



## **THE CHILD WITH NEUROFIBROMATOSIS TYPE 1 (NF1):**

### **A GUIDE FOR PARENTS**

NEUROFIBROMATOSIS (or NF for short) is the name for a group of conditions that cause lumps to grow on the covering of nerves. Neurofibromatosis occurs in all parts of the world. It affects boys (and therefore men) and girls (and therefore women) equally.

There are two main types of Neurofibromatosis: Neurofibromatosis Type 1 (NF1) and Neurofibromatosis Type 2 (NF2). They are two completely separate conditions that bring different health problems. People diagnosed with NF will have either Type 1 or Type 2, not both — Type 1 cannot change into Type 2.

This information is about Neurofibromatosis Type 1 (shortened to NF1 in this factsheet). It has been written as a general information guide to help families to understand what the diagnosis means. NF1 is a very variable and so, for specific advice, you need to talk to the doctor who knows your child best and is caring for their particular health needs.

NF1 is one of the most common genetic conditions. A genetic condition is one that can be passed on (inherited) in families. About 1 person in 2,500 has NF1. Some people have a very mild form of NF1 and may not even realise that they have it; for example they may just have some changes on their skin. Other people can have more serious problems that affect their health.

#### **How the diagnosis of NF1 is made**

The way a doctor can tell if a child has NF1 is to examine them carefully to look for certain signs or features. To make a diagnosis a doctor needs to find two of the following.

- Six or more café au lait patches (flat milky-brown marks) on the skin that are about 5mm in diameter or larger. These usually appear early on in life and can be referred to as “birthmarks”. Children tend to get more café au lait patches as they get older. This does not mean they will have more problems. These brown marks are harmless and never cause any health problem, nor do they need extra protection in the sun.
- Children with NF1 develop another change in the pigment of their skin: freckles in the armpit, around the base of the neck or in the groin. These are harmless. It is unusual to have freckles in these areas, but in NF1 children do. The freckles usually start to develop around the age of three and become more obvious as the child grows.

The unusual areas of freckling have, until very recently, been considered to be characteristic of just NF1. However they are also present in a related condition called Legius syndrome (discussed below). Therefore freckling is no longer used as separate criteria for the diagnosis of NF1.

- The lumps growing on nerves are called neurofibromas. Doctors may refer to these lumps as “tumours”. The word tumour does not mean cancer in this instance. Most adults with NF1 will have these on their skin. They may first appear as a purplish mark on the skin. In time a fibrous lump appears in the same area. Neurofibromas usually start to appear from the age of ten onwards and may increase in number during a lifetime. Some children develop a few skin neurofibromas at an earlier age. The number of neurofibromas adults with NF1 develop is very variable. In most adults neurofibromas are limited to the trunk of the body and so may not be obvious when someone is dressed.
- Some children have a special kind of neurofibroma called plexiform neurofibroma. These often develop earlier in childhood than the skin neurofibromas. They are called “plexiform” because they involve swelling to the whole section of a nerve and its branches. The doctor may be alerted to check for them by the appearance of the skin overlying them which may be thickened or hairy in appearance.
- Lisch nodules. These are small clumps of pigment cells that develop on the coloured part of the eye (iris). They are harmless. They can usually only be seen during a special eye examination performed by an eye doctor (ophthalmologist). They usually start to develop from the age of four or five and increase in number with age. Children with very dark brown eyes may not develop them.
- Sometimes the diagnosis of NF1 is made because a child may have very specific bone changes. One of these is called pseudarthrosis and causes the bone below the knee or in the arm below the elbow to curve more than normal. Another rare bone change is called sphenoid wing dysplasia that affects the bones forming the eye socket. This may cause the eye on the affected side to be more obvious compared to the other side.
- A diagnosis of NF1 may be suspected after an eye examination reveals that a child is having problems with their vision. If this problem is investigated and shown to be caused by a swelling on the nerves that supply the eye, NF1 may subsequently be confirmed. This swelling on the eye nerve is called optic nerve glioma.
- A Parent with NFI

Some of these features of NF1 take time to appear and that is why a doctor is cautious when suggesting the diagnosis, especially where no-one else in the family has this condition. It can feel very frustrating as a parent when the doctor seems hesitant about making a definite decision about the diagnosis.

If one of the parents has NF1 then making the diagnosis in their child is usually straightforward. Around half the children will be the first person in the family to have NF1. In this situation it can take time for the diagnosis to be confirmed. During this time of uncertainty, the paediatrician may suggest that you take your child for the sort of regular health checks that are appropriate to a child that definitely

has NF1. This is sensible health care and will enable the doctor to check for any changes.

The paediatrician may also refer your child to a clinical geneticist asking this doctor whether they think the child has NF1. The geneticist may discuss using a genetic blood test for NF1. This is discussed in more detail later in this information sheet.

### Why does my child have NF1?

NF1 is a genetic condition. This means there is a problem that is caused by a change in a particular gene. Our body is made up of cells. Genes contain the instructions within the cells that tell the body how to work. Genes determine things like eye colour or hair colour. NF1 is caused by a change or miscopy in the structure of the gene.

The genetic miscopy can be passed from a parent with NF1 to their child. A parent with NF1 has an equal chance (or 50% risk) of passing on the NF1 gene fault to each child they have. If a child is born with an NF1 gene fault, when they decide to have children, there is the same equal chance (50% risk) of passing on the condition.

Where no-one else in the family has NF1, then the miscopy in the gene that has caused a child to have NF1 has most likely occurred by chance. There is no known reason why it has happened. It is not something that could have been prevented by, for example, diet or environmental factors.

NF1 varies in the way it affects people. Even members of the same family who have NF1 may not have the same features. Explaining genetic inheritance is complicated. If you wish to know more about this, you could ask to be referred to your nearest Regional Genetics Service. There is a helpful website at [www.geneticalliance.org.uk](http://www.geneticalliance.org.uk) where information is available about all UK genetic centres together with other useful information about genetic conditions.

### Some important points

- The severity of problems in NF1 is not related to the number of café au lait patches. So it is not significant whether a child has 6 or 60 café au lait patches.
- NF1 does not skip generations. There are 2 ways in which it occurs:
  - \* It crops up out of the blue as a first event in a child, or
  - \* It is inherited from an affected parent.
- You cannot catch NF1. Nor are the lumps infectious. You cannot pass them on to someone else by contact.

About a third of people with NF1 may have some difficult health problems, but for many people it is just “skin deep”. Everyone with NF1 should have a health check once a year. This is particularly important for children, so that their development and learning can be monitored and because some of the health difficulties that are associated with NF1 can crop up in childhood.

### Childhood NF1 health complications

Most childhood complications in NF1 are rare. All children with NF1 should have a health check with a paediatrician until they are grown up. The purpose of this check is to monitor health and to ensure that any potential problem is identified early and therefore can, wherever possible, be treated. It is also an opportunity for parents (and their child) to ask questions and learn about NF1.

These are some of the complications that can occur in children with NF1:

### Eye problems (ophthalmology)

All children who have NF1 should have their eyes checked annually by a hospital based eye doctor (paediatric ophthalmologist) until they reach their 7<sup>th</sup> birthday. They then need annual eye checks with a reliable high street optician. Children, who are at risk of inheriting NF1 from a parent, but where the diagnosis has not been confirmed, should have their eyes checked at least once by a hospital eye doctor (ophthalmologist). The reason for these checks is because some children with NF1 can develop a lump at the back of the eye. This is called an optic nerve tumour or sometimes an optic pathway glioma.

An optic nerve tumour is a non-cancerous growth that involves the cells of the optic nerve. The optic nerve connects the eye to the brain and carries messages about sight. NF-related problems with this nerve can reduce the field of vision or the accuracy of vision.

Rarely, it can lead to loss of sight. Sometimes this nerve can become thickened but without any problem affecting vision. NF1 eyesight problems of this sort tend to crop up in early childhood, before the age of 7 years. However, it is wise to continue to have annual eye checks by an optician once discharged from hospital care. (More details about NF1 and eye problems can be found in our information sheet on this.)

### Bone problems (orthopaedic)

There are a number of bone problems that can occur in NF1 but each of them is rare. As a matter of routine a child with NF1 should have their back checked annually by their paediatrician or GP.

Scoliosis or curvature of the spine can occur around the growth spurt in perhaps 10% of children with NF1. If a curve is identified this will need specialist supervision and checks by an orthopaedic doctor with expert knowledge about the spine.

The doctor may monitor the spine for changes by a series of X-rays or scans. Occasionally surgery is offered.

A mild curve may cause no obvious problem and might not require treatment.

Once children have stopped growing and reached their final height at about 18 years of age, if the spine is straight, then it will remain straight.

Some children with NF1 have a change in the shape of their chest; it may be described as having a "pigeon-chest" or "hollow-chest". These are variations of what is the normal shape and do not usually cause any problem.

There can be a problem with the development and growth of the bone itself. This is called a pseudarthrosis. Where this occurs, it affects the long bones of the leg (most often the tibia) or the arm. Children will be referred to a specialist doctor (orthopaedic surgeon) for checks and offered treatment such as splinting to protect the bone, or a temporary plaster cast, or surgery.

Only about 1 or 2 children in 100 have this particular bone problem in NF1. It occurs in infancy (before a child is 3 years old) and will not crop up out of the blue later. More details about bone problems and NF1 can be found in our information sheet on this.

The other bone problem in NF1 is caused by the bones in one limb (usually the leg) growing faster than the other. This can be associated with a plexiform neurofibroma on the same side. Many people in the general population may have a slight difference in the length of their limbs but NF1 children with differences of 1cm or more should be monitored in a children's orthopaedic clinic. If the difference keep increasing then an operation to arrest the overgrowth may be considered to try to restore equilibrium.

### Skin problems (dermatology)

Skin changes are part of having NF1. Most people with NF1 will develop some lumps and bumps on their skin. These lumps are called neurofibromas. Doctors often call lumps "tumours". This does not mean cancer. The number of lumps a person gets will vary. It is not something that can easily be predicted<sup>1</sup>.

Neurofibromas are fibrous lumps that grow on the covering of nerves. The most common neurofibromas develop on the skin. This kind is not usually associated with any medical problems such as pain. Less commonly a child will develop neurofibromas on deeper-seated nerves that run inside the body. These deeper lumps can be painful if that area is knocked. If they develop in the neck they can be mistaken for lymph nodes (part of the immune system that helps us fight off infections).

As a general guide, the neurofibromas tend to start to appear in adolescence, occasionally earlier. Some people get only a few neurofibromas, others will get more.

At this time there is no treatment to prevent the lumps appearing, although scientists and doctors, both nationally and abroad, are working hard to find a suitable drug. The lumps can be a cosmetic problem but surgery or laser treatment is now more readily available.

Some neurofibromas can grow in a wide-spreading manner and can feel like a bunch of grapes or knots under the skin surface. These types of lumps are called plexiform neurofibromas. They can grow on different areas of the body and very rarely on the face. They can be painful if knocked. Surgery to remove these lumps requires a skilled and experience plastic surgeon. Sometimes it is not possible to surgically remove them because of the damage this would cause.

If a child does not have any signs of a swelling on the face by the age of 2 years, then they are most unlikely to go on to develop a plexiform neurofibroma that involves the face. Only 1 in 100 children with NF1 will develop this sort of large disfiguring lump on the face.

### High blood pressure (hypertension)

This is a very rare complication of NF1 but all children (and adults) with the diagnosis will have their blood pressure (BP) routinely checked once a year. This is because there are two NF1-related problems that may cause BP to rise: firstly, a lump can grow on the adrenal gland, the gland that secretes a hormone called adrenalin; secondly, there may be a narrowing of the artery to the kidney. This latter is the usual cause of high blood pressure in children with NF1 but it is an extremely rare problem. These are both normally treatable conditions.

## Epilepsy

Epilepsy is noted a little more frequently in children with NF1 compared to the general population. Where seizures are suspected in a child, they may be referred to a neurologist who will investigate the problem and run a series of tests. Epilepsy is a treatable condition and a child with suspected NF1 and epilepsy will share the same management as anyone else with this diagnosis.

## Other characteristics

Children with NF1 tend to be a little shorter than you would expect given the height of their parents. They may have a larger head circumference but this is not thought to indicate any particular problem.

## Speech and Language

A variety of speech and language problems occur in some children with NF1. Children can be slow to develop fluent speech. Speech may be difficult for others to understand with words poorly articulated, the pace of speech rapid and “gabbled” and with poor pitch control. Children may also have poor receptive and expressive language skills.

Early identification of potential speech problems is important to ensure access to Speech and Language Therapy to support children with speech difficulties. Where possible the strategies should be practiced both at home and in school to ensure a consistent approach.

If speech problems persist it may be helpful to consider referral to ENT to ensure there is no underlying hearing or palate problem.

## Learning and Behaviour

Some children with NF1, but not all, may find learning and school work difficult. They are often quite bright and able children but have difficulty with skills such as concentration, co-ordination, visuo spatial skills, and following a sequence of instructions. The work they produce may not reflect their ability.

Day to day, performance (the work that they produce) can be inconsistent so it is difficult for teachers to make a judgement about their true ability. Progress made one day seems to be lost the next, so it is difficult to build on basic concepts.

Behaviour can be challenging particularly when competing for attention in a busy home environment. Children with NF1 can be impulsive, restless and sometimes do not understand social cues or the need to take turns or be “fair”.

Identifying learning and behaviour difficulties is important to ensure that any child does not fall behind in their work or become isolated from possible friendships. This is the same for the child with NF1. Early assessment is important so that appropriate resources from the Special Needs range of support in school are made available. Talking to your child’s teachers about NF1 can be helpful.

The learning and behaviour difficulties found in children with NF1 are not unique; they can occur in many other children who do not have this diagnosis. The strategies for helping are the same. Learning and behaviour problems often respond well to support. The learning difficulties are not progressive, that is they do not get worse over time.

Children with NF1 have an increased frequency of being diagnosed with ADHD (Attention Deficit Hyperactivity Disorder) or ASD (Autistic Spectrum Disorder) or both. It is important to be aware of this so appropriate assessments can be made. ADHD can be missed as the problem in NF1 is usually one of poor concentration (so ADD or Attention Deficit

Disorder without hyperactivity). Children with NF1 usually respond well to a small dose of medication for ADHD/ADD. For children with NF1 and autistic spectrum problems there are a range of behaviour strategies and techniques that are helpful. The important thing is that these difficulties do not go unrecognised as it can impede progress in school and cause unhappiness socially.

More details about learning and behaviour in NF1 can be found in the following information sheets: "Information for Teachers" and "About learning disabilities". It may be helpful to involve a Neurofibromatosis Specialist Advisor when addressing this issue if there is one available in your area (please contact us for information).

### What checks should my child have when a diagnosis of NF1 has been made?

All children with NF1 should be checked annually by a paediatrician (children's doctor). The regular health checks include:

- recording the height and weight of your child and checking that these are within the normal range
- checking that your child's spine is straight
- looking carefully at the skin
- checking vision
- checking blood pressure
- asking questions about your child's general health and development

Doctors may refer your child to another doctor if they consider another specialist opinion will be helpful. The doctor will talk with you about any concerns you may have about your child's health and development.

The appointment is an opportunity for you to ask questions and to learn more about NF1. As children get older, the appointments will help to build up their knowledge of NF1 so that they too learn to keep a check on their health and know when to seek help.

### What changes do I need to look for? When do I need to seek help?

If your child is generally unwell and has any unusual symptoms it is sensible to see your GP (family doctor) and ask for advice. Remind the doctor that your child has NF1 and ask if the symptoms could be anything to do with that.

Some examples of unusual symptoms could be:

- sudden loss of consciousness, faints or "funny turns"
- increasing headaches that do not settle
- any rapid increase in the size or hardness of an existing lump or changes in the usual appearance of a lump
- unexplained pain
- changes in your child's eyesight that you notice
- any other symptom that is different from your child's normal state of health.

This is not an exhaustive list but a general guide.

All these symptoms would be unusual. There could be several explanations for them occurring that have nothing to do with NF1. However, it is appropriate to seek medical

advice in these circumstances. If the symptoms do not settle down even after you have been to see your doctor, then do return for further advice.

Most people who have NF1 are able to lead a normal life. You can help your children by encouraging them to find things they are good at, helping them to grow in confidence and giving them opportunities to succeed.

Clear and consistent rules help to guide behaviour and so teach your child how to fit in. By boosting confidence, you will encourage your child to accept that NF1 is just part of who they are and not the whole person.

### Does my child need a scan?

Children with NF1 do not need routine scans as part of their regular health care. However if they develop unusual symptoms then a scan can help to guide doctors in their investigations.

### Genetic testing in NF1

Doctors are usually able to make the diagnosis of NF1 relatively easily by a careful skin examination. The signs for NF1 appear during early childhood, in most cases before a child has reached the age of 5 years. The majority of children with multiple café au lait marks on their skin will, in time, develop features associated with NF1 and thus confirm a suspected diagnosis.

If your child is the first person in the family suspected of having NF1, a doctor called a Clinical Geneticist may suggest a test to confirm the diagnosis of NF1. This will show the gene change in around 95% of people with NF1. About 1% of children with multiple café au lait spots will not have NF1 but a related condition called Legius syndrome. This differs from NF1 in so far as people have the same café au lait spots and freckles but they do not go on to develop neurofibromas. Legius syndrome was only identified in 2007 and so we are only relatively recently starting to learn more about it. It is a very mild condition and does not seem to be associated with any of the severe health problems that can occur in NF1.

Sometimes the signs for NF1 may be limited to a particular area of the body, either a strip of skin or perhaps a segment of the body. This is called segmental or mosaic NF1. For more information on this please refer to our factsheet called "Information about localised, segmental or mosaic NF1".

### National Neurofibromatosis Centres

A nationally funded specialist service started in 2009 to look after the healthcare needs of the most complex NF1 patients in England. This includes some children with rare NF related health problems to ensure access to specialist expertise and high quality care. There are guidelines as to which patients are eligible. It is important that these children remain under the care of a local paediatrician as well to ensure they can access relevant community based services if needed.

The Neurofibromatosis centres are located at Guy's Hospital, London and St Mary's Hospital, Manchester.

### NF Specialist Advisor

The Neuro Foundation funds jointly with the NHS a number of NF Specialist Advisor posts. The NF Specialist Advisor service offers support and information which is available to families and individuals who have NF1. The NF Specialist Advisor can speak with you by phone or can sometimes visit you at home.



The service also has links with many other health and education professionals. The NF Specialist Advisor can, if they are based in your region, visit your child's school to give information to teachers on learning and NF1, or contact other health professionals who are working with you to give them information about NF1.

The Neuro Foundation also runs a helpline service. In the absence of direct access to a Specialist Advisor, the Helpline Specialist Advisor can speak to you or your child's school by phone.

The service is offered to all families in the UK and you are welcome to make contact with the charity to access this service. You will find contact numbers below.

For more information and a full list of publications please contact:

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