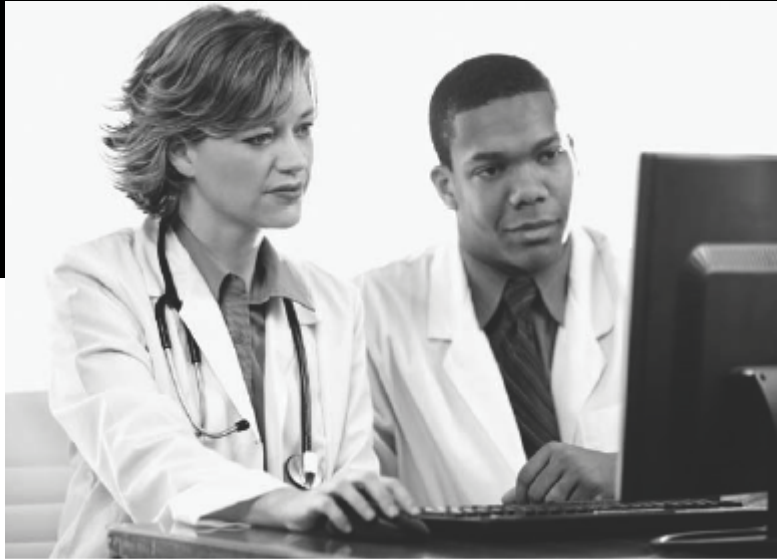


A Diagnostic Tool for Physicians



*Unexplained Hepatic, Neurologic or Psychiatric Symptoms?*

*Think.....*

**WILSON DISEASE**



**TABLE 1. CLINICAL FEATURES IN PATIENTS WITH WILSON DISEASE**

**Hepatic**

- Asymptomatic hepatomegaly
- Isolated splenomegaly
- Persistently elevated serum aminotransferase activity (AST, ALT)
- Fatty liver
- Acute hepatitis
- Resembling autoimmune hepatitis
- Cirrhosis: compensated or decompensated
- Acute liver failure

**Neurological**

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

**Psychiatric**

- Depression
- Neurotic behaviours
- Personality changes
- Psychosis

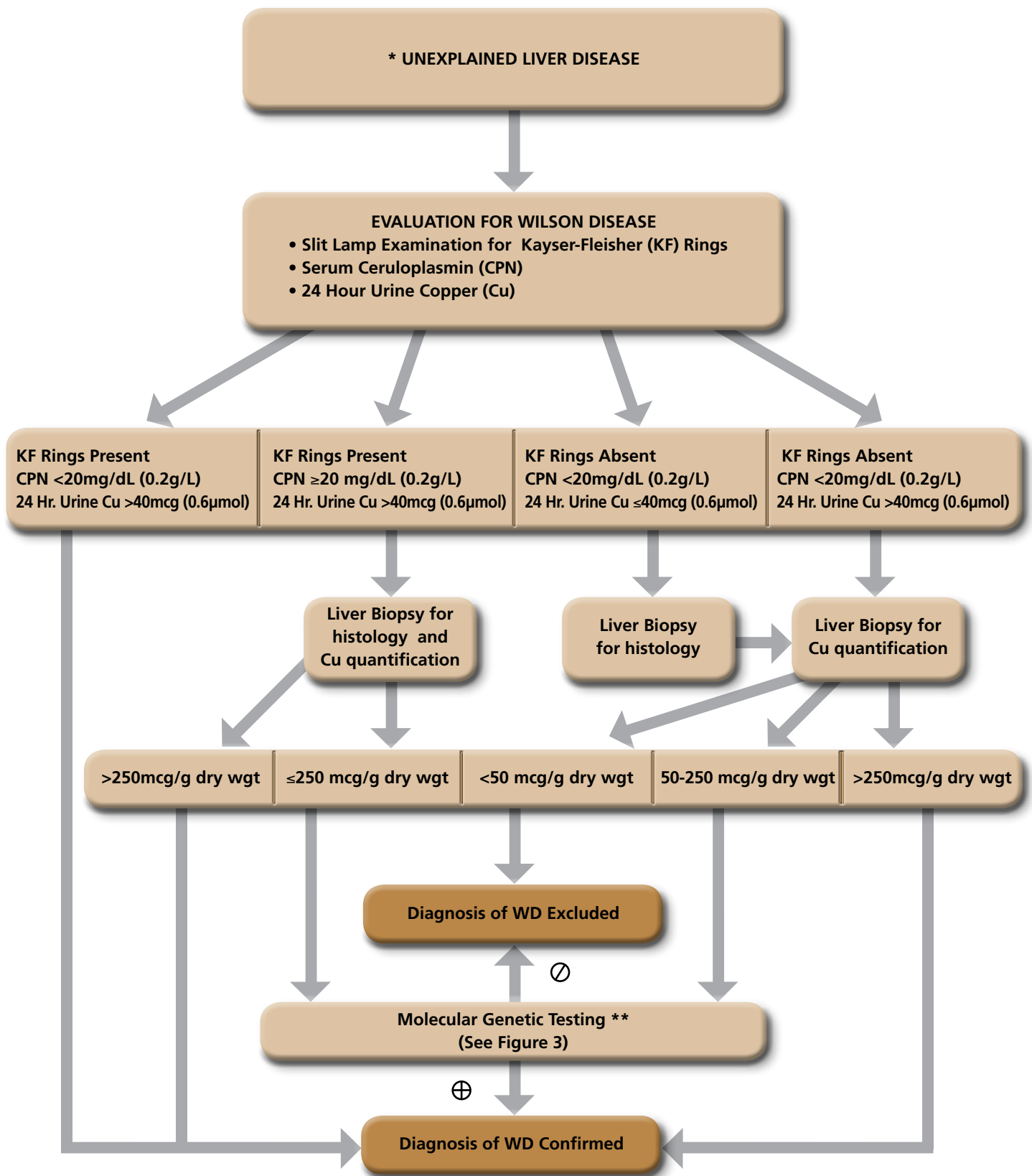
**Other Systems**

- Ocular: Kayser-Fleischer rings, sunflower cataracts
- Cutaneous: lunulae ceruleae
- Renal abnormalities: aminoaciduria and nephrolithiasis
- Skeletal abnormalities: premature osteoporosis and arthritis
- Cardiomyopathy, dysrhythmias
- Pancreatitis
- Hypoparathyroidism
- Menstrual irregularities; infertility, repeated miscarriages



**Figure 1.**

Approach to diagnosis of Wilson disease (WD) in a patient with unexplained liver disease.

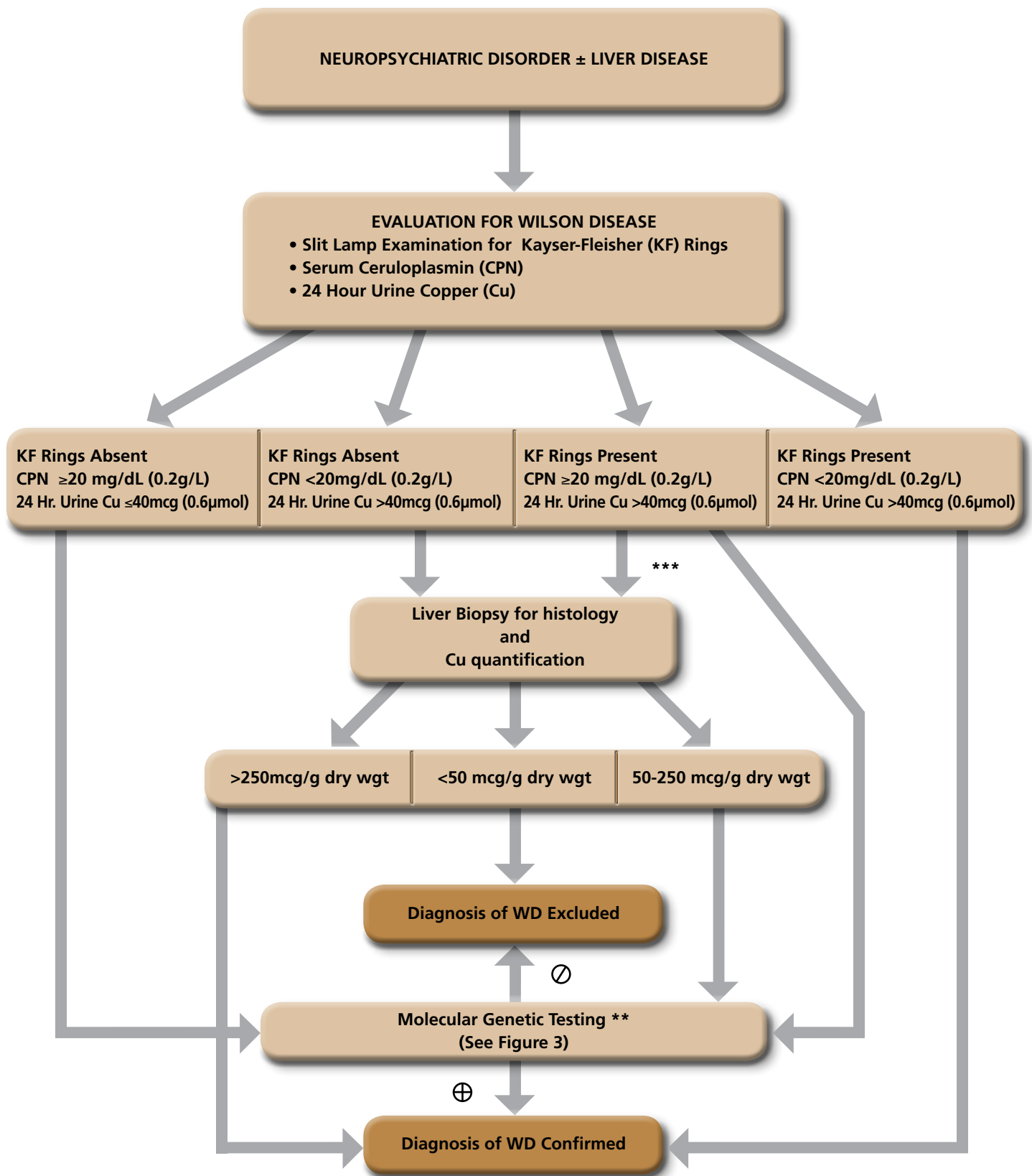


\* Abnormal liver histology and/or liver function tests without clear etiology.

\*\* Molecular testing means confirming homozygosity for one mutation or defining two mutations constituting compound heterozygosity.

**Figure 2.**

Approach to diagnosis of Wilson disease (WD) in a patient with a neurological disorder or psychiatric disease with or without liver disease.

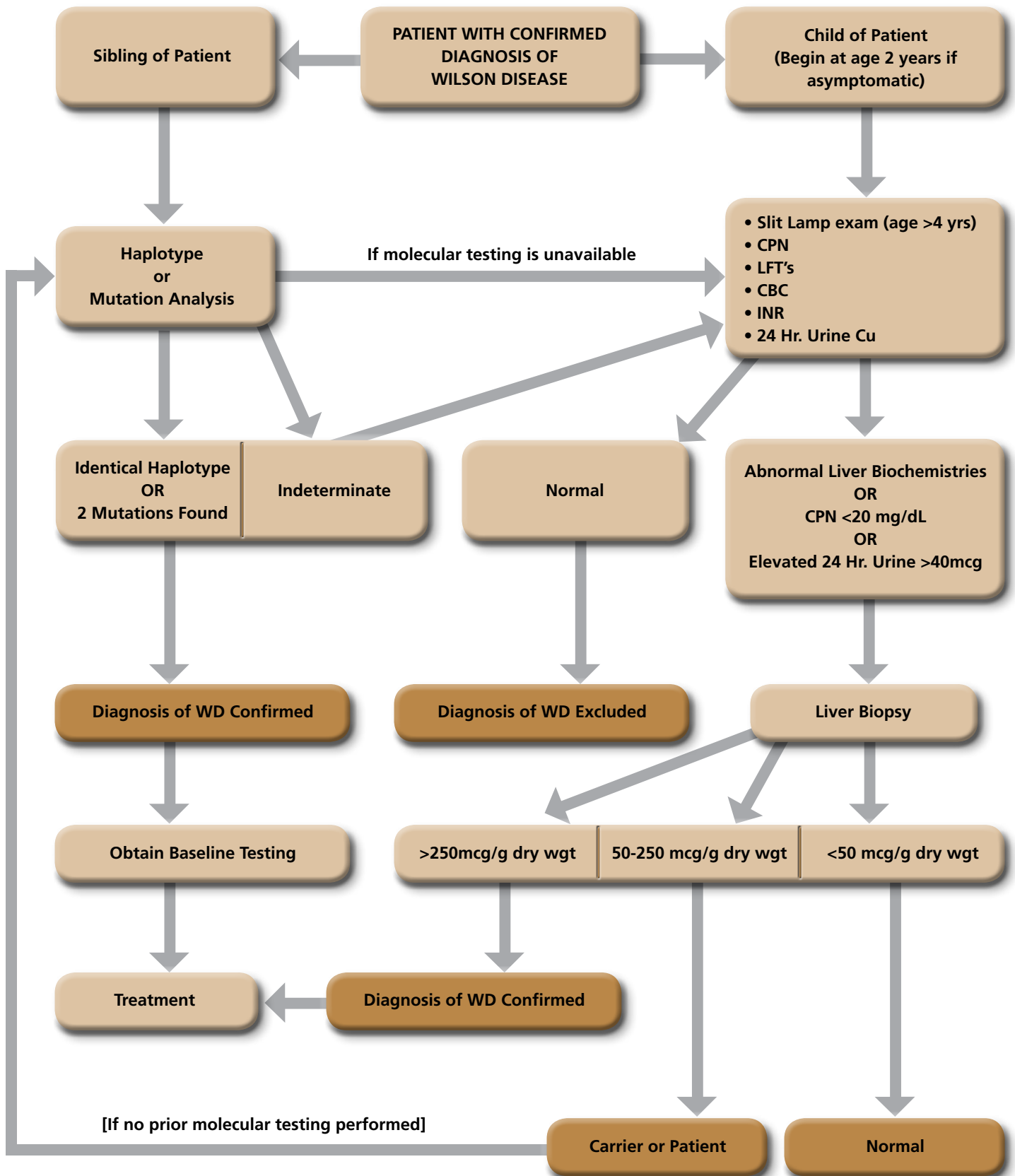


\*\* Molecular testing means confirming homozygosity for one mutation or defining two mutations constituting compound heterozygosity.

\*\*\* If molecular testing unavailable or limited testing performed.

**Figure 3.**

Screening for Wilson disease (WD) in a sibling or child of a patient with secure diagnosis of WD. If molecular testing is available in the index patient, then this is the most efficient screening strategy. If initial screening by blood biochemistries and urine testing is normal, then consider repeat screening in 2 – 5 years.



*“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 disease.”*

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