Wilson's Disease

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Neurological dysfunction constitutes the initial clinical manifestation in 40–60% of individuals with Wilson's disease2

Psychiatric features were evident at the time of initial presentation in 65% of individuals with Wilson's disease2

Estimation of serum copper, serum ceruloplasmin and 24 h urinary copper is recommended for suspected Wilson's Disease (WD)[1]

- Wilson's disease should be considered and excluded in any young person who develops unexplained psychiatric dysfunction, especially when any signs of neurological dysfunction are also present
- Poor school performance, especially if coupled with abdominal symptoms, should prompt consideration of Wilson's disease
- The possibility of Wilson's disease should also be considered in young persons suspected of drug abuse, because the symptoms can be similar

Recommendations for Diagnostic Testing :

Ceruloplasmin

- An extremely low serum ceruloplasmin level (<50 mg/L or < 5 mg/ dL) should be taken as strong evidence for the diagnosis of WD
- Modestly subnormal levels suggest further evaluation is necessary. Serum ceruloplasmin within the normal range does not exclude the diagnosis

1) Neurology India, Sep-Oct 2009, Vol 57, Issue 5
2) Seminars in Neurology Volume 27, Number 2, 2007

Urinary Copper Excretion

- Basal 24-hour urinary excretion of copper is typically > 100 microgram (1.6 micromols) in symptomatic patients

- Finding of > 40 microgram (> 0.6 micromol) may indicate WD and requires further investigation

Monitoring Patients on Pharmacotherapy :

Serum 'Free' Copper, calculated by subtracting ceruloplasmin bound copper (3.15 x ceruloplasmin in mg/L) from the total serum copper, is of more value in monitoring pharmacotherapy than in the diagnosis of Wilson's Disease

(American Association for Study of Liver Diseases (AASLD). Diagnosis and treatment of Wilson's Disease: An Update. Hepatology 2008 Jun; 47 (6): 2089-111)