Clinical Genetics

AB058. Prader-Willi syndrome: clinical and genetic features

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Background: Prader-Willi syndrome (PWS) is a complex multisystem genetic disorder that results from the lack of expression of paternal inherited imprinted genes on chromosome 15q11-13. PWS is characterized by severe infantile hypotonia; hypogonadism; obesity and hyperphagia; developmental delay, characteristic facial features, short stature; a distinctive behavioral phenotype. This study was performed to delineate the clinical and genetic features of patients diagnosed PWS at National Children's Hospital, Hanoi, Vietnam.

Methods: A total of 118 patients were collected in the descriptive study from 2007 to 2017, they were diagnosed PWS by Holm's criteria. All patients had karyotype and fluorescence *in situ* hybridization (FISH) performed at National Children's Hospital, Hanoi. For the patients with no deletion detected by FISH, their specimens [10] were sent to other laboratories for SNRPN methylation-specific PCR (MS-PCR).

Results: All patients were found to have normal karyotype and 88/118 patients had deletion as demonstrated by FISH. Among 10 patients with MS-PCR performed, 8 patients revealed only maternal part of 15q11-13. Male:female ratio was 64%:36%. In regards to age at diagnosis, 10 (8.5%) patients were diagnosed in neonatal period; 68 (58.3%) patients diagnosed before 24 months. The mean age at diagnosis was 42.3 months. 86.6% of patients were found to have hypotonia. Of those 108 patients diagnosed after neonatal period, 33.3% of patients had obesity and 82% of patients had hyperphagia. Total of 88% of patients had hypogonadism.

Conclusions: Our study showed good sensitivity and specificity of Holm's criteria in diagnosing PWS caused by 15q11-13 deletion. Based on clinical features more PWS patients should be diagnosed early.

Keywords: Prader-Willi syndrome (PWS); clinical features; fluorescence *in situ* hybridization (FISH); methylation-specific PCR (MS-PCR)

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