WHAT IS NEUROFIBROMATOSIS?

Neurofibromatosis (NF) refers to a group of complex genetic conditions that cause tumours to form on nerves, under the skin and deep in the body – affecting **1 in every 2,000 births** or about 4 million people worldwide.

NF includes neurofibromatosis type 1 (NF1) and all types of schwannomatosis (SWN), including NF2-related schwannomatosis (NF2), formerly known as neurofibromatosis type 2.

The signs, symptoms, and management of each type of NF are different and **there is no way to predict how mildly or severely someone will be impacted.**

NF can lead to a range of significant health issues including blindness, deafness, bone abnormalities, disfigurement, chronic pain, learning difficulties and even cancer.

Roughly half of all cases arise in families with no history of NF and it can affect anyone regardless of ethnicity, gender, or family history.

A SINGLE GENE CHANGE CAN LEAD TO A LIFETIME OF TUMOURS

NF is a lifelong medical condition requiring surveillance and management by a General Practitioner (GP) and/or a team of specialists.

Although NF is often misdiagnosed and misunderstood, the more informed you are about how it affects you personally, the better you will be at managing your own health.

This knowledge will provide you with a sense of control and will allow you to focus only on those things you need to be concerned about.

The impact of NF changes with each stage of life.

We are called the Children's Tumour Foundation because NF is most often diagnosed in childhood, but NF is a lifelong condition, and we are committed to supporting and finding treatments for everyone who lives with NF, at any age.

FACTS ABOUT NEUROFIBROMATOSIS

There are more than 13,000

Australians living with neurofibromatosis, and millions worldwide. There is currently no cure and treatment options are limited.

NF1 is the most common form of NF

as it affects 1 in 2,500 people and is characterised by cafe-au-lait (light brown) spots and neurofibromas (small benign tumours) on or under the skin.

Approx 20 per cent of children

with NF1 will develop an optic pathway glioma (brain tumour). Over half will experience some form of learning difficulties.
Incidence of breast cancer is five times more likely in women with NF1 aged 30-60 years.

NF2-related schwannomatosis

(NF2-SWN) occurs in 1 in 25,000 births characterised by the development of tumours called vestibular schwannomas on the eighth cranial nerve - the nerve that carries sound and balance information to the brain.

Schwannomatosis (SWN)

is an umbrella term for the following conditions, which are named according to the gene change causing the condition (with gene names are underlined):

- NF2-related schwannomatosis
- SMARCB1-related schwannomatosis
- LZTR1-related schwannomatosis
- 22q-related schwannomatosis
- schwannomatosis (NOS), not otherwise specified
- schwannomatosis (NEC), not elsewhere classified

SWN (excluding NF2-SWN) occurs in approximately 1 in 70,000 births.

Signs and symptoms of NF2-SWN

usually develop during the late teen or early adulthood years, although around 10% of people with NF2-SWN develop symptoms in late childhood.

Spinal tumours are seen

in 60-80 per cent of people with NF2-SWN.

Being diagnosed and living with NF can be difficult. It is natural to feel worried, stressed and upset at times, but having NF does not mean that you cannot live a happy and fulfilling life.