Your next steps

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

Follow medical guidelines
Annual control by a specialist, MRI, hearing tests and annual visit to paediatric NF2 specialist for <16 years old are advised to be scheduled regularly.

Monitor symptoms
Keep track of new or changing health issues.

Consider treatment options
Discuss available therapies and surgical interventions.

Stay updated on NF2 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

www.genturis.eu
E-mail: genturis@radbodumc.nl

European Reference Network

Funded by the European Union

Patient information

NF2-related schwannomatosis: what this diagnosis means for your health and next steps

If you or a family member has been diagnosed with *NF2*-related schwannomatosis 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 *NF2*-related schwannomatosis?

***NF2*-related schwannomatosis** is a hereditary tumour predisposition syndrome. Hallmark signes are vestibular schwannomas predominantly leading to hearing decline and balance problems and can be passed down in families. Symptoms mostly occur after the age of 10 years and may vary widely from person to person.

***NF2*-related schwannomatosis** can cause development of noncancerous tumours along the nerves called schwannomas, which nearly always occur on the hearing and balance nerves of the central and peripheral nervous system.

How is *NF2*-related schwannomatosis inherited?

The condition can be passed from a parent to a child. In about 4 out of 10 cases, a person with NF2-related schwannomatosis inherits it from a parent who also has the condition.

In the other cases, the genetic change starts in that person. This can happen in two ways:

1. All cells affected – the genetic change was already present in the egg or sperm, meaning it's in every cell of the body from the start.
2. Some cells are affected (called mosaicism) – the genetic change happened after the baby started developing, so only some cells are affected.

What causes *NF2*-related schwannomatosis?

***NF2*-related schwannomatosis** is an inherited genetic condition caused by a genetic change in one copy of the *NF2* gene on chromosome 22.

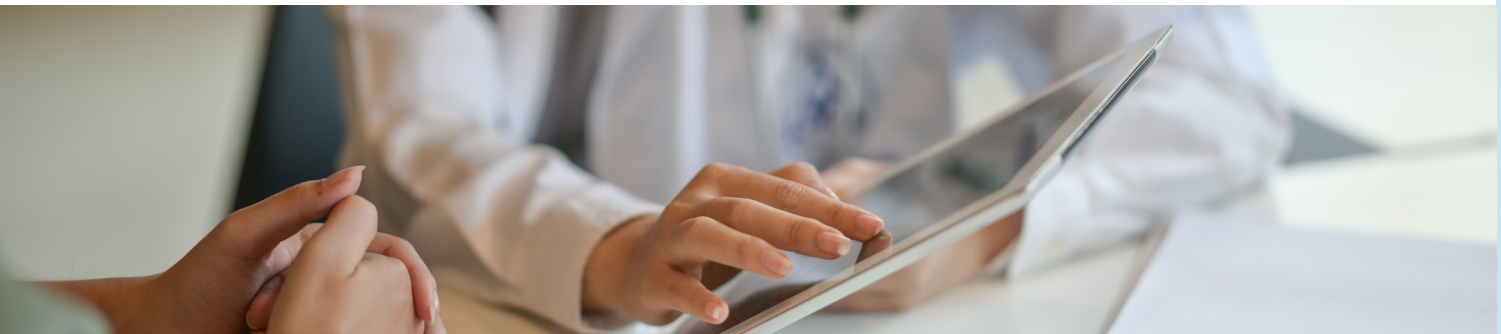
What are the surveillance options?

For families with NF2-related schwannomatosis, genetic testing can be done in relatives before they show any symptoms, to see if they carry the genetic gene change.
Once someone is diagnosed, screening is usually based on their symptoms. However, for most adults, regular check-ups include:

- A head MRI once a year
- A spinal MRI every 3 years
- Auditory assesment

Treatment options include:

- Surgery to remove tumours
- Bevacizumab, a drug that can slow the growth of fast-growing schwannomas
- Targeted radiation therapy, but this is usually not used in children.



How this diagnosis affects you?

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

How will *NF2*-related schwannomatosis impact your health?

- The condition varies from person to person; regular medical evaluations are crucial.
- You may need annual specialist control, MRI scans, hearing tests, and neurological evaluations over time.
- Since NF2 is a genetic disorder, family members should also consider genetic testing and surveilance.

What medical steps should I take?

- **Step 1:** schedule an appointment with an expert centre.
- **Step 2:** discuss genetic testing and options for relatives.
- **Step 3:** develop a personalised 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