

JOINT EVENT ON

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Genotypes of Vietnamese patients suspected to congenital adrenal hyperplasia**Nguyen Thi Phuong Mai**

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Congenital adrenal hyperplasia (CAH) is an autosomal recessive disease which is characterized by a deficiency of one of the enzymes involved in the synthesis of cortisol from cholesterol by the adrenal cortex. More than 90% of CAH cases are due to 21-hydroxylase deficiency (21OHD). Mutations in the *CYP21A2* gene are affected to 21-hydroxylase activity deficiency. The frequency of mutations in the *CYP21A2* gene is due to different populations and there is a correlation between genotype and phenotype. This study was performed in 60 Vietnamese children (120 alleles) suspected with congenital adrenal hyperplasia. Total DNA extraction from whole blood and we use sequencing and MLPA techniques to identified mutations in *CYP21A2* gene. We identified 10 cases (16.7%) with heterozygous of mutations; 9 cases (15%) with homozygous of mutations, 19 cases (31.7%) with compound heterozygous of mutations and 22 patients (36.7%) have no mutations. The mutation 30kb deletion is the most common mutation with 17 alleles (14.2%), I172N with 14 alleles (11.7%), I2G with 12 alleles (10%), promoter conversion with 5 alleles (4.2%), R356W with 4 alleles (3.3%), the others were rare mutations such as V281L, V304M, Q318X Sequencing and MLPA techniques were accurate for screening mutations in *CYP21A2* gene. The genotype datas will be helpful for the treatment and prenatal testing.

Biography

Nguyen Thi Phuong Mai has completed her Master's of Science from University of Technology and Science in Hanoi, Vietnam. Currently, she is a PhD student of Institute of Genome Research, Vietnam Academic of Science and Technology in Hanoi, Vietnam. She is a Vice-Head of Human Genetics Department in National Children's Hospital. She has published more than 10 papers in reputed journals.

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