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Genetic Variation in *ATP7B* Promotor and 5' UTR in Han Chinese Patients with Wilson's Disease

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Abstract:
Wilson's disease (WD) is an autosomal recessive disorder of copper transport characterized by the accumulation of intracellular copper in the liver and extrahepatic tissues. The WD gene (*ATP7B*) encodes a copper transporting P-type ATPase with 50-70% similarity to the Menkes disease (MNK) protein, and is primarily expressed in the liver, while the MNK gene (*ATP7A*) is almost ubiquitously expressed in a variety of tissues, but not in the liver. The *ATP7B* gene, but not *ATP7A*, contains several metal response elements (MRE) and MRE-like sequences (MLS) in the promotor region that seem to play important roles during the *ATP7B* gene expression. We previously reported that in 40 WD patients mutations were found in 83.8% (67/80) of alleles on direct sequencing of polymerase chain reaction products of all exons of the *ATP7B* gene. It is possible that disease-caused mutations may exist in the promotor, 5' untranslated region, or introns. So far, only two mutations have been reported in the promotor region of the *ATP7B* gene. In the present study we identified one mutation and seven polymorphisms in the promotor region of the *ATP7B* gene in Chinese population. The mutation -215 A to C, occurred close to MREd, between MREd and the E-box. The results of the present study suggest

that mutations in the regulatory elements of *ATP7B* that result in WD are rare in Chinese patients with WD. These results will be very useful in facilitating the molecular diagnosis and counseling of WD patients in the Chinese population.

Key words: Wilson's disease, copper, the ATP7B gene promoter, metal response elements, mutation, polymorphism

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