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# 3q13.31 deletion syndrome in a patient with autistic features and global developmental delay

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## Abstract

Copy-number variants have been shown to be involved in the etiology of neurodevelopmental disorders like autism spectrum disorders, developmental delay and other neuropsychiatric disorders.

We report on a 2-year-old patient with autistic features, global developmental delay, craniofacial dysmorphism, and cryptorchidism carrying a large proximal deletion of chromosome 3q, detected by chromosomal microarray. The proband belongs to a group of patients referred to our clinic and laboratory with autism as main feature. A complete clinical evaluation was performed with focus on neurologic, psychiatric and psychological evaluation with specific autism tests (ADOS, ADI-R). Array-based comparative genomic hybridization (array-CGH) was performed using 180K platform (Agilent Technologies). A proximal deletion of 3q12.3-q13.33, with a length of ~16.8 Mb was detected in our patient. The deleted region contains 105 genes and overlaps the relatively newly defined 3q13.31 microdeletion syndrome. The critical region for this syndrome spans approximately 0.6 Mb and includes the following genes: DRD3, ZNF80, TIGIT, MIR568, and ZBTB20, also deleted in our patient. Among these genes, DRD3 and ZBTB20 are considered candidates for some of the main clinical findings: developmental delay, characteristic facial features, and abnormal male genitalia. Autism is reported in some of the patients with 3q13.31 deletion syndrome, GAP43 being the candidate gene for the autistic phenotype. GAP43 gene is among the genes deleted in our patient. Thus, our patient was classified as having a 3q13.31 deletion syndrome, a rare neurodevelopmental disorder. Our data illustrates the utility of array-CGH in the investigation of patients with autism, specifically in the context of complex phenotypes. Acknowledgment: The research leading to these results has received funding from the EEA Grant 2014-2021, under the project contract No 6/2019.

## Biography

Aurora Arghir has completed his master's degree and currently working as professor in Victor Babes National Institute of Pathology, Bucharest, Romania.

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