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AB068. Characteristic of *ATP7B* gene mutation in Vietnamese Wilson's disease patients and presymptomatic diagnosis for their siblings

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Background: Wilson's disease (WD) is an autosomal recessive disorder of copper metabolism. The disease is caused by mutation in the copper-transporting P-type ATPase encoded by *ATP7B* gene. The mechanism of this disease is the failure of hepatic excretion of copper into bile compartment, leading to copper deposits in the liver and other organs. This study aimed to investigate the characteristics of *ATP7B* gene mutation in Vietnamese patients with WD, and provided pre-symptomatic diagnosis for their family members.

Methods: Forty-three WD patients and their 67 siblings were enrolled in the study. All 21 exons and its exon-intron boundaries of the *ATP7B* gene were analyzed by PCR followed by direct sequencing.

Results: We identified 2 novel *ATP7B* mutations, L902P and D1027H, in the sum of 18 detectable mutations. Mutation S105X was determined to have a high rate (34.9%) in this study. The hotspot regions of *ATP7B* mutations were exon 2 (40.7%), exon 16 (11.6%), exon 8 (9.3%), intron 14 (7%) and exon 18 (5.9%). Among 11 homozygote/compound heterozygote siblings of the patients with WD, 4/67(6%) individuals were found asymptomatic at the time of being tested.

Conclusions: A total of 18 different mutations were detected in the present study. Of this number, 2 novel mutations were explored, including L902P and D1027H. The mutation S105X is the most prevalent and has been considered as a biomarker that can be used in a rapid detection assay for diagnosis of WD.

Keywords: *ATP7B*; mutation; presymptomatic diagnosis; Wilson's disease (WD); Vietnamese

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