Muscular Dystrophy New Zealand

Hereditary Spastic Paraplegia/Familial Spastic Paraparesis

What is Hereditary Spastic Paraplegia?

Hereditary spastic paraplegia (HSP), also called familial spastic paraparesis (FSP), refers to a group of inherited disorders that are quite different in their cause and presentation but all share the trait of weakness and spasticity (stiffness) and mild reduction of vibration sensation of the lower extremities and urinary urgency may occur. Initial symptoms that make be experienced are mild difficulties in walking style (causing what is called a spastic gait) and mild leg stiffness. This condition has a slow progression which may result in the person requiring the assistance of a cane, walker, or wheelchair. The number of people who have HSP varies from population to population with it being as low as 1.3:100,000 people in Ireland and up to 9.6:100,000 people in Spain.

Varieties of HSP

Pure or uncomplicated HSP presents with the primary features of HSP. Other forms can be more complicated and these present with other symptoms of a systemic or neurological nature. These can include different severity in presentation of ataxia (lack of muscle coordination), epilepsy, impaired vision due to cataracts, problems with the optic nerve and retina of the eye, cognitive impairment, peripheral neuropathy, and deafness.

Symptoms may begin in childhood or adulthood, depending on the particular HSP gene involved. When symptoms begin after childhood they usually progress slowly and steadily. If symptoms start in very early childhood they may not progress and therefore resemble spastic diplegic cerebal palsy. The severity of the condition varies with some people being severely disabled and others only mildly. Life expectancy for uncomplicated HSP is not affected.

Causes of HSP

More than 30 genes that are responsible for several forms of HSP have been identified, and many more will likely be identified in the future. These genes generally encode proteins that normally help maintain the function of axons (which conduct nerve impulses) in the spinal cord. Loss of the conduction of information in these cells causes the symptoms experience.

Muscular Dystrophy New Zealand

Inheritance

HSP has several forms of inheritance. Not all children in a family will necessarily develop symptoms, although they may be carriers of the abnormal gene

Depending on the specific gene defect that is causing the condition it is inherited in three ways which are described below:

X-linked recessive: The sex chromosomes X and Y determine if a baby will be a boy or a girl. X-Linked HSP is caused by a defect in a gene on the X chromosome. One functioning copy is enough to prevent X-Linked HSP. Girls receive an X from mum and an X from dad and are described as XX. Boys receive an X from mum and a Y from dad and are described as XY. As boys have only one X chromosome if they inherit an X chromosome with a defective gene then they will have X-Linked HSP. The mother is described as a 'carrier' and with one functioning EMD gene is usually unaffected. A carrier mother has a 25% chance in each pregnancy of having an affected male child.

Autosomal dominant: This means that one defective copy is enough for the disease to present. The gene with the defect is found on one of the 22 pairs of chromosomes not involved in sex determination. Men and women are equally likely to be affected. A person with an autosomal dominant condition has a 50% chance in each pregnancy that their child will also be affected.

Autosomal recessive: This means that both copies of the abnormal gene must be defective for the disease to develop fully. In this situation each parent is a carrier of one defective gene. Each child they have has a 25% chance of inheriting the disease.

Spastic gait is a walking style where the legs are held together and move in a stiff manner, with the toes seeming to drag and catch.

Genetic counseling is available to families who have had a diagnosis of hereditary spastic paraplegia (as there are several different inheritance patterns it is important that the diagnosis is correct). This service provides information, helps families understand inheritance patterns and what this means in their family, as well as enabling people to make more informed family-planning decisions. You can access this via your GP, self-refer or talk to an MDA Fieldworker.

Muscular Dystrophy New Zealand

Diagnosis

The diagnosis of HSP is primarily by neurological examination and observation of a spastic gait as well as testing to rule out other disorders. A family history may be taken by the Doctor as there is often a family history. MRI abnormalities, such as a thin corpus callosum (largest midline structure of the brain), may be seen in some of the complicated forms of HSP and can help the Doctor to come to a diagnosis.

Management

There are no specific treatments to prevent, slow, or reverse HSP. There is a multidisciplinary team approach to management of the symptoms that present during the course of this condition and to improve balance, strength and agility. A neurologist, physiotherapist, occupation therapist, and a dietitian may all be needed at some point.

Current recommendations are:

- Daily physical therapy aiming towards improving cardiovascular fitness and maintaining and improving muscle strength and gait and reducing muscle tightness.
- Referral to an Occupational Therapist to make sure any assistive walking devices or ankle-foot orthotics are used if appropriate.
- Drugs to reduce muscle spasticity (including Botox injections) and reduce urinary urgency.
- Annual or as needed evaluations by a neurologist and physiotherapist to monitor any progression of the condition and to make sure treatment programmes are relevant.
- Make sure you are given referrals as things change so that the new symptoms are getting the best treatment available.
- Avoid exposure to medications or chemicals that cause neuropathy if possible.

During Pregnancy:

HSP symptoms generally do not change significantly during pregnancy, unless a medication treating symptoms is stopped during the course of the pregnancy. In general, uncomplicated HSP does not pose increased risk for pregnancy, labour, or delivery. In general, having uncomplicated HSP does not increase risk associated with obstetric anaesthesia.

Muscular Dystrophy New Zealand

Research

Current research is looking at the following methods of treating muscular dystrophies:

• Gene therapy: a mechanism for supplementing defective genes with healthy genes in the tissues affected by neuromuscular disease;
• Gene silencing: turning off genetic instructions that cause the production of toxic proteins; and
• Cell therapy: transplanting new muscle cells, using stem cells or immature muscle cells from a donor or genetically corrected cells from the patient's own body.

Others are looking at ways to preserve muscle despite the presence of a degenerative disease.

Support

The MDA Fieldworkers are available for support. They have in-depth knowledge of a range of neuromuscular conditions, and will have a better understanding of your needs and challenges. Have a chat over the phone or they can come to you for a kanohi ki te kanohi/face-to-face visit. They may have some real practical suggestions that have worked for others to offer as well. This service is offered free of charge to MDA members and is funded through donations and grants. Contact your local MDA Branch to be put in contact with your fieldworker.

The MDA Support Network allows people with similar circumstances or challenges to come together to share their experiences and provide each other with emotional and moral support in addition to practical advice and information. By bringing together people with common experiences, support networks can provide an invaluable addition to medical care. The MDA of New Zealand Support Network currently has over 600 members throughout New Zealand who want to be in touch with others livings with neuromuscular conditions. Please see the MDA website www.mda.org.nz for contact details and more information that you might find relevant for you and your whanau.

Useful Websites

http://sp-foundation.org/
http://rarediseases.org/

References

http://www.ninds.nih.gov/disorders/hereditary_spastic_paraplegia/hereditary_spastic_paraplegia.htm
https://umaine.edu/edhd/research/acc/what-is-agenesis-of-the-corpus-callosum-acc/
http://www.ncbi.nlm.nih.gov/books/NBK1509/