Autosomal Recessive Hereditary Spastic Paraplegia (AR-HSP)

Record Status
This is a bibliographic record of a published health technology assessment. No evaluation of the quality of this assessment has been made for the HTA database.

Citation

Authors' conclusions
Hereditary spastic paraplegia (HSP), also known as familial spastic paraparesis or Strümpell-Lorrain syndrome, is a clinically and genetically heterogeneous group of neurodegenerative disorders. HSP, which has an estimated prevalence that ranges from approximately 2 to 10 per 100,000 individuals, comprises more than 40 different subtypes, typically referred to as SPGs (for spastic paraplegia). The clinical presentation of each subtype may be classified as either pure or complex. Pure (or uncomplicated) HSP is characterized by progressive lower limb weakness and spasticity (stiffness) in the absence of additional neurological symptoms, although some patients with pure HSP may experience mild sensory disturbance and/or bladder dysfunction. In contrast, complex (or complicated) HSP is characterized by the presence of progressive lower limb weakness and spasticity in combination with additional neurological and/or non-neurological symptoms, such as cognitive impairment, dementia, seizures, signs of cerebellar dysfunction, neuropathy (peripheral nerve dysfunction), and amyotrophy (muscle wasting). HSP may be inherited in an autosomal recessive (i.e., both copies of the causative gene need to be altered in order to have the condition), autosomal dominant (i.e., only 1 copy of the causative gene needs to be altered in order to have the condition), or X-linked (i.e., caused by variants in a gene located on the X chromosome and occurring primarily in males) manner. To date, at least 24 autosomal recessive HSPs (AR-HSPs) have been described. Clinically, AR-HSP subtypes are more likely to have a complex presentation, although there is significant overlap in the clinical features of the various SPGs, and some subtypes may manifest as either pure or complex HSP. As a result, the clinical diagnosis of a specific ARHSP subtype may be difficult. For 8 AR-HSP subtypes (SPG5A, SPG7, SPG11, SPG15, SPG20, SPG21, SPG30, and SPG35), there is a clinically available gene test, which may be used to help establish a specific diagnosis and confirm autosomal recessive inheritance. Finally, depending on the subtype, the age at which AR-HSP symptoms first appear is highly variable. Moreover, while all AR-HSP subtypes are progressive and often result in an inability to walk unassisted, the speed at which the condition progresses may vary significantly. There is no cure for AR-HSP and no way to prevent or delay onset. Treatment is based on the symptoms present and may include physical and occupational therapies, medication to treat spasticity or other complications, and the use of medical devices to assist with ambulation in the later stages of disease.

Final publication URL
The report may be purchased from: http://www.hayesinc.com/hayes/crd/?crd=15726

Indexing Status
Subject indexing assigned by CRD

MeSH
Spastic Paraplegia, Hereditary; Cognition Disorders

Language Published
English

Country of organisation
United States

English summary