

AB094. Characteristic of *ATP7B* gene mutation in Vietnamese Wilson patients and asymptomatic diagnosis for their siblings

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Background: Wilson disease (WD) is an autosomal recessive disorder of the copper metabolism, which is caused by *ATP7B* gene mutation in the copper-transporting P-type ATPase. The mechanism of this disease is the failure of hepatic excretion of copper to bile, and leads to copper deposits in the liver and other organs.

Methods: This study aimed to investigate the characteristic of *ATP7B* gene mutation in the Vietnamese patients with WD, and make an asymptomatic diagnosis for their familial members.

Results: Forty-three WD patients and their 67 siblings were identified as having *ATP7B* gene mutations; 21 exons and exon-intron boundaries of the *ATP7B* gene were analyzed by direct sequencing. We recognized 2 novel mutations L902P and D1027H in the sum of 18 detectable

mutations, accounting for 91.9 % of the total. Mutation S105X was determined to have a high rate (34.9%) in this study. The hotspot regions of *ATP7B* were found at exon 2 (40.7%), exon 16 (11.6%), exon 8 (9.3%), intron 14 (7%), exon 18 (5.9%). Among 11 homozygote/compound heterozygote siblings of the patients with WD, 4/67(6%) individuals were identified as asymptomatic by screening mutations of the probands.

Conclusions: Eighteen different mutations were detected. Of this number, two 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. Exons 2, 8, 16, 18 and intron 14 should be screened initially for WD patients in Vietnam. Four asymptomatic Wilson patients would be treated soon after diagnosis to prevent the onset of WD and till now are healthy. Based on risk profile for WD, genetic testing is also useful in asymptomatic diagnosis and treatment.

Keywords: *ATP7B* gene mutation; asymptomatic diagnosis; Wilson disease (WD); Vietnamese Wilson patient

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