

Birth Defects, Dysmorphology, Skeletal Dysplasia, Craniofacial Anomalies

AB020. Chromosome rearrangement in patients with 46,XY disorders of sex development

Dung Chi Vu¹, Khanh Ngoc Nguyen¹, Ngoc Bich Can¹, Thao Phuong Bui¹, Maki Fukami²

¹Medical Genetics, Metabolism and Endocrinology, National Children's Hospital, Hà Nội, Vietnam; ²National Research Institute for Child Health and Development, Tokyo, Japan

Background: Disorders of sex development (DSD) is defined by congenital conditions in which development of chromosomal, gonadal, or anatomical sex is atypical. Causative mutations have not been identified in more than 50% 46,XY DSD cases. We aimed to identify chromosomal rearrangement in the development of 46,XY DSD in Vietnamese patients.

Methods: Case series report including clinical presentations and data from array-based comparative genomic hybridization analysis for six genetic males with genital abnormalities combines with mental disability and other congenital anomalies.

Results: Heterozygous submicroscopic deletions and/or duplications were identified in six cases. A 7.2 Mb terminal deletion at chromosome 9 including deletion of *DMRT1* gene and a 2.7 Mb terminal duplication at chromosome 17 were detected in case 1 that presented with prematurity, dysmorphism and ambiguous genitalia. A terminal deletion

affects *DMRT1-3* at 9p22-23 was identified in case 2 with ambiguous genitalia, mental disability and dysmorphism. An 18 Mb terminal duplication at chromosome 5 was detected in case 3 with DSD, growth retardation, microcephaly and dysmorphism, ptosis, ventricular septal defect and craniosynostosis. An interstitial deletion including deletions of *WT1*, *PAX6*, and *PRRG4* genes at chromosome 11 was detected in case 4 with WAGR syndrome. A terminal duplication at chromosome 7 was detected in case 5 with DSD, severe hypospadias, small phallus size (1 cm at 3 years of age), and no testis found clinically. A 5 Mb terminal deletion at chromosome 4 and a 6 Mb terminal duplication of chromosome 16 were detected in case 6 with severe motor-mental retardation, microcephaly (head circumference -3.5 SD), micrognathia, and DSD.

Conclusions: The results indicate that chromosomal rearrangements constitute an important part of the molecular bases of 46,XY DSD and that submicroscopic deletions and/or duplication can lead to various types of 46,XY DSD combined with other congenital anomalies and/or mental disability.

Keywords: Disorders of sex development (DSD); array-based comparative genomic hybridization; congenital anomaly; chromosome rearrangement

doi: 10.21037/atm.2017.s020

Cite this abstract as: Vu DC, Nguyen KN, Can NB, Bui TP, Fukami M. Chromosome rearrangement in patients with 46,XY disorders of sex development. *Ann Transl Med* 2017;5(Suppl 2):AB020. doi: 10.21037/atm.2017.s020