Friedreich Ataxia (FA)

Processing a new diagnosis

A diagnosis of FA can be a huge shock for the person with the condition, parents, siblings, extended family members and friends. It is normal to feel an overwhelming mix of grief, confusion, anxiety, loneliness and helplessness as your family's life has been forever changed. Everyone will have different ways of getting through this time but it's important to know that help and support are available for you, your child and family. Hear advice on processing your diagnosis from a community member with shared experience.

The video is available at **https://youtu.be/A9XM71knDVw.**

Getting help processing a new diagnosis

There are many ways you can seek support as you process a new diagnosis:

- A psychologist is a university-qualified health professional who can help you talk about your thoughts and feelings to understand and cope with the challenges you and your family are facing. Visit our page on **Psychology** for more information.
- A counsellor is a trained professional who can help you talk about and work through problems. Visit our page on **Counselling** for more information.
- A social worker can provide information and support to people experiencing a range of issues including family problems, anxiety, depression, crisis and trauma. Visit our page on **Social Work** for more information.
- A genetic counsellor can help you understand how FA is inherited and whether there are any implications for other members of your family. Your specialist will be able to advise whether a genetic counsellor is required. Visit our page on **Genetic Counselling** for more information.
- Your state or territory neuromuscular organisation: can provide support, advice and information about living with FA. Visit our page on **state or territory neuromuscular organisations** for more information.
- Your GP can talk to you about a mental health plan and how you can use this to access the help and services you need. Visit our page on **Wellbeing** for more information.
- Other people and families living with FA have also experienced the emotional rollercoaster that comes with a diagnosis of FA and are able to understand exactly how you're feeling. You are not alone. Connect with other people and families living with FA in The Loop Community on our **Forum**.

How to talk to your child and others about a new diagnosis

Having a conversation with a loved one about a diagnosis of FA is not easy. In fact, it will probably be one of the hardest talks you will have. But, like all difficult conversations, it is important and necessary.

There are a lot of reasons why parents may not want to have this conversation, mostly centred on wanting to protect their child.

But avoiding the topic is not helpful. Children are often aware of the differences between themselves and their siblings and/or peers and may hear their condition being discussed during medical appointments.

So it is crucial that you have a role in your child or loved ones learning about their/your diagnosis. This will enable you to support them as they process the information and to be on hand to answer any questions they may have. If you are the person with FA it will provide you with support and someone to talk to. Together, you can learn about the journey ahead.

It will also allow you to:

- Provide answers to questions in an age appropriate way
- Correct any misinformation your child or loved one has heard or read
- Show them that you have faith in their ability to handle difficult conversations.

Ultimately, these conversations are an important step in providing your child with the tools to succeed, navigate their world and develop independence and self-advocacy skills.

There is no right time to have this conversation. Research suggests the earlier you talk to your child or loved ones about their/your condition, the more natural the conversation will become. The important part is to pick a time and commit to it.

Some important do's and don'ts:

- Do be positive but also realistic.
- Do reassure them that they have done nothing wrong and this is not their fault.
- Do tell them they will do many wonderful things in their/your life – they/you may just do them differently.
- Do tell the truth. Answer all their questions.
- Do consider your language. Try to avoid negative or emotive words, such as 'suffering from a condition'.
- Do use daily living examples that they can relate to.
- Don't avoid answering questions and don't shut the conversation down.

If you are having difficulty starting the conversation, try asking a couple of questions such as 'How are you feeling today?' or 'Do you know why you are tired?'

Remember, it's okay to be upset during the conversation but try to avoid breaking down as this will only cause greater distress. And if speaking with a child, remember children are resilient and they generally handle information of this nature far better than adults.

Understanding FA and how it's diagnosed

About FA

Friedreich Ataxia (FA) is a neuromuscular condition that mainly affects the nervous system and the heart. It is commonly diagnosed between 5 and 18 year of age. The condition progresses slowly but its symptoms and severity can vary from person to person.

What causes FA?

FA is a hereditary disease, meaning it is caused by a change or fault (also called a mutation) in a gene (the parts of our cells that tell our cells what to do). The faulty gene involved in FA is the FXN gene. This gene carries the instructions for production of a protein called frataxin. Frataxin appears to act as a storage depot for iron inside the mitochondria, the engines that produce energy for nearly all our cells. Frataxin cleans up excess iron and releases it when it is needed.

It is not yet fully understood what causes FA but it appears to be the result of a problem managing iron. The altered FXN gene greatly reduces the amount of frataxin being produced in the cells. Without frataxin, excess iron is left inside mitochondria. This damages mitochondria and leaves them unable to produce the required amounts of energy. Cells without enough frataxin are also particularly sensitive to harmful molecules (called free radicals) that can damage and destroy cells. The cells that are damaged or can't produce enough may not function properly, leading to the signs and symptoms of FA. FA mostly affects cells in the nervous system, heart and sometimes other tissues.

How is FA diagnosed?

Most people with symptoms of FA will be referred by their GP to a neurologist (a doctor who specialises in conditions that affect the nervous system) who will use several tests to reach a diagnosis of FA. Diagnosis usually begins with a physical examination and an assessment of personal and family history. During the physical exam, the neurologist will test your reflexes, including the knee-jerk reflex. Loss of reflexes occurs in most people with FA. The neurologist will also be looking at balance difficulty, loss of proprioception (joint sensation) and other signs of neurological problems.

FA can also be diagnosed from a genetic test using a blood sample. Tests for mutations frataxin mutations are highly reliable

and can be used to confirm or exclude a diagnosis of FA in almost all cases. The tests can also be used prenatally to determine carrier status. Further information about genetic testing can be found on the **Healthdirect website**.

Other tests that can be done for diagnosis or management of FA include:

- Electromyogram (EMG), which measures the electrical activity of muscle cells
- Nerve conduction velocity test which measures the speed with which nerves transmit impulses
- Electrocardiogram (ECG), which gives a graphic presentation of the electrical activity or beat pattern of the heart
- Echocardiogram, which records the position and motion of the heart muscle
- Blood tests to check for elevated glucose levels and vitamin E levels (vitamin E deficiency can cause similar symptoms to FA)
- Magnetic resonance imaging (MRI) or computed tomography (CT) scans, tests which provide brain and spinal cord images that are useful for ruling out other neurological conditions.

The genetics of FA

FA is inherited in an autosomal recessive pattern. This means that for a child to have FA, they must inherit two faulty copies of the FXN gene, one from each parent. The parents, who each have one abnormal copy of the FXN gene are known as ‘carriers’. Carriers do not have any symptoms of FA and usually don’t know that they have one faulty copy of the gene. If both parents are carriers, they have a 1 in 4 chance of having a child with FA, with each pregnancy.

Long-term outlook

Although there's no cure for FA, there are treatments for cardiac (heart) symptoms and ways to manage symptoms, such as ataxia (a loss of balance and coordination) and muscle weakness. Many people with FA lead active lives, working, studying, travelling married and starting families.

For more information about living with FA, overcoming some of the day-to-day challenges and where to get the right support, visit:

- **Living Life**
- **Counselling**
- **Social Work**
- **Psychology**

Understanding and planning for changes

Learning about FA and surrounding yourself with the right healthcare providers, services and support can help you feel more in control of what lies ahead. Although every person’s journey with FA will be unique, the information below will help you understand this journey and how to prepare and plan for changes in the future.

What are the main symptoms of FA?

FA affects people in different ways and the severity of symptoms can also vary from person to person. Symptoms of FA include:

- Ataxia - a loss of balance and co-ordination
- Fatigue - tiredness
- Vision impairment
- Hearing loss
- Slurred speech
- Scoliosis - curvature of the spine
- Diabetes
- Serious heart conditions.

FA typically presents in childhood, usually between 10 and 15 years of age, although it has been diagnosed in people from ages 2 to 50 years. FA that presents at an older age is usually associated with a less severe course.

Community Advice

Hear from a community member who has walked the path before you.

The video is available at **https://youtu.be/_6oJ9Mc0UO8.**

Life stuff

To find out more about living life with a neuromuscular condition and to access stories and peer-advice from the community, visit our **Living Life** section.

Where to find more information about living with FA

Friedreich Ataxia fact sheet
From Muscular Dystrophy New South Wales, 2017. This fact sheet includes considerations for future planning.

Friedreich Ataxia
This web page from Muscular Dystrophy Association, United States covers signs and symptoms, diagnosis, causes/inheritance, medical management and research of FA.

Consensus Clinical Management Guidelines for Friedreich's Ataxia and User Guide
From Friedreich's Ataxia Research Alliance, November 2014. These guidelines provide information about the diagnosis, treatment and management of FA written by international doctors and researchers. The guidelines cover nearly all symptoms and issues related to FA so not all chapters will be applicable to every person. It is recommended to read the User's Guide first for instruction on how best to use the guidelines.

Support for FA

Therapies and support

Clinical management guidelines

The **Consensus Clinical Management Guidelines for FA** provide recommendations that are designed to cover every aspect of the management of FA. When reading the guidelines and the list below it is it important to remember that not all areas covered in this document will apply to all individuals with FA. This information can be overwhelming but the guidelines are designed so that you an easily identify the chapters that are relevant to you. You might also find it useful to work through the information in the guidelines with your healthcare team to help you gain maximum benefit. It is recommended that the user guide is read first to explain the use of the guidelines.

The guidelines provide recommendations covering:

- Physiotherapy, exercise and rehabilitation to help with balance, flexibility, stretching and maintaining strength
- Occupational therapy for prescription of equipment and review of activities of daily living and use of ankle foot orthotics
- Speech pathology and swallowing evaluation
- Orthopaedic care
- Medications that may be helpful
- Vision screening and testing
- Muscle tightness and spasms
- Mobility
- Restless legs
- Bladder and bowel function
- Sexual function
- Hearing evaluation
- Cognition

- Cardiac evaluation
- Sleep
- Pain management and anaesthesia
- Scoliosis
- Diabetes
- Genetic counselling and pregnancy
- Mental health
- Advance care planning and end of life care
- Palliative care.

Other helpful support services

The video is available at **https://youtu.be/BDaCNsd9R_s.**

- Your **state or territory neuromuscular organisation** can provide information and advice on what support they can offer such as local support groups, camps, programs, services in the local area, advocacy or assistance in times of crisis. They can also provide an ear to listen if you need someone to talk to or guide you to get the assistance you are needing.
- Seeing a psychologist, counsellor or social worker can be incredibly helpful if you, your child or other members of the family are having a tough time or struggling with negative thoughts and feelings. Learn more about what services are available and how to access them here. Visit our pages on **Psychology**, **Counselling**, **Social Work** and **Wellbeing** for more information.
- Living with FA may mean that you or your child may need some assistance for everyday activities. Find out more about how a disability support worker could help and how to engage this type of support. Visit our page on **Disability Support Workers** for more information.
- Other people and families living with FA have also experienced the emotional rollercoaster that comes with a diagnosis of FA and are able to understand exactly how you're feeling. You are not alone. Connect with other people and families living with FA in The Loop Community on our **Forum**.

How to have better conversations when communicating your needs

To learn how to have better conversations when communicating your needs, visit the following pages:

Employers

Educators

Living Life: Education

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Friedreich Ataxia
This web page from Muscular Dystrophy Association, United States covers signs and symptoms, diagnosis, causes/inheritance, medical management and research of FA.

Treatment & Care Guidelines

Consensus Clinical Management Guidelines for Friedreich's Ataxia and User Guide
From Friedreich's Ataxia Research Alliance, November 2014. These guidelines provide information about the diagnosis, treatment and management of FA written by international doctors and researchers. The guidelines cover nearly all symptoms

and issues related to FA so not all chapters will be applicable to every person. It is recommended to read the User's Guide first for instruction on how best to use the guidelines.

Networks and Organisations

FAN Friedreich Ataxia Network
Queensland's FA Support Network is a support and information sharing network for anyone impacted by Friedreich Ataxia (FA), family, carers and friends. The network is based in Queensland and although much of the information provided is relevant to people in QLD the general FA information is relevant to everyone and anyone affected by FA is welcome.

Friedreich Ataxia Research Association (fara)
Is a not for profit Australian organisation that supports research into treatments and a cure for Friedreich Ataxia.

Friedreich's Ataxia Research Alliance
Is a US-based organisation dedicated to the pursuit of scientific research leading to treatments and a cure for Friedreich's Ataxia.

BabelFAmily
Is a non-profit organisation headquartered in Spain. We are a worldwide group of volunteers that unite our skills to support the greater FA community of patients, doctors, researchers, scientists and associations in their mission to find treatments and a cure for Friedreich's Ataxia.

Registry

FA Global Patient Registry
If you have FA or a parent of a child with FA you can register on the FA Global Patient Registry. The registry will allow you to be informed about opportunities to participate in clinical research studies and kept updated on the progress of clinical trials.