Wilson disease (WD) is an autosomal recessive disorder in ATP7B gene which encodes a copper-transporting ATPase. This protein involves in the transport of copper into the plasma protein ceruloplasmin and in excretion of copper from the liver. Large numbers of mutations in ATP7B gene were found to have a role in the pathogenesis of WD. The variation in the clinical and biochemical features of the disease renders the diagnosis difficult task. This study aimed to determine the mutations in exon14 and 21 of ATP7B gene in Iraqi patients with WD to be used for molecular diagnosis.A total of 35 patients with WD and other 10 apparently healthy individuals were recruited for this study. Blood sample was obtained from each subject from which DNA was extracted and exon 14 and 21 were amplified with amplification refractory mutation system (ARMS) and conventional polymerase chain reaction using specific primers. Direct sequencing was used to find out the mutations in these exons.Five novel mutations and one single nucleotide polymorphism (homozygous or heterozygous) were recorded, among which 3 silent (c.3133C>T, c.4194T>C and c.4302G>A), 1 missense (c.3181G>C) and 2 splice site mutations (IVS21+40delA and IVS21+23G>A). The heterozygous mutation c.3133C>T was the most prevalent one among WD patients (40%).
For diagnostic purpose, these results strongly suggested the heterozygous mutation c.3133C>T could be exploited in combination with the other high prevalent mutations.