ANALYSIS CYP21A2 GENE MUTATIONS TECHNIQUE IN PATIENTS WITH CONGENITAL ADRENAL HYPERPLASIA

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The technique of CYP21A2 gene mutation analysis, which can be applicable for pre- and postnatal diagnosis of congenital adrenal hyperplasia various types was developed.

The analysis of extended CYP21A2 gene deletions and mutations 655A/C>G, 999T>A, 1994C>T, 2108C>T, which lead to congenital adrenal hyperplasia simple virilysing and salt wasting forms, and mutations 89C>T and 1683G>T, which lead to non-classical forms of the illness was proposed. This technique is based on analysis of the allele specific polymerase chain reaction and restriction fragment length polymorphism.

The technique has passed quality control on available DNA samples. Thus this may be suggested for DNA diagnostics (including prenatal) in high-risk families and for analysis of heterozygous carriers illness. Using the developed technique nine alleles with CYP21A2 gene deletion, 4 alleles with 655A/C>G mutation and 1 allele with 2108C>T mutation were detected in seven patients and their parents.

Key words: congenital adrenal hyperplasia, enzyme 21-hydroxylase, CYP21A2 gene.

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