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Gaze Palsy: A Genetic Autosomal Recessive Disease with Progressive Scoliosis

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Opinion

Horizontal Gaze Palsy with Progressive Scoliosis (HGPPS) is a rare autosomal recessive congenital disorder which is characterized by the absence of conjugate horizontal eye movements, and progressive debilitating scoliosis during childhood and adolescence.

Horizontal Gaze Palsy with Progressive Scoliosis (HGPPS) is a disorder which affects vision and also causes an abnormal curvature of the spine (scoliosis). People with this condition are unable to move their eyes side-to-side. As a result, affected individuals must turn their head instead of moving their eyes to track moving objects. Up-and-down eye movements are typically normal.

Scoliosis develops in infancy or childhood and worsens over time. Scoliosis can be painful and may interfere with movement so it is often treated with surgery early in life. HGPPS is caused by changes (mutations) in the *ROBO3* gene and is inherited in an autosomal recessive manner. Treatment is based on the signs and symptoms present in each person.

Horizontal Gaze Palsy with Progressive Scoliosis (HGPPS) is a rare congenital autosomal recessive disease, presenting in children and adolescents, and characterized by progressive scoliosis along with the absence of conjugate horizontal eye movements and associated with failure of the somatosensory and corticospinal neuronal tracts to decussate in the medulla.

HGPPS is caused by the mutations in the *ROBO3* gene. This gene provides instructions for making a protein which is important for the normal development of certain nerve pathways in the brain. These include motor nerve pathways, which transmit information about voluntary muscle movement, and sensory nerve pathways, which transmit information about sensory input such as touch, pain and temperature. These nerve pathways must cross from one side of the body to the other in the brainstem, a region that connects the upper parts of the brain with the spinal cord for the brain and the body to communicate effectively.

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

HGPPS is an autosomal recessive neurologic disorder which is characterized by eye movement abnormalities apparent from birth and childhood-onset progressive scoliosis. These features which are associated with a developmental malformation of the brainstem include hypoplasia of the pons and cerebellar peduncles and defective decussation of certain neuronal systems.

Although several conventional brain Magnetic Resonance Imaging (MRI) studies have described its anatomical features and clinical relevance studies incorporating Diffusion Tensor Imaging (DTI) tractography and electrophysiological assessment are limited. Herein, we describe a case with clinical-radiological features typical for HGPPS, and further explore its relevant pathogenesis on a neuroanatomic basis.

Patients with other conditions including Möbius syndrome and Duane retraction syndrome may also have similar clinical presentations of congenital horizontal gaze paresis. They frequently exhibit additional neurological signs related to agenesis of other cranial nuclei, but not to have a spilt pons sign pathognomonic to HGPPS.

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