**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% |