Analysis and application of *ATP7B* gene mutations in 35 patients with hepatolenticular degeneration

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ABSTRACT. We investigated the genetic mutations involved in Wilson’s disease to improve prenatal genetic diagnosis and presymptomatic diagnosis. The polymerase chain reaction (PCR) was used to amplify the exons and exon-intron boundaries of the *ATP7B* gene in 35 Wilson’s disease pedigrees. The PCR products were further analyzed by Sanger sequencing. Prenatal genetic diagnoses were performed by chorionic villus sampling after the genotypes of parents of the probands were identified. The overall mutation detection frequency was 92.9%. A total of 24 distinct mutations were detected, seven of which are novel: A1291T (c.3871G>A), c.2593_2594insGTCA, c.2790_2792delCAT, c.3661_3663delGGG, c.3700delG, c.4094_4097delCTGT, and IVS6+1G>A. Three mutations, R778L (c.2333G>T) (45.7%), A874V (c.2621C>T) (7.1%), and P992L (c.2975C>T) (7.1%) are relatively frequent. Two presymptomatic patients were detected through familial screening, and they began taking medicine after diagnosis. Of the subjects with Wilson’s disease pedigrees who had received a prenatal genetic diagnosis, three fetuses were normal and one was a carrier. Twenty-four distinct mutations were identified, and our knowledge of the population genetics of Wilson’s disease in China has therefore improved. For pedigrees with the Wilson’s disease, genetic
counseling, prenatal diagnosis, and presymptomatic diagnosis by Sanger sequencing and haplotype analysis are feasible.

**Key words:** Wilson’s disease; ATP7B gene; Mutation analysis; Prenatal diagnosis; Presymptomatic diagnosis