Pediatric Neurology Part III: Chapter 195. Hereditary spastic paraplegias: one disease for many genes, and still counting (Handbook of Clinical Neurology)

Paola S. Denora, Filippo M. Santorelli, Enrico Bertini

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Hereditary spastic paraplegias (HSPs) are genetically heterogeneous Mendelian disorders characterized by spastic gait with stiffness and weakness in the legs and an associated plethora of neurological or extraneurological signs in “complicated” forms. Major advances have been made during the past two decades in our understanding of their molecular bases with the identification of a large number of gene loci and the cloning of a set of them. The combined genetic and clinical information obtained has permitted a new, molecularly-driven classification and an improved diagnosis of these conditions. This represents a prerequisite for better counseling in families and more appropriate therapeutic options. However, further heterogeneity is expected and new insight into the possible mechanisms anticipated.

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