FACT SHEET

FRIEDREICH ATAXIA

MUSCULAR DYSTROPHY NSW BUILDING STRENGTH REACHING POTENTIAL

Friedreich Ataxia (FA) is a degenerative condition caused by damage to the nervous system.

INCIDENCE: 1 : 50,000 and 1 : 90 per head of population are carriers of the gene
AVERAGE AGE OF DIAGNOSIS: 5 - 15 years of age
OCCURRENCE: affects males and females
LIFE EXPECTANCY: early to late adulthood depending on severity and significant heart complications

Friedreich Ataxia (FA) is an inherited condition that comes from both parents. FA is caused by a fault in the frataxin gene which leads to degeneration of the nerves of the spinal cord and peripheral nerves. The parts of the brain that control movement and balance become affected as they also degenerate but at a slower rate compared to the other muscles. FA causes the person to have awkward movements and impaired sensory function. FA causes heart and spine problems and some people may develop diabetes. FA does not affect cognitive function.

A young person affected by FA presents with walking difficulties and impaired muscle coordination known as ataxia. The ataxia becomes very debilitating as it spreads to the arms and trunk. Muscle weakness of the trunk leads to scoliosis or curvature of the spine. Speech and swallowing are also affected over time and this can become quite distressing for the person with FA as they lose the ability to communicate and eat. Hearing and vision loss may also develop in the later stages of FA. People with FA experience heart palpitations, chest pain and shortness of breath as the heart enlarges. Heart complications are the largest cause of death in FA.

There is currently no cure for FA. Treatments for FA focus on keeping the child/adult healthy and active for as long as possible. Regular physiotherapy aims to prolong function, walking and maintain the muscles used for breathing. Occupational therapy improves the skills of everyday living and focuses on equipment to enhance mobility. The person with FA will need an experienced physiotherapist and occupational therapist to ensure their needs are being met.

As FA progresses, a person's needs will change, and the amount of support and care needed will increase. It is important that they are assessed at regular intervals to plan their future medical, physical, social and psychological supports.

As children become adolescents and adults they will have goals and desires similar to their peers. This may include relationships, work options, study or planning for holidays and so on. Access and inclusion are paramount to ensure independence for education and community involvement. Issues related to puberty, relationships and sexuality may require professional intervention.

Planning ahead for services and equipment is essential, particularly given the degenerative nature of FA

The rate of progression varies, however the earlier the onset, the more severe the symptoms. Children diagnosed with FA will often lose the ability to walk in their teenage years. In adulthood, the weakness associated with FA can progress significantly and may affect the person's abilities to function at home, school and or at work. Adults with FA who experience increased physical difficulties will require support to help with issues such as physical access, transport, social exclusion, financial constraints, health risks, low self-esteem and daily support needs.

Given their changing needs, people with FA are at risk of becoming depressed and socially isolated, so it is important that a variety of opportunities are encouraged, for example accessible work options, sport and recreation. A person with FA may need to rely on others for a significant level of care support, which may impact family dynamics. Enough support needs to be provided to the person with FA so that their carer can have a break.

As FA progresses, medical complications can arise, therefore the following areas should be monitored: tightening of the joints and feet deformities; respiratory care including prevention and treatment of chest infections; sleep studies; non-invasive mechanical

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respiratory support; heart and annual reviews; speech and swallowing and eating; continence issues; curvature of the spine and possible surgery; and psychological issues as they grapple with the loss of independence and increased reliance on others such as family, friends or paid carers.

People with FA and their families may experience grief and loss dealing with the changes in function. There may come a time for referral to palliative care services for symptom management of pain and discomfort. The need for "end-of-life" care planning should be discussed when deemed appropriate for management of pain and discomfort.

It is important that service provision is centered around quality of life outcomes and meets the needs of the individual

Genetic testing and counselling should be offered to people with FA, parents and family members for future pregnancy planning. Families and individuals affected by chronic life limiting conditions benefit most when empowered to have control over their life choices. Through seeking information about what is available, people will be empowered to make the right decisions to achieve their goals. Despite the challenges a person with FA faces, there are many opportunities available to support people to live fulfilled and productive lives. Goal setting and positive attitudes contribute to achieving success in many facets of life.

CONSIDERATIONS FOR PLANNING SHOULD INCLUDE BUT ARE NOT LIMITED TO THE FOLLOWING:

- A consistent General Practitioner (GP)
- Specialists for monitoring and medical care planning eg. neurologist, respiratory, heart physicians
- Support coordination due to degenerative nature of FA and complex care needs
- Physiotherapy eg. hydrotherapy, stretching, chest physio
- Occupational therapy eg. to increase independence, equipment assessment
- Support for speech problems eg. speech therapy
- Assistive technology eg. scooter, manual wheelchair, power wheelchair, shower chair, hoist, slings, voice command devices, alternative augmentative communication devices (AAC)
- Orthotics eg. leg splints and ankle-foot orthoses (AFO's)
- Comfort/sleeping eg. air pressure relieving mattress, electronic bed with high/low options
- Continence management eg. urinary bottles, pans
- Nutrition support eg. dietician, supplements, straws, feeding equipment
- Opportunities to socialise and be away from home eg. MDNSW Camps, accessible travel
- Assistive ventilation devices eg. CPAP or BiPAP machines, cough assist machines
- Home modifications for access to all areas eg. ramps, lifts, bathrooms, bedrooms, kitchen, outdoor areas
- Vehicle modifications/transport eg. taxis to and from school or work and the community
- Adapted sporting and recreation equipment eg. boccia equipment, sports wheelchair
- Support workers eg. showering, toileting, meal preparation, feeding, community access
- Professional psychological support eg. counsellor, clinical psychologist, palliative care
- Future goal planning eg. independent living, study, work choices
- Financial entitlements eg. Centrelink, Medicare, Companion Card

USEFUL RESOURCES

MDNSW www.mdnsw.org.au
MDNSW NDIS Toolkit www.mdnsw.org.au/ndis
Brain Foundation www.brainfoundation.org.au/disorders/friedreichs-ataxia
FA Research Association www.fara.org.au

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