Prenatal Genetics, Reproductive Genetics

AB044. Prenatal diagnosis of congenital adrenal hyperplasia due to 21-hydroxylase deficiency

Mai Thi Phuong Nguyen, Mai Nguyen

Human Genetics Department, National Children's Hospital, Hanoi, Vietnam

Background: Congenital adrenal hyperplasia due to 21-hydroxylase deficiency (21OHD-CAH) is an autosomal recessive disorder. Around 95% of CAH cases are caused by deficiency of the enzyme 21-OH. *CYP21A2* gene located on the short arm of chromosome 6 (6p21.3) encodes the protein enzyme 21-OH.

Methods: We analyzed 9 samples (8 female fetuses and 1 male fetus) of amniotic fluid from at-risk pregnancies of women who had their first child with CAH. Their family members including the fetuses, their husbands and their CAH children were analyzed for mutations of the *CYP21A2* gene by using MLPA and sequencing.

Results: Three out of nine (3/9) fetuses were found to have compound heterozygous mutations in *CYP21A2* gene; 4/9 fetuses were carrier for mutations in the *CYP21A2* gene, and 2/9 fetuses had no mutation. Compound heterozygous of 30kb deletion with other mutations were common genotype. One of the fetus was found to be heterozygous for 5 mutations (I172N; Exon 6 clusters; V281L; R307fs; R356W) in one allele. All heterozygous and healthy fetuses were confirmed postnatally.

Conclusions: Sequencing and MLPA techniques were accurate for screening all mutations in *CYP21A2* gene. DNA testing is the basis progress for the diagnosis and preventive treatment before birth. Prenatal diagnosis process is only done on the family who had known mutations.

Keywords: Congenital adrenal hyperplasia (CAH); 21-hydroxylase deficiency (21-OHD); *CYP21A2*; prenatal diagnosis

doi: 10.21037/atm.2017.s044

Cite this abstract as: Nguyen MT, Nguyen M. Prenatal diagnosis of congenital adrenal hyperplasia due to 21-hydroxylase deficiency. Ann Transl Med 2017;5(Suppl 2):AB044. doi: 10.21037/atm.2017.s044