

Name of Journal:World Journal of Clinical Cases

Manuscript NO: 65790

Manuscript Type:CASE REPORT

SATB2-associated syndrome caused by a novel *SATB2* mutation in a Chinese boy: A case report and literature review

Zhu YY *et al.* A novel *SATB2* mutation for SAS

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Abstract

BACKGROUND

Special AT-rich sequence binding protein 2 (*SATB2*)-associated syndrome (SAS; OMIM 612313) is an autosomal dominant disorder. Alterations in the *SATB2* gene have been identified as causative.

CASE SUMMARY

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Jul 24, 2018 · **SATB2** is associated with schizophrenia and is an important transcription factor regulating neocortical organization and circuitry. Rare mutations in **SATB2** **cause** a **syndrome** that includes developmental delay, and mouse studies identify an important role for **SATB2** in learning and memory.

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Common Chromosomal Disorders (Chromosomes 1-5 and X and Y)

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Jun 10, 2019 · The **SATB2-associated syndrome** leads to developmental delay, intellectual and behavior problems, head and face anomalies. ... The condition is **caused by mutations**, ... Researchers **report** rare case ...

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May 01, 2011 · It has also been shown to be the important determinant in regulating skeletal development, particularly craniofacial patterning and osteoblast differentiation.²³ **SATB2** is also a target for SUMOylation (small ubiquitine relative modifier), a reversible modification of the protein that modulates its activity as a transcription factor.²⁴ Murine in vivo experiments have suggested that **SATB2** haploinsufficiency affects jaw development.²⁵ Nonsense **mutation** of **SATB2** ...

Cited by: 54 **Author:** M Balasubramanian, K Smith, L Basel-Vana...
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Dr. Nathaniel H. Robin, MD - U.S. News & World Report

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Natural history and genotype-phenotype correlations in 72 individuals with **SATB2-associated syndrome**. ... is **caused by a novel COL1A2 mutation**. ... a **case report** and review of the literature.

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Chromosome abnormality

A chromosome abnormality, chromosomal anomaly, chromosomal aberration, chromosomal mutation, or chromosomal disorder, is a missing, extra, or irregular portion of chromosomal DNA. These can occur in the form of numerical abnormalities, where there is an atypical number of chromosomes, or as structural abnormalities, where one or more individual chromosomes are altered. Chromosome mutation was formerly used in a strict sense to mean a change in a chromosomal segment, involving more than one gene. Chromosome anomalies usually occur when there is an error in cell division following meiosis or mitosis. Chromosome abnormalities may be detected or confirmed by comparing an individual's karyotype, or full set of chromosomes, to a typical karyotype for the species via genetic testing.

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