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

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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

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