IS THERE ANY TREATMENT FOR SCHWANNOMATOSIS?

Treatment will always depend on the signs and symptoms present for each person affected. Like the other forms of NF, there is no cure for Schwannomatosis.

While no medication has been shown to be effective in reducing the size of schwannomas, treatments typically involve surgery where possible and through the management of pain.

Because Schwannomatosis can cause pain, neurological symptoms and other potentially serious complications, it's important to seek treatment from a neurologist or neurosurgeon who has experience with the condition.

Pain Management may include the use of drugs such as Gabapentin, Pregabalin, short-acting opioids or non-steroidal anti-inflammatory drugs.

If these prove unsuccessful, surgery may be considered, but due to the risk of further neurological complications, it is often used as a last resort.

Surgical Intervention may be considered for people who have tumours that are causing neurologic or organ-related pain that have not responded to the above medications.

It may also be utilised to help lessen nerve pain; however, it can sometimes lead to an increase in painful symptoms.

Finding the right specialists can be challenging considering how rare the condition is, particularly in Australia.

ABOUT US

The Children's Tumour Foundation (CTF) is the only dedicated support service for families living with Neurofibromatosis (NF) in Australia.

NF is a life-long genetic condition with few treatment options and no cure.

The CTF exists to provide a pathway from fear to hope by investing in promising research, advocating for better resources and empowering individuals and their families with knowledge, connections and support needed at every stage of their journey.

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UNDERSTANDING

SCHWANNOMATOSIS

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WHAT IS SCHWANNOMATOSIS?

Schwannomatosis is a rare form of neurofibromatosis that has only recently been identified. It causes tumours - called *schwannomas* - to form on the spinal and peripheral nerves. Occasionally these tumours may form in the brain as well.

Schwannomas develop when Schwann cells (which form the insulating cover around nerve fibres), grow abnormally.

Schwannomatosis is more complicated than the other forms of NF and an accurate diagnosis can take some time.

However, it is thought to affect around 1 in 40,000 people worldwide.

WHAT CAUSES SCHWANNOMATOSIS?

Schwannomatosis is a genetic condition like all other forms of Neurofibromatosis (NF), but current research suggests it involves more than one gene.

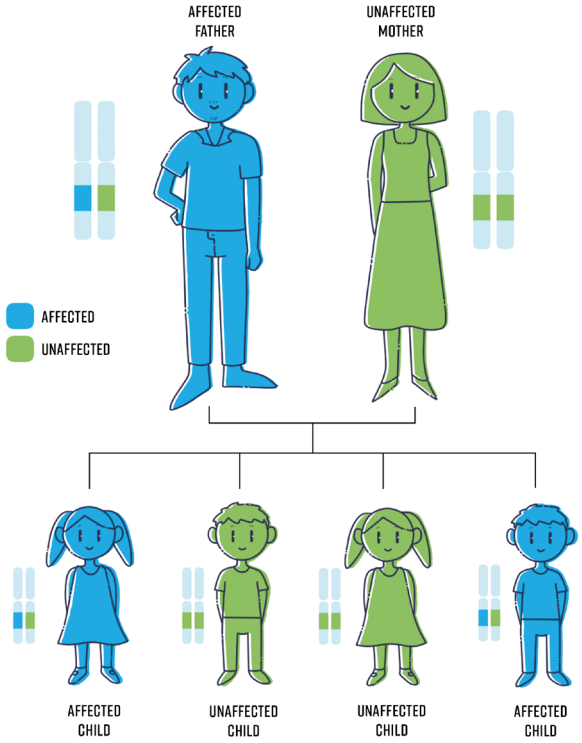
Tumour development appears to be primarily related to a change in certain genes that help regulate cell growth in the nervous system.

These genes (SMARCB1 and LTZR1) are located on Chromosome 22.

Most people with Schwannomatosis have a spontaneous change in one of at least two of the genes known to cause the condition.

Any child born to a parent who is affected by Schwannomatosis is thought to have a 50% chance of inheriting the condition.

However, not everyone with the gene change will develop tumours or symptoms related to the condition.



Infographic: example of how Schwannomatosis can be inherited.

Monitoring is vital for people with a diagnosis of Schwannomatosis.

Each person's condition will develop differently and may change quickly so prompt review and appropriate action, based on observed changes over time, is very important in managing the condition.

WHAT ARE THE SYMPTOMS OF SCHWANNOMATOSIS?

Whilst symptoms can occur at any age, they are usually detected between the ages of 30 and 60.

The most common symptom is chronic pain, which can occur anywhere in the body and does not always relate to a tumour. It is thought to be caused by tumours pressing on nerves.

In some cases, the pain that people experience is disproportionate to the size of the tumours that are present.

The intensity and frequency of pain varies significantly among individuals who are affected.

Symptoms may include:

- Numbness or Tingling
- Weakness, including facial weakness
- Bowel disfunction and difficulty urinating
- Vision changes
- Headaches

People may also see swollen areas under the skin where tumours have formed.