Other NF team members and support
Your NF management team may also include speech pathologists, occupational therapists, psychologists, genetic counsellors and/or social workers who assist in managing the effects of NF1 on you and your family. Some families may be able to access funding for some of these services through GP Chronic Illness Management Plans, Disability Support or the new NDIS.

Genetics and reproductive implications
A geneticist or genetic counsellor can assist in confirming a diagnosis, providing information and counselling about the condition, its management and risks, and considerations and available options when planning a family.

Please note: The media can sensationalise the condition, when the reality is that the majority of people with NF1 are mildly affected and live a normal healthy life.

How can the Children's Tumour Foundation of Australia support you?
The Children's Tumour Foundation of Australia (CTF) can provide information and support. We employ support coordinators and host support groups around the country in conjunction with local peer support contacts and organise a range of community events including camps and information seminars.

For all NF news, subscribe to the national e-bulletin at
http://bit.ly/ctfsubscribe
Testimonial from a reader of the e-bulletin (subscription above): "I love the NF Bulletin – it's great to know what is happening around the country with NF awareness."

Contact Us
National Support: 02 9713 6111
VIC/TAS Support: 03 9936 6268
Email: info@ctf.org.au
Web: www.ctf.org.au

Published: 2017

Neurofibromatosis Type 1

Children's Tumour Foundation
CONQUERING NF

What is Neurofibromatosis Type 1 (NF1)?

Neurofibromatosis Type 1 (NF1) is a common genetic disorder and it affects 1 in 2,500 people men and women from all races and ethnicities. About 50% of people with the condition have inherited it from a parent, while the other 50% are the first in their family.

Common Features of NF1

Every person with NF1 will have a different experience of the condition. It is very variable even within families, and at least two of the following must be present to receive a diagnosis:

- **Café-au-lait spots:** usually 6 or more coffee coloured "birthmarks" varying in shape and size, with no medical significance.
- **Freckling:** in the armpits or groin.
- **Lisch nodules:** small, benign lumps of pigment on the coloured part of the eye.
- **Neurofibromas:** at least 2 small benign lumps that develop in or under the skin on small nerves, OR one **plexiform neurofibroma:** benign tumours involving a group of nerves in or under the skin or within the body.
- **Optic glioma:** a benign tumour affecting the optic nerve.
- **Bony abnormalities:** bowing or fracture of the long bones or of a small bone in the eye called the sphenoid.
- **An immediate family member** with a confirmed diagnosis of NF1.
- Another feature is **a positive genetic test** that shows a spelling mistake (mutation) in the NF1 gene.

While many people will remain healthy throughout life and only display the skin features of the condition, some people will be affected more severely with some of the features mentioned above, and may also have some of the features below, which can be well managed by regular monitoring and treatment:

- **Malignant Peripheral Nerve Sheath Tumour (MPNST):** usually arises in a benign plexiform neurofibroma but occasionally may develop without a known pre-existing neurofibroma.
- **Learning and cognitive impairments:** difficulties with learning, attention and behaviour are commonly reported in children.
- **Scoliosis:** curvature of the spine.
- **Hypertension:** raised blood pressure.
- **Glomus Tumours:** pain around the nail bed of fingers and toes, often likened to being 'hit by a hammer'.
- **Gastrointestinal Stromal Tumour (GIST):** symptoms include abdominal pain, anaemia and upper or lower gastrointestinal bleeding.
- **Epilepsy:** seizures can occur anywhere from infancy to late middle age.
- **Neurofibromatous Neuropathy:** some people may have a non-progressive weakness in their limbs and/or changes in touch and pain sensation.
- **Depression:** some people with NF1 report feelings of isolation, fear and anxiety, difficulty obtaining employment and some children are bullied at school.
- **Breast Cancer** risk is moderately increased in women with NF1; they should commence breast screening from aged 40
- **Headaches, migraines and pain** can be associated with benign tumours.

Regular Reviews

For children it is recommended that they have an annual NF1 check-up with a paediatrician, geneticist or neurologist that will include a skin examination, eye, spine and blood pressure check. Regular eye assessments are guided by clinical features and children with NF1 should be seen by an ophthalmologist (not an optician) regularly, up to the age of 10 years. The family should also maintain close links with the child's school to aid in decisions on additional help that may be required.

Adults with uncomplicated NF1 can be managed by their GP and should have their blood pressure checked at least annually and a skin check.

Adults with ongoing issues (complex) will require regular monitoring by an appropriate specialist who is familiar with the condition.

There may be times that the doctor will need to send you or your child to see another specialist such as an orthopaedic surgeon, a plastic surgeon, a neurosurgeon or a neuropsychologist for a child's learning assessment.