SATB2 Syndrome (Glass syndrome)

https://pubmed.ncbi.nlm.nih.gov/29023086/

Clinical characteristics:

SATB2

-associated syndrome (SAS) is a multisystem disorder characterized by significant neurodevelopmental compromise with limited to absent speech, behavioral issues, and craniofacial anomalies. All individuals described to date have manifest developmental delay / intellectual disability, with severe speech delay. Affected individuals often have hypotonia and feeding difficulties in infancy. Behavioral issues may include autistic features, hyperactivity, and aggressiveness. Craniofacial anomalies may include palatal abnormalities (cleft palate, high-arched palate, and bifid uvula), micrognathia, and abnormal shape or size of the upper central incisors. Less common features include skeletal anomalies (osteopenia, pectus deformities, kyphosis/lordosis, and scoliosis), growth restriction, strabismus/refractive errors, congenital heart defects, genitourinary anomalies, and epilepsy. While dysmorphic features have been described in individuals with this condition, these features are not typically distinctive enough to allow for a clinical diagnosis of SAS.

Diagnosis/testing:

The diagnosis of

SATB2-

associated syndrome (SAS) is established in a proband by detection of one of the following:

A heterozygous intragenic

SATB2

pathogenic variant (61%)

A heterozygous deletion at chromosome 2q33.1 that includes

SATB2

(22%)

An intragenic deletion or duplication of

SATB2

(9%)

A chromosome translocation with a chromosome 2q33.1 breakpoint that disrupts

SATB2

(8%)

Management:

Treatment of manifestations:

Treatment is symptomatic. Nutritional support for feeding difficulties and management by a cleft/craniofacial team for those with palatal anomalies early in life. Early referral for developmental support/special education; standard treatment for dental anomalies, sleep disturbance, skeletal anomalies, seizure disorders, genitourinary anomalies, strabismus and refractive errors, and congenital heart defects.

Surveillance:

Evaluation of nutritional status, growth, and developmental progress at each visit; routine monitoring by a neurologist for those with epilepsy; annual sleep study in those with a history of sleep disturbance; evaluation for scoliosis/spine deformity at each visit and consideration of screening for osteopenia; routine evaluations by dentistry and ophthalmology.

Genetic counseling:

SATB2

-associated syndrome (SAS) is an autosomal dominant disorder. Almost all probands with SAS reported to date have the disorder as the result of a

de novo

genetic event. In two families, parental mosaicism seemed likely (given recurrence of SAS in sibs and failure to detect the genetic alteration in parental blood samples).To date, individuals with SAS are not known to reproduce. Once an

SATB2

intragenic pathogenic variant, a 2q33.1 deletion that includes

SATB2

, or a chromosome translocation affecting

SATB2

has been identified in an affected family member, prenatal testing for a pregnancy at increased risk and preimplantation genetic testing are possible.