Molecular Genetics, Genomics, Mechanisms of Diseases

AB051. Chromosomal microarray analysis in a large cohort of Thai patients with autism spectrum disorder

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Background: Chromosomal microarray (CMA) is now recommended as the first-tier clinical diagnostic test for detecting copy number variations (CNVs) in patients with autism spectrum disorder (ASD) of unknown cause. The aim of this study was to identify ASD associated-CNVs in a large cohort of Thai patients with ASD using CMA.

Methods: CMA was performed in 130 normal-karyotype Thai patients with ASD, and negative results from Fragile X and *MECP2* DNA tests using the Illumina HumanCytoSNP-12 v2.1 array (n=16 cases) and the Illumina CytoSNP-850K BeadChip (n=114 cases).

Results: We identified 8 (6.1%) pathogenic CNVs and 7 (5.4%) variants of unknown clinical significance (VOUS) with likely pathogenic CNVs. The overall diagnostic yield of abnormal CNVs in the Thai ASD patients was 11.5% (15/130), which is comparable to the reported yield in studies in other populations. Among the pathogenic CNVs, there were 7 patients with well-known microduplication or microdeletion syndromes (i.e., 1q21.1 duplication, 15q13.3 microdeletion and16p13.11 microduplication syndrome). In addition, CMA also revealed uniparental disomy (UPD) of chromosome 15 in one case. Methylation-specific polymerase chain reaction (MS-PCR) and haplotype analysis were then performed on this patient, confirming that he had Angelman syndrome with autistic features caused by paternal UPD of chromosome 15. Interestingly, we identified a new and *de novo* 1p35.2 duplication encompassing *SERINC2* gene that are highly expressed in the brain but have not yet been implicated in ASD.

Conclusions: This study is the first CMA testing in a large cohort of Thai patients with ASD. Our findings support the usefulness of CMA as a diagnostic test in patients with ASD of unknown cause. In addition, we propose considering the *SERINC2* gene as a potential novel ASD candidate gene that warrants further investigation.

Keywords: autism spectrum disorder (ASD); autism; copy number variation (CNV); microarray; Thailand

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