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.
