Wilson Disease Testing Algorithm

Begin with:
- AST, ALT, ALP, total and conjugated bilirubin, albumin, CBC
- Serum ceruloplasmin (CP)
- Serum copper (Cu)
- 24-Hour urine Cu
- Slit-lamp exam for Kaiser-Fleischer (K-F) ring
- Brain MRI for neurologic symptoms

All siblings and first-degree relatives of affected patients

- Neurological or psychiatric symptoms ± liver disease
- Unexplained liver disease (elevated AST, ALT)

Normal CP and serum Cu
Normal 24-hour urine Cu
Normal liver function tests
K-F ring absent

Age ≥15 years
Wilson disease excluded

No mutations

WDZ / Wilson Disease, Full Gene Analysis
OR
Continue follow-up

Any of the following combinations:
- Two mutations identified
- Two mutations identified AND consistent histology regardless of Cu level
- No mutations identified AND increased Cu >250 mcg/g dry weight and consistent histology in the absence of long-standing (>1 year) liver failure or obstruction

Normal CP and serum Cu
Increased 24-hour urine Cu
K-F ring present

Age <15 years

Wilson disease excluded

No mutations

WDZ / Wilson Disease, Full Gene Analysis

If histology is required for confirmation
If liver Cu quantitation is required

No mutations identified AND Clinical picture consistent with WD

Liver biopsy with histology and Cu quantitation.

Diagnosis established
Initiate treatment
Initiate family screening

No mutations identified AND Clinical picture supports an alternative diagnosis

Continue evaluation for alternative diagnosis

No mutations identified AND Clinical picture supports an alternative diagnosis

Continue evaluation for alternative diagnosis

No mutations identified AND Cu <250 mcg/g dry weight and inconsistent histology

Continue evaluation for alternative diagnosis