**Case Report:**

**Wilson’s disease: Case report from Maharashtra**

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**ABSTRACT**

Wilson's disease is an autosomal recessive disorder of copper metabolism resulting from the absence or dysfunction of a copper transporting P-type ATPase encoded on chromosome 13. Presentation in childhood may include chronic hepatitis, asymptomatic cirrhosis, or acute liver failure. In young adults, neuropsychiatric symptoms predominate and include dystonia, tremor, personality changes, and cognitive impairments secondary to copper accumulation in the central nervous system. The laboratory diagnosis of Wilson's disease is confirmed by decreased serum ceruloplasmin, increased urinary copper content, and elevated hepatic copper concentration. Molecular genetic analysis is complex as more than 100 unique mutations have been identified and most individuals are compound heterozygotes.

**Keywords:** Wilson's disease, copper – liver