



[Authoritative facts](#) about the skin from the [New Zealand Dermatological Society Incorporated](#).

[Home](#) | [Skin signs of systemic disease](#)

## Neurofibromatosis

### What is neurofibromatosis?

---

Neurofibromatosis (NF) is a genetic disorder that affects the bone, soft tissue, skin and nervous system. It is classified into 2 distinct types, neurofibromatosis 1 (NF1) and neurofibromatosis 2 (NF2). NF1 occurs in about 1 in 3000 births whilst NF2 only occurs in about 1 in 50,000 births.

NF1, also known as von Recklinghausen disease, is characterised by the presence of:

- 6 or more [café-au-lait spots](#) (defined oval-shaped light brown patches greater than 0.5cm in diameter)
- Multiple neurofibromas (tumours on, under, or hanging off the skin)
- Freckling (under the armpits and areas of skin folds such as the groin)
- Lisch nodules (tiny tumours on the iris of the eye)

NF2, also known as bilateral acoustic neurofibromatosis, is characterised by multiple tumours and lesions on the brain and spinal cord. Tumours growing on the auditory nerves that lead to hearing loss is usually the first symptom of the disease. Often this is not apparent until the late teens or early 20's.

### What causes neurofibromatosis?

---

NF1 and NF2 occur as a result of defects in different genes. NF1 is caused by a mutation on a gene located on chromosome 17 and NF2 on chromosome 22. The mutated gene can be inherited from a parent who has NF or in some cases you could be the "founder" of a spontaneously mutated gene. A parent with NF has a 50% chance of passing the gene on to each of their children.

### What are the signs and symptoms of neurofibromatosis?

---

The extent and severity of manifestations of NF vary greatly from person to person and varies within the same family.

Although isolated café-au-lait spots can be found in many people without NF, individuals with more than 5 of these have a good chance of also having NF1, particularly if they appear on the skin within the first 5 years of life. More than 5 café-au-lait spots are found in 1.8% of newborns, 25–40% of children and 14% of adults with NF1. Freckling under the armpits is a clear sign of NF1.

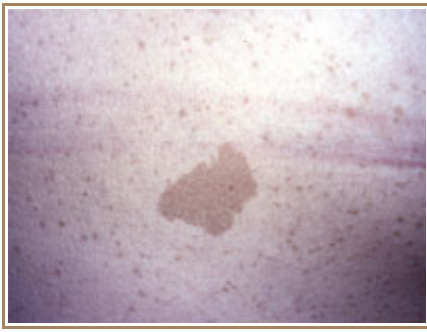
After puberty, Lisch nodules are present in 97–100% of patients with NF1. Clinically, they do not cause any problems but help to confirm diagnosis.

There are basically 4 types of neurofibromas found in NF1:

- Cutaneous: superficial, soft button-like tumours with no malignant potential
- Subcutaneous: tumours in the dermis that may cause localised pain or tenderness
- Nodular plexiform: large network of tumours involving the dorsal nerve roots
- Diffuse plexiform: invasive tumours that may involve all layers of skin, muscle, bone and blood vessels

### Neurofibromatosis

---



Café-au-lait mark



Café-au-lait mark



Neurofibroma



Neurofibromas



Freckling in the armpit



Plexiform neurofibroma

The severity of cutaneous involvement in NF1 is not an indicator of the extent of the disease as internal manifestations are common and are often more serious. Problems may occur in other parts of the body including:

- Malformation of the long bones (below the knee and elbow) and curvature of the spine (scoliosis)
- Short stature and growth hormone deficiency
- Learning difficulties (speech problems) and behavioural problems (25–40% have learning disabilities, 5–10% may have mental retardation)
- Tumours on the optic nerve which can cause visual loss
- High blood pressure and other blood problems
- Tumours on the spine and brain: increase risk of epilepsy
- Tumours or lesions on the gastrointestinal tract that may cause bleeding or obstruction
- Hearing defects

NF2 does not have as many outwardly signs as NF1 and in most instances substantial hearing loss is the first sign of possible NF2. The main problem of NF2 is the development of tumours on the brain and spinal cord.

Most tumours in both NF1 and NF2 are non-cancerous (benign). But benign tumour enlargement can interfere with vital functions. It is estimated that a person with NF1 has a 3–15% increased risk for developing cancerous tumours.

### What treatment is available?

There is no cure for NF. The main goal of treatment is to monitor its development and intervene when necessary. Healthy children with NF should be followed-up and examined every 6–12 months by a paediatrician.

Neurofibromas that become large and painful can be cut out to reduce the risk of malignancy and other complications.

Genetic counselling and education about NF is important. One concern that should not be overlooked is the risk of isolation or loneliness in people with NF. People with NF are often anxious about future complications and sometimes disfiguring lesions can lead to withdrawal from society.

## Related information

---

### References:

Book: Textbook of Dermatology. Ed Rook A, Wilkinson DS, Ebling FJB, Champion RH, Burton JL. Blackwell Scientific Publications.

### On DermNet NZ:

- [Birthmarks](#)
- [Congenital melanocytic naevi](#)

### Other websites:

- [Neurofibromatosis](#) - emedicine dermatology, the online textbook
- [The National Neurofibromatosis Foundation, Inc.](#)
- [The Neurofibromatosis Association](#)
- [Neurofibromatosis Association of New Zealand](#) (email)

**Author:** Vanessa Ngan, staff writer

DermNet does not provide an on-line consultation service.

If you have any concerns with your skin or its treatment, see a [dermatologist](#) for advice.

Created 2003. Last updated 26 Dec 2006. © 2008 NZDS. Disclaimer.