**Features**

- Significant neurodevelopmental disorders in all affected individuals, which may include: infantile hypotonia and feeding difficulties, global developmental delay including severe speech delay (speech apraxia, commonly absence of speech), gross and fine motor delays (developmental dyspraxia), cognitive delay.
- Behavioral issues: autistic tendencies, hyperactivity, sleep disturbances, aggressiveness, frustration due to lack of communication.
- Palatal anomalies: cleft palate, bifid uvula, or high-arched palate.
- Dental anomalies: prominent upper incisors, other anomalies.

**Diagnosis**

Established in a proband by detection of one of the following:
- heterozygous intragenic SATB2 pathogenic variant.
- heterozygous non-recurrent deletion at 2q33.1 that includes SATB2.
- intragenic deletion or duplication of SATB2 detectable by chromosomal microarray analysis (CMA).
- chromosomal translocation with a 2q33.1 breakpoint that disrupts SATB2.

Molecular genetic testing approaches can include a combination of CMA, a multi-gene panel, comprehensive genome sequencing, and exome array.

**Resources**

For additional medical and scientific information, as well as registry information, please visit www.satb2gene.com.

For more information about the SATB2 Gene Foundation, please visit www.satb2gene.org.

Closed Facebook group for families to connect, search for “SATB2 Syndrome (2q33.1)”.

Additional Resources: