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A common underdiagnosed highly penetrant monogenic cause of Autism affecting sensory processing and language

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ABSTRACT

Background: Mutations affecting SHANK3 gene accounts for about 1-2% of ASD cases. SHANK 3 is crucial for ensuring appropriate responses of the intracellular machinery to glutamatergic stimulation, suggesting an association between glutamatergic functioning and sensory processing. Moreover, several studies suggest that SHANK3 differentially affects the development and expression of human language and speech. The current study aimed to assess SHANK3 CNVs in a group of ASD Egyptian patients. In addition, to assess their language and sensory processing patterns. Subjects and methods: 70 children with ASD were assessed using the Autism Diagnostic Interview-Revised. The Arabic Preschool Language Scale and sensory profile were used to evaluate language and atypical sensory behavior respectively. SHANK3 CNVs were assessed in these children using Multiplex Ligation-dependent Probe Amplification technique.Results: Three of the 70 autistic cases showed de novo duplications at 22q13.33. The duplications included SHANK3 in two of the cases and only the distal flanking region of SHANK3 in the third case. The 1st case was a 3-year-old boy (one of twin) with moderate non-verbal autism, mild intellectual disability (ID) and showing symptoms of hyperactivity. The 2nd case was a 5-year-old girl diagnosed with mild verbal autism. Her speech showed stereotyped utterance, neologisms with comorbid ADHD and epilepsy. Finally, the 3rd case was a 4-year-old boy with moderate to severe non-verbal autism, showing symptoms of hyperactivity and EEG changes. As regard, sensory scores in the three patients, the total SSP scores ranged from typical (normal) performance in the case with mild autism to severe sensory symptoms in the case with severe autism. However, the three patients showed definite difference from normal in low-energy/weak domain of sensory profile. Conclusion: Studies propose that not only SHANK3 deletions, but also SHANK3 duplications are associated with ASD. This study contributes to better understanding of the genotype and phenotype correlation of CNVs at 22q13.3 to language and sensory issues in ASD.