Severe Expressive-Language Delay and Congenital Malformations in A Boy with Microduplication 7q11.23 Diagnosed by Molecular Cytogenetic Analysis

We report a patient with speech delay and hydronephrosis ascertained with array-CGH screening of patients with abnormal neuronal migration and intellectual disability, having a 7q11.23 duplication reciprocal to the Williams-Beuren critical region (WBCR) deletion. Similarly to the reported patients, he showed serious difficulties in expressive language in the absence of severe intellectual disability and marked dysmorphic features. In addition to the abnormal development of the cerebral cortex with simplified gyral pattern and hydronephrosis. Chromosomal analysis showed normal karytoptype, so Micro-array comparative genomic hybridization was used to search for a possible cryptic anomaly. An interstitial duplication of 1.46 Mb was detected in 7q11.23. Fluorescence in situ hybridization confirmed the diagnosis, revealing a tandem duplication of the Williams-Beuren critical region. Our study provides an additional case for better understanding and delineating the duplication 7q11.23. We thus performed a genotype-phenotype correlation analysis to ascertain the contribution of 7q11.23 to the clinical features of our patient