Gene test information

WILSON DISEASE
(ATP7B H1069Q MUTATION)

- **Background**

  Wilson's disease is an autosomal-recessive disorder caused by mutation in the ATP7B gene, with resultant impairment of biliary excretion of copper. Subsequent copper accumulation, first in the liver but ultimately in the brain and other tissues, produces protean clinical manifestations that may include hepatic, neurological, psychiatric, ophthalmological, and other derangements.

  The most common mutation in patients from Central, Eastern, and Northern Europe is the point mutation H1069Q. About 50–80% of Wilson disease (WD) patients from these countries carry at least one allele with this mutation with an allele frequency ranging between 30 and 70%.

- **Indications for testing**

  Suspected diagnosis of Wilson's disease

References:

Ferenci, Regional distribution of mutations of the ATP7B gene in patients with Wilson disease: Impact on genetic testing. Hum Genet. 2006;120:151-9.

Laboratory for Molecular Genetics
Univ.-Doz. Prof. Dr. Wilfried Renner

Schaftal 50
8044 Kainbach bei Graz

Tel +43 (0) 664 124 2483
Fax +43 (0) 316 231123 7030

www.labor-renner.at
office@labor-renner.at