SATB2-associated syndrome

SATB2-associated syndrome is a condition that affects several body systems. It is characterized by intellectual disability, severe speech problems, dental abnormalities, other 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 overfriendly 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).

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 mutations 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 mutation 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*.

Inheritance Pattern

SATB2-associated syndrome is not typically inherited. It results from new (de novo) mutations 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

Diagnosis & Management

Genetic Testing Information

- What is genetic testing?
  /primer/testing/genetictesting
- Genetic Testing Registry: Chromosome 2q32-q33 deletion syndrome
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C2676739/

Other Diagnosis and Management Resources

- GeneReview: SATB2-Associated Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK458647
- SATB2gene.com: Making the Diagnosis
  https://satb2gene.com/making-the-diagnosis/

Additional Information & Resources

Health Information from MedlinePlus

- Encyclopedia: Intellectual Disability
  https://medlineplus.gov/ency/article/001523.htm
- Encyclopedia: Speech Disorders - Children
  https://medlineplus.gov/ency/article/001430.htm
- Health Topic: Craniofacial Abnormalities
  https://medlineplus.gov/craniofacialabnormalities.html
- Health Topic: Developmental Disabilities
  https://medlineplus.gov/developmentaldisabilities.html
- Health Topic: Speech and Language Problems in Children
  https://medlineplus.gov/speechandlanguageproblemsinchildren.html

Genetic and Rare Diseases Information Center

- SATB2-associated syndrome
  https://rarediseases.info.nih.gov/diseases/13206/satb2-associated-syndrome

Educational Resources

- Centers For Disease Control and Prevention: Developmental Disabilities
  https://www.cdc.gov/ncbddd/developmentaldisabilities/
- Centers For Disease Control and Prevention: Facts About Intellectual Disability
  https://www.cdc.gov/ncbddd/actearly/pdf/parents_pdfs/intellectualdisability.pdf
- Foundation for Faces of Children
  https://facesofchildren.org/
- KidsHealth from Nemours: Delayed Speech or Language Development
  https://kidshealth.org/en/parents/not-talk.html
- MalaCards: satb2-associated syndrome
  https://www.malacards.org/card/satb2_associated_syndrome
- Merck Manual Consumer Version: Intellectual Disability
  https://www.merckmanuals.com/home/children-s-health-issues/learning-and-developmental-disorders/intellectual-disability
- Unique: 2q33.1 Deletions and Other Deletions Between 2q31 and 2q33
  https://www.rarechromo.org/media/information/Chromosome%20%202/2q33.1%20deletions%20and%20other%20deletions%20between%202q31%20and%202q33%20FTNW.pdf
- Unique: Rare Chromosome Disorder Support Group (UK)
  https://www.rarechromo.org/media/singlegeneinfo/Single%20Gene%20Disorder%20Guides/SATB2%20syndrome%20QFN.pdf

Patient Support and Advocacy Resources

- American Association on Intellectual and Developmental Disabilities (AAIDD)
  http://aaidd.org/
- American Cleft Palate-Craniofacial Association
  https://cleftline.org/
- American Speech-Language-Hearing Association
  https://www.asha.org/
- Children's Craniofacial Association
  https://ccakids.org/
- Foundation for Faces of Children
  https://facesofchildren.org/
- Resource List from the University of Kansas Medical Center: Developmental Delay
  http://www.kumc.edu/gec/support/devdelay.html
- Resource List from the University of Kansas Medical Center: Facial Anomalies/ Craniofacial Conditions
  http://www.kumc.edu/gec/support/craniofa.html
- SATB2gene.com
  https://satb2gene.com/

Clinical Information from GeneReviews

- SATB2-Associated Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK458647

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?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

Catalog of Genes and Diseases from OMIM

- GLASS SYNDROME
  http://omim.org/entry/612313

Medical Genetics Database from MedGen

- Chromosome 2q32-q33 deletion syndrome
  https://www.ncbi.nlm.nih.gov/medgen/436765

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  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1592575/
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