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ASSOCIATION OF IRELAND

Neurofibromatosis Type 1 for Teens

NF1

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Contents

Professor Green
Director, Centre of Medical Genetics

"I am delighted to support and endorse the new information sheets for people with Neurofibromatosis 1 and their families. The information will be of great help to the many families in Ireland with NF1, and will help those families to understand better the many ways in which Neurofibromatosis 1 can affect people. The National Centre for Medical Genetics is delighted to be associated with the Neurofibromatosis Association of Ireland, and that the Neurofibromatosis Association of Ireland has funded a genetic counsellor to run a specialised NF clinic in the NCMG. The NCMG has a wealth of experience with Neurofibromatosis and sees many families with the condition throughout Ireland. The NCMG holds genetics clinics in Dublin, Cork, Limerick and Galway, and is happy to see families with NF1, with a referral from their own doctor.

Prof. Andrew Green

NEUROFIBROMATOSIS CLINIC

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INTRODUCTION

You may have just learned that you have Neurofibromatosis (NF), or perhaps you have been going to doctors for years because of the condition. You may only have minor signs of NF – maybe some light brown patches on your skin – or you may be experiencing more serious complications. One of your parents may also have NF, or you may be the only one in your family who has the condition. In any case, this booklet was written to explain some of what is known about NF and what can be done to help you deal with it.

NF can affect the body in many different ways. It also can affect different people in very different ways. For some, NF may be nothing more than a nuisance; for others, it can cause challenging medical problems. It is natural to have lots of questions about NF. *How might it affect my health? Why did it happen? What can be done about it? Could it change my appearance? What should I tell my friends?*

We hope this booklet will answer many of your questions. In time, you may have other questions about NF; we encourage you to seek answers from your parents and doctor whenever a question arises. Also, NFA Ireland is a place you can turn to for information and support.

ACKNOWLEDGEMENT

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What is NF?

There are actually several kinds of Neurofibromatosis, two of which are named NF1 and NF2. Although these sound similar, they are in fact quite different. NF1, which is the subject of this booklet, is far more common than NF2. NF1 and NF2 do not occur in the same person, or even in the same family. NF1 does not turn into NF2, and vice-versa. They are totally separate conditions.

The term "neurofibromatosis" is put together from two words: "neuro" and "fibroma". "Neuro" means nerve, and a "fibroma" is a growth in the form of a lump. Nerves are like wires – carrying instructions from the brain to the muscles, and bringing messages and sensations back to the brain. Just like wires, nerves are surrounded by a protective coating. This coating is made up of cells, and it is the excessive growth of some of these cells that cause neurofibromas to grow. Therefore, a "neurofibroma" is an abnormal growth of these cells and are generally harmless, or benign, tumours.

Individuals born with NF1 can develop neurofibromas on, just under, the skin, or deeper in the body. Not all teens with NF1 have neurofibromas, and even those who do may not have neurofibromas that are easily seen or felt.

How Serious is NF1?

You should expect to live a long life and enjoy good health in spite of having NF1. The majority of people who have NF1 go through life with relatively few medical problems related to the condition. We estimate that two-thirds of all individuals with NF1 have a mild form of the condition.

Although there is a long list of things that can happen as a result of NF1, it is important to remember that no one experiences them all – and most of the serious complications are fairly rare. Many of these problems would already be present by early childhood if they were going to happen at all. On the other hand, there are some problems that can occur at any age. Part of dealing with NF1 is realising that it is a highly unpredictable condition. No two people are affected by NF1 in the exact same way, even if they are in the same family.

What Are Some Signs and Symptoms of NF1?

One of the most common, and earliest appearing, signs of NF1 is the presence of flat, light brown patches on the skin called café-au-lait patches. The term "café-au-lait" comes from the French word for "coffee with milk" because its colour is similar to that of these skin patches. Café-au-lait patches are completely harmless and serve only as a clue that someone has NF1. Individuals with NF1 typically have six or more spots, and many have dozens. There is no connection between the number of spots a person with NF1 has and the severity of the condition, or between the location of a café-au-lait patch and the location of a neurofibroma. Café-au-lait patches may darken during the summer on exposure to sunlight, and fade in winter. Too much exposure to sunlight is not good for anyone's skin, but the tanning of the

café-au-lait patches will not cause any harm. They sometimes fade later in life, and there are ways of treating them if their appearance is a problem to you.

Neurofibromas (caused by a growth of the cells that surround nerves) are usually noticed as a small bump on the skin, often like a mosquito bite that doesn't go away. Sometimes there is a slight pinkish or purplish colour over the site of a neurofibroma. Most neurofibromas on the skin, known as dermal neurofibromas, are small from the size of a pinpoint to that of a pencil eraser. They are usually soft and painless. They can occur anywhere on the skin and can appear at any time in life. There is no telling how many neurofibromas a person with NF1 will get; some people just get one or two, while others get many more. Just as with café-au-lait patches there are ways of treating neurofibromas on the skin, if their appearance is a problem to you.

Neurofibromas can occur not only on the skin, but also in most parts of the body where there are nerves. Those under the skin are firmer than those on the skin, but they, too, are usually painless. Sometimes these can be felt as pea-sized or larger bumps. Others may be so deep in the body that you won't know they are there. Occasionally, if a neurofibroma puts pressure on a nerve, symptoms will develop.

Some people with NF1 may have what is called a plexiform neurofibroma, which is a growth around large nerves. These are thought to form while a person is developing before they are born, so they are usually present at birth. Sometimes they will be obvious early in life as an area of swelling, but other times they can be deep inside the body and harder to detect. Unless they cause symptoms, plexiform neurofibromas do not need treatment. You should always tell your parents and doctor if you feel any pain, numbness, or weakness, since these can be

signs of a neurofibroma that requires the attention of a doctor. When present near the surface of the body, plexiform neurofibromas sometimes grow to a rather large size and can affect one's appearance. Usually, if this is going to happen, it does so in the first few years of life. If these large neurofibromas are not present by adolescence it is unlikely that they will ever appear. Surgery may be performed when a plexiform neurofibroma is causing a problem.

One characteristic of NF1 that can involve the eye is an optic glioma – a growth of the cells that surround the optic nerve, which connects the eye to the brain. Most of the time this presents no harm, but in some rare cases it can cause problems with vision or with the abnormal production of hormones. The diagnosis of optic gliomas is usually confirmed by performing a scan such as MRI (Magnetic Resonance Imaging) which takes a detailed picture of the nerve that connects the eye and the brain. Treatment options are available if an optic glioma causes problems. Optic gliomas that cause problems usually do so in young children between the ages of two and four; it would be unusual for problems to first appear much after age five or six. If you are a teenager and had an optic glioma identified when you were younger, your doctors will continue to watch for any changes.

Some people with NF1 develop scoliosis, or abnormal curvature of the spine. This is something your doctor can check for and treat if a problem develops. Less often, it is possible to have bone abnormalities in the leg bone or the bone that surrounds the eye. These problems typically are present at by one year of age, and will not develop later in life. People with NF1 have a slightly higher chance of developing something called osteoporosis later in life. Your doctor may talk with you about other risks for this including your diet and your family history.

Is NF a Form of Cancer?

NF is *not* a form of cancer. Cancer is a disease in which some of the cells of the body grow in an uncontrolled manner, and then spread throughout the body. While neurofibromas do tend to grow some, normally they do not grow like a cancerous (malignant) tumour and they do not spread throughout the body.

There are some exceptions, but fortunately these are very rare. Neurofibromas that grow on the skin almost never become cancerous. But larger neurofibromas under the skin, or plexiform neurofibromas, occasionally can become cancerous. *Signs of a tumour that becomes cancerous might be*:

- Sudden growth of a neurofibroma that was either not growing before, or was only growing slowly;
- Unexplained pain in a neurofibroma, especially if it wakes you at night. Pain that occurs in a neurofibroma after it is bumped is not worrisome, but if the pain appears for no apparent reason and does not go away, it should be checked by a doctor;
- If the feel of the neurofibroma changes from soft to hard.

There are other cancerous tumours that can occur in NF1, but these also are rare.

Should I Limit My Activities Due to NF1?

Just having NF1 does not mean you should limit your normal activities in any way. People with NF1 are not especially fragile or prone to injury. Of course, if you have a particular complication such as a problem with the shin bone or spine, this may require special protection and can interfere with some physical activities. You should ask your doctor if you have any questions about participating in physical activities.

Cosmetic Effects

Many people with NF1 have no change in appearance due to the condition, but it is common to have at least some features that are visible on the skin. If café-au-lait patches are bothersome, they can be covered with makeup. On the other hand, it is possible to have a café-au-lait patch "removed" by treatment with a special laser, but it is generally not practical to do this for every patch.

Neurofibromas on the skin, when present, may cause more obvious cosmetic effects. A single neurofibroma can be removed by surgery, although there is no guarantee that it will not grow back. Usually, surgery is reserved for a particular skin bump that is in a visible place, or is located where it causes discomfort, such as by rubbing against clothing. Some people with a large number of neurofibromas have chosen to have many removed at the same time. Sometimes surgery can leave a thickened scar, and no technique for treating dermal neurofibromas has been proven to have a lasting effect on improving appearance. There are several different treatment methods for removing neurofibromas, so ask your doctor if you have questions about this. If you choose to have surgery, it is best to find a surgeon who has specific experience in removing neurofibromas for patients with NF1.

The most serious cosmetic effects of NF1 result from the growth of plexiform neurofibromas around the eye or elsewhere on the face, arms, legs or torso. Again, this problem first appears in very young children; *if a teenager does not already have a large plexiform neurofibroma it is very unlikely that he or she will ever have one*. If you do have one of these plexiform neurofibromas, you know they can be difficult to deal with. The only treatment available at present is surgery, and it may require a number of surgical procedures to remove some of the

neurofibroma. Unfortunately, often there is no way to completely remove a plexiform neurofibroma because it is not always possible to separate it from surrounding nerves and tissue. In those cases, these tumours have a tendency to slowly grow back after surgery. On the other hand, plexiform neurofibromas may grow for a while and then stop growing on their own. So far there is no way to stop the growth of plexiform neurofibromas using medication, but this is a major area of research as scientists look for effective drug treatments as alternatives to surgery.

Pain

Fortunately, neurofibromas are not usually painful. If one is bumped, there may be pain that in some cases can last several days. Neurofibromas located under the skin can sometimes cause a tingling or mild shock-like sensation if pushed on. Only rarely does the pain become severe enough to require treatment. *Again, you should talk with your doctor about any neurofibromas that are painful, so that appropriate examination and treatment can be provided.*

While most people do not experience pain all the time as part of NF1, some individuals do develop occasional headaches or stomach pains as a result of the condition. In some cases, these can occur as a form of migraine. Your doctor can suggest medications to prevent or treat this pain, after examining you to make sure it is not the result of a more serious complication. If you experience prolonged back pain, this should also be reported to your doctor.

Growth and Development

Some people with NF1 find themselves a few inches shorter than others their age and than others in their family. As long as growth is occurring at a consistent

rate, there is nothing to worry about medically. Hormone treatments have been used to stimulate growth if it is occurring very slowly, but that usually is not necessary. Likewise, many with NF1 have slightly larger head size than other people. Head size in NF1 has no relationship to intelligence, and generally is not associated with problems.

Puberty is a time of many body changes, and these usually occur normally in people with NF1. Occasionally, however, these changes will begin earlier or later than usual. Your doctor may suggest some medical tests to find a reason for early or late puberty, if it occurs. This problem can usually be treated with hormonal therapy. It is often the case that neurofibromas first appear, or grow, during the teen years – possibly due to the influence of hormones. It is important to realise that not everyone with NF1 experiences a change in neurofibromas during adolescence.

Learning Difficulties

Many people with NF1 have some difficulty with learning. Studies show that many people with NF1 have some form of learning difficulty, which is usually mild. *Having a learning difficulty does not mean that a person is less intelligent*. Rather, it means that a person has trouble with some particular aspect of learning even though they may have average or above average intelligence.

Learning difficulties associated with NF1 vary from person to person, and they also vary in severity. They are not caused by tumours, and they do not become worse over time. In fact, with extra help people can make great progress in overcoming a learning difficulty. Learning difficulties are also common in people who do not have NF1.

Some people with a learning difficulty may have trouble paying attention, or remembering a sequence of instructions. Others may have difficulty reading, or doing maths. For some, handwriting can be a problem, due to poor muscle tone in the hands. Other individuals may have a speech deficit. Some people have a harder time using spatial information, such as making sense of maps or diagrams. Those who do may have difficulty learning to drive and navigate a car. In many areas of the country, special driver education programs are available for students with learning difficulties.

Many accommodations can be made at school for students with learning difficulties. These may include providing a tutor or special education services, giving more time on tests, or reducing the amount of handwriting required (sometimes by allowing use of a laptop computer, instead of requiring pencil and paper work). Speech and occupational therapy can be very helpful for those who need it. Occasionally, a medication may be prescribed to treat people with attention deficit.

Unfortunately, sometimes children with learning difficulties are misunderstood in school and are thought either to have bad behaviour or to not be working hard enough. Some teachers and parents may push to have these children work harder – not realising that they are trying as hard as they can, but may be unable to perform certain tasks in school as well or as quickly as others. Fortunately, awareness about learning difficulties is increasing, and it is getting easier to arrange special help for those who need it. When resources and special help are given, students with learning difficulties often can do very well in school. Another booklet is available from NFA Ireland about specific kinds of learning difficulties and practical ways that teachers and parents can help students with learning difficulties succeed in school.

People who have learning difficulties can go to college and hold any kind of job. Most colleges offer special services for students with learning difficulties. Many famous and highly successful people struggled with learning difficulties in school and found ways to overcome them.

Medical Follow-Up

Regular medical follow-up, ideally scheduled once a year, are important in order to catch any complications of NF1 as early as possible. A blood pressure check should be part of this follow-up, because even young people with NF1 can develop high blood pressure. You can use these doctor visits to ask any questions you might have about the condition, and to call attention to any changes you have noticed in your body. It is especially important to ask about problems such as pain (including headaches that are unusual for you), growth of neurofibromas, or any weakness, numbness or tingling sensations. Usually, your doctor will be able to reassure you with a simple physical examination. Sometimes special tests will be done to check out any symptoms you may be experiencing.

How Did I Get NF?

NF is not contagious; it is always the result of a genetic change that occurs before birth. NF does not occur as the result of any known environmental factor, such as exposure to X-rays, drugs, alcohol, or other chemical substances.

NF1 is caused by a change (mutation) in the NF1 gene. Genes are tiny structures inside our cells that control our growth and development. They determine characteristics such as hair colour, how tall we are, and our blood type. Most of our genes come in pairs

– we inherit one from our mother and one from our father. A change in a gene that prevents it from functioning properly may result in an altered trait or a genetic condition. NF1 is such a condition. Someone with NF1 can get the condition in one of two ways. About half of all people born with NF1 inherited this change in the NF1 gene from one of their parents who also has the condition. Often it is clear which parent has NF1. But sometimes a parent may be mildly affected and unaware of having NF1 until a special medical examination is performed. The other half of all people born with NF1 got the condition as the result of a random "new mutation." In these cases, neither parent has NF1. The gene change resulting in NF1 occurred for the first time in the egg or sperm that produced the affected individual.

Although parents may worry that they did something to cause this new mutation, we know this is not the case. Since passing on our genetic material is such a complicated process, requiring the copying of a vast amount of genetic code information, it is not uncommon for changes, or copying errors, to occur by chance and result in a condition like NF1.

Dealing with NF Emotionally

Adolescence can be a challenging time of life for anyone. In addition to experiencing hormonal changes, this is a time when you are trying to figure out who you are and what you believe in. This can cause your emotions to intensify. Sometimes you may feel confused or out of control, and you may think that nobody understands you or knows how you feel. Having NF1 may add to these emotions and can make an already stressful period seem even more so. Peer pressure, the demands of schoolwork, or perhaps feeling different from others can sometimes seem overwhelming.

In truth, everyone has something that makes them feel different at times. You can take heart in the fact that once one enters adulthood these tensions typically decrease and other people are more appreciative of individual differences. It is important to remember that you are far from alone in dealing with NF1.

NF1 can cause a number of physical changes to occur during adolescence, such as the growth of neurofibromas, right at a time when teens often feel the most self-conscious about their body. You may begin to be more involved in your own medical care. At the same time, doctor visits may frustrate you. You may not want to share information about your body or changes that have occurred. You may see it as an invasion of privacy. This is a common feeling for teens, whether or not they have NF1. If you are not experiencing any problems, you may feel these visits are a waste of time. It is important to realise, however, that seeing a doctor on a regular basis can help to keep an eye on changes that may need medical attention. It also gives you an opportunity to learn more about NF1 so that you can become a more active participant in your own healthcare.

Adolescence also is a time of change in your relationships with your family. It is natural for teens to want more control over their lives and to make more of their own decisions. Your parents are aware that you might develop more symptoms of NF during this time and they are, naturally, watchful. This difference in perspective can create tension, as teens sometimes resent the scrutiny of their parents at a time when they are becoming more independent. Ultimately, you will benefit a great deal from the support of your parents. By listening to one another, parents and teens can work together to handle challenges – including any that may arise in the management of NF1.

Most people will not experience serious cosmetic complications of NF1. If you do have cosmetic effects, however, just like all teens you may wonder how your appearance will affect dating and finding a boyfriend or girlfriend. While it is true that our culture often places too much importance on physical appearance, you will find that the more people get to know you the less they will care about any physical differences. True friends, particularly as people mature care much more about a person's character. *Even those adults who have serious cosmetic effects from NF1 are able to experience loving relationships, marriage, fulfilling jobs, and all the same opportunities that others have*.

At times you may feel angry about having NF1, since there is nothing you can do to prevent changes from occurring. You may wonder why you have NF, rather than a brother or sister. It is normal to feel angry or overwhelmed at times. It is important to remember that all people – with or without NF1 – will have to deal with some kind of difficulty in their lives. Accepting challenges as part of life's journey can make you a stronger and more understanding person. NF1 may be one aspect of your life, but it is only a small part of who you are.

Helpful Steps You Can Take

- Find someone you trust and share your feelings – including your fears, hopes, and frustrations. Be sure to talk to your parents or another adult if you feel depressed, anxious, or lonely. Counselling can be extremely helpful.
- Look for new social activities that promote meaningful friendships, such as volunteer work and after-school clubs.
- Join activities in which you excel. Feeling competent and getting positive feedback will increase your self-confidence and help to put your NF in perspective.

- Talking to other teens with NF1 can be especially helpful. NFA Ireland can help connect you with other affected teens in your geographic area and throughout the country.
- Having a positive attitude will help you deal with NF, and with life's everyday challenges. Naturally, no one wishes to have NF, yet many people report that coping with the condition has in some ways enriched and inspired their lives and they were able to make valuable contributions to others. Over time, look for ways this might be true for you.
- When you take an active role in dealing with NF, you likely will feel better about yourself and your situation. Many people find that becoming more involved helps them to feel more in control of their condition. Helping to educate others about NF, for example, gives many people feelings of inner strength and optimism. Taking more responsibility for your own medical care, and asking questions of your doctor, is another way that teens can take action toward staying healthy.

Telling Others That You Have NF

Deciding whether to tell other people that you have NF can be difficult sometimes. Whether you have few visible signs and no one knows you are affected, or if your condition is more obvious, having NF is a personal matter – and whom you share this information with is your choice.

You may want to tell your **friends** about NF in order to grow closer, yet even the best of friends may ask difficult questions. They may wonder about those "things" on your skin. They may not understand what a genetic condition is and worry about "catching" NF – which is, of course, impossible. They may make you feel vulnerable, or treat you differently. You need not share information that you feel uncomfortable sharing. On the other hand, many people find their

"true" friends to be great sources of support in dealing with NF. If you do decide to share this information with them, you may be able to teach them something about genetics and NF, which will help them understand your situation better.

It may be helpful to rehearse (ahead of time) responses that you might want to give your friends, so that you feel more prepared for their reactions and questions. With acquaintances that ask about café-au-lait patches or neurofibromas, for example, you might want to give a very brief answer such as, "It's like a birthmark," or "It's just a little skin problem." With closer friends, you might choose to explain more about the genetic condition. If you think that some terms might cause undue worry, such as the word "tumour," you might instead choose to use the word "growth" in your explanations to peers.

Whether to tell your **teachers** that you have NF can present another dilemma. The most important reason for telling them is if you have a learning difficulty, physical disability, or emotional need that could be affecting your self-confidence or academic performance. Most likely you will benefit greatly from some additional help, such as tutoring or special accommodations like being given extra time on tests. Many people don't want to tell their teachers they have NF because either they are doing fine in school or they worry that they may be treated differently. However, teachers typically want to know about any problems so they can provide assistance to help students succeed. School personnel can make sure you receive proper testing to identify your strengths and weaknesses, if recommended, and take specific steps to improve your education experience. Talking about this decision with your parents can help you make the choice that is best for you.
If you find yourself on the receiving end of bullying or teasing by your **peers** – perhaps due to looking

different or having some form of disability as a result of NF – it can be very helpful to seek assistance or guidance from school personnel. It is part of their job to try to prevent this kind of hurtful behaviour. Tell your favourite teachers and ask them for help. Many students find that educating their peers about NF, in class together with their teachers, can help. Your parents are also helpful resources for learning to deal effectively with this situation – as are the many good books and websites now available about teasing and bullying.

If you have a job, you may wonder whether to tell your **employer** that you have NF. If you have a learning difficulty you may benefit from additional training in a particular area or special work accommodations, such as receiving written instructions or reminders for complex tasks. If you have a specific medical complication, you may want your employer to know in case a health issue arises while you are on the job. However, this is a very personal decision that depends on your own unique situation. We encourage you to seek guidance from your parents before deciding how much personal information to share with your boss or co-workers. If you have a physical disability or medical problem that might be made worse by your job, such as heavy lifting if you have a back problem (like scoliosis), be sure to discuss this with your parents and doctor right away. In some cases, your employer will need to find different work activities for you that meet your health requirements.

Thinking About The Future

As you move closer to adulthood, you may begin to think more about the future implications of having NF1. As you gain more information about the condition, remember that not all of the complications you read about will happen to you! Again, most

people who have NF1 experience relatively mild or treatable symptoms.

It may seem a long way off, but when you approach an age that you start wondering about marriage and planning a family of your own it will be important to consider the genetic aspects of having NF. Regardless of whether a person has NF because it was inherited from a parent or through a new mutation, everyone with NF has a 50% chance of passing on the condition to their child. In other words, when one parent has NF there is a 50-50 chance with each pregnancy that his or her child will inherit the condition – the same odds as flipping a coin.

There is no way to predict how severely a child might be affected by NF1. It is possible for a parent who is mildly affected by NF1 to have a child with serious complications of the condition – just as a parent with serious problems due to NF1 can have a child with mild symptoms. It is also important to mention that some women with NF1 report an increase in growth of dermal neurofibromas during pregnancy, but for other women with NF1 this is not the case.

Recent advances in understanding the genetics of NF have made it possible to determine if an egg, sperm, or fetus (a baby developing in the womb) has the NF1 gene mutation, for those who wish to know. When the time comes, you may speak with your doctor or a genetic counsellor to learn the most up-to-date testing information.

It is helpful to take things one step at a time and not worry about decisions or potential problems too far in advance. Scientific understanding of NF1 is advancing rapidly, and with each new year important discoveries are made that move us closer to our ultimate goal – finding effective drug treatments, and a cure.

What Research is Being Done on NF1?

We are fortunate to live in a time of great hope that new treatments for NF1 will be developed through research. The gene responsible for NF1 has been identified and is being studied by scientists all over the world. For the first time in the more than 100 years since NF1 was fully recognised, we have the tools necessary to understand this condition. Already we have made extraordinary progress toward the goal of treating tumours and other potential symptoms of NF1, including learning difficulties. As new treatments are developed, they may be tested for safety and effectiveness in a clinical trial. You can expect exciting advances to come faster every year, as we work hard to "solve the NF puzzle".

Children's Tumor Foundation

Finally, The American Children's Tumor Foundation supports persons with NF and their families by providing thorough and accurate information. It also offers a YouthCONNECT program which includes an online youth chatroom. **www.ctf.org**

NF1 BROCHURES

- OVERVIEW OF NEUROFIBROMATOSIS TYPE 1
- A GUIDE FOR EDUCATORS
- LEARNING & COGNITIVE DIFFICULTIES
- NEUROFIBROMATOSIS TYPE 1 FOR TEENS
- THE CHILD WITH NEUROFIBROMATOSIS TYPE 1
- TALKING TO YOUR CHILD
- READERS 100 QUESTIONS ANSWERED

LEAFLETS

- NF1 REVIEW CHECKLIST FOR CHILDREN & ADULTS
- NEUROFIBROMATOSIS – A BRIEF INTRODUCTION
- SCHWANNOMATOSIS
- CONTACT FORM

CLINICAL GUIDELINES FOR MANAGING NF1
- FOR ADULTS
- FOR HEALTH PROFESSIONALS

NEUROFIBROMATOSIS TYPE 2 BROCHURES
- FOR FAMILIES
- FOR HEALTH PROFESSIONALS

HANDBOOK
- NF IRELAND HANDBOOK

NF1 For Teens

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Neurofibromatosis is a Little Known Genetic Condition and Can Manifest Itself in a Whole Lot of Different Ways

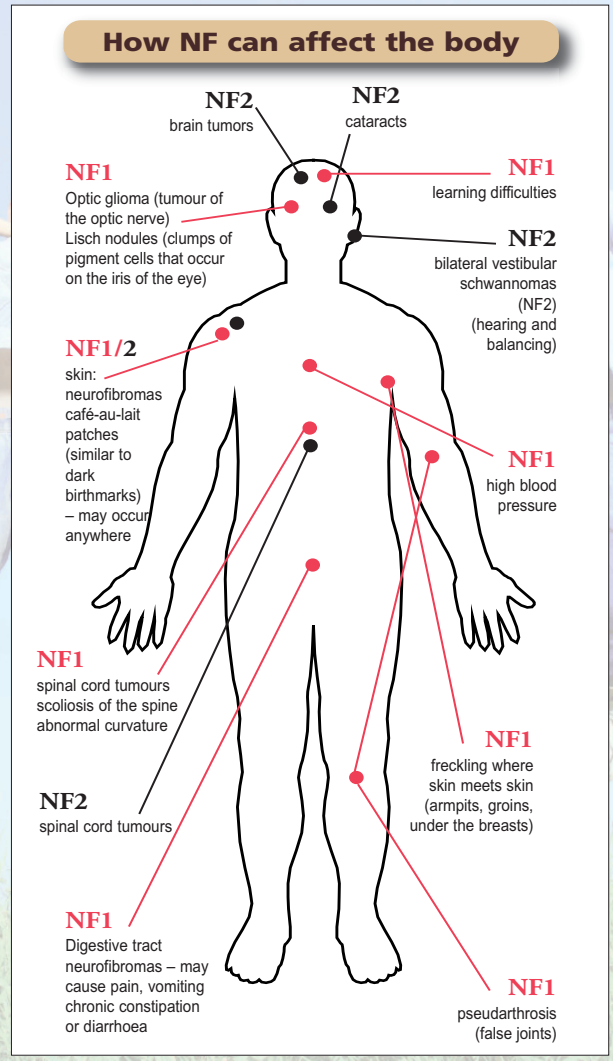
The care of persons with NF is made complex by the wide range of expression of the disorder. It is difficult to predict the specific problems that will occur in a particular individual. Diagnosis is made if an individual has two or more of the following features.

The diagnosis is based on the following clinical criteria:

1. **Six or more (café au lait)** coffee coloured patches sized 5mm or over in pubertal individuals and over 15mm in size in post pubertal individuals.
2. **Freckling** under the **arm** or in the **groin** area.
3. **Two or more Neurofibromas** of any type (growth of tumours on nerve tissue anywhere on the body) usually first seen on the skin.
4. **Plexiform Neurofibromas –** large bundle of nerves are thickened and appear as a soft tissue mass under the skin, these growths often large, can change the normal shape of the body.
5. **Optic Glioma –** Thickening of the optic nerve.
6. **Lisch Nodules –** clumps of pigment cells that occur on the iris of the eye.
7. **Orthopaedic** problems include **scoliosis** (curvature of the spine) **abnormal bone development**, such as overgrowth in long bones causing bowing and deformity that result in fractures, which fail to heal.
8. **First-degree relative with NF** e.g. parent, sibling, offspring.
9. **Learning Difficulties.** As many as 50% of children with NF have short attention span, appear clumsy and uncoordinated. Problems particularly with arithmetic and spelling are common.

Neurofibromatosis Type 2

Another rarer type of Neurofibromatosis and distinct in its clinical feature is **NF2.** The gene for **NF2** is located on chromosome 22, Features include: **Vestibular schwannomas** (tumour on hearing nerve). **Schwannoma** (type of tumour of the substance that covers nerve fibres). **Meningomas** (tumour of the covering of the brain). **Cataract**.



NEUROFIBROMATOSIS: Tell-Tale Signs
Café-au-lait patches,
6 or more consult your Doctor

Neuro
fibroma
tosis
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