

Complex Genetic Disorders, Genetic Susceptibility to Infections

AB090. Preliminary results of array CGH test in Vietnamese children with autism spectrum disorder

Huy Duong Do, Thi Phuong Hoa Bui, Thi Thanh Ha Ly, Thi Dieu Linh Pham, Trung Kien Tran, Thi Thanh Huong Le, Sy Vinh Le, Thanh Liem Nguyen

Vinmec Research institute of Stem cell and Gene technology, Hanoi, Vietnam

Background: Autism spectrum disorder (ASD) is highly prevalent accounting for around 1% of children worldwide. Chromosomal microarray is widely recommended as the first-tier test for autistic children with diagnostic yield of around 10%. However, this test is still new with limited clinical study and practice in Vietnam. Our study aims to identify genetic patterns of ASD in the Vietnamese children by using Microarray-based Comparative Genomic Hybridization (aCGH).

Methods: We recruit 100 children with ASD and their parents at Vinmec International Hospital in Hanoi. Children are diagnosed with ASD by the Diagnostic and Statistical Manual V (DSM-V), Autism Diagnostic Observation Schedule (ADOS) and Childhood Autism

Rating Scale (CARS). Rett and Fragile X tests are applied to exclude probands with Rett and Fragile X syndromes. We perform aCGH test on probands and their parents to detect copy number of variants (CNVs).

Results: Here we report our current results on 33 probands. We identified 14 CNVs containing autism-related genes in 11 probands. Results from parents of these 11 probands shows that 4 CNVs on three probands are *de novo* mutations (one proband has two *de novo* CNVs). In addition, one proband has a loss CNV consisting of *NPHP1* gene that is related to ASD in the autosomal recessive inheritance model. The initial findings suggest the specific etiology of ASD in the Vietnamese children with a diagnostic yield of 12%.

Conclusions: This is the first empirical study on the Vietnamese children with ASD using aCGH. This work will be expanded to whole exome sequencing and promising findings would be expected.

Keywords: Autism Spectrum Disorder (ASD); Microarray-based Comparative Genomic Hybridization (aCGH); Vietnam

doi: 10.21037/atm.2017.s090

Cite this abstract as: Do HD, Bui TP, Ly TT, Pham TD, Tran TK, Le TT, Le SV, Nguyen TL. Preliminary results of array CGH test in Vietnamese children with autism spectrum disorder. *Ann Transl Med* 2017;5(Suppl 2):AB090. doi: 10.21037/atm.2017.s090