# A Look at Upper Motor Neuron Diseases
## Hereditary Spastic Paraparesis and Primary Lateral Sclerosis

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Sources:


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| | **Hereditary Spastic Paraplegia** | **Primary Lateral Sclerosis** |
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| **What is it?** | A group of degenerative, neurological disorders chiefly affecting upper motor neurons and principally causing progressive spastic weakness of the legs. Also known as familial spastic paraplegia or paraparesis (FSP) and Strumpell-Lorrain syndrome. | A group of degenerative, neurological disorders chiefly affecting upper motor neurons and principally causing progressive spastic weakness of the legs as well as the arms and bulbar muscles. |
| **Incidence rate** | Estimated at 10,000–20,000 individuals in the U.S. It may be higher, as it is frequently misdiagnosed or undiagnosed. | Estimated at 300-500 individuals in the U.S. It may be lower or higher, due to misdiagnosis or changing diagnosis. |
| **Predominant features** | Insidious, progressive spasticity and weakness of the legs that often gets severe, requiring assistive devices. There is also difficulty with balance, clumsiness, and often muscle spasms. | Insidious, progressive spasticity and weakness of the legs that often gets severe, requiring assistive devices. There is also difficulty with balance, clumsiness, and often muscle spasms. In time, weakness and spasticity in the arms and hands also occurs, as well as slurred speech, drooling and difficulty swallowing. |
| **Secondary features** | Urinary urgency and frequency is common and high arched feet are often present. Very rare types can present speech problems, ataxia, mental retardation, dementia, visual or hearing dysfunctions, extrapyramidal dysfunctions, adrenal insufficiency, or ichthyosis. | |
| **What causes it?** | HSP is hereditary, with some 30 genes thought to cause different types of HSP. Most forms are autosomal dominant, others are X-linked or autosomal recessive. | PLS is thought to be spontaneous. There is a rare, autosomal-recessive, childhood-onset form. |
| **What is going wrong?** | The upper motor neurons in the brain and spinal cord degenerate.<br><br>Upper motor neurons control voluntary movement. They deliver signals to lower motor neurons, which carry messages to the muscles. Because upper motor neurons degenerate, nerve impulses cannot adequately reach the lower motor neuron, and the lower motor neuron | The upper motor neurons in the brain and spinal cord degenerate.<br><br>Upper motor neurons control voluntary movement. They deliver signals to lower motor neurons, which carry messages to muscles. Because upper motor neurons degenerate, nerve impulses cannot adequately reach the lower motor neuron, and the lower motor |

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| | cannot relay the correct message out to the muscles.<br><br>This causes spasticity (increased muscle tone/stiffness) and weakness, which increase as the degeneration progresses. | neuron cannot relay the correct message out to the muscles.<br><br>This causes muscle spasticity (increased muscle tone/stiffness) and weakness, which increase as the degeneration progresses. |
| **How is it diagnosed?** | HSP is a clinical diagnosis made through exclusion of other possibilities and examining family history. Absence of documented family history cannot rule out HSP. It is estimated some 30% of individuals with HSP do not have documented family history. Gene testing can confirm dominantly inherited HSP in 45% of patients.<br><br>Early stages of HSP can mimic PLS or ALS. In the absence of family history, neurologists watch for upper body symptom development to indicate PLS or lower motor neuron involvement to indicate ALS. | PLS is a clinical diagnosis made through exclusion of other possibilities and examining family history. Absence of documented family history cannot rule out HSP as a possible diagnosis.<br><br>Early stages of PLS can mimic HSP or ALS. Neurologists watch for upper body symptoms to confirm PLS or lower motor neuron involvement to indicate ALS. EMG, nerve conduction tests and symptoms of lower motor neuron involvement distinguish PLS from ALS. |
| **Age of onset** | Symptoms can begin at any age from childhood through late adulthood. Most patients experience onset of symptoms in the second through fourth decades of life. | The reported age of onset ranges from 35-66 years with a median of 50.5 years. A rare, child-onset form has been reported. |
| **What is the prognosis?** | It affects the quality of life. Difficulty walking usually gets slowly worse, often requiring canes, walkers, or wheelchairs. However, some individuals with childhood-onset of symptoms experience very little worsening.<br><br>There is currently no cure. | It affects the quality of life. Difficulty walking usually gets slowly worse, often requiring canes, walkers, or wheelchairs. Speech and swallowing difficulty may become severe, as well as weakness of the arms.<br><br>There is currently no cure. |
| **What is the treatment?** | There is no treatment to prevent, retard or reverse the degenerative process. Treatment is focused on symptom relief (medications for spasticity), physical therapy and exercise, assistive devices and supportive therapy. | There is no treatment to prevent, retard or reverse the degenerative process. Treatment is focused on symptom relief (medications for spasticity), physical therapy and exercise, assistive devices, speech therapy and supportive therapy. |
| **What research is being done?** | There have been few researchers working on HSP. Fortunately, there are more today and research is accelerating.<br><br>Six HSP genes have been discovered and the search continues for more. Scientists are working to understand the genes and how mutations lead to upper nerve degeneration. Mouse models are now underway. There is also research being conducted regarding spasticity treatments and understanding neurological functioning. It is also hopeful treatments or cures discovered for other neurological conditions may be applicable to HSP. | There have been few researchers working on PLS. Fortunately, there are more today and research is accelerating.<br><br>PLS research is currently done in conjunction with research on related disorders. A gene for a very rare, familial form of PLS has been identified. Scientists are working to understand this gene and how mutations lead to upper nerve degeneration. There is also research being conducted regarding spasticity treatments and understanding neurological functioning. It is hopeful that treatments or cures discovered for other neurological conditions may be applicable to PLS. |