

## Information for young people with Neurofibromatosis type 1 (NF1)



### **Finding out and speaking up...**

Growing up means many things in your life will start to change. You may want to try out new looks and styles. You are becoming more independent. You could be thinking about leaving school, choosing a college course, or starting a job. You are taking more responsibility and making decisions for yourself.

If you have Neurofibromatosis (NF) it means that now it will be you, rather than your parent (or carer) to think about things to do with your health. The questions you have now may be different compared to when you were younger. You may find you rely less on your parents for advice and information but want to put questions directly to doctors yourself.

This information sheet offers you some facts and advice to help you. We can't cover everything here and for that reason it is important that you also talk to your own doctor. Your doctor is there to help. If your doctor does not know the answer to the question you ask, they will usually try to find out for you.

Some people with NF1 will have other members of their immediate family who also have NF1. This may be your mum, dad, brother or sister, or grandparents. It can be helpful to talk with them too. But the way NF1 affects them may be quite different to how it affects you. NF1 varies from person to person even within the same family.

If you are the first and only person in the family to have NF1 it can feel a bit lonely: as if you're the only person on the planet with NF1. In fact you're not! About 25000 people in the UK have the same condition as you. One baby is born with NF1 every day.

So if you decide to find out a bit more about NF1 then this information is a starting point. It will help you to feel you have more control over your health. Understanding what you need to be concerned about (and what you don't) can be reassuring.

## What is Neurofibromatosis type 1 (NF1)?

Neurofibromatosis is a genetic condition. This means it is a condition that can be passed on from a parent to a child.

Neurofibromatosis type 1 is a condition that causes lumps called neurofibromas to grow on the covering of nerves. Although doctors sometimes call the lumps tumours, they are **not** cancer.

There are different conditions called Neurofibromatosis. NF1 should not be confused with NF2. NF2 is a **completely different** condition and brings different health problems.

**NF1 cannot turn into NF2.  
NF2 cannot turn into NF1.**

**This information sheet is about NF1.**

## How do I check what type of NF I have if I'm not sure?

If you are unsure whether you have NF1 or NF2 ask your doctor. That information will be written down in your medical records.

Your GP will be able to tell you the correct diagnosis. Your GP will have received copies of letters written by any specialist doctors who have cared for you. This means that every doctor who needs to know shares important medical information affecting your health.

## What happens now I am a young adult...who looks after my NF1?

You do! It is important that you get to know your own body. At this age (the teenage years) it changes and getting used to the new you takes time. Understand what is "normal" for you. If things change or you notice an unusual problem (new symptoms) check it out with your doctor.

Young adults outgrow children's services (or paediatric care) and will transfer to adult care services at the age of 16 -18 years. This will be a different setting with no single doctor overseeing your care. There will be more emphasis on **you** taking responsibility for your health. So you may find that you need to do things such as:

- arrange your own appointments
- see your doctor if you have new or unusual health changes
- making sure that you get your eyes checked out at a good optician
- if you don't see any NF1 specialist doctor then each year remember to make an appointment with your GP for an NF1 health check

Now you are an adult, you must make sure that you attend all appointments for health checks. If you miss one or if an appointment you are expecting does not arrive, you will have to ask for another one to be sent out to you. In adult care it is unlikely that you will be sent further appointments in the way you would have been when you were a child.

Some patients will continue to see a hospital specialist doctor if they have a particular problem with their health. For example if you have epilepsy you will continue to see a neurologist. If you have a problem affecting your bones you will continue to have appointments with an orthopaedic surgeon.

Make sure you let hospitals/doctors know your new address if you move house. That way they can keep in touch with you.

## What are the health changes I need to look out for?

Your GP is not likely to know about NF1 in great detail as they may only see a few people in their surgery with that condition. So it is partly up to you to help your GP to give you the best possible care for your health.

Talk to your GP if you have new or unusual health changes (symptoms) such as:

### a neurofibroma (lump) that

- has changed from feeling soft to feeling hard
- has grown rapidly
- feels painful all the time or wakens you at night

### If you notice new symptoms including:

- difficulty breathing or swallowing
- changes in your bladder or bowel (going to the toilet)
- headaches that are getting worse and not better
- changes in your vision
- numbness, tingling, weakness in an arm or leg

Any other pain or health change that has lasted for more than a week if there is no obvious cause.

If you have any of these health changes, remind your doctor that you have NF1 and ask the question “can this change be linked to my NF1?”

If the health changes you notice continue to be a problem, **go back to your doctor** for more advice. More tests might be needed to work out what’s going on. Your doctor may refer you to see a specialist to get their opinion.

If you still don’t feel you are getting help in this situation after you have discussed your worries with your doctor, please contact the Neuro Foundation who will put you in touch with one of their Specialist Advisors.

## What do most people have with NF1?

Everyone, whether they have a condition like NF1 or not, has differences. That's what makes us who we are.

NF1 has some common signs that almost everyone with NF1 will have. Some people have some unusual things that are linked to NF1 but these only occur rarely.

One of the first signs of NF1 is brown birthmarks (café au lait patches) on the skin. People with NF1 have 6 or more of these birthmarks. Some people notice that as they get older their café au lait patches seem to fade and are less obvious.

People with NF1 also have freckling in the armpit and groins. These are another sign of having NF1.

As you reach your teens you may already have some lumps and bumps on your skin. This is part of having NF1. Doctors call these lumps neurofibromas. Some people will get just a few, others will get more. It varies from one person to another. At the moment there is no way these lumps can be stopped from appearing (say by a medicine or treatment or a drug). Doctors across the world are working hard to find a drug or treatment that will prevent lumps from appearing

There are 3 main types of neurofibroma:

- **Dermal** where the lump develops on the surface of the skin
- **Subcutaneous** where the lump grows on a deeper nerve lying under the surface of the skin
- **Plexiform** where the lump grows along a bundle of nerves and causes a larger swelling in the area affected.

Skin (dermal) neurofibromas start as a purplish area on the skin.



If you pass your finger over the purplish area it feels different to the surrounding area. Almost like a “buttonhole” in the skin. Later the area here will become more raised as the neurofibroma develops and forms a lump.



Sometimes lumps can be removed. The reason for removing a lump could be that the lump is sore, rubbing on clothing or in an awkward place. If you want a lump removed you should first of all discuss this with your own doctor (GP).

If your doctor agrees, they can refer you for a discussion with a specialist doctor such as a plastic surgeon or to a dermatologist to decide what treatment, if any, can be offered.

The choice may be removal by laser treatment (a type of burn) or by surgery (cutting). It is important to understand what can realistically be achieved after this treatment. It is not a magic solution ...and may leave scars. Talking this over with the specialist doctor can help you decide if you want to go ahead with this...or not. The appointment with the surgeon should help you to make a decision based on the best advice and information and an understanding of the likely outcome.

It is not realistic to remove all neurofibromas.

**Plexiform neurofibromas** can be difficult to manage. It may not be possible to remove the lump completely because it can be tangled up with other important structures such as nerves. Removing them may cause other problems such as excessive bleeding during the operation itself or nerve damage so the feeling in that area is lost. So surgery is best undertaken by a doctor who has experience of managing this type of lump. Sometimes the surgeon will try to reduce the size of the lump or “de-bulk” it. This means removing some but not all of it. Some people may have several operations because the lump re-grows or it is too difficult to remove completely without causing damage.

**Remember if you notice a lump is growing fast...is painful...has changed from feeling soft and squishy to feeling hard, tell your doctor and get it checked out.**

### **What can happen to me now?**

This varies from person to person. It can be almost impossible to predict.

Some of the problems with NF1 occur during childhood. These include:

- problems affecting the eye nerve (optic pathway glioma)
- problems with long bones (pseudarthrosis)
- scoliosis which is a curve that develops in the spine

If you have not had a problem like this during childhood, then it is very unlikely to occur later in your life as an adult.

Eating well can help to protect your bones. So having a diet that includes milk, cheese and yoghurt will ensure your body has a diet that includes a supply of calcium which is necessary for bone health and strength.

Some people have problems with their blood pressure. This should be checked once a year by your GP/practice nurse or hospital doctor.

There are still some problems that can crop up when you are an adult and this is the reason people with NF1 should keep a check on their health. If you notice things that are worrying you, don't just ignore them...get yourself checked out with your doctor.

Most problems with NF1 will cause a symptom...that is a change in your normal health: how your body feels or behaves. Know what is "normal" for you.

If you need to see your doctor, prepare for the appointment before you go. Think about what you want to know. If you think you may have trouble remembering your questions, write them down. If the doctor says something you don't understand ask them to explain it again. Or you could take someone with you who you trust. If there is still a problem ask the doctor to write it in a letter for you.

## NF1 and problems with learning

Some people have found school difficult. This can be because you may struggle with learning, or you may have trouble making and keeping friends. School can be a hard place whether you have NF1 or not.

Once you leave school the good thing is that mostly you won't have to take part in the lessons that you found so difficult. Hopefully you now know yourself: what you are good at and what you find very difficult. You may discover that in an adult education setting, life is easier to manage than it was in school. All colleges offer support for students who have health factors that affect how they learn and this support is easily available...as long as you ask. The department will be called Learning Support (or similar) and you can ask for help there.

You may still find calculating things difficult or struggle to work out things that need a mathematical skill, but often there are ways round this.

When deciding about courses at college, university or getting a job think about what your strong points are. Ask yourself what you are good at? For example:

- good at talking
- music or singing
- photography

- cooking
- caring for other people or animals
- design
- computer technology
- sports
- outdoor work, gardening and landscape

Make the most of your skills. It is important to find happiness and a sense of achievement in whatever you choose. There are people who can help you to make these important decisions about the future. These include your parents, teachers and specialist careers advisors.

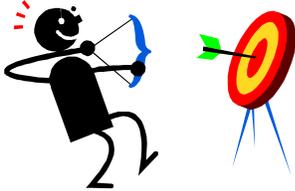
If you go on to college or university discuss your learning needs with the professionals there. They will ensure you get the right support to enable you to do your best and make the most of your course.

If you are working, look at our information sheet called [NF1 and employment](#) for some useful suggestions.

## Looking and feeling good

We all feel unsure about ourselves at times. If you are not a confident person, try to focus on the things that you can do rather than you can't. Things you are good at and enjoy.

Remember NF1 is a part of you but it doesn't change who you are or what you might achieve in your life. Find activities that you enjoy and make the most of the skills that you have. Have a go at new things. Even if you find they are not for you, it is a chance to meet other people with the opportunity for new friendships. It is not always the activity itself that is important but the chance it gives you to meet new people, have fun ...and so feel good about yourself.



If you are anxious or feeling down ask for help...from someone you trust. This might be a parent or relative, a good friend, a teacher or another professional. If you are feeling sad and low most of the time go to see your GP and talk to them. They will be able to support you.

If you are worried about your appearance or feel very self-conscious this can affect how you feel about yourself and stop you doing things. For some really good ideas about how to tackle this look at [www.changingfaces.org](http://www.changingfaces.org) . Changing faces is a charity that helps people who look different. They have a lot of great ideas to help and they also run confidence building courses.

### Some questions you might have at this time in your life

#### **If I have NF1 can I have the vaccine to protect me against cervical cancer?**

Yes as long as you are generally well and have no significant health problems at that time.

#### **Should I tell my friends I have NF1?**

This is a private matter and needs thought. Some people choose not to share their diagnosis outside their family. Some people think they might be treated differently because of their diagnosis. Some people don't care about who knows. If you have obvious

signs of NF1 it may prompt other people to ask questions. Being prepared for this is helpful so you are ready with a reply.

It is sensible to be careful about who you trust with private information. If you give personal information out readily it can soon become public knowledge through social media and cause you upset and hurt.

### **I don't like my skin and how it looks...what can I do?**

It can be upsetting if you have obvious signs of NF1 whether this is the skin changes, cosmetic effects or something else. Your feelings about this can be difficult to manage or feel overwhelming. It is important to talk to someone if this is worrying you.

Some problems cannot be changed but some can. Talk with your doctors to see what can realistically be offered to help. There are specialist doctors who can remove neurofibromas, either by surgery or by laser. There is a cosmetic camouflage service offered by the Changing Faces charity.

### **Is NF1 affected by the sun?**

Doctors will advise you that exposure to sunlight is best avoided in the heat of the day. You should protect your skin with high factor sun cream. However the sun does not have a harmful effect on the neurofibromas. You just need to take the same precautions as anyone else.

Don't avoid sunlight altogether. Sunlight can be beneficial as it is a source of vitamin D, a vitamin essential for healthy bones.

### **Genetic Testing and planning a family**

In the UK genetic testing is not offered routinely for NF1. This is because the signs on the outside of the body are usually obvious so a diagnosis can be easily made

Very occasionally a test may be offered: for example if the diagnosis is uncertain. A genetic test involves taking blood, removing the genetic material and sending it to a special laboratory. In most cases the gene change or mutation can be found.

For adults the main reason to have a genetic test is if you are planning to have children. If you have NF1 there is a 1 in 2 chance (or 50%) of passing it on to any children you have.

It is the same chance whether you are a man or a woman.

The problem with NF1 is that usually doctors cannot predict how mild or severe NF1 will be. If you are only mildly affected yourself it does not mean that your children will also follow that same pattern of mild NF1. The chance of having a child with more severe NF1 is about 1 in 12.

Before starting a family you may want to discuss this with a doctor who knows about NF1. You can ask your GP to refer you and your partner to Regional Genetics so you can learn more about NF1. You can ask the geneticist what choices there are when planning a pregnancy. This may include testing in pregnancy to see if the unborn baby has the NF1 gene change. Or you might want to ask about something called PGD (pre implantation genetic diagnosis). This is a process using IVF techniques (in vitro fertilization) when the cells that make the baby are tested in a laboratory and the unaffected embryo re-implanted.

This procedure is difficult and needs a lot of preparation. If this is something you want to know more about, you should ask your doctor for an appointment with your Regional Genetics Service. The referral needs to be made at an early stage of planning and before an established pregnancy.

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