NF1 AND ME…..
A GUIDE TO THE BASICS

LOVE ME

LOVE MY SPOTS

LADYBUG FOUNDATION

RAISING AWARENESS FOR NEUROFIBROMATOSIS

Newly Diagnosed?

You are not alone……

The Ladybug Foundation knows that receiving a diagnosis of neurofibromatosis (NF) can be overwhelming and a lot to digest all at once. Everyone deals with difficult news in different ways. Some like to take the information piece by piece so as not to be overwhelmed and so that they can have time to digest everything. Others prefer to delve in immediately and get as much information as they can. Both are perfectly normal approaches.

People also have different emotional reactions to being diagnosed with neurofibromatosis. Some may become anxious, overwhelmed, depressed, or fell a sense of loss or shock from unexpected news. Instead of fighting and trying to take control of the unpredictable, one of the most helpful mental strategies can be to simply try to accept the diagnosis. Once acceptance begins, it becomes much easier for the negative feelings to lessen and to cope effectively.

What is most important is that you understand that you are not alone. It is estimated that 1 in 3000 New Zealanders have NF, making it more common than cystic fibrosis, Duchenne muscular dystrophy and Huntington's disease combined. The Ladybug Foundation has many resources for you and would like to think of us as your safe haven. These resources include facebook and various events on the horizon that you can attend to help you meet more of the NZ NF Community.

Dealing with a diagnosis of a genetic disorder such as neurofibromatosis can be hard. We know that the lack of a cure and the unpredictability of the disorder do not make things any easier. However, there are things that you can do to help make this feel a little more manageable.

How to make it feel a little more manageable:

* **Get the facts:**

Read the "NF Basics" in this package

* **Get support:**

You can find social support by getting in touch with your health care provider or by visiting the Ladybug Foundation on facebook

* **Get involved**

Annual NF awareness walks being held during NF awareness month of May. Also on the horizon is the Cupids Undie Run a great way to empower yourself and the community in the fight for the awareness for NF. It is also a great way to begin meeting others in your local NF community.

LOVE ME

LOVE MY SPOTS

NF Basics

Neurofibromatosis (NF) is caused by a genetic change that makes people more likely to develop benign (non-cancerous) tumours around the nerves and on the skin. NF can also affect bones, vision, and other body systems. NF1 is a lifelong condition often diagnosed in childhood. People with NF can lead full lives, but require a specialist's care. Neurofibromatosis has been classified into three distinct types: NF1, NF2 and schwannomatosis. One type cannot turn into another type.

Neurofibromatosis 1 (NF1):

Also known as on Recklinghausen NF or Peripheral NF, is the most common form of NF. Occurring in one out of every 3,000 births, it is characterized by multiple café-au-lait (light brown) spots and neurofibromas (small benign growths) on or under the skin. About 50% of people with NF also have learning challenges. Softening and curving of bones and curvature of the spine (scoliosis) occur in some patients with NF1. Occasionally , tumours may develop in the brain, on cranial nerves, or on the spinal cord. While NF tumours are not cancerous, they may cause health problems by pressing on nearby body tissues. In a minority (10%) of cases, a benign NF1 tumour may become malignant (cancerous). But 90% of people with NF1 will never develop a malignant tumour from NF1.





LOVE ME

LOVE MY SPOTS

NF Basics continued…..

One of the first and most pressing concerns a person might have after a diagnosis of NF1 is how it affects life expectancy. You might see that some research shows that affected people have a shorter-than-average life expectancy. Although two-thirds of people with NF will never develop any major medical symptoms, some people with NF1 may have shortened life expectancies because of uncommon but serious NF1 related complications, mainly malignancies (cancers) and problems with blood vessels. These situations may be rare, but are concerning and screening for them is an important part of NF1 management.

NF1 symptoms are different for each individual. It is impossible to predict how mildly or severely affected someone will be, or what medical issues will develop. This can be frustrating for patients and their families. That is why it is very important for patients with NF1 to be treated by a group of healthcare providers knowledgeable about the disorder.

"Around two-thirds of kids with NF will never develop any major medical symptom."

- Nicole Ullrich, MD, Boston Children's Hospital





LOVE ME

LOVE MY SPOTS

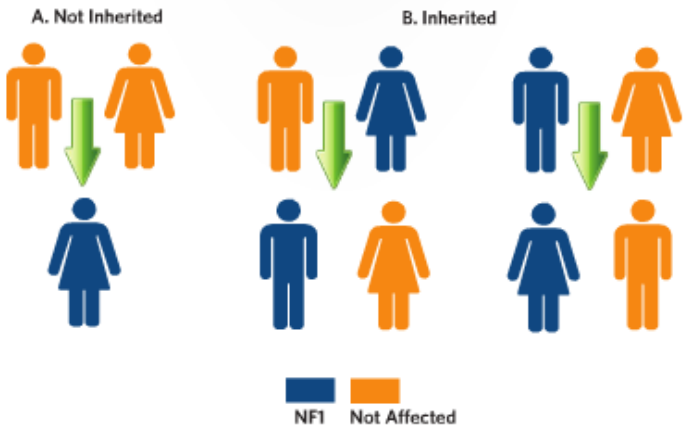
How Does Someone Get NF1?

When someone has NF, they are born with it. Some people inherit NF1 from a parent, while others are the first case in their family (a spontaneous case). It does not result from anything that occurred during pregnancy. One out of 3,000 people are born with NF1. The cause of NF1 is a change in a gene. A gene is a sequence of DNA. The NF1 gene has a very long sequence, and even a small change can shut down its normal activity. The NF1 gene is responsible for the production of neurofibrimin, a protein that keeps cells from growing too quickly. A person with NF1 has a genetic change in the NF1 gene that interferes with the normal production of neurofirbromin.

In 50% of cases, patients inherit a changed NF1 gene from a parent with NF1 (seeB, below). In the other half of cases, patients are born with a change in the NF1 gene even though neither one of their parents has NF1 (see A, below).

If the change is inherited from a parent, each time that parent has a child, the chance of that child having NF1 is 50/50. Whether or not any particular child develops the disorder is irrelevant to whether their other children develop it. For each child, the probability is still 50%.

If the change is a spontaneous case, the parent couple are very unlikely to have other children with NF1.



LOVE ME

LOVE MY SPOTS

How is the Diagnosis made?

The diagnosis of NF1 is most often based on a medical examination of a person's body. This type of diagnosis is called a clinical diagnosis. Foe example, light brown "café au lait" pigmented areas of the skin are often visible in the newborn period and parents may be told that their new born may have NF1. Other times it is not until additional signs emerge that a diagnosis is suspected or confirmed.

Additional signs that may emerge in childhood can include Lisch nodules (harmless "dots" in the coloured part of the eye), freckling (especially under the arms or in the groin region), learning difficulties, or bowing of the long bones in the leg or arm. Sometimes NF1 is not diagnosed until puberty, when typically more neurofibromas appear on the skin.

A clinical diagnosis of NF1 is made based on a medical evaluation finding any TWO of the following:

* Six or more café-au-lait spots that are 5 mm or larger in pre-pubertal individuals or 15 mm or larger in poet-pubertal individuals.
* Two or more neurofibromas of any type of one or more plexiform neurofibromas.
* Freckling in the underarm or groin areas.
* Optic gliomas (tumour of the optic pathway).





LOVE ME

LOVE MY SPOTS

How is the Diagnosis made? Continued...

* Two or more Lisch nodules (benign coloured spots in the eye).
* A distinctive bony lesion: dyspasia (abnormal growth) of the sphenoid bone behind the eye, or dysplasia of long bones, often in the lower leg.
* Having a close relative (parent, sibling, or child) with NF1.

People who have a suspected but not confirmed diagnosis of NF1 are usually offered regular medical follow-ups as if they have a diagnosis.

Genetic testing:

NF1 is caused by a genetic change. This change can be detected by a specialised blood test. Genetic testing ("DNA testing") for NF1 may be done in certain circumstances, but this test is not done routinely because a clinical diagnosis (based on observable signs of NF1) is considered reliable in most cases. In some instances, genetic testing can help to confirm the diagnosis if it is uncertain, and can be useful to establish a diagnosis in a young child who has not yet developed enough features to make a definite clinical diagnosis. For the most part, genetic testing does not predict the severity or specific complications of NF1. Genetic testing may also be informative for other family members or for deciding about reproductive options. The decision to have genetic testing is a personal one. A genetics healthcare provider or genetic counselor can help you with this decision.





LOVE ME

LOVE MY SPOTS

What Are the Possible Symptoms of NF1?

No one person will have all the possible symptoms of NF1. Many people are quite mildly affected. There is a range of severities, and we currently have no way to predict which symptoms will appear in an individual. People with NF1 may experience the following:

Café Au Lait Spots:

These flat, light brown spots on the skin are not harmful.

Neurofibromas:

These are the lumps or bumps on and under the skin that are typical of NF1. They can be on the skin surface (cutaneous neurofibroma) or under the skin (subcutaneous neurofibroma). Most are not a medical issue, but can cause pain and itching. The number of neurofibromas can be different between each person. They can be mild and only a few in number or more severe and be in the thousands. *Please be aware that photos on the internet of neurofibromas tend to show severe cases. These images are not accurate picture of what most patients with NF1 look like.*

A less common type of neurofibroma is called a plexiform neurofibroma. They occur in 25% of people with NF1. They can grow to be large and can cause a number of problems such as pressure on nerves and organs. These types of neurofibromas can become malignant in a minority of cases (about 10%).





LOVE ME

LOVE MY SPOTS

What Are the Possible Symptoms of NF1?

Continued ……..

Cognitive Differences:

Intelligence in NF1 is usually in the normal range, but children may need extra help in school. Learning challenges occur in about 50% of children with NF1.

Optic Pathway Tumours:

An optic gliomas is a tumour that develops in the cells surrounding the optic nerve. About 15% of children with NF1 will develop an optic gliomas. Children are most at risk for optic gliomas when they are under the age of six years. Most of the time, optic gliomas do not cause any symptoms and do not require treatment but sometimes can affect vision and may require chemotherapy.

Delayed or Early Puberty:

Most people with NF1 will start puberty at the expected age range, but some may have precocious (early) or delayed puberty.





LOVE ME

LOVE MY SPOTS

What Are the Possible Symptoms of NF1?

Continued ……..

Small Stature:

Affects about 30% of people with NF1. May be treated with growth hormones.

High Blood Pressure:

For a small percentage of people, hypertension is one of the most serious complications of NF1. People with NF1 should have their blood pressure checked at least annually.

Bone Issues:

Tibial bowing (curved lower leg bone) is seen in about 5% of patients. Scoliosis (curvature of the spine) occurs in 12-120% of patients.

Epilepsy (seizure disorder):

According to the most widely quoted NF studies, epilepsy in NF1 occurs with a frequency of 3.5-7.3%. The seizures associated with NF1 do not differ from seizures seen in the general population and, when not associated with a brain lesion, are generally well controlled with routine antiepileptic medications.





LOVE ME

LOVE MY SPOTS

Medical Management of NF1

Because NF1 involves many different systems of the body, doctors and staff from many different specialties may be involved in your care.

Your doctor may refer you to healthcare professionals who are experts in different NF1-related issues. You may be referred to the following:

* **Genetics:**

Medical Geneticists are physicians with expertise in the diagnosis and management of genetic disorders including NF. Genetic counsellors are healthcare professionals with training in medical genetics and personal counseling. Their job is to explain the genetic contribution to a medical condition and the various choices that can be made to deal with that condition.

* **Dermatology:**

Dermatologists are trained to evaluate and treat conditions of the skin, hair, and nails.

* **Neurology:**

Neurological specialists are trained in the treatment and diagnosis of conditions affecting the brain and nerves.





LOVE ME

LOVE MY SPOTS

Medical Management of NF1 continued…..

* **Ophthalmology:**

These specialists are trained to diagnose, monitor, and treat (through medicine or surgery) conditions affecting the eyes and vision-related structures in the body.

* **Orthopedics:**

This branch of medicine deals with conditions involving the muscles and bones.

* **Oncology:**

Oncologists specialise in the diagnosis and treatment of different types of benign and malignant tumours.

* **Psychology:**

Psychologists diagnose and treat emotional and behavioral issues.

* **Cardiology:**

Cardiologists specialise in the heart and major blood vessels. The manage cardiac conditions such as heart abnormalities.

* **Pediatrics:**

Pediatricians are experts in the medical care of children and adolescents.





LOVE ME

LOVE MY SPOTS

Breaking the news

How to Share the News

One of the more difficult parts of dealing with a new diagnosis is telling close friends and family, or even the child who has the condition. Although everyone has their own strategies and personal preferences for what and how to tell others, the following suggestions may help make the process a little bit easier.

WHO TO TELL

It is not uncommon for this question to come up when talking with a family member, a close friend, or maybe even a teacher—especially if the child has visible signs on the body, or NF related learning difficulties that affect their school performance. Many people reveal their diagnosis to others only if it seems important to their relationship (for example, to a close friend, or a teacher). Letting the child also have a say in who to tell can be helpful as well.

WHAT TO TELL

When you decide to share your or your child's diagnosis with others, you must also decide how much information to share. You might share in a limited way, such as discussing the learning difficulty aspects of NF1 with your child's teacher. Other times, it is helpful to have someone with whom you can share more details, including all of the emotional ups and downs that go along with the diagnosis.





LOVE ME

LOVE MY SPOTS

Breaking the news continued…..

SOME TIPS FOR TELLING CHILDREN

Children prefer routine and certainty. Although neurofibromatosis is filled with uncertainty, the more you can establish certain routines and normalcy for the child, the more comfortable the child will be. It is also important that you tell the truth. Creating a trusting and honest relationship is very important. Keeping a child in the dark can sometimes create more anxiety.

How much you choose to tell your child will vary depending on the child's age and maturity level. Many parents allow the level of information that they tell their child to grow with them as they get older. This is not a one-time talk, but instead an ongoing conversation that evolves over time. Some studies suggest that until a child reaches age eight, they only need basic information such as the name of the disorder, the parts of the body affected, how it will be treated (if at all), and how their lives will be affected in the short-term.

Another tip to keep in mind is the way in which you are reacting to the diagnosis in front of your child. Before you begin the conversation with your child, it may be helpful to make sure that both parents, or caregivers, are on the same page and have come to terms with their own feelings. Children often depend on a parent or adult's reaction to decide how they themselves should react. Although neurofibromatosis is a serious matter, the calmer you remain, the less anxious the child will be.





LOVE ME

LOVE MY SPOTS

Potential Learning Challenges Associated with NF1

The most common problems in children with NF1 are learning challenges and related problems. These affect about 50% of children with NF1. Most children with NF1 have normal intelligence; however, they may have issues in the processing of information. Problems with working memory, attention, visual-motor function, and spatial orientation are common issues. Keep in mind that these in NF1 are NOT universal.

Early intervention to address any learning issues can make a big difference. Working with your child's school and teachers to address there issues is vital.

What is known about cognitive and developmental issues in NF1:

1. For babies, delayed crawling, sitting, walking and talking may be observed.
2. For school-age children, difficulties in handwriting, focus and attention, and verbal memory may occur.
3. At all ages, patients with NF1 often have difficulties in organization and time management.
4. Learning challenges in NF1 do not get worse over time, but it may appear that way as school tasks become more complex at each level.





LOVE ME

LOVE MY SPOTS

Potential Learning Challenges Associated with NF1 continued……

Some general recommendations for identification and intervention of these problems:

1. Talk to your doctor about your concerns. There are many things that you can do to help your child with daily school activities.
2. Talk to the school. Educate them about NF1 and learn about what they do for children with learning issues.
3. Learn about your educational rights. Get in touch with parent support groups. They can provide additional resources. Learn about school support, special education laws, and additional resources in the school system.
4. An Individualised Education Programme (IEP) for your child may include extra time, shorter tests, or being assigned a desk near the front of the classroom.
5. Keep an eye on your child's self-esteem. The better they feel about themselves, the more willing they will be to put in the hard work needed to overcome and learning difficulties.
6. Check in periodically with the Ladybug Foundation. As we learn more, new interventions may become available.





LADYBUG FOUNDATION

RAISING AWARENESS FOR NEUROFIBROMATOSIS

The Ladybug Foundation is dedicated to our beautiful daughter Mikayla Mullins

The Ladybug Foundation is a charity focussed on supporting families dealing with Neurofibromatosis. Our personal journey with NF has given us a good understanding of what a roller coaster ride NF really is.

This Foundation is dedicated to making a real difference for not just the NF community but the wider community of New Zealand as *awareness is the key to finding a cure* so please join us as we head on this journey of awareness…….

We may have NF but NF doesn't have us!!!

16 McCutchan Road
RD3
WHAKATANE

PHONE: 073049251
CELL: 027 525 7303
EMAIL: ladybugfoundation@xtra.co.nz

www.ladybugfoundation.co.nz