Table 1 CLINICAL PATTERNS

Clinical Patterns of Hepatic, Neurologic, and Psychiatric Disease in Patients with Wilson's Disease

Hepatic

- Asymptomatic hepatomegaly.
- · Isolated splenomegaly
- Persistently elevated AST, ALT
- Fatty Liver
- Acute hepatitis
- Resembling autoimmune hepatitis
- Cirrhosis (compensated or decompensated)
- · Fulminant hepatic failure

Neurological

- Movement disorders (tremor, involuntary movements)
- Drooling, dysarthria
- Rigid dystonia
- Pseudobulbar palsy
- Seizures
- Migraine headaches
- Insomnia

Psychiatric

- Depression
- Neuroses
- Personality changes
- Psychosis

Other systems

- Renal abnormalities: aminoaciduria and nephrolithiasis
- Skeletal abnormalities: premature osteoporosis and arthritis
- Cardiomyopathy, dysrhythmias
- Pancreatitis
- · Hypoparathyroidism
- Menstrual irregularities; infertility, repeated miscarriages

Table 2 FAMILY SCREENING

- · Medical history
- · Physical examination
- AST, ALT, ALP, total and conjugated bilirubin, albumin, INR, CBC Ceruloplasmin
- · Slit lamp exam for K-F Rings
- 24 hour basal urinary Cu
- · Genotyping/haplotype studies based on proband

"The Wilson's Disease Association funds research and facilitates and promotes the identification, education, treatment, and support of patients and other individuals affected by Wilson's Disease."

For more information please contact the:

WILSON'S DISEASE ASSOCIATION



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A Diagnostic Tool for Physicians

Unexplained

Hepatic,

Neurologic or Psychiatric Symptoms?

Think.....

WILSON'S DISEASE

WILSON'S DISEASE ASSOCIATION



ALGORITHM FOR · Normal Ceruloplasmin ASSESSMENT OF SUSPECTED · K-F Rings absent CONTINUE · Normal 24 hour basal urine Cu **EVALUATION FOR** WILSON'S DISEASE Normal Liver Function Tests **ALTERNATIVE** OR DIAGNOSIS · Decreased Ceruloplasmin · K-F rings absent · Normal 24 hr. urine Cu excretion · Normal Liver Function Tests · Absent neuro/psych symptoms Unexplained • Cu <250μg/d dry weight liver disease or histology typical of (elevated AST, ALT; another hepatic disease hepatomegaly) · Decreased Ceruloplasmin · K-F Rings absent · AST, ALT, ALP, · Abnormal Liver Function Tests or total and 24 hour urine Cu >40 µg/d or conjugated 600 nmoles/d bilirubin, OR albumin, INR, **EVALUATION** · Liver Biopsy with histology, CBC · Decreased Ceruloplasmin **FOR** histochemistry and Cu Serum WILSON'S · K-F Rings absent ** quantification Ceruloplasmin DISEASE Normal Liver Function Tests 24 hour basal Persisting significant symptoms *** urinary Cu OR Slit lamp exam · Normal serum Ceruloplasmin for K-F rings · K-F Rings present · Liver Function Tests abnormal • Elevated Cu >250 μg/d Neurological or dry weight and consistent psychiatric histology symptoms ±liver disease Decreased Ceruloplasmin · K-F Rings present Patients referred for unexplained liver disease ** For patients under 18 years, a penicillamine **DIAGNOSIS ESTABLISHED** typically have liver enlargement or abnormal challenge test may be performed prior to liver **INITIATE TREATMENT AND** serum aminotransferases. biopsy. **FAMILY SCREENING** * Those with neurologic or psychiatric symptoms *** Persisting significant symptoms include: (See Table 2) consistent with Wilson Disease should have an hemolysis, unexplained splenomegaly, extra-

hepatic manifestations of WD, and neurologic

or psychiatric disorders. (See Table 1)

MR scan of the head performed prior to the

hepatologic evaluation.