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NERVE TUMOURS UK

"Don't feel you're alone. There is help and support out there."

Catherine, NF1 patient

Neurofibromatosis Type 1 (NF1)

Some helpful information
and resources

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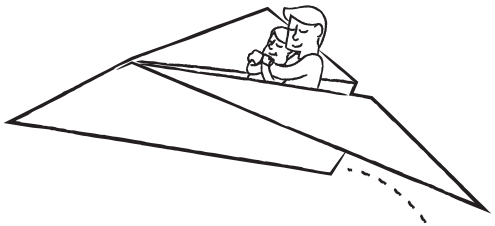


Introduction

Neurofibromatosis Type 1 is indiscriminate; it affects all races, age and genders equally. It is a dominant genetic condition, which means that it can be passed on in families from one generation to the next through genetic inheritance. About half of NF1 cases recorded have no previous family history of the condition. This is called a "spontaneous gene mutation".

NF1 is an incredibly variable condition, and might vary from one person to another even within the same family.

Some people may be affected very mildly and have very few health complications. Others may have many more complications that can seriously impact daily life, and restrict what they can do.

With a condition that varies so much, it is important to learn some basic facts about NF1, and to understand when you need to seek help from your doctor.

This is a large part of why Nerve Tumours UK exists; to support people with Nerve Tumours, and to help you manage your condition and live your best life.

"I have learnt through all of this is that I am a lot stronger than I ever thought possible."

Catherine, NF1 patient

Michelle, NF1 patient

Kieran's Story

"I was a very happy active boy with no known illnesses. When I was 5 years old, I became very unwell with vomiting and lethargy. My parents had noticed some freckling under my arms and groin but had no idea of the significance. In A&E, the consultant took one look at me and put me straight into a scanner. He explained to my parents that I had a large tumour sitting on the left side of my brain behind my left eye, and that I had Neurofibromatosis Type 1.

"I am so lucky as the surgeon removed the entire tumour. Today I have some small neurofibromas on my stomach, arm and head but they don't bother me. As a child, I joined urban scouts where we would play laser tag in the woods, and learn survival skills, which I did for 7-8 years. At age 11, I was able to go out and ride a motorbike under my dad's supervision.

I could still do nearly anything I wanted. I've been playing rugby since I was 12 and I have also recently passed my driving test and I am out on the roads.

"Throughout my life of having NF1 and my scar, I've had people stare, be rude and bully me, but I don't let that bother me. I'm proud of my scar."

> **"Living with NF doesn't bother me, I just take things as they come."**

NF1 is a common genetic condition that causes nerve tumours to grow where they shouldn't. The 'spelling mistake' in the gene is found on chromosome 17 and occurs in 1 in 2,500 of the population. There are over 25,000 people in the UK diagnosed with NF1.

What is NF1?

NF1 varies widely in how it affects those who have the condition. Many people with the disorder will be affected very mildly and may have nothing more than skin changes. A minority of people (around a third) who have NF1 will have medical problems related to the disorder at some time in their life. Some of these problems will be mild and easily treatable and others will be more severe.

NF1 is normally easy to diagnose, but if there is doubt, a genetics department can usually help to clear up any uncertainty.

How is it diagnosed?

The way the doctor can tell if a person is affected with NF1 is to examine him or her to see if there are signs of the disorder, so it's important to keep an eye out for signs that might help a doctor diagnose Neurofibromatosis.

There are specific features that the doctor will look for. NF1 usually appears in childhood and the diagnosis is confirmed if two or more of the following signs are present:

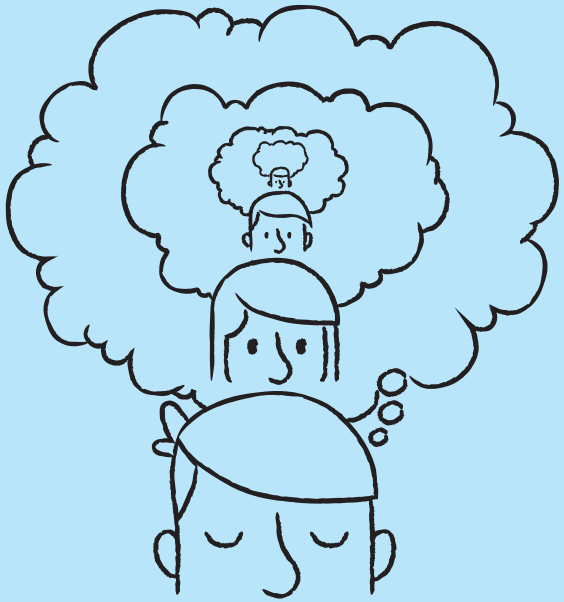
- Six or more flat, café-au-lait patches (brown coffee-coloured skin marks), which are the size of a pencil top or bigger. These usually appear during the first year of life and can increase in size and number. Café-au-lait patches are generally harmless and there is no connection between the number or size of café-au-lait patches and the severity of the condition. They sometimes fade later in life.
- Neurofibromas (small benign pea-sized growths or lumps) on or under the skin. These are a common feature of NF1. They usually appear during adolescence and tend to increase in number throughout life.
- Freckles under the arm or in the groin. Again, these are harmless but can be another sign of NF1.
- Plexiform neurofibromas. These represent growths around large nerves and can first appear as a swelling.
- Lisch nodules – these are small pigmented areas in the iris (coloured part) of the eye. They are often not visible except during examination with an eye microscope called a slit lamp. Lisch nodules are harmless and never cause symptoms, but can help in making a diagnosis of NF1.
- Another close family member has NF1.

How does it affect me?

If someone has NF1, he or she will have it for life. There is as yet no specific medical treatment or gene therapy to cure, prevent or reverse the features of NF1.

Many people who have been diagnosed with NF1 never experience health problems. However, some people who have NF1 can develop some complications that are very common with Neurofibromatosis.

The development of some of these features is related to age and some complications can first appear in childhood. Specific complications can be treated medically or surgically depending on the problem. Research may lead to new treatments in the future. Overall, the vast majority of the complications faced by people with NF1 are not life-threatening.



Remember, NF1 is a complex disorder and the effects of the condition can vary from person to person. However, research suggests that an estimated two thirds of people with NF1 are mildly affected and live a normal healthy life.

Here are some specific complications that can occur in NF1:

- **Ophthalmic** – NF1 can cause problems at the back of the eye.
- **Orthopaedic** – NF1 can sometimes create problems with bone development.
- **Dermatological** – Neurofibromas are benign tumours / growths that develop on or under the skin or along a nerve.
- **Plexiform Neurofibromas** – Some neurofibromas can grow in a wide, spreading fashion around large nerves and may feel like a bunch of knots or cords beneath the skin.
- **Tumours & Cancer** – Everyone, whether they have NF1 or not, has a risk of developing cancer during their lifetime. It is known that a small number of individuals with NF1 will develop malignant (cancerous) tumours. However, the lifetime risk of someone developing cancer related to NF1 is around 10%. Research in this area is continually being updated.

What health changes should you look out for?

You should seek medical advice if you experience any new, significant or unusual changes in body habits which do not go away, just as you would if you did not have NF1. This could include a new or persistent pain, or a change in physical activities.

- **Hypertension (High Blood Pressure)** – Occasionally NF1 can cause high blood pressure. It is recommended that everyone with NF1 should have his or her blood pressure checked once a year throughout life.
- **Learning & Behavioural Problems** – Although intelligence (IQ) studies have shown that the vast majority of people who have NF1 are within the normal intelligence range, somewhere between a third and two thirds of people who have the condition will experience some problems with learning. It is important that these problems are recognised as early as possible.
- **Issues with Self Image** – Many individuals with NF1 have more obvious signs of NF1 than others and they can find the outward signs of the condition an extremely distressing burden to live with. It is important that these problems are recognised as early as possible.
- **Pain & Itching** – Itching sometimes happens in NF1 and no one knows why this is so. Medication such as antihistamines or a simple emollient can sometimes help – ask your GP for guidance.

What support can I get?

Nerve Tumours UK Specialist Support

Nerve Tumours UK helps fund a team of Support Specialists in a number of regions across the United Kingdom. These Specialists work to improve the lives of those affected by Neurofibromatosis and provide crucial support to patients and families.

National Helpline

Another service we work to maintain is our national helpline. Open Mondays and Wednesdays 9am – 5pm, if you need someone to talk to or some help getting to the right place. Call **07939 046 030** or email **helpline@nervetumours.org.uk**

Nerve Tumours UK Website

Head over to our website site to find out more information on Neurofibromatosis Type 1 (NF1) and where you can find more help.

nervetumours.org.uk

"My advice is to just stay positive, live your life as you would do."

Kieran, NF1 patient

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Do you have nerve tumours? We're here to help.

nervetumours.org.uk
info@nervetumours.org.uk
020 8439 1234

/NerveTumoursUK **@NerveTumoursUK** **@NerveTumoursUK**

Our Nerve Tumours UK Specialists are available to offer support to anyone living with nerve tumours.

Call our helpline on 07939 046 030
Monday and Wednesday 9am-5pm

Nerve Tumours UK
First Floor, 44 Coombe Lane, London SW20 0LA

Never Tumours UK is the trading name of the Neurofibromatosis Association.
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