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Genotype-Phenotype Analysis of Mutation R778L in the *ATP7B* Gene

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Abstract:
Wilson's disease (WD), an autosomal recessive disorder of copper transport, usually presents with symptoms of the liver or central nervous system. The known disease-caused mutations identified in the responsible gene, *ATP7B*, exceed 200 variations. The H1069Q mutation is the most common mutation in European and North American patients with WD, while the R778L mutation is found in most Asian patients. In the present study we identified 12 homozygotes and 22 heterozygotes for R778L. We summarize our results here along with those of previous studies. There were a total of 46 homozygotes and 66 heterozygotes for R778L. The phenotypes of R778L homozygotes and heterozygotes were grouped as hepatic presentation, neurologic presentation, or other presentation according to the most recent standards. No significant differences were found in three clinical features (mean age of onset, number of patients with hepatic presentation, and number of patients with neurologic presentation) between R778L homozygote and heterozygote groups, suggesting that the phenotype of WD is influenced by a series of factors, rather than only by the *ATP7B* gene.

Key words: genotype-phenotype, mutation, ATP7B, Wilson's disease

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