Friedreich's Ataxia

It is my joy in life to find
At every turning of the road
The strong arm of a comrade kind
To help me onward with my load.

F.D. Sherman

Friedreich's Ataxia (FA) is a debilitating, life-shortening, degenerative neuro-muscular disorder. A neuro-muscular disorder results in muscle weakness and fatigue. Although muscle wasting isn't painful, the resultant weakness can cause cramping, stiffness, joint deformities, chronic aches and pain, and sometimes the tightening and freezing of joints. Onset of symptoms can vary from childhood to adulthood.

What are the signs and symptoms of the condition?

- loss of coordination (ataxia) in the arms and legs.
- fatigue - energy deprivation and muscle loss.
- vision impairment, hearing loss, and slurred speech.
- aggressive scoliosis (curvature of the spine).
- diabetes mellitus (insulin-dependence, in most cases).
- a serious heart condition (enlarged heart - hypertrophic cardiomyopathy).

Not all these symptoms are present in people who have FA. The rate of deterioration and incapacitation affects each person differently and at varying times in their lives. The progressive loss of coordination and muscle strength leads to motor incapacitation and eventually the full-time use of a wheelchair. Most young people diagnosed with FA require mobility aids such as a cane, walker, or wheelchair by their teens or early twenties.

What is Friedreich's Ataxia?
FA is a genetic disorder. FA patients have gene mutations that limit the production of a protein called frataxin. This important protein called frataxin functions in the mitochondria (the energy producing factories) of the cell. Frataxin helps to move iron and is involved with the formation of iron-sulphur clusters, which are necessary components in the function of the mitochondria and thus energy production. We also know that specific nerve cells (neurons) degenerate in people with FA, and this is directly manifested in the symptoms of the disease.

Treatment for Friedreich's Ataxia
At present, there is no cure for FA — although the day is rapidly approaching when genetic and drug therapies may change that situation. Medical interventions have increased the life-span and improved the quality of life for many children. These interventions focus on treating or delaying symptoms, enhancing physical mobility and social interactions, and preventing heart and lung complications.

Common interventions include assistive equipment (computer equipment and software, walkers, wheelchairs etc.), physical and occupational therapy, surgery, nutritional support, cardiac and respiratory care.

Tips for Learning and Teaching

Students with FA demonstrate a range of cognitive abilities, and many have higher-than-normal intelligence. However muscle weakness and fatigue can make it difficult for students to keep up with the physical demands of handwriting, completing assignments, organising books and materials, etc.

Although each student has different needs, many students with FA experience numbness in their hands and/or feet. and may have great difficulty knowing where their hands and feet are 'in space'. Bearing this is mind darkness can be very confusing and disorientating for the student with FA. The following are some areas that require particular attention and awareness in relation to the student with FA:

- challenges in keeping up with class/home work.
- difficulty with written language and writing tasks.
- fatigue.
- difficulty paying attention and concentrating.
- participating in classroom activities/physical education.
- the importance of balancing the need to withdraw the student from whole-class instruction to avail of additional resource teaching.

- physical positioning and special seating.
- health concerns, such as infections and the slowed recovery for students with FA.
- social skills and peer relations.
- physical accessibility of the school, including doorway widths, stairs, heavy doors, water fountains, distances between classes, curbs, aisles between desks and accessible bathrooms.

Possible accommodations that can be made to assist the student with FA:

- special transportation.
- curriculum modifications .
- allocating special needs assistant support to meet student's care needs
- provision of additional resource teaching.
- creating opportunities to engage in un-timed tests.
- adapted keyboards, software and calculators.
- adaptive physical education.
- including rest-time in student's time-table.
- assistance on class outings/trips.
- providing for extra time to transition between classes.
- organising for an extra set of textbooks to be kept at home in order to minimise the need to carry heavy loads.
- classroom and school campus accessibility modifications.
- creating structured opportunities to assist student in making/accessing friends and social relationships.
- developing students' independence and self-care skills, especially as they move to senior classes in primary school and in secondary school through ensuring appropriate management of special needs assistant support, requesting required accommodations, developing whole-school awareness of students' needs and enabling students to make choices and decisions that assist students in developing emotional and social independence as their physical dependence increases.
- allowing alternative ways to demonstrate understanding of a concept, such as making an oral rather than a written report. As tape recording work is already available as an accommodation for public exams, students could use this where they feel comfortable with it.
- assigning work buddies and using computer-assisted learning.
- e-mailing homework to student who may not be able to come to school and using available technology to maintain contact with the student.
- including students in social and extracurricular activities.
- emphasising the student's strengths and abilities.

TEACHER TIPS

When teaching students with FA remember that:

- Students experience physical, academic and social challenges.
- Fatigue is a feature of the student's school day.
- Students with FA may have a general learning disability, however most students with FA are within the normal range of intelligence.
- Students may need to be provided alternative methods of demonstrating their understanding.
- Curriculum activities should be differentiated to ensure accessibility.
- Helping students make friends enhances their school experience and performance, and eliminates the possibility of bullying and teasing. Not all students enjoy large group physical activity and students should therefore be encouraged to engage in other small group games and to share their interests (indoors or outdoors) at break time by affording them the facility of a supervised room if necessary.
- Teachers should maintain high expectations of the student's school performance.
- Informed, co-operative, accessible schools and strong family-school links are vital in helping students overcome challenges and optimise their educational experiences.

When a teacher shows sincere interest and feels comfortable speaking with the student about his or her abilities and challenges, the student will more likely feel comfortable asking questions and expressing feelings.

Additional Resources/References

1. Central Remedial Clinic, Ireland: provides a range of specialised services for children and adults with physical disabilities, website: http://www.crc.ie/
2. Enable Ireland: provides services for children and their families, which address aspects of a child's physical, educational and social development from early infancy through to adolescence. Enable Ireland also provides services for adults, website: http://www.enableireland.ie/
3. Friedreich's Ataxia Society Ireland (FASI): The Society provides ongoing help and support to the families of those with Friedreich's and other Genetic Ataxias, website: www.ataxia.ie
4. Ataxia UK: provides support for people affected by ataxia and funds research into causes and potential treatments, website: www.ataxia.org
5. National Ataxia Foundation: provides information on ataxia to persons with ataxia, ataxia families, caregivers, the medical community, researchers, and others, who are interested in ataxia, website: www.ataxia.org
6. Friedreich's Ataxia Research Alliance (FARA): serves as a catalyst in public, private, scientific and patient-family communities, to build worldwide support and collaborations that drive medical research advances for Friedreich's ataxia, website: www.curefa.org
7. Friedreich's Ataxia Parents Group (FAPG): provides support and advice for parents, who have children with Friedreich's Ataxia, website: www.fortnet.org/fapg

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Notes