FOXP1 syndrome

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FOXP1 syndrome is a neurodevelopmental disorder caused by mutations or deletions that disrupt the forkhead box protein 1 (FOXP1) gene, which encodes a transcription factor important for the early development of many organ systems, including the brain. Numerous clinical studies have elucidated the role of FOXP1 in neurodevelopment and have characterized a phenotype. FOXP1 syndrome is associated with intellectual disability, language deficits, autism spectrum disorder, hypotonia, and congenital anomalies, including mild dysmorphic features, and brain, cardiac, and urogenital abnormalities. Here, we present a review of human studies summarizing the clinical features of individuals with FOXP1 syndrome and enlist a multidisciplinary group of clinicians (pediatrics, genetics, psychiatry, neurology, cardiology, endocrinology, nephrology, and psychology) to provide recommendations for the assessment of FOXP1 syndrome.