Friedreich Ataxia (FRDA)

Division of Human Genetics
University of Cape Town

What is Friedreich ataxia?

Friedreich ataxia is named after the German doctor Nikolaus Friedreich who described the disorder in 1863. Friedreich ataxia (FRDA) is a genetic condition that damages nerve tissue in the area of the brain responsible for coordination, muscle movement, and some sensory functions (cerebellum and spinal cord, Fig. 1). Uncoordinated movements (ataxia) worsen over time. The condition usually begins between the ages of 8 and 15, but it can occasionally begin at birth. Friedreich ataxia mostly affects the nervous system involved in muscle control and coordination but may also affect heart function, this is illustrated in Figure 1. Friedreich ataxia does not affect parts of the brain involved in thinking.

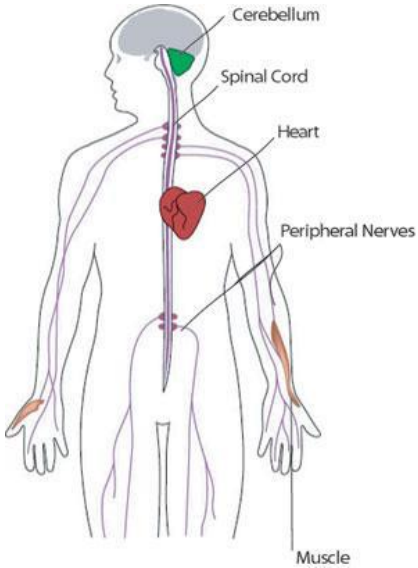

Figure 1: Friedreich ataxia affects the nervous system and may also affect heart function. **(Picture from the Muscular Dystrophy Association website: http://www.m dausa.org/)**

What is the genetic fault that causes FRDA?

95% of FRDA is caused by a genetic mistake that involves the repetition of a particular DNA region over and over again. The scientists refers to this as a 'triplet repeat'. The remainder is due to a combination of faults.

What are the symptoms of Friedreich ataxia?

The age at which symptoms will first appear varies. Generally, the symptoms appear before age 25 but they may also appear as early as age two or as late as 30 to 40 years. Longer triplet repeats are associated with more severe disease. Initial weakness and unsteadiness of the legs results in difficulties in standing and walking. An affected child might, for example, trip frequently over low obstacles. The difficulty in walking is usually followed by uncoordinated movements (ataxia) of the arms and hands causing difficulties in writing and other manipulative tasks. Other symptoms that appear early in the course of the disease are loss of knee and ankle tendon reflexes and speech difficulties.

Abnormal muscle control and tone leads to problems such as scoliosis (curvature of the spine) and foot deformities such as high-arched feet. Arm weakness, if it occurs, develops later in the course of the disorder. Loss of muscle control eventually makes it necessary to use a wheelchair.

Heart muscle enlargement with or without an abnormal heartbeat is present in about two-thirds of cases. About one–third of patients develop diabetes, most of whom will require insulin. Other symptoms of FRDA include optic nerve atrophy (wasting), eye tremor, loss of muscle mass, hearing loss, difficulty swallowing, and incontinence.

How common is Friedreich ataxia?

Friedreich ataxia is known to be the most common of the inherited ataxias, affecting approximately 1 in 50 000 individuals in Caucasian (white) populations, but is rare among sub-Saharan Africans and does not exist in the Far East.

How is Friedreich ataxia inherited?

FRDA is an autosomal recessive condition, which means that an affected child has inherited two faulty FRDA genes, one from each parent. In other words, each parent has at least one faulty FRDA gene, which has been passed on to the affected child. (Refer to Autosomal Recessive sheet).

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Is there treatment or cure for Friedreich ataxia?

Not at present. There is also no treatment to slow the progression of the disease. Treatment is aimed at supporting the patient and his/her family. Treatment includes psychological support, prostheses, walking aids, wheelchairs, physical therapy, and speech therapy -- all important for maintaining an active lifestyle. Orthopaedic interventions for scoliosis and foot deformities may be necessary. Treatment of associated cardiac disease and diabetes may help improve the quality of life in individuals with FRDA.

Testing

How does the test work?

The Division of Human Genetics at UCT provides genetic testing for people affected with FRDA (DNA diagnostic testing) and for their family members if they are over 18 years old (DNA predictive testing). This test has been shown to be highly reliable in determining if a person has inherited the mutated (faulty) genes that cause FRDA. The test detects the abnormally expanded triplet repeats that occur in 95% of FRDA patients.

FRDA Predictive Testing Protocol

The predictive test for FRDA allows testing of family members that may be at risk, before any clinical signs present. A programme or protocol has been recommended by doctors and geneticists based on their experiences in dealing with individuals and families at risk for the disease. If, after careful consideration, you decide to take the test, you will be requested to come to the Groote Schuur Hospital Neurogenetic Clinic or the Department of Human Genetics on at least four occasions to see the doctors involved in running the programme.

The full protocol for predictive testing is available at the following website: **http://www.uct.ac.za/depts/genetics.**

How soon will I have the results?

Results are available within 4 weeks of the test. The results will be communicated to you personally via your general practitioner, neurologist or by the staff of the Division of Human Genetics at the University of Cape Town.

Genetic counselling

As this is a genetic condition, genetic counselling is strongly recommended. Genetic counselling provides information on the condition, its inheritance pattern, risks to other family members and the prognosis. The Division of Human Genetics at the University of Cape Town (UCT) can be contacted in this regard.

Where can I read more about Friedreich ataxia?

You may find the following recourses about Friedreich ataxia helpful.

Genetics Home Reference website:
http://ghr.nlm.nih.gov/

Muscular Dystrophy Association website:
http://www.mdausa.org/

Muscular Dystrophy Foundation (MDF) of South Africa:

http://www.mdsa.org.za

Is there a Friedreich ataxia support group?

The Friedreich ataxia support group falls under the Muscular Dystrophy Foundation of South Africa. Please contact your local MDF office for information.

- **National Office:**

P.O. Box 1535, Pinegowrie, 2123

Tel: (011) 789-7634, Fax: (011) 789-7634

Email: national@mdsa.org.za

- **Cape Branch:**

P.O. Box 13449, Mowbray, 7705

Tel: (021) 448-8766, Fax: (021) 448-8766

Email: cape@mdsa.org.za

Who do I contact for more information regarding testing?

Division of Human Genetics Molecular Laboratory:

Prof. Jacquie Greenberg: (021) 406-6299

Genetic Nurses: (021) 406-6304