ISSN 0972-3757
International Journal of
HUMAN GENETICS

PRINT: ISSN 0972-3757 ONLINE: 2456-6360
Int J Hum Genet, 12(2): 125-132 (2012)
DOI: 10.31901/24566330.2012/12.02.08

Mutation Profile in Wilson's Disease from North Indian Patients

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KEYWORDS Wilson's Disease. ATP7B. P-type ATPase. SSCP, H1069Q. R778L

ABSTRACT Wilson disease (WD) is an autosomal recessive disorder caused by defects in the ATPase, Cu2transporting, polypeptide gene (*ATP7B*) resulting in accumulation of copper in liver and brain. The study was conducted in the Department of Neurology, of a tertiary care center in India from 2004 to 2009. DNA samples of 90 WD patients, their unaffected first degree relatives and 90 unrelated healthy controls were analyzed for mutations in ATP7B gene *(ATP7B)*. The researchers screened the entire coding region of *ATP7B*. The desired sequence of the gene was amplified using PCR followed by SSCP. Samples showing shifts in the banding pattern on SSCP were sequenced commercially. Out of 90 WD patients 41 variations in 55 WD patients were observed in DNA samples for *ATP7B*. Of these 24 patients were homozygous, 6 were compound heterozygous and 25 were heterozygous. Of these 6 were known mutations and the rest 33 was novel. The researchers observed exon 2, 13 and 18 as hot spot exons of *ATP7B* with large number of variations. Lack of common dominant mutations prevented correlation of individual mutations with WD phenotype. The researchers did not observe common mutations reported in *ATP7B* in other countries.