HEREDITARY SPASTIC PARAPLEGIA

Definition and prevalence

Hereditary spastic paraplegia (HSP), also called familial spastic paraplegia or Strümpell-Lorrain disease, is a genetically and clinically heterogeneous group of inherited neurodegenerative disorders, characterised primarily by progressive spasticity and hyperreflexia in lower limbs as a result of nerve damage. NSP affects approximately 1 in 20 000 individuals, regardless of gender. In clinical aspect, NSP can be divided into pure and complex forms. The first are defined by slowly progressive weakness, spasticity in lower limbs, and occasional hypertonic urinary disturbances and reduction of lower extremity vibration sense. Complex forms feature additional neurological and non-neurological damages.

Etiology

Etiology and pathogenesis of this disease are not fully understood yet. Patoanatomy shows degeneration of the ends of the corticospinal tracts within the spinal cord. The ends of the longest fibers, which supply the lower extremities, are affected to a much greater extent than the fibers to the upper body. The disease is inherited as an autosomal dominant (over 70% of cases), autosomal recessive or X-linked recessive trait, and multiple recessive and dominant forms exist. Currently, over 40 genetic loci associated with various forms of HSP have been defined. However, only eleven autosomal and two X-linked genes have been identified to date, the genetic basis for most forms is to be further studied.

Diagnosis

The initial diagnosis is based on family history and clinical examination. Physical examination shows decreased sensitivity in the distal parts of limbs, hyperactive reflexes, and increased muscle tone. In general, the peripheral nerves are not affected in patients with pure forms of the disease, but there are reported rare cases of reduced perception of acute stimuli in below knees. Deep tendon reflexes are pathologically increased in lower extremities. Further examination is required (image diagnosis, electroencephalography, long chain fatty acids, electromyography, and human T-cell lymphotropic virus type 1 serology), so differential diagnosis could exclude other syndromes. For example, MRI is an important procedure for the exclusion of other common neurological diseases, such as multiple sclerosis, but also for detection of concomitant disorders. Final confirmation of diagnosis can be provided by genetic tests for HSP identified genetic mutations.

Clinical picture

The main symptom is progressive, often severe spasticity of lower extremities. In uncomplicated autosomal dominant HSP after normal gestation, delivery, and early childhood development, subjects develop leg stiffness and gait disturbance (eg., stumbling, tripping) because of difficulty in dorsiflexing the foot and weakness in hip flexion. Classic symptom of HSP is progressive difficulty in walking, but severity varies. Some patients eventually may require the use of a wheelchair, while others may never need any type of assistive device. Other features of the disease include:

- decreased sense of balance;
- urinary problems;
- visual disturbance;
- fasciculations;
- dementia and seizures;
- peripheral neuropathy, cerebellar ataxia, and sensory disturbances.

Listed symptoms are not mandatory and may not be developed in all patients. Neurologic examination reveals no evidence of reduced mentation and cranial nerve dysfunction.

Treatment

Currently, no specific treatment exists to prevent, retard, or reverse the progressive disability in patients with HSP. Therapies mainly consist of symptomatic medical management and promotion of physical and emotional well-being. Medicinal therapies include muscle relaxants and spasmolytic agents. Botulinum toxin may be applied to reduce muscle overactivity. Antidepressants are used in patients, experiencing clinical depression. Orthoses may be required in case of footdrop. Surgical treatment targets frequent contractures and tendon problems.

Although it is a progressive disorder, the prognosis for people with NSP varies considerably. The disease primarily affects lower limbs, although upperbody may be damaged in some individuals too. There are reported cases of serious disability, as well as cases that are fully compatible with a productive and fulfilling life. The majority of patients with HSP have a normal life expectancy.

Rehabilitation and follow-up cares

Regular physical therapy is important to maintain and improve range of motion and muscle strength. Although it does not reduce the degenerative process within the spinal cord, individuals with HSP must maintain an exercise regimen at least several times each week, guided by their physical therapist. Exercise can help retain or improve muscle strength, minimize atrophy of the muscles caused by disuse, increase endurance and reduce fatigue, help prevent spasms and cramps.

Physiotherapy is aimed at maintaining the overall functional status, as well as managing accompanying symptoms of the disease such as pain, stiffness, poor coordination and motor difficulties. Physical rehabilitation uses 3 complementary approaches towards NPS. These are manual therapy, exercises and electrotherapy. They adjust and strengthen muscles and joints, increasing overall operational capacity of the body.

Last but not least, this rehabilitation complex has a positive psychological effect on the patient, and acts as a functional readjustment instrument. Physiotherapy promotes the ability to independently perform daily activities.

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