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

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Objective To explore the method for prenatal diagnosis of congenital adrenal hyperplasia (CAH) with 212hydroxylase deficiency (212OHD). **Methods** DNA mutations of 21-hydroxylase were detected by Southern hybridization, single strand conformation polymorphism (SSCP) and amplification created restriction site (ACRS) analysis in the proband with 21-OHD and his parents. During the fifth pregnancy, amniocentesis was carried out at the 16th week of gestation, and prenatal gene diagnosis was performed. **Results** Both point mutations int2, nt 656 and a deletion were confirmed in the proband. Each parent carried one of the two mutations. ACRS analysis on amniocytes showed that neither of the two defects existed in fetus. This prenatal diagnosis was confirmed by the normal clinical manifestation and laboratory tests after the infant's birth. **Conclusion** DNA analysis of amniocytes was a feasible method for the prenatal diagnosis of congenital adrenal hyperplasia with 21-hydroxylase deficiency.

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