Snyder-Robinson Syndrome

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Snyder-Robinson syndrome is a condition characterized by intellectual disability, muscle and bone abnormalities, and other problems with development. It occurs exclusively in males.

Keywords: genetic conditions

1. Introduction

Males with Snyder-Robinson syndrome have delayed development and intellectual disability beginning in early childhood. The intellectual disability can range from mild to profound. Speech often develops late, and speech difficulties are common. Some affected individuals never develop any speech.

Most affected males are thin and have low muscle mass, a body type described as an asthenic habitus. Weakness or "floppiness" (hypotonia) typically becomes apparent in infancy, and the loss of muscle tissue continues with age. People with this condition often have difficulty walking; most have an unsteady gait.

Snyder-Robinson syndrome causes skeletal problems, particularly thinning of the bones (osteoporosis) that starts in early childhood. Osteoporosis causes the bones to be brittle and to break easily, often during normal activities. In people with Snyder-Robinson syndrome, broken bones occur most often in the arms and legs. Most affected individuals also develop an abnormal side-to-side and back-to-front curvature of the spine (scoliosis and kyphosis, often called kyphoscoliosis when they occur together). Affected individuals tend to be shorter than their peers and others in their family.

Snyder-Robinson syndrome is associated with distinctive facial features, including a prominent lower lip; a high, narrow roof of the mouth or an opening in the roof of the mouth (a cleft palate); and differences in the size and shape of the right and left sides of the face (facial asymmetry). Other signs and symptoms that have been reported include seizures that begin in childhood and abnormalities of the genitalia and kidneys.

2. Frequency

Snyder-Robinson syndrome is a rare condition; its prevalence is unknown. About 10 affected families have been identified worldwide.

3. Causes

Snyder-Robinson syndrome results from mutations in the *SMS* gene. This gene provides instructions for making an enzyme called spermine synthase. This enzyme is involved in the production of spermine, which is a type of small molecule called a polyamine. Polyamines have many critical functions within cells. Studies suggest that these molecules play roles in cell growth and division, the production of new proteins, the repair of damaged tissues, the function of molecules called ion channels, and the controlled self-destruction of cells (apoptosis). Polyamines appear to be necessary for normal development and function of the brain and other parts of the body.

Mutations in the *SMS* gene greatly reduce or eliminate the activity of spermine synthase, which decreases the amount of spermine in cells. A shortage of this polyamine clearly impacts normal development, including the development of the brain, muscles, and bones, but it is unknown how it leads to the specific signs and symptoms of Snyder-Robinson syndrome.

3.1 The gene associated with Snyder-Robinson syndrome

- SMS

4. Inheritance

This condition is inherited in an X-linked recessive pattern. The gene associated with this condition is located on the X chromosome, which is one of the two sex chromosomes. In males (who have only one X chromosome), one altered copy of the gene in each cell is sufficient to cause the condition. In females (who have two X chromosomes), a mutation would have to occur in both copies of the gene to cause the disorder. No cases of Snyder-Robinson syndrome in females have been reported.

5. Other Names for This Condition

- mental retardation, X-linked, syndromic, Snyder-Robinson type
- Snyder-Robinson X-linked mental retardation syndrome
- spermine synthase deficiency
- SRS

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