SATB2-associated syndrome

Description

SATB2-associated syndrome is a condition that affects several body systems. It is characterized by intellectual disability, severe speech problems, dental abnormalities, abnormalities of the head and face (craniofacial anomalies), and behavioral problems. Some of the common features can be described using the acronym SATB2 (which is the name of the gene involved in the condition): severe speech anomalies, abnormalities of the palate, teeth anomalies, behavioral issues with or without bone or brain anomalies, and onset before age 2.

Individuals with *SATB2*-associated syndrome typically have mild to severe intellectual disability, and their ability to speak is delayed or absent. Development of motor skills, such as rolling over, sitting, and walking, can also be delayed. Many affected individuals have behavioral problems, including hyperactivity and aggression. Some exhibit autistic behaviors, such as repetitive movements. A happy or overly friendly personality is also common among individuals with *SATB2*-associated syndrome. Less common neurological problems include feeding difficulties and weak muscle tone (hypotonia) in infancy. About half of affected individuals have abnormalities in the structure of the brain.

The most common craniofacial anomalies in people with *SATB2*-associated syndrome are a high arch or an opening in the roof of the mouth (high-arched or cleft palate), a small lower jaw (micrognathia), and dental abnormalities, which can include abnormally sized or shaped teeth, extra (supernumerary) teeth, or missing teeth (oligodontia). Some people with *SATB2*-associated syndrome have other unusual facial features, such as a prominent forehead, low-set ears, or a large area between the nose and mouth (a long philtrum). People with this disorder may also have a shortage of minerals, such as calcium, in bones (decreased bone mineral density), which makes the bones brittle and prone to fracture.

Less-commonly affected are the heart, genitals and urinary tract (genitourinary tract), skin, and hair.

Frequency

SATB2-associated syndrome is a rare condition. Its prevalence is unknown.

Causes

SATB2-associated syndrome is caused by genetic changes that affect the *SATB2* gene. These include changes within the *SATB2* gene itself and deletions of large pieces of DNA from chromosome 2 that remove the *SATB2* gene and other nearby genes. The *SATB2* gene provides instructions for making a protein that is involved in the development of the brain and structures in the head and face. The SATB2 protein directs development by controlling the activity of multiple genes in a coordinated fashion.

Researchers suspect that genetic changes affecting the *SATB2* gene reduce the amount of functional SATB2 protein. Reduction of SATB2 function likely impairs normal development of the brain and craniofacial structures, leading to intellectual disability, delayed speech, craniofacial anomalies, and other features of *SATB2*-associated syndrome.

The signs and symptoms of *SATB2*-associated syndrome are usually similar, regardless of the type of alteration that causes it. However, uncommon features of the condition, such as problems with the heart, genitourinary tract, skin, or hair, tend to occur in individuals with large deletions. Researchers suspect these features are related to the loss of other genes near *SATB2*.

Learn more about the gene and chromosome associated with SATB2-associated syndrome

- SATB2
- chromosome 2

Inheritance

SATB2-associated syndrome is not typically inherited. It results from new (de novo) changes in the gene that occur during the formation of reproductive cells (eggs or sperm) or in early embryonic development. Affected individuals have no history of the disorder in their family.

Other Names for This Condition

- 2q32 deletion syndrome
- 2q33.1 microdeletion syndrome
- Chromosome 2q32-q33 deletion syndrome
- Glass syndrome
- SAS

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Chromosome 2q32-q33 deletion syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C2676739/)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- GLASS SYNDROME; GLASS (https://omim.org/entry/612313)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28SATB2-associated+syndrome%5BTIAB%5D%29+OR+%28SATB2+haploinsufficiency%5BTIAB%5D%29+OR+%282q33.1+microdeletion+syndrome%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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