people with Neurofibromatosis notice no symptoms.

## 1 Introduction

Neurofibromatosis (NF) is one of the most common genetic disorders and can affect anyone, regardless of race, gender, ethnic background or family history. NF causes tumors to form on nerve tissues and these tumors can develop anywhere in the nervous system, including the brain, spinal cord and nerves. NF is usually diagnosed in childhood or early adulthood.

## 2 Types of NF

There are three types of neurofibromatosis, each with unique signs and symptoms.

- **NF1** usually appears in childhood, with symptoms often noticeable at birth or shortly thereafter. Symptoms are typically mild to moderate, but can vary. It is characterized by patches of tan or light brown skin and soft fleshy growths (neurofibromas) on or under the skin.
- **NF2** is less common than NF1 and symptoms generally appear in the late teen to early adult

years. Because tumors tend to grow in the ears, NF2 can affect hearing and balance.

- **Schwannomatosis** is the most rare form of NF and typically affects people between the ages of 25-30. Tumors develop on the cranial, spinal and peripheral nerves, leading to chronic pain, weakness and loss of muscle.

## 3 Treatment

Although NF cannot be cured, treatments are available to manage the symptoms.

- **Careful Monitoring** – regular medical exams are important to check for symptoms and abnormalities.
- **Surgery** – your physician may recommend surgery or other procedures to treat severe symptoms or complications
- **Cancer Treatment** – NF tumors can sometimes become malignant. In these cases, standard cancer therapies would be indicated.
- **Pain Management** – drugs for nerve pain, as well as certain antidepressants, SSRI's and epilepsy drugs can help relieve pain.

---

## Did you know?

NF1 is the most common neurological disorder caused by a single gene, occurring in 1 in every 3,000 children born.

For more information on Neurofibromatosis and supportive resources, please visit <https://nfpittsburgh.org/>.

---

## References:

<https://www.mayoclinic.org/diseases-conditions/neurofibromatosis/symptoms-causes/syc-20350490>

<https://www.aans.org/en/Patients/Neurosurgical-Conditions-and-Treatments/Neurofibromatosis>

<https://www.webmd.com/pain-management/neurofibromatosis#>