**Table I. Scoring system** **for the diagnosis of Wilson disease**

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| **Liver copper (in absence of cholestasis)** | | | | **Serum caeruloplasmin** | | | |
| Normal (<50 µ/g) | | -1 | | Normal (>0.2 g/L) | | | 0 |
| <5xULN (50-250 µ/g) | | 1 | | 0.1-0.2 g/L | | | 1 |
| >5xULN (250 µ/g) | | 2 | | <0.1 g/L | | | 2 |
| **Rhodanine** **stain (in absence of quantitative liver copper determination)** | | | |  | | | |
| absent | | 0 | |  | | |  |
| present | | 1 | |  | | |  |
| **Mutation analysis** | | | | **Clinical symptoms and signs** | | | |
| 2 chromosome mutations | | 4 | | KF rings | | |  |
| 1 chromosome mutation | | 1 | | present | | | 2 |
| no mutation detected | | 0 | | absent | | | 0 |
| **Urinary copper (in absence of acute hepatitis)** | | | |  | | | |
| normal (<0.9 µmol/d or <100 mg/d) | | 0 | | severe | | | 2 |
| 1-2x ULN | | 1 | | mild | | | 1 |
| >2x ULN | | 2 | | absent | | | 0 |
| normal but >5x ULN after penicillamine | | 2 | | Coomb's negative hemolytic anemia | | |  |
|  | |  | | present | | | 1 |
|  | | 2 | | absent | | | 0 |
|  | | | | | | | |
| **Score** | | | | | | | |
|  | **≥4** | | **2-3** | | **≤1** | **Total** | |
| **Wilson disease patients** | 50 | | 3 | | 0 | 53 | |
| **Other diagnosis** | 5 | | 40 | | 45 | 90 | |
|  | **True+** | | **False-** | | **False+** | **True-** | |
| **Wilson disease patients** | 50 | | 3 | |  |  | |
| **Other diagnosis** | 94% | | 94% | | 91% | 97% | |
|  | **Sensitivity** | | **Specificity** | | **+Predictive value** | **Predictive value** | |
|  | 94% | | 94% | | 91% | 97% | |