

White-Sutton syndrome

Description

White-Sutton syndrome is a disorder that causes intellectual disability, specific facial features, and other signs and symptoms affecting various parts of the body. Most affected individuals have features of autism spectrum disorder (ASD), a varied condition characterized by impaired social skills, communication problems, and repetitive behaviors. However, in White-Sutton syndrome these features can occur along with other characteristics that are unusual in people with ASD, such as an overly friendly demeanor.

People with White-Sutton syndrome have delayed development, with speech and language usually being more delayed than motor skills such as walking. Intellectual disability can range from borderline normal to severe.

Most people with White-Sutton syndrome have mild abnormalities of the head and face, which can include an unusually small head (microcephaly); a wide, short skull (brachycephaly); wide-set eyes (hypertelorism); a flat or sunken appearance of the middle of the face (midface hypoplasia); and a small mouth with a thin upper lip.

A wide variety of additional signs and symptoms can occur with White-Sutton syndrome. Among the more common are hyperactivity; sleeping difficulties; vision defects, especially farsightedness; gastrointestinal problems; obesity; and short stature. Some individuals with White-Sutton syndrome are born with a hole in the muscle that separates the abdomen from the chest cavity (the diaphragm), which is called a diaphragmatic hernia.

Frequency

The prevalence of White-Sutton syndrome is unknown. Researchers estimate that changes in the gene associated with White-Sutton syndrome may account for up to 1 in 700 cases of intellectual disability, autism spectrum disorder, or both. However, most of these affected individuals have not been diagnosed with White-Sutton syndrome and may not exhibit all the features of this disorder.

Causes

White-Sutton syndrome is caused by mutations in the *POGZ* gene. This gene provides instructions for making a protein that is found in the cell nucleus. The POGZ protein

attaches (binds) to chromatin, which is the network of DNA and proteins that packages DNA into chromosomes. Binding of the POGZ protein is part of the process that changes the structure of chromatin (chromatin remodeling) to alter how tightly regions of DNA are packaged. Chromatin remodeling is one way gene activity (expression) is regulated; when DNA is tightly packed gene expression is lower than when DNA is loosely packed. Regulation of gene expression by the POGZ protein is thought to be important to brain development, but the specific function of POGZ in the brain is not well understood.

POGZ gene mutations are thought to impair the ability of the POGZ protein to bind to chromatin, leading to abnormal gene expression that affects development of the brain and other body systems. However, little is known about the specific changes in gene expression and how they lead to the development of intellectual disability and other signs and symptoms of White-Sutton syndrome.

[Learn more about the gene associated with White-Sutton syndrome](#)

- POGZ

Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Most cases of this condition occur in people with no history of the disorder in their family, and result from new (de novo) mutations in the gene that occur during the formation of reproductive cells (eggs or sperm) or in early embryonic development.

Other Names for This Condition

- Mental retardation, autosomal dominant 37
- MRD37
- WHSUS

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Intellectual disability-microcephaly-strabismus-behavioral abnormalities syndrome (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C4225351/>)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

Clinical Trials

- ClinicalTrials.gov ([https://clinicaltrials.gov/search?cond=%22White-Sutton syndrome%22](https://clinicaltrials.gov/search?cond=%22White-Sutton%20syndrome%22))

Catalog of Genes and Diseases from OMIM

- WHITE-SUTTON SYNDROME; WHSUS (<https://omim.org/entry/616364>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28pogz%5BTIAB%5D%29+AND+%28intellectual+disability%5BTIAB%5D%29%29>)

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