What is Hereditary Spastic Paraplegia?

Hereditary Spastic Paraplegia (HSP) is a medical term for a condition that affects muscle function. The terms spastic and paraplegia comes from several words in Greek:

- ‘spastic’ means afflicted with spasms (an alteration in muscle tone that results in affected movements)
- ‘paraplegia’ meaning an impairment in motor or sensory function of the lower extremities (from the hips down)

What are the signs and symptoms of HSP?

Muscular spasticity

- Individuals with HSP commonly will have lower extremity weakness, spasticity, and muscle stiffness.
- This can cause difficulty with walking or a “scissoring” gait.

Other common signs or symptoms include:

- urinary urgency
- overactive or over responsive “brisk” reflexes
HSP is usually a chronic or life-long disease that affects people in different ways. HSP can be classified as either “Uncomplicated HSP” or “Complicated HSP”.

Types of Hereditary Spastic Paraplegia

1. Uncomplicated HSP:
   - Individuals often experience difficulty walking as the first symptom.
   - Onset of symptoms can begin at any age, from early childhood through late adulthood.
   - Symptoms may be non-progressive, or they may worsen slowly over many years.
   - Typically, individuals retain normal strength of the upper extremities and have no involvement of speech, chewing, or swallowing.

2. Complicated HSP:
   - In addition to the common symptoms of HSP, individuals with Complicated HSP have other system involvement. This may include other neurologic findings such as ataxia (balance issues), seizures, intellectual disability, dementia, muscular atrophy, vision changes, or peripheral neuropathy.

If you have any questions about DM1, please speak with your doctor, genetic counsellor, or nurse at the Neuromuscular and Neurometabolic Centre.

Notes and questions
How will having HSP affect my life?

Each person responds differently to HSPDM1. Outcome depends on the type and severity of your condition.

Living with a chronic condition can be challenging and emotional. Here are some ideas for living well with a chronic disease.

**Educate yourself**
- Learn as much as you can about the disease and how to manage symptoms. If you have questions, ask your health care providers. This will help you make informed decisions about your care.

**Take part in your care**
- Work closely with your health care providers, as a team. Follow your treatment plan. Keep track of your symptoms and your response to treatment. Tell your health care providers how you are doing.

**Take care of yourself**
- Learn to listen to your body. Rest and conserve your energy when you feel tired. When you are active, go at your own pace.

**Talk about your feelings**
- It is normal to have mixed emotions when you are living with a chronic illness. At times, you may feel sad, angry or helpless. It can help to talk about your feelings with your partner, someone close to you or another person who has faced a similar situation. Remember that your health care providers can also help.

What causes HSP?

HSP is a genetic condition that can inherited from your parents (see inheritance patterns below) or happen spontaneously (also known as ‘de novo’ gene change).

1. **Autosomal dominant inheritance:** this is when the condition is seen in several relatives across multiple generations (for example, your parents, aunts, uncles, children).

   ![Autosomal dominant inheritance diagram](image)

2. **Autosomal recessive inheritance:** this is when the condition occurs from a combination of two genetic changes, one inherited from each parent. This condition would likely only be observed in one generation (for example, siblings).

   ![Autosomal recessive inheritance diagram](image)

3. **X-linked inheritance:** this is when a woman carries the mutation in her genetic code but either does not have symptoms, or just has mild symptoms. Male relatives who inherit this genetic mutation have symptoms.

   ![X-linked inheritance diagram](image)
How do you know I have HSP?

Your doctor or specialist can tell if you have HSP from:

- talking with you about your symptoms and family history
- checking the strength of your muscles
- neurological exam
- reviewing the results of a genetic test for HSP

Genetic tests

- Based on the observed inheritance pattern running through your family, genetic testing can be ordered to sequence known, autosomal dominant, autosomal recessive, and/or x-linked genes associated with HSP.
- Genetic testing is performed through special bloodwork and can take a few months to get results.
- Genetic testing does not always find a disease-causing mutation and a clinical diagnosis is enough to confirm someone has HSP.

Electromyography (EMG)

- This test measures and listens to the electrical activity in your muscles and nerves. An acupuncture needle is inserted into the muscle and used to listen for distinct sounds and patterns of the electrical activity of the muscle.

How is HSP treated?

HSP is usually treated with a combination of methods. There are many different types of HSP and each subcategory may be associated with its own clinical symptoms. Your specialist will work with you to identify your particular subtype and design your treatment plan to meet your needs. The goal of treatment is to prevent or lessen your symptoms.

Mobility

- It is important to have neurological and strength assessments regularly to monitor the progression of your HSP.
- Assistive devices may be considered, depending on your abilities. This can include ankle-foot orthoses, Rollator walker, wheelchairs, or other assistive devices.

Exercise

- We suggest that people slowly increase exercise tolerance starting off slowly and gradually to eventually attain 3 to 4 sessions each week of mixed endurance (such as biking, rowing, walk, jog, elliptical) and strength (such as weights, elastic bands, pilates) exercise.
- We provide strength monitoring and exercise advice with a DVD and exercise hand-outs.

Nutrition

- Refer to Eating Well with Canada’s Food Guide to make sure you have enough protein and calories in your diet. Ask your family doctor for a copy or visit Health Canada’s website.
- We recommend that you take a multivitamin and Vitamin D (2000 IU a day for adults and 400 IU a day for children).
- A supplement of Creatine Monohydrate can also help improve strength in conjunction with exercise. The amount you should take is based on your weight. You can add it to a glass of juice or sprinkle it on cereal or yogurt.
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