



FRIEDREICH'S ATAXIA

Useful Information for Lay People

FOREWORD

The Friedreich's Ataxia Society Ireland, which was founded in 1980, has produced this information booklet for lay people generally, but especially for the benefit of people disabled with Friedreich's Ataxia and any of the other genetic ataxias.

While all the general symptoms relating to FA are listed, it must be stressed that the presence and progression of these symptoms may vary from one FA person to another.

If you require further information, or if you wish to become a member of the Friedreich's Ataxia Society, please write to:

The Friedreich's Ataxia Society Ireland,
San Martino,
Mart Lane,
Foxrock,
DUBLIN 18.

Or ring: (01) 289 4788
Text: 086-6004285
E-Mail: info@ataxia.ie
Website: www.ataxia.ie

PART ONE

THE DISORDER

FA was first classified as a specific disorder in 1863 by a German physician named Nicholas Friedreich. The name of the disorder is derived, therefore, in part from the surname of its discoverer and partly from the ataxia symptoms that stem from it.

‘Ataxia’ means a breakdown in co-ordination and balance. There are a number of other disorders similar to Friedreich’s Ataxia that are not as common as FA, but they are embraced in the umbrella organisation. These include Cerebellar Ataxias, Ataxia Telangiectasia, AOA1 and AOA2.

THE SYMPTOMS

FA is a genetic disorder of the central nervous system that generally first becomes noticeable in children between the ages of six and sixteen, but in some cases there may be a much later onset.

The first sign is a breakdown in the person’s co-ordination, which slowly develops to a very marked unsteadiness. This may affect how a person walks, talks or writes. Trying to balance and control their body can make them very tired. As Friedreichs Ataxia is progressive the symptoms get worse as time goes on; this is usually a gradual process, and takes place over a number of years and eventually it may be necessary to use a wheelchair. The person will know when it is time to use a wheelchair as it will become easier, faster and less stressful to get around with one. It is very important that the wheelchair is fitted to the person’s individual needs by an Occupational Therapist.

Some other changes may occur as the disorder develops. The feet can become high-arched with a clawing of the toes and can also feel cold. Scoliosis, or curvature of the spine, may occur in some cases, and this in turn can cause breathing difficulties. In some instances, diabetes and cardiomyopathy (heart palpitations) may develop. The intellect is not affected.

INHERITANCE

Friedreich's Ataxia is a genetically transmitted disorder: that is, the tendency to develop it is passed on to a child by the genes (the basic units of inheritance) transmitted to him/her by both parents. Both the mother and the father are involved in the transmission. Therefore they are both carriers.

The body cells contain thousands of genes which always occur in pairs (except in the germ cells). Each pair determines one particular characteristic (e.g. eye colour or a bodily chemical process); these are commonly known as hereditary factors. Of the two genes in any pair, one will come from the mother via the genes in the ovum or egg cell, and one from the father via the genes in the male sperm cell.

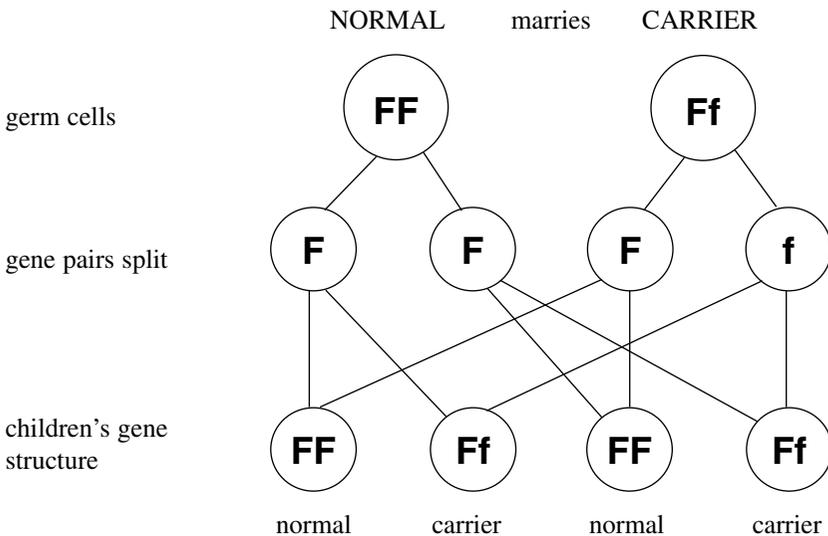
Each of the genes in any one pair can be either 'dominant' (that is, powerful) or 'recessive' (that is, weak relative to the dominant gene). Friedreich's Ataxia is transmitted by what is called a recessive mode of inheritance.

This is best illustrated by some diagrams. Suppose we call the dominant gene 'F', and the recessive (defective) gene 'f'. A completely normal person will have the gene pairing 'FF'. A person who has the gene pairing 'Ff' does not himself/herself have Friedreich's Ataxia, but is what is called a 'carrier' of the condition. A person who has the gene pairing 'ff' is a victim of Friedreich's Ataxia.

Classical Friedreich's Ataxia is carried on Chromosome 9 and the location of most of the other genetic ataxias has been discovered also. It is, therefore, possible to confirm the presence of a genetic ataxia and to identify it by a simple blood test. Similarly, it is also possible to identify if a person is a 'carrier' of the defective gene.

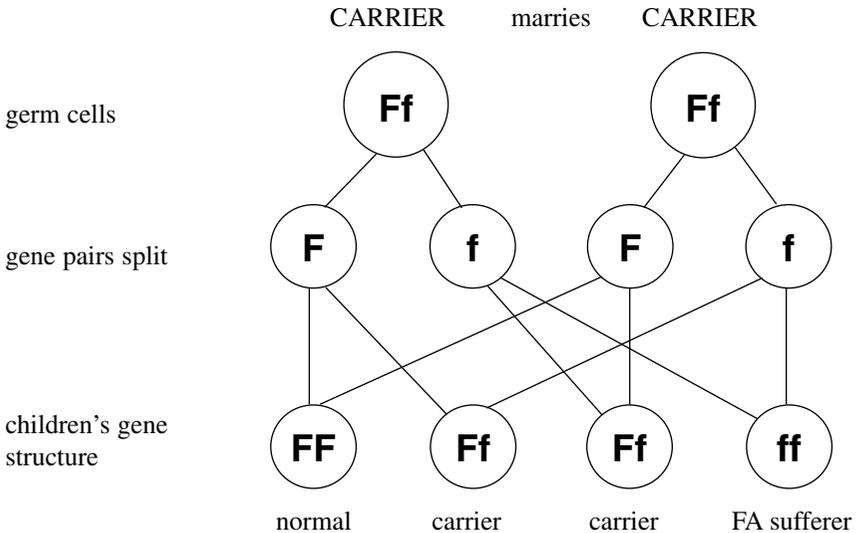
Let us consider two kinds of marriage

Marriage 1 – A normal person marries a "carrier"



There is a 50/50 chance that each child will be either normal or a carrier.

Marriage 2 – A “carrier” happens to marry another “carrier”



There is one chance in four of it being a normal child, two chances in four of it being a carrier, and one chance in four of it having FA. This is much the commonest way in which FA is inherited.

Similarly, it can be shown that, if an FA sufferer marries a normal person, all of the children will be carriers ($ff+FF=Ff,Ff,Ff,Ff$).

If an FA sufferer marries an FA carrier, there is a 50/50 chance of the children being either carriers or sufferers of FA.

Finally, in the event of two FA sufferers marrying, all of the children will themselves suffer from FA ($ff+ff=ff,ff,ff,ff$).

Note that the FA carrier is apparently quite normal. To test if one is a carrier, it is necessary to obtain a referral letter from your GP to Professor Andrew Green, Director at the National Centre for Medical Genetics at Our Lady's Hospital for Sick Children, in Crumlin, Dublin 12.

The carrier simply has the potential, if by chance he or she marries another carrier, of having children with FA.

So long as this does not happen, the carrier state may be passed on unknowingly for generation after generation in a family (as in Marriage 1 illustrated above).

It is very important to also note that, if two FA carriers marry, the 1 in 4 chance of them having an FA child is true for each child born. In other words, should they have, say, two FA children to start with, the chance of a third child having FA is unaffected – that is, the chance is still 1 in 4.

It should be remembered that FA is only one of many recessive genes that may, in a double dose, i.e. inherited from both parents, cause some form of illness and that everyone carries some deleterious genes.

The most common Recessive Ataxias are:

Friedreich's Ataxia (FRDA)

Ataxia Telangiectasia (AT)

Ataxia + Ocular Apraxia + Hypoalbuminemia (AOA1)

Ataxia + Ocular Apraxia + Elevated u-FP (AOA2)

DOMINANT ATAXIA

Dominant ataxia is passed on as a dominant hereditary disease. Each child born of a parent with dominant ataxia has a 50-50 chance of getting ataxia. It is passed directly from parent to child. If the child does not inherit ataxia, his or her children in turn cannot become affected, and future generations will be free of the disease. Children of either sex are equally likely to inherit the abnormal gene and to thus

develop ataxia. By chance, all offspring in one family may have ataxia while all offspring in another family may be free

The most Common Dominant Ataxias are:

Spinocerebellar Ataxia 1	(SCA1)
Spinocerebellar Ataxia 2	(SCA2)
Spinocerebellar Ataxia 3	(SCA3)
Spinocerebellar Ataxia 6	(SCA6)

RESEARCH

Since the early 90's, many research teams throughout the world have been involved in intensive research in an effort to locate and isolate the rogue gene. In 1996, it was finally discovered on chromosome 9 – by Massimo Pandolfo (an Italian scientist working in Houston, Texas) and confirmed by Michel Koenig (Strasbourg, France). The scientists discovered that a lack of hitherto unknown protein – which they have named FRATAXIN – in the spinal cord, heart and pancreas, is the likely primary cause of neuronal degeneration, cardiomyopathy and increased diabetes. Since then, Dr. Pandolfo and a number of laboratories in Canada, the United States, France, Italy and Belgium have clarified how lack of FRATAXIN, the protein expressed by the FA gene X25, appears to cause FA and how this cause might eventually be treated.

Recently there has been a greater surge in the research programme due to increased investments and co-ordination of effort on a worldwide basis. As a result of this, the medical scientists are very close to producing a suitable treatment for the disorder. However, some time will be needed for suitable controlled trials to be undertaken to ensure that the treatment is effective and without serious side effects.

The Friedreich's Ataxia Society Ireland is already giving strong support to this research programme and will continue to do so until a suitable treatment for the disorder becomes available. Please check our website www.ataxia.ie for any research updates.

MANAGEMENT OF FRIEDREICH'S ATAXIA

In the meantime, the Society will continue to provide help and support to the ataxia families in Ireland and to the medical people who serve these families. Apart from the help and information available from the FA Society, the family doctor may wish to call upon the services of a number of other experts. Thus a neurologist may be required to help in making the initial diagnosis and for periodic re views; an orthopaedic surgeon can sometimes help with operative intervention of appliances for foot and spinal deformities; an expert in physical medicine in the matter of exercises and mechanical aids; and a psychiatrist for emotional disturbances in the patient or relatives.

Diabetes may require treatment with insulin or oral anti-diabetic agents; congestive cardiac failure with diuretics, in-optrics etc. It is most desirable for the patient to keep up his/her walking for as long as possible, and the skills of occupational therapist, social workers etc. may all be very useful in that context.

How do we deal with Ataxia:

At present there is no known cure for the ataxias.

- But it is possible to manage ataxia with the help of medicines, as prescribed by your doctor.
- Physical exercise; a physiotherapist can recommend different exercises to keep your body strong.
- Regular visits to a qualified speech and language therapist to find ways of coping with speaking and swallowing (eating) difficulties (if they occur).
- Support from family, friends and other people, such as FASI.
- Swimming, any outdoor games or exercise within the person's limits.

PART TWO

THE FRIEDREICH'S ATAXIA SOCIETY OF IRELAND

The Friedreich's Ataxia Society Ireland (FASI) was established in 1980, and its address is:

San Martino,
Mart Lane,
Foxrock,
DUBLIN 18.
Tel: (01) 289 4788
Fax: (01) 289 8845
Text: 086-6004285

E-Mail: info@ataxia.ie
Website: www.ataxia.ie

The objectives of the Society are:-

1. To provide a forum in which people with Friedreich's and other hereditary ataxias, and their relatives, can meet, exchange views, make and comment on suggestions, and discuss their difficulties and aspirations.
2. To provide information on all aspects of the condition.
3. To provide help and support for people with FA (and their relatives) by advising and helping them in obtaining suitable accommodation and other necessary amenities. Advice is also given in relation to financial and other benefits to which they are entitled.

4. To keep in touch with similar groups throughout the world and with other support organisations in Ireland.
5. To act as a pressure group on behalf of people with FA and their families.
6. To fund research into FA, and investigations of methods of treatment and management of the condition, and to monitor and assess the research being undertaken in other countries.

Membership of the Society is available to any interested person and there are no entrance fees.

A good deal of emphasis is put on the help the Society can provide to relatives of those with FA, particularly when the disorder is first diagnosed. It helps to remedy their uncertainty and sense of loneliness at that difficult time, and can provide genetic information as well as support and general information, enabling them to adjust to their new situation.

The Society also arranges social and welfare services for its members. These include:-

SOCIALS: – a chance to meet up with fellow members for a meal, where members get together to discuss problems, hopes and ambitions;

RESPIRE CARE:– for disabled and their carers;

EQUIPMENT:– such as computers and suitable fixtures and fittings;

INFORMATION:– such as social welfare entitlements, grants, council house applications, personal assistants etc.;

- when required the Society will visit schools, teachers and also attend case conferences to represent our members as advocates;

COUNSELLING:– we organise counselling for members and their families;

ATAXIA
CLINICAL
SERVICES:– we run a special Ataxia Clinic every Tuesday in The Adelaide & Meath Hospital, Tallaght where we have an Ataxia Registrar and Ataxia Nurse who assess our members medical needs and where necessary organise, speech & language therapy, physiotherapy etc and then liaise with community services.

We would encourage anyone with an Ataxia to contact the Society to register for this Specialist Clinic. Once a treatment or cure is found, it will be very important to have a correct diagnosis so that the appropriate treatment can be given to every Ataxian.

If you need help or further information, please contact us.

Also, information on Entitlements for people with Disabilities is available from your local Health Centre OR

The Citizens Information Board

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George's Quay House

43 Townsend St

Dublin 2

Ireland

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Web: www.citizensinformationboard.ie

Updated: August 2008

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