IS THERE ANY TREATMENT FOR NFI?

While there is no simple cure for the symptoms that having NF1 causes, there is a great deal to know about how to manage and treat them when they are identified.

Most importantly, even if a person with NF1 seems to have little or no symptoms, it is important that there are a team of health professionals monitoring them so as to ensure prompt treatment should it become necessary.

Monitoring may include having a doctor examine the skin and movement, eye checks, x-rays and specialised imaging such as MRI.

If tumours cause severe symptoms such as pain or impact or function, there may be options for surgical and non-surgical treatments.

These will depend on the tumour location, size and what the managing specialist and the person with NF1 feel are the best options. There is often not a *one size fits all* treatment even for different tumours affecting the same person with NF1.

MEET HANNAH
Diagnosed at 16 years

ABOUT US

The Children's Tumour Foundation (CTF) provides information and support to individuals and families impacted by all forms of NF.

We have a dedicated support team who are here to answer your questions, provide reliable information and help strengthen the NF community in Australia.

WHERE CAN I FIND OUT MORE?

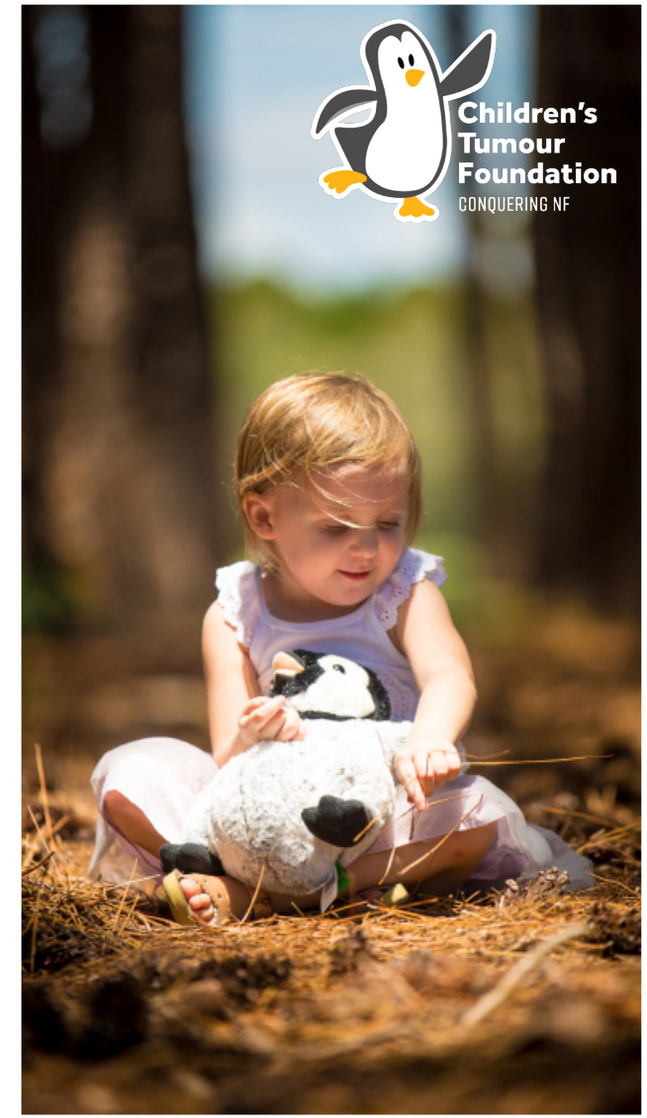
There are a number of useful resources available on the CTF website (www.ctf.org.au) or contact your healthcare professional.

There are also some online support groups for those impacted by NF.

Follow us on Facebook
@ctfaustralia
and join a group that suits your needs.

CONTACT US

National Support:	(02) 9713 6111
Email:	info@ctf.org.au
Website:	www.ctf.org.au
Social Media:	@ctfaustralia



UNDERSTANDING

NEUROFIBROMATOSIS TYPE I

(NFI)

WWW.CTF.ORG.AU

WHAT IS NEUROFIBROMATOSIS?

Neurofibromatosis or NF refers to several variable conditions which cause tumours to grow on nerves in the body. NF can affect both males and females and is diagnosed in people from all ethnicities and backgrounds.

NF type 1 (NF1) is quite common and is thought to affect around 1 in every 2,500 people. NF type 2 or NF2 is much less common and affects around 1 in 35,000 people. Schwannomatosis is also a type of NF which is rare in the community.

This brochure provides an overview of NF1 only.

WHAT CAUSES NFI?

NF1 is a genetic condition which means that it comes about due to a genetic change (mutation) in an important gene which is needed for tumour protection.

The gene involved in NF1 is located on chromosome number 17 and is called *neurofibromin*. Some people with NF1 (about 50%) will be the first in their family to have this gene change. This means it is not inherited, but caused by a "new mutation" in the *neurofibromin* gene.

For the other 50% of people with NF1, the gene change will be inherited from one of their parents. When a person with NF1 has children, there is a 1 in 2 chance that they will pass on the NF1 causing gene.

MEET EDWARD
Diagnosed at 2 years

WHAT ARE THE SYMPTOMS OF NFI?

NF1 is a condition which is variable, and no two people will have the same number of symptoms or signs. Listed below are some of the most common signs of NF1; however, not all people will necessarily develop these.

Cafe-au-Lait spots are light brown birthmarks on the skin. These do not cause any health issues.

Freckles in the armpits, groin or other skinfolds not usually exposed to the sun are a sign of NF1, but do not cause any health issues.

Neurofibromas are small lumps or bumps, often around the size of a pea, that often develop on or under the skin. These are typical of NF1 and do not usually cause health issues, though some people may find they become itchy or painful if located in an awkward spot. If neurofibromas cause pain or cosmetic concerns they may be removed. Neurofibromas can also grow internally and in those cases may impact on the function or structure of internal organs.

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

Lisch Nodules are freckles or coloured lumps that can be seen on the iris (coloured part of the eye). Often an eye doctor (ophthalmologist) needs a specialised lamp (slit lamp) to check for these. They do not cause any vision or other health issues, but are one of the distinct features of NF1.

Optic Pathway Glioma (OPG) is a tumour that develops on the nerve at the back of the eye. They are usually diagnosed in childhood, and most do not require treatment, but some cause vision problems and require chemotherapy or other treatment like surgery.

Bone dysplasia refers to bending of the long bones, most commonly in the lower leg. In rare cases this can result in repeated fractures. Incomplete healing may lead to the formation of a "false joint" or **pseudoarthrosis**. Less commonly, a small bone behind the eye socket called the sphenoid may develop unusually or not at all causing a bulging of the skin around the eye. **Scoliosis** or curvature of the spine may also occur.

Learning difficulties, attention span and behaviour issues are areas many people with NF1 struggle with. Learning assessments with tailored interventions can be a help with these issues.

It is impossible to predict how mildly or severely signs of NF1 will be expressed in each person who has it. Some people do not get diagnosed until they are adults, while others show many of the features listed above in early childhood.