## عنوان مقاله:

Approaches in Genetic testing in Autism: Applications of Copy Number Variation (CNV) analysis and Next generationSequencing (NGS), in some Iranian Patients with Autism


## محل انتشار:

دومین کنگره بین المللی و دهمین همایش ملی نوروژنتیک ایران (سال: 1396)

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## خلاصه مقاله:

Introduction: The autism spectrum disorders (ASDs) are commonneurodevelopmental disorders estimated to affect 1 in 68 children. ASD is acomplex multifactorial and a highly heterogeneous condition. Genetics playsan important role. Copy number variations account for 10-15% in sporadicASD cases. Next generation sequencing (NGS) is a valuable technique inidentifying causative mutations. Variation in about 100 genes have so far beenindicated in ASD. Use of NGS in familial ASD, where a monogenic inheritance issuggestive, can be of great value in unravelling the genetic causes.Materials and Methods: We used karyotyping, MLPA (Subtelomeric andAutism kits) and array CGH to detect CNVs in 50 Iranian patients withsporadic non syndromic autism with additional clinical features includingintellectual disability, seizure, and craniofacial anomalies. We have alsorecruited 15 familial non syndromic ASD cases, and are using SNP Array andWhole Exome Sequencing. The latter study is ongoing.Results: In the sporadic cases, two 0f 50 (4%) patients showed chromosomeabnormality including 16p duplication (16p13.11-p13.3) and 15q deletion(15q11.2q13.1). MLPA detected CNVs in 5 patients (10%) both in subtelomeric and interstitial regions, one in supporting the cytogenetic resultfor 15q11.2q13.1 region. Array CGH was performed for 15 patients. Six out of15 patients (40%) showed significant CNVs including two pathogenic losses(15q24 and Xq28 microdeletions), and 4 likely pathogenic gains(15q13.3, 7q36.3, Xp22.33 and 10q21.2-21.3 microduplications). Weperformed genotype-phenotype analysis and compared our results withother studies. In the familial cases, SNP array have been carried out in eightfamilies demonstrating the homozygous regions, and Whole ExomeSequencing (WES) and co segregation analysis are ongoing.Conclusion: We recommend testing patients with non-familial autism andadditional features for CNVs. The CNV analysis in such patients could lead tothe discovery of novel syndromes as well as unraveling the etiology of autism.The NGS analysis for familial ASD patients where monogenic inheritance isindicated, in particular in extended families with affected branch members,can be of great value in identifying the causative mutation in the family.

**کلمات کلیدی:**
Autism Spectrum Disorder, Sporadic, Familial, CNV, Karyotype, MLPA, Array-CGH, NGS, WES