IS THERE ANY TREATMENT FOR NF1?

Whilst there is currently no cure for NF1, there are several options that may be considered in managing NF1 symptoms.

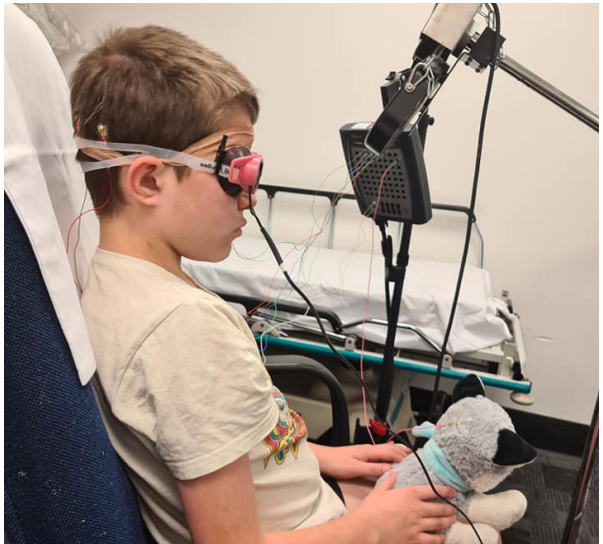
The cornerstone of NF1 management is *surveillance*. This is because the condition affects everyone so differently and the signs and symptoms that can be experienced are so varied.

Surveillance may consist of:

- Clinical examination or eye exams
- Cognitive assessments
- Ultrasound, CT or PET scans
- Magnetic Resonance Imaging (MRI) scans
- X-rays
- Blood tests

The treatment provided will depend on the symptoms and a person's individual circumstances.

- Surgery
- Chemotherapy
- Laser Treatments
- Learning and behavioural interventions
- New drug treatments

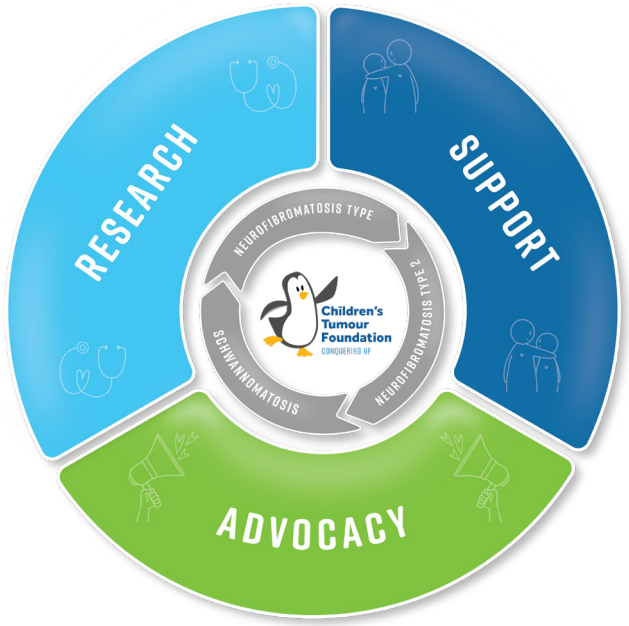


ABOUT US

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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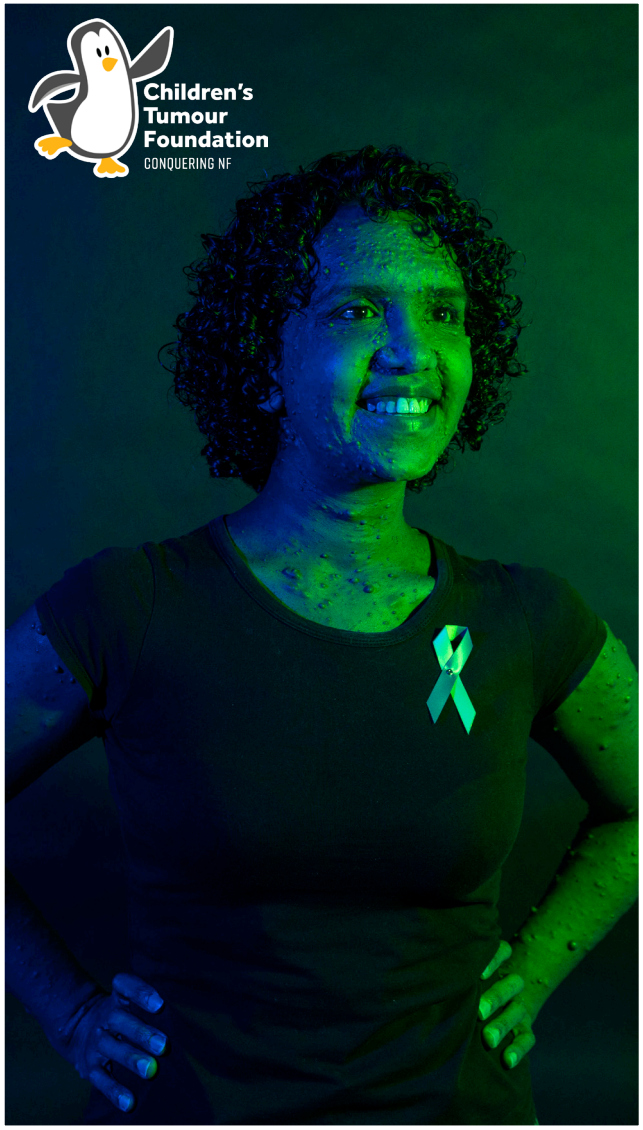


UNDERSTANDING

NEUROFIBROMATOSIS TYPE 1

(NF1)

CTF.ORG.AU

WHAT IS NEUROFIBROMATOSIS?

Neurofibromatosis (NF) is a set of three complex genetic conditions that cause tumours to form on nerves throughout the body.

NF affects more than 10,000 Australians.

While the signs and symptoms of each condition are distinct, the way they present and impact on someone's life is variable.

There is no way to predict how mildly or severely someone with NF will be impacted and can affect anyone regardless of gender, ethnicity or family history.

Neurofibromatosis Type 1 (NF1) is th most common form of NF and affects around 1 in every 2,500 people in Australia.

WHAT CAUSES NFI?

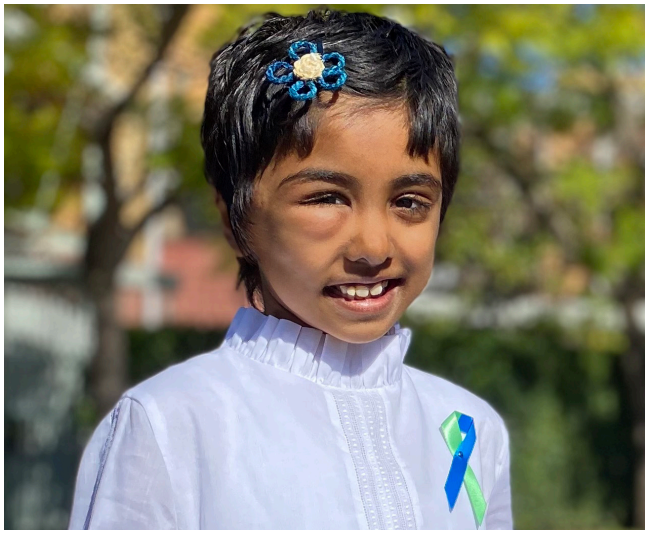
NF1 is a genetic condition caused by an alteration of the NF1 gene found on chromosome 17.

The NF1 gene is responsible for protecting cells from developing tumours, and if it is not functioning correctly, tumours can form and grow on nerves throughout the body.

Around half of all people living with NF1 will be the first in their family to have the gene change, meaning it is *spontaneous*.

The other half of people will have inherited the condition from one of their parents.

When a person with NF1 has children, there is a 1 in 2 chance (50%) they will pass the condition onto their children.



WHAT ARE THE SYMPTOMS OF NFI?

The first signs and symptoms of NF1 usually develop in early childhood. A clinical diagnosis will be made when at least two of these common symptoms appear.

- **Six or more cafe-au-lait marks:** light brown, coffee coloured birthmarks on the skin. These do not cause any health issues, but must be of a certain size to meet criteria.

- **Bilateral Freckling:** in the armpits or groin; areas that are not usually exposed to the sun. These do not cause any health issues.

- **Lisch Nodules:** small brown lumps that form on the iris of the eye. They do not interfere with vision in any way.

- **Two or more neurofibromas:** benign tumours found on or under the skin or within the body. When on the skin, they often appear as pea-sized lumps that are mobile under the skin. They do not tend to cause health issues, but some people find them itchy or painful, and they can bleed if bumped.

Plexiform Neurofibromas: tumours that involve a group of nerves and often grow to a larger size than a neurofibroma. In general, plexiform neurofibromas grow under the skin and may impact on nerve or organ function.

Optic Pathway Glioma (OPG): a tumour that develops on the nerve behind the eye. Usually diagnosed in childhood, those that cause issues with vision will require treatment (commonly chemotherapy), but most will simply need to be monitored.

Bone Dysplasia: can affect the sphendoid wing or long bones. The sphenoid is a small bone in the eye socket which can be deformed or absent in a small number of people with NF1.

The long bones, most commonly in the lower leg (tibia) can also be thinner, causing bowing and in rare cases can result in repeated fractures. Incomplete healing may lead to the formation of a *false joint* called **pseudoarthrosis.**

Along with these symptoms, there are two other criteria than may lead to a diagnosis.
- **A parent with NF1**
- **A confirmed genetic variant**

Learning and behavioural difficulties: Whilst not included as a diagnostic indicator, many people with NF1 experience specific learning difficulties or struggle with attention and social interactions.

Learning assessments and tailored interventions can be put in place.