Test Definition: WDKM

Wilson Disease Known Mutation, ATP7B DNA Sequencing

Reporting Title: Wilson Disease Known Mutation
Performing Location: Rochester

Specimen Requirements:

This test can only be performed if a mutation has previously been identified in a family member of this individual.

Specimen must arrive within 96 hours of collection.

Specimen Type: Whole blood
Container/Tube:
Preferred: Lavender top (EDTA) or yellow top (ACD)
Acceptable: Any anticoagulant
Specimen Volume: 3 mL
Collection Instructions:
1. Invert several times to mix blood.
2. Send specimen in original tube.
Specimen Stability Information: Ambient (preferred)/Refrigerated
Forms:
1. Molecular Genetics-Congenital Inherited Diseases Patient Information Sheet (Supply T521) in Special Instructions
2. New York Clients-Informed consent is required. Please document on the request form or electronic order that a copy is on file. An Informed Consent for Genetic Testing (Supply T576) is available in Special Instructions.
3. If not ordering electronically, submit a Molecular Genetics Request Form (Supply T245) with the specimen.

Specimen Type	Temperature	Time
Varies	Varies	

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
22696	Specimen	Alphanumeric		N/A
22697	Specimen ID	Alphanumeric		N/A
22698	Source	Alphanumeric		31208-2
22699	Order Date	Alphanumeric		N/A
22700	Reason For Referral	Alphanumeric		42349-1
22701	Method	Alphanumeric		49549-9
22702	Result	Alphanumeric		In Process
22703	Interpretation	Alphanumeric		69047-9
22704	Amendment	Alphanumeric		In Process

Result ID	Reporting Name	Type	Unit	LOINC®
22705	Reviewed By:	Alphanumeric		N/A
22706	Release Date	Alphanumeric		N/A

CPT Code: 1 × 81403

Reference Values:

An interpretive report will be provided.