SATB2 gene

SATB homeobox 2

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*.

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 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=23314[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28SATB2%5BTIAB%5D%29+OR+%28SATB+homeobox+2%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- SPECIAL AT-RICH SEQUENCE-BINDING PROTEIN 2; SATB2 (https://omim.org/entry/608148)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/23314)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SATB2[gene])

References

- Bengani H, Handley M, Alvi M, Ibitoye R, Lees M, Lynch SA, Lam W, Fannemel M, Nordgren A, Malmgren H, Kvarnung M, Mehta S, McKee S, Whiteford M, Stewart F, Connell F, Clayton-Smith J, Mansour S, Mohammed S, Fryer A, Morton J; UK10KConsortium; Grozeva D, Asam T, Moore D, Sifrim A, McRae J, Hurles ME, Firth HV,Raymond FL, Kini U, Nellaker C, Ddd Study, FitzPatrick DR. Clinical and molecularconsequences of disease-associated de novo mutations in SATB2. Genet Med. 2017Aug;19(8):900-908. doi: 10.1038/gim.2016.211. Epub 2017 Feb 2. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/28151491)

- Britanova O, Akopov S, Lukyanov S, Gruss P, Tarabykin V. Novel transcriptionfactor Satb2 interacts with matrix attachment region DNA elements in atissue-specific manner and demonstrates cell-type-dependent expression in thedeveloping mouse CNS. Eur J Neurosci. 2005 Feb;21(3):658-68. doi:10.1111/j.1460-9568.2005.03897.x. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15733084)
- Britanova O, de Juan Romero C, Cheung A, Kwan KY, Schwark M, Gyorgy A, VogelT, Akopov S, Mitkovski M, Agoston D, Sestan N, Molnar Z, Tarabykin V. Satb2 is apostmitotic determinant for upper-layer neuron specification in the neocortex. Neuron. 2008 Feb 7;57(3):378-92. doi: 10.1016/j.neuron.2007.12.028. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18255031)
- Britanova O, Depew MJ, Schwark M, Thomas BL, Miletich I, Sharpe P, TarabykinV. Satb2 haploinsufficiency phenocopies 2q32-q33 deletions, whereas loss suggestsa fundamental role in the coordination of jaw development. Am J Hum Genet. 2006Oct;79(4):668-78. doi: 10.1086/508214. Epub 2006 Aug 30. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16960803) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1592575/)
- Dobreva G, Chahrour M, Dautzenberg M, Chirivella L, Kanzler B, Farinas I,Karsenty G, Grosschedl R. SATB2 is a multifunctional determinant of craniofacialpatterning and osteoblast differentiation. Cell. 2006 Jun 2;125(5):971-86. doi:10.1016/j.cell.2006.05.012. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16751105)
- Zarate YA, Fish JL. SATB2-associated syndrome: Mechanisms, phenotype, andpractical recommendations. Am J Med Genet A. 2017 Feb;173(2):327-337. doi:10.1002/ajmg.a.38022. Epub 2016 Oct 24. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/27774744)
- Zarate YA, Kalsner L, Basinger A, Jones JR, Li C, Szybowska M, Xu ZL, VerganoS, Caffrey AR, Gonzalez CV, Dubbs H, Zackai E, Millan F, Telegrafi A, Baskin B, Person R, Fish JL, Everman DB. Genotype and phenotype in 12 additionalindividuals with SATB2-associated syndrome. Clin Genet. 2017 Oct;92(4):423-429.doi: 10.1111/cge.12982. Epub 2017 Mar 7. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/28139846)

Genomic Location

The *SATB2* gene is found on chromosome 2 (https://medlineplus.gov/genetics/chromosome/2/).

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