Wilson Disease (ATP7B) Sequencing
ARUP test code 2010716

ATP7B Sequencing Specimen: Whole Blood

ATP7B Sequencing Interpretation:

Negative

TEST PERFORMED – 2010716
TEST DESCRIPTION – Wilson Disease (ATP7B) Sequencing
INDICATION FOR TEST – Not Provided

RESULT
No pathogenic variants were detected in the ATP7B gene.

INTERPRETATION
No pathogenic variants were detected in the ATP7B gene by sequencing all coding regions, promoter, and intron-exon boundaries. This result significantly decreases the likelihood that this individual is affected with, or a carrier of, Wilson disease. Please refer to the background information included in this report for the clinical sensitivity and limitations of this test.

RECOMMENDATIONS
Medical management should rely on clinical findings and family history. Genetic consultation is recommended.

COMMENTS
Reference Sequence: GenBank # NM_000053.3 (ATP7B)
Nucleotide numbering begins at the "A" of the ATG initiation codon.
Likely benign and benign variants are not included in this report.

This result has been reviewed and approved by Pinar Bayrak-Toydemir, M.D., Ph.D.

H=High, L=Low, *=Abnormal, C=Critical
BACKGROUND INFORMATION: Wilson Disease (ATP7B) Sequencing

CHARACTERISTICS: Wilson disease is a disorder of copper metabolism caused by mutations in the ATP7B gene. Toxic accumulation of copper in body tissues, particularly the liver and central nervous system, causes progressive disease that is eventually lethal if untreated. The clinical presentation of Wilson disease is highly variable and age-dependent. Symptoms, including Kayser-Fleisher rings, liver disease, neurologic findings, and psychiatric disease, may present at any time from early childhood to late adulthood.

INCIDENCE: 1/30,000-1/50,000

INHERITANCE: Autosomal recessive.

PENETRANCE: Age-dependent.

CAUSE: Pathogenic ATP7B gene mutations.

CLINICAL SENSITIVITY: 98 percent.

METHODOLOGY: Bidirectional sequencing of the entire ATP7B coding region and intron/exon boundaries.

ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.

LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. Regulatory region mutations, deep intronic mutations, and large deletions/duplications will not be detected. Mutations in genes other than ATP7B are not evaluated.

See Compliance Statement C: www.aruplab.com/CS

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