

## Using Genetic Testing for Diagnosis and Risk-Assessment for Wilson Disease (WD)

### Indications:

- Suspicion of Wilson disease based on biochemical testing
- Presence of Kayser-Fleischer rings
- Unexplained liver disease, neurological, or psychiatric symptoms typical of WD
- Known familial mutation in *ATP7B*

### Benefits:

Genetic testing for WD

- Can confirm a suspected clinical or biochemical diagnosis of WD and identify the familial mutation(s).
- Can enable an accurate diagnosis of WD in very young children or in cases of equivocal biochemical findings.
- Can identify presymptomatic family members of patients, enabling preventative treatment.
- Can distinguish presymptomatic family members from heterozygous carriers, who do not need and should not receive preventative treatment.

### Background:

- Wilson disease is an autosomal recessive disorder of copper metabolism characterized by abnormal accumulation of copper in the liver, brain, and other organs.<sup>1</sup>
- Wilson disease affects approximately 1 out of every 30,000 persons worldwide.<sup>2</sup>
- Wilson disease most typically first presents between ages 6 and 50 years with acute or chronic liver disease (40% of affected), neurological movement disorders (40% of affected) or psychiatric disturbances such as depression or neurotic behaviors (20% of affected).<sup>1,3</sup>
- Wilson disease has been linked to mutations in the gene *ATP7B*. Mutations in *ATP7B* that cause complete lack of function have been associated with earlier disease onset and severity than mutations that allow residual activity.<sup>3</sup>
- Early or prophylactic treatment can preserve liver function and prevent development of neuropsychiatric illness.<sup>3</sup>
- Biochemical testing may give equivocal results in young children and cannot reliably distinguish presymptomatic individuals at high risk of becoming affected with Wilson disease from unaffected carriers of Wilson disease.<sup>3</sup>
- **References:** 1. Hamosh A et al (2010) OMIM 277900 2. de Bie et al (2007). J Med Genet 44:673-688. 3. Kniffin CL (2007) OMIM 606882.

**Ordering Information:** Please see other side.

## Ordering Information for Wilson Disease (WD) Testing

### Indications for Testing

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### Ordering Information

Gene(s)	Test Code
<i>ATP7B</i>	252803

### Family Testing (single amplicon)

Family Testing is available. Please contact Client Services at 1-866-647-0735 for requirements.

### Test Methodology

- Amplification by polymerase chain reaction (PCR); sequencing of entire protein-coding region

NOTE: Specimens must be accompanied by a completed consent form. In the case of family tests (ie, known mutations), a copy of the result of the first patient tested in the family (the index case) must be submitted unless that test was performed at Correlagen. Other family members are subsequently tested for the specific mutation found in the first patient tested.

**For test information, sample requirements, or to request a sample shipping kit, please contact Client Services at 1-866-647-0735 or visit us on the web at [www.correlagen.com](http://www.correlagen.com).**