Your next steps

- **Find a specialist**
  Neurologists, geneticists, or NF centres can provide further testing.
- **Follow medical guidelines**
  Above 20 years old MRI is 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 non-*NF2*-related schwannomatosis 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

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

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

Non-*NF2*-related schwannomatosis is a rare condition characterised by the development of benign (noncancerous) tumours called schwannomas along their nerves. These tumours usually appear on nerves outside the brain and spinal cord, especially in the arms, legs, or along the spine.

Non-*NF2*-related schwannomatosis usually becomes symptomatic after age 30. Some people with this condition only have tumours in one area of the body, such as a single arm or part of the spine - this is called segmental schwannomatosis.

A major symptom of non-*NF2*-related schwannomatosis is chronic nerve pain, which can be severe and doesn't always match the size or number of tumours. This pain can really affect daily life and varies from person to person. Other symptoms may include muscle weakness, numbness, or tingling, depending on which nerves are affected.

How is non-*NF2*-related schwannomatosis inherited?

The genetics of schwannomatosis is complicated and not yet fully understood.

Three genes are most commonly involved: *SMARCB1*, *LZTR1*, and *DGCR8*. A mutation in one copy of the *SMARCB1*, *LZTR1 or DGCR8* genes in each cell greatly increases the risk of developing schwannomas.

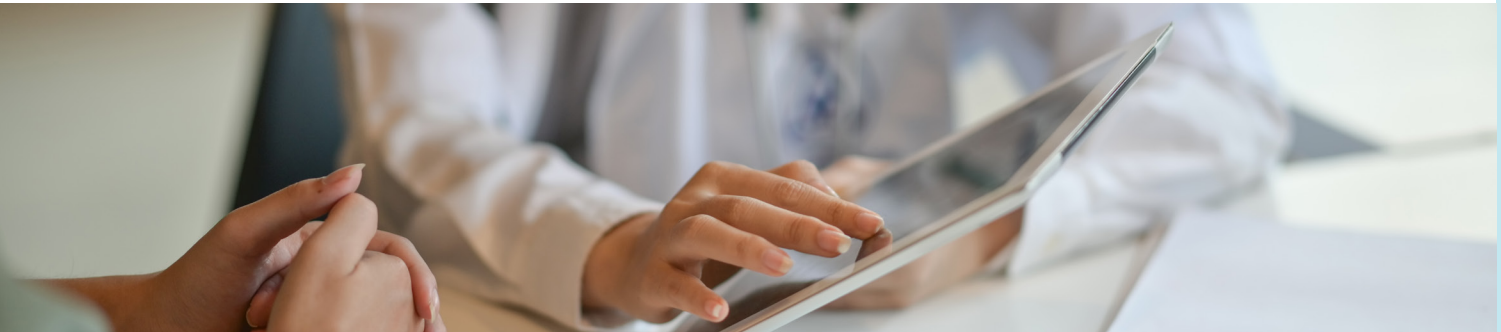
What causes non-*NF2*-related schwannomatosis?

Pathogenic variants in *SMARCB1*, *LZTR1* and possibly the *DGCR8* genes can cause non-*NF2*-related schwannomatosis. All three genes are located on chromosome 22q, near the *NF2* gene. Variants in the *SMARCB1* gene can cause multiple meningeomas.

What are the surveillance options?

People with schwannomatosis usually need regular check-ups to keep an eye on their tumours. Surgery may be needed if a tumour causes serious pain or nerve damage.

Pain is the most common and challenging symptom. Managing it usually requires a comprehensive approach, which may include medication, physical therapy, and other strategies.
Mental health support is also important - counselling or therapy can help if anxiety or depression develops.



How this diagnosis affects you?

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

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

- The condition varies from person to person; regular medical evaluations are crucial.
- You may need MRI scans to monitor brain and spine.
- Since schwannomatosis is genetic, family members should also consider screening.

What medical steps should I take?

- **Step 1:** schedule an appointment with an expert cenre.
- **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