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FRIEDREICH ATAXIA

Friedreich Ataxia (FA or FRDA) is a slow, progressive disorder of the nervous system and muscles, which results in an inability to co-ordinate voluntary muscle movements (ataxia). This condition is caused by the degeneration of nerve tissue in the spinal cord and of nerves that extend to peripheral areas such as the arms and legs. FA affects the upper and lower limbs as well as the head and neck. There is a loss of sensations in the arms and legs, but mental capacity is not affected.

Friedreich Ataxia with Retained Reflexes (FARR) is a condition related to FA, where individuals still have normal deep tendon reflexes, such as the knee jerk reflex. All other diagnostic criteria are the same as for FA.

The onset of symptoms of FA usually occurs in childhood between the ages of 5 to 15 years, however may appear as early as 18 months or as late as 40 years of age. It is a rare condition, yet is the most common form of inherited ataxia, with 1 in 50,000 affected. FA affects males and females equally.

FA reduces normal life expectancy of the individuals, usually due to associated conditions, such as heart disease and diabetes. However, some people with less severe symptoms of FA often live through into their sixties or seventies.

What are the Features of Friedreich Ataxia?

The first symptom to appear in FA is usually difficulty walking (gait ataxia). This gradually worsens and eventually spreads to the arms and the trunk. Individuals may over- or under-extend the leg when it is brought forward in walking, and feet may be lifted higher than necessary and brought down too hard. The use of a cane or other walking aids may be required.

Over time, muscles begin to weaken and waste away, particularly in the feet, lower legs, and hands. Frequent falls and difficulty controlling the hands will result in increased clumsiness. It may become increasingly difficult to perform tasks such as writing, getting out of chairs and climbing stairs.

Foot deformities such as club foot, involuntary bending (flexion) of the toes, hammer toes (curled toes), or foot inversion (turning inward), high arches of the foot (pes cavus) may develop. These usually do not pose a problem in themselves, though if problems do arise, bracing or surgery can be beneficial.

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Although progression varies from individual to individual, the ability to walk is often lost within eight to ten years from the onset of symptoms, making it necessary to get a wheelchair. Options can be assessed by an occupational and/or seating therapist.

Other symptoms include loss of tendon reflexes, especially in the knees and ankles. There is often a gradual loss of sensation in the fingers and toes, which may spread to other parts of the body.

The muscles controlling speech can be affected, resulting in slow and slurred speech (dysarthria). Speech therapists may provide beneficial support for this. Rapid, rhythmic, involuntary movements of the eye (nystagmus) can also be common.

Scoliosis, an abnormal curvature of the spine, can become an issue and can impair respiration. Spinal bracing may be required, and in more severe cases spinal fusion surgery. An orthopedic specialist is essential in monitoring the scoliosis.

Conditions Associated With Friedreich Ataxia

There are conditions associated with FA that do not result from the degeneration of nerves. Cardiac problems are common and are present in approximately 80% of FA individuals and arise from various forms of heart disease that often accompany FA. These include cardiomyopathy (enlargement of the heart), myocardial fibrosis (formation of scar tissue in heart muscle) and heart failure. Symptoms can include chest pain, shortness of breath, and heart palpitations. Other heart rhythm abnormalities may be present, such as a fast heart rate (tachycardia) or a heart block, which is the impaired conduction of electrical impulses, necessary for the contraction of the heart. Cardiac problems can be treated with medication, though severe forms of heart disease can be fatal.

Diabetes mellitus is a condition characterized by abnormally high blood and urinary sugar levels. About 10% of individuals will develop this and will experience increased thirst, hunger and urination. This can be managed with diet and medication, such as insulin.

What Causes Friedreich Ataxia?

FA is an autosomal recessive disorder. For further information on genetics and how disorders are inherited, please refer to the Muscular Dystrophy Association Genetics Factsheet.

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Humans have 46 chromosomes made up of genes. Each chromosome, which is a tightly coiled chain of DNA (deoxyribose nucleic acid) contains millions of chemicals called bases. The four bases are adenine, thymine, cytosine and guanine (A, T, C and G), which pair together in sets of three to form coded messages. These messages are instructions for producing proteins that make the body function. Many disorders are a result of a mutation to these bases.

In FA, there is a defect in a gene on chromosome 9. In 98% of cases there is a triplet repeat expansion of the GAA sequence of bases. A greater number of repetitions is related to earlier onset and faster progression of the disorder. The corresponding protein that is altered due to this repeat is called frataxin and is produced in diminished amounts. Frataxin is found in energy-producing parts of cells called mitochondria. When frataxin levels are low, cells (particularly in the brain, spinal cord, and muscle cells) cannot produce energy properly and the build up of toxic by-products leads to "oxidative stress", which has the potential to destroy cells.

In FA, this "oxidative stress" affects nerve cells in the spinal cord and the peripheral nerves, which connect the spinal cord to muscles and sensory organs. This results in failure to stimulate some muscles, which will eventually weaken and waste away (atrophy). There is also damage to the cerebellum, which is a small structure are the back of the brain which helps to plan and co-ordinate movements. Combined, these problems lead to the progressive losses of muscle strength, sensation, balance and coordination that characterize FA.

Diagnosis of Friedreich Ataxia

Diagnosis usually commences after the identification of key characteristics of FA. Several tests are available to confirm diagnosis of FA and associated conditions:

- Physical Examination – test of reflex and sensory responses
- Electromyography (EMG) – observes the electrical activity of muscles and its consistency with activity typical of individuals with FA
- DNA Testing – can identify the presence of the abnormal gene in the individual with FA as well as carriers
- Nerve or Muscle Biopsy – can confirm diagnosis
- Nerve Conduction Studies – measure the speed with which nerves transmit impulses
- MRI (magnetic resonance imaging) or CT (computed tomography) scan, which maps the brain and the spinal cord

• Spinal Tap – to evaluate cerebrospinal fluid
• Electrocardiogram (ECG) – which gives a graphic presentation of the electrical activity or beat pattern of the heart to look for heart abnormalities
• Blood and Urine Tests – to check for elevated glucose levels

Soon after a diagnosis of FA in the family, it is essential that genetic counselling is arranged, for one or both of two issues. The first is the probability of Mum or Dad having the disorder and the second is whether testing for FA in pregnancy can be offered and with what degree of accuracy.

Genetic counselling provides information about possible diagnostic tests, including prenatal testing. Genetic services in NZ are available and a referral can be made by the MDA.

Management of Friedreich Ataxia

As with many degenerative diseases of the nervous system, there is currently no known cure for FA. Treatment focuses on the prevention and management of symptoms and accompanying complications to help maintain optimal functioning as long as possible.

From an early stage, it is important to undergo regular exercise and stretching programmes, with the help of a physiotherapist, to maintain muscle strength and flexibility. Swimming is an excellent option to exercise and mobilize all muscles and joints.
A good diet with plenty of fresh fruit and vegetables is very important in ensuring excessive weight does not impede mobility. Contact with a physician and/or a nutritionist is valuable for this

Research into Friedreich Ataxia

Scientists hope that recent advances in understanding the genetics of FA may lead to breakthroughs in treatment. Research is being carried out to further understand the interactions between muscles and nerves to uncover the pathological process of FA, and as the exact mechanisms become better understood, it will be possible to direct treatment strategies to the cause, rather than simply towards the symptoms. Additionally, in-depth studies of heart disease and diabetes mellitus are being conducted to determine their relationship with this neurological disorder.

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Support for People with Friedreich Ataxia

Support is available from the MDA who can offer information, support, advocacy and referrals to other providers. There is also a nationwide Support Network for those interested in meeting with others.

There is no reason why individuals with FA should be disadvantaged in terms of receiving full education. For more information, request the Education Pack available from the MDA.

Disability should not hinder employment possibilities, though it is wise to choose a career that does not require physical activity. Any individual has the right to equal pay and equal rights for employment. For more information contact the Employment Relations infoline on 0800 800 863 or visit www.ers.dol.govt.nz.

The government promotes equal employment opportunities in private sector and can be contacted on (09) 525 3023 or visit www.eeotrust.org.nz

Workbridge provides a professional employment service for people with all types of disabilities and administers support funding on behalf of Work and Income. Contact on 0508 858 858 or visit www.workbridge.co.nz

More Information Muscular Dystrophy Association can be contacted for further information, assistance, advice, support and referrals, on 0800 800 337 or by e-mail at info@mda.org.nz.

The Muscular Dystrophy Association Website also contains information on services available within NZ, our quarterly magazine, contacts, membership details, news and links to other sites - www.mda.org.nz

Further Resources

www.nzord.org.nz – the New Zealand Organisation for Rare Disorders website provides information on a number of rare disorders, a directory of support groups, practical advice, health and disability resources, research information, news and issues.

www.mdausa.org – the MDA USA website has an extensive site with plenty of further information on any muscular dystrophy conditions as well as research news.

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www.fara.org.au – the Friedreich Ataxia Research Association is a New Zealand and Australian organisation which carries out continuous research for the causes and possible treatments for Friedreich Ataxia.

www.ninds.nih.gov – the National Institute of Neurological Disorders and Stroke provides further information on Friedreich Ataxia and other neurological disorders, as well as current research findings.

Information in this fact sheet was primarily sourced from:

Muscular Dystrophy Association of Australia (2003) Friedreich Ataxia Fact Sheet. www.mda.org.au

National Institute of Neurological Disorders and Stroke (2006) Friedreich Ataxia Fact Sheet. www.ninds.nih.gov

National Ataxia Foundation (2006) Ataxia. www.ataxia.org