G.M. Rice, G. Raca, K.J. Jakielski, J.J. Laffin, C.M. Iyama-Kurtycz, S.L. Hartley, R.E. Sprague, A.T. Heintzelman, & L.D. Shriberg. (2012). Phenotype of *FOXP2* haploinsufficiency in a mother and son. *American Journal of Medical Genetics*, Part A 158A, 174–181.

Abstract

Disruptions in *FOXP2*, a transcription factor, are the only known monogenic cause of speech and language impairment. We report on clinical findings for two new individuals with a submicroscopic deletion of *FOXP2*: a boy with severe apraxia of speech and his currently moderately affected mother. A 1.57 Mb deletion on chromosome 7q31 was detected by array comparative genomic hybridization (aCGH). In addition to *FOXP2*, the patients' deletion involves two other genes, *MDFIC* and *PPP1R3A*, neither of which has been associated with speech or language disorders. Thus, findings for these two family members provide informative phenotypic information on *FOXP2* haploinsufficiency. Evaluation by a clinical geneticist indicated no major congenital anomalies or dysmorphic features. Evaluations by a clinical psychologist and occupational therapist indicated cognitive-linguistic processing and sensorimotor control deficits, but did not support a diagnosis of autism spectrum disorder. Evaluation by clinical and research speech pathologists confirmed that both patients' speech deficits met contemporary criteria for apraxia of speech. Notably, the patients were not able to laugh, cough, or sneeze spontaneously, replicating findings reported for two other *FOXP2* cases and a potential diagnostic sign of nonsyndromic apraxia of speech. Speech severity findings for the boy were not consistent with the hypothesis that loss of maternal *FOXP2* should be relatively benign. Better understanding of the behavioral phenotype of *FOXP2* disruptions will aid identification of patients, toward an eventual understanding of the pathophysiology of syndromic and nonsyndromic apraxia of speech.