

Application of ATP7B Gene Mutation Analysis in Prenatal Diagnosis of Wilson's Disease

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Abstract : Wilson's disease is an autosomal recessive disorder of copper metabolism, which is caused by mutation in copper-transporting P-type ATPase (ATP7B). The mechanism of this disease is a failure of hepatic excretion of copper to the bile, and it leads to copper deposits in the liver and other organs. Most clinical symptoms of Wilson's disease can present as liver disease and/or neurologic disease. Objective: The goal of the study is prenatal diagnosis for pregnant women at high risk of Wilson's disease in Northern Vietnam. Material and method: Three probands with clinically diagnosed liver disease were detected in the mutations of 21 exons and exon-intron boundaries of the ATP7B gene by direct Sanger-sequencing. Prenatal diagnoses were performed by amniotic fluid sampling from pregnant women in the 16th-18th weeks of pregnancy after the genotypes of parents with the probands were identified. Result: A total of three different mutations of the probands, including of S105*, P1052L, P1273G, were detected. Among three fetuses which underwent prenatal genetic testing, one fetus was homozygote; two fetuses were carriers. Conclusion: Genetic testing provided a useful method for prenatal diagnosis, and is a basis for genetic counseling.

Keywords : ATP7B gene, genetic testing, prenatal diagnosis, pedigree, Wilson disease

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