Your next steps

Find a specialist
Paediatrician, neurologists, geneticists, or NF centres can provide further testing.

Follow medical guidelines
Regular specialist checkup, eye test, and imaging or MRI are advised to be scheduled regularly.

Monitor symptoms
Keep track of new or changing physical, cognitive and mental health issues.

Consider treatment options
Discuss available therapies and surgical interventions.

Stay updated on NF research
Clinical trials may offer new treatment possibilities.

Find a patient organisation
They can offer support.

Visit **www.genturis.eu** for medical guidelines and NF treatment centres.

About ERN GENTURIS

ERN GENTURIS is a European expert network for hereditary tumour syndromes. While we do not offer direct treatment, we provide patients and healthcare professionals with trusted information, guidance, and connections to specialists.

More info and contact

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European Reference Network

Funded by the European Union

Patient information

Neurofibromatosis type 1 (NF1): what this diagnosis means for your health and next steps

If you or a family member has been diagnosed with NF1, this guide provides clear medical information and practical next steps.

European Reference Network
for rare or low prevalence complex diseases
Network Genetic Tumour Risk Syndromes (ERN GENTURIS)

ERN GENTURIS
With every diagnosis we can help an entire family

What is NF1?

Neurofibromatosis type 1 (NF1) is a complex multisystem-tumour predisposition disorder recognized as a neurocutaneous disease because of typical cutaneous stigmas. Symptoms can occur from infancy or childhood and may vary widely from person to person.

It is characterised by macules (café-au-lait spots), freckling in unusual areas (such as the armpits or groin), and can cause the growth of benign nerve tumours (neurofibromas). Some individuals may experience neuopsychological issues or bone abnormalities. Other malignancies can occur.

How is NF1 inherited?

The condition can be passed from a parent to a child. Each child of a parent with NF1 has a 1 in 2 (or 50%) chance of inheriting the condition.

However, about half of the people diagnosed with NF1 do not have a family history of the condition. In these cases, the genetic change happened for the first time in that individual.

To confirm the diagnosis and rule out other conditions that may also cause spots on the skin, a genetic test using a blood sample can be helpful.

Moreover, genetic mosaicism is common in NF1. Mosaicism describes a condition that is not present in all cells of the body. Inheritance might be reduced.

What causes NF1?

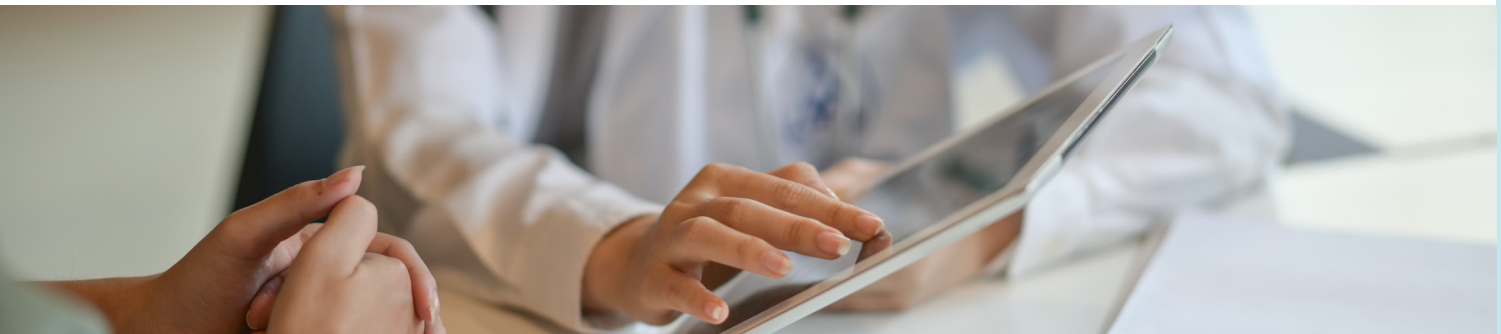
NF1 is caused by a genetic change in one of the two copies of the *NF1* gene on chromosome 17.

What are the surveillance options?

Children should be monitored on a yearly basis for vision, growth, blood pressure, skeletal problems, development and growth of (plexiform) neurofibromas, development and school career.

Depending on the presence or absence of tumours as assessed by a whole body MRI at the latest at the age of 16-18 years, adults might need a specific yearly follow-up.

Otherwise a follow-up every 3 years is strongly recommended. It is recommended that females have yearly breast MRI (preferred) or mammograms preferably starting at the age of 30 years.



How this diagnosis affects you?

Receiving a genetic diagnosis requires careful medical and psychosocial supervision. Here are key points to consider:

How will NF1 and the risk of tumour predisposition impact your health?

- The condition varies from person to person; regular medical evaluations are crucial.
- You may need (neurologic) specialist evaluation, vision tests and MRI scans over time.
- Since NF1 is genetic, family members should also consider screening.

What medical steps should I take?

- **Step 1:** schedule an appointment with an NF1-expertise centre.
- **Step 2:** discuss genetic testing and family screening options.
- **Step 3:** develop a personalised longterm surveillance plan with your doctor.

ERN GENTURIS provides information and resources to help you to understand this condition and guide your medical team toward the best possible treatment.