SATB2 Syndrome (Glass syndrome)

https://pubmed.ncbi.nlm.nih.gov/27774744/

The SATB2-associated syndrome is a recently described syndrome characterized by developmental delay/intellectual disability with absent or limited speech development, craniofacial abnormalities, behavioral problems, dysmorphic features, and palatal and dental abnormalities. Alterations of the SATB2 gene can result from a variety of different mechanisms that include contiguous deletions, intragenic deletions and duplications, translocations with secondary gene disruption, and point mutations. The multisystemic nature of this syndrome demands a multisystemic approach and we propose evaluation and management guidelines. The SATB2-associated syndrome registry has now been started and that will allow gathering further clinical information and refining the provided surveillance recommendations. © 2016 The Authors. American Journal of Medical Genetics Part A Published by Wiley Periodicals, Inc.