SATB2 Syndrome (Glass syndrome)

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The goal of this study was to investigate the medical, communication, activities of daily living (ADLs), and mental health concerns affecting adolescents and adults with SATB2-associated syndrome (SAS). A comprehensive questionnaire was administered to the caregivers of 49 individuals 12 years or older with SAS (mean age was 19.4 years, range 12-37 years). For all individuals, medical records, including laboratory results, were reviewed. Most individuals required some degree of assistance for ADLs and none of the adults were able to live independently. Health status was qualified as excellent or very good in 61% of individuals. The most common medical problems were dental anomalies, with a significantly higher frequency of hypotonia and gastroesophageal reflux in younger individuals. Medical and surgical interventions were often required. Sixty-nine percent (n = 33) of individuals spoke 10 or fewer words. Autism (41%), anxiety (37%), and attention deficit disorder (37%) were common with one third of individuals receiving medical treatments for these diagnoses. While medical and developmental problems in individuals with SAS were similar to those previously reported, many of these are persistent into adolescence and adulthood. This study provides better guidance for the challenges facing adults with SAS and their families.