Troyer Syndrome

National Institute of Neurological Disorders and Stroke (NINDS)

Source

National Institute of Neurological Disorders and Stroke (NINDS). Troyer Syndrome Information Page.

Troyer syndrome is one of more than 40 genetically-distinct neurological disorders known collectively as the hereditary spastic paraplegias. These disorders are characterized by their paramount feature of progressive muscle weakness and spasticity in the legs. Additional symptoms of Troyer syndrome (also called SPG20) include leg contractures, difficulty walking, speech disorders, drooling, atrophy of the hand muscles, developmental delays, fluctuating emotions, and short stature. Onset is typically in early childhood, and symptoms gradually worsen over time. Troyer syndrome is an autosomal recessive disorder (meaning that both parents must carry and pass on the defective gene that produces the illness) that results from a mutation in the spastic paraplegia gene (SPGP20) located in chromosome 13 that results in loss of the spartin proteins. The disease was first observed in Amish families in Ohio. Diagnosis is made by specialized genetic testing.