Muscular Dystrophy New Zealand

Spinocerebellar ataxia (SCA)

Spinocerebellar ataxia (SCA) is an umbrella term for a group of genetic disorders that results in slowly progressive loss of coordination of gait, causing clumsy and unsteady motion, and often loss of coordination of the hands, speech, swallowing and eye movements . Frequently, atrophy (wasting) of the cerebellum in the brain occurs. The cerebellum is where movement, posture, and balance are coordinated. The symptoms of the condition vary with the specific type of SCA (there are several), and with the individual patient. Signs and symptoms of the disorder typically begin in early adulthood but can appear anytime from childhood to late adulthood, depending on the genetic mutation that has caused this. Early onset forms of the conditions generally tend to be more severe, and progress faster.

Over time, individuals with SCA may develop a variety of other symptoms such as cognitive impairment or dementia, numbness, tingling, or pain in the arms and legs (sensory neuropathy); uncontrolled muscle tensing (dystonia); muscle wasting (atrophy), muscle twitches (fasciculations), rigidity, tremors, seizures, tinnitus, vertigo, and involuntary jerking movements (chorea). The condition may be complicated by vision disorders and eye movement paralysis, or have association with heart disease, breathing problems, bone abnormalities and diabetes depending on the type.

Diagnosis

SCA can be misdiagnosed as another neurological condition, such as multiple sclerosis (MS). To obtain a diagnosis, physical and neurological examination, review of family history and exclusion of non-genetic causes is carried out. One means of identifying the disease is with imaging such as an MRI to view the brain. Once the disease has progressed sufficiently, the cerebellum (a part of the brain) can be seen to have visibly shrunk. The most precise means of identifying SCA, including the specific type, is through molecular DNA analysis. Nomenclature and classifications for this condition and its various types has varied over the years and can still cause confusion and debate due to the overlap of symptoms with other conditions.

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Causes

The Spinocerebellar Ataxias are genetic, which means they are caused by a defect in a certain gene that is present from the start of a person's life. All of us have genes that have little alterations or variations but most of these do not cause disease, when they do they are called mutations. There are various genes that when mutated cause ataxia. What they all have in common is that they make abnormal proteins that affect the function of nerve cells, primarily in the cerebellum (and other parts of the brain) and the spinal cord. Some types also cause additional symptoms.

Inheritance Patterns

Autosomal dominant inheritance refers to an inheritance pattern where the gene that causes the ataxia is located on one of the autosomes and will affect males and females equally. An autosome is any of the chromosomes that are not sex chromosomes. All genes come in pairs and dominantly inherited means that the gene with the disease mutation dominates over the normal copy of the gene. Each child of a parent with an autosomal dominant ataxia gene has a 50/50 chance of whether they will inherit the ataxia gene or not.

Autosomal recessive inherited diseases also affect males and females equally but it takes a "double dose" of the ataxia gene to result in disease symptoms. Both parents must be carriers of the disease gene and each must pass on the ataxia gene to their child for the double dose that is needed to produce symptoms of the recessive disease. Each child of parents who are carriers of a recessive disease has a 25% chance of inheriting two ataxia genes so will develop the disease, a 50% chance of inheriting just one of the ataxia genes and, therefore, be a carrier and a 25% chance of inheriting no ataxia gene and be completely free of ataxia. Because a single recessive ataxia gene does not cause symptoms, it can be passed on in a family for generations without being recognized. Therefore, there is often no "family history" of ataxia if the disease is inherited as a recessive gene.

Ataxia Gene Identified in 1993

The first ataxia gene was identified in 1993 for a dominantly inherited type. It was called "Spinocerebellar Ataxia type1 (SCA1)". Subsequently, as additional dominant genes were found they were called SCA2, SCA3, etc. Generally the number behind the SCA refers to the order in which the gene was found. At this time, 36 different gene mutations have been found. In other words, there are dominant ataxia classifications from SCA1 to SCA 36. Genes have also been located for some of the

recessive ataxias, the most common being Friedreich's ataxia (FRDA). The most common type of dominantly inherited ataxia is SCA type 3.

Ataxia Results in the Degeneration of Nerve Cells

Eventually the affected nerve cells begin to function poorly and ultimately degenerate. As the disease progresses, muscles become less and less responsive to commands from the brain, causing coordination problems to become more pronounced. Those affected by poor
coordination will notice poor balance when walking, inability to run, clumsiness of the hands, a change in speech, or abnormal eye movements.

Treatment and prognosis

There is no known cure for spinocerebellar ataxia and treatments are generally to manage symptoms. SCA is progressive and a person with one of these conditions may eventually require the use of a wheelchair, and need assistance to perform daily tasks. Modification of the home with things such as grab bars, raised toilet seats, and ramps may be necessary. Speech therapy and communication devices such as writing pads and computer-based devices may benefit those affected with slurring speech. Weighted eating utensils and dressing hooks can help maintain independence. Weight control is important because obesity can exacerbate difficulties with ambulation and mobility. Individuals experiencing swallowing difficulties (dysphagia) may suffer significant weight loss and will benefit from seeing a speech language therapist and dietician.
Other symptoms in addition to the ataxia could include tremor, stiffness, pain, depression, spasticity, and sleep disorders, among others and these can often be treated with medication and / or therapy. Substances that have a neurotoxic effect, including alcohol, are best avoided. People with SCA should be followed by a neurologist annually with visits to physiotherapists, occupational therapists and other specialists as needed. Genetic counseling will be of benefit for patients and families affected by the hereditary ataxias.

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References

http://www.ataxia.org

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