Making the Diagnosis

So far, no specific criteria have been outline to make the diagnosis of *SATB2*-associated syndrome (SAS). However, the diagnosis of SAS should be considered in anybody that has developmental delay with severely compromised or absent speech and that also has palatal and dental abnormalities.

Regardless of the medical history and findings on physical exam, any of the potential mechanisms that result in the alteration of the *SATB2* gene needs to be documented through genetic testing to make the diagnosis of SAS. The testing that could show changes in the *SATB2* gene include:

- **Chromosomal microarray (CGH):** also known as array CGH or SNP array, this test is a test designed to look for deletions (pieces missing) or duplications (pieces extra) throughout all the chromosomes. This test is available at many laboratories in the United States and around the world and is considered to be a first-line test in the evaluation of developmental delay and/or birth defects. The majority of deletions or duplications in chromosome 2 that include the *SATB2* gene should be able to be detected by this test.

- **Sequencing of *SATB2*:** sequencing of a gene refers to looking at all the individual letters in the gene for misspellings. This test, however, is unlikely to be commonly ordered since it tends to be more cost-effective to look at several genes at once rather than just this one.

- **Next Generation Sequencing testing:** in this test, all the individual letters in several genes (ranging from a handful to several hundred) are looked at simultaneously. Because developmental delay and severe speech delay are main features of the SAS, testing for misspellings of the *SATB2* gene is often conducted by this type of test when no other more distinctive medical issues are present. The *SATB2* gene is included in panels that are offered by several laboratories targeting autism, seizures, developmental delay, or intellectual disability.

- **Clinical Exome/Whole Exome Sequencing:** In this test, the coding regions of several thousands (4000-5000) or all our 20,000 genes are looked at simultaneously. This test is also quickly becoming widely available worldwide.