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P27. THE SMALLEST 7Q11.23 DUPLICATION ENCOMPASSING GTF2I AND GTF2IRD1 GENES IN AN INDIVIDUAL WITH INTELLECTUAL DISABILITY.

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Microduplication of 7q11.23 region has been recognized as a syndrome presenting with speech delay, cognitive impairment, and behavioral abnormalities. It typically involves 1.5 Mb region including approximately 26 genes that is deleted in Williams-Beuren Syndome (WBS) deletion. The majority of reported patients have recurrent size and breakpoints because the duplication is mediated by nonallelic homologous recombination between low copy repeats flanking the critical region. GTF2I and GTF2IRD1 have been proposed as responsible for the neurobehavioral phenotype. However, to date there have been no reported cases with duplications involving only these two genes. Here we present a 9-yeat-old boy with language delay, cognitive impairment, obesity and dysmorphic facial features. Array-CGH identified a maternally inherited 184-kb duplication of the 7q11.23 region including GTF2I and GTF2IRD1 genes. The evaluation of intellectual functioning by WISC-IV resulted in a Full-Scale QI of 61, with homogeneous scores for subcategories. The adaptive behavior tested by VABS was in line with intellectual functioning. No evident behavioral and emotional problems were highlighted by CBCL 6-18. His facial dysmorphisms were not specific. Her mother had learning difficulty and is presently unemployed. Neither the patient nor the mother showed behavioral abnormalities consistent with autism spectrum disorder. GTF2I and GTF2IRD1 encode for the ubiquitous TFII-I transcription factor and for a DNAbinding protein, respectively and have been proposed as responsible for the distinctive neurobehavioral phenotypes of both 7q11.23 deletion and duplication syndromes. The individual we reported is the first case supporting the involvement of GTF2I and GTF2IRD1 in intellectual disability.