SATB2 Syndrome (Glass syndrome)

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Normal Function

The SATB2 gene provides instructions for making a protein that helps control the development of certain body systems. The SATB2 protein attaches to special regions of DNA called matrix attachment regions (MARs). These regions help determine the structure of chromatin, which is the complex of DNA and proteins that packages DNA into chromosomes. The structure of chromatin is one way that gene expression is regulated during development.By organizing chromatin structure, the SATB2 protein coordinates the activity of multiple genes involved in the development of certain body systems. In particular, the SATB2 protein promotes the maturation of cells that build bones (osteoblasts) and directs development of structures in the head and face. The protein also plays roles in the maturation and function of different types of nerve cells (neurons) in the brain.

Health Conditions Related to Genetic Changes

SATB2-associated syndrome

Mutations in the SATB2 gene have been found to cause SATB2-associated syndrome. Individuals with this condition have intellectual disability and severe speech problems. They may also have an opening in the roof of the mouth, dental abnormalities, or other abnormalities of the head and face (craniofacial anomalies). Some of these mutations are deletions of large pieces of DNA that remove several genes, including SATB2. Other mutations add, remove, or rearrange smaller pieces of DNA within the SATB2 gene. Still other mutations change single DNA building blocks (nucleotides) in the SATB2 gene. It is likely that these genetic changes reduce the amount of functional SATB2 protein. Reduction of SATB2 function is thought to impair 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, some individuals with large deletions that include additional genes have uncommon features of the condition, such as problems with the heart, genitals and urinary tract (genitourinary tract), skin, or hair. These features are thought to be related to loss of other genes near SATB2.

More About This Health Condition

Other Names for This Gene

DNA-binding protein SATB2 FLJ21474 GLSS KIAA1034 SATB family member 2 special AT-rich

sequence-binding protein 2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

Tests of SATB2

Scientific Articles on PubMed

PubMed

Catalog of Genes and Diseases from OMIM

SPECIAL AT-RICH SEQUENCE-BINDING PROTEIN 2; SATB2

Gene and Variant Databases

NCBI Gene

ClinVar

References

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Genomic LocationThe SATB2 gene is found on chromosome 2.

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Genetics

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What is a gene?

What is a gene variant and how do variants occur?

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