ARUP LABORATORIES | aruplab.com
500 Chipeta Way, Salt Lake City, Utah 84108-1221
phone: 801-583-2787, toll free: 800-522-2787
Tracy I. George, MD, Chief Medical Officer

Patient Report

Patient Age/Gender: 1 day Male

Specimen Collected: 27-Oct-21 16:19

Wilson Disease (ATP7B) by NGS | **Received: 28-Oct-21 13:01** **Report/Verified: 28-Oct-21 14:13**

Procedure	Result	Units	Reference Interval
Wilson Disease (ATP7B) Specimen	Whole Blood		
Wilson Disease (ATP7B) Interp	See Note [i1]		

Test Information

i1: Wilson Disease (ATP7B) Interp

BACKGROUND INFORMATION: Wilson Disease (ATP7B) Sequencing

CHARACTERISTICS: Wilson disease is a disorder of copper metabolism caused by pathogenic variants 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-Fleischer rings, liver disease, neurologic findings, and psychiatric disease, may present at any time from early childhood to late adulthood.

PREVALENCE: 1 in 10,000 to 1 in 30,000.

CAUSE: Pathogenic germline variants in ATP7B.

INHERITANCE: Autosomal recessive.

PENETRANCE: Age dependent.

CLINICAL SENSITIVITY: 98 percent.

GENE TESTED: ATP7B (NM_000053)
Deletion/duplication analysis is not available for this gene.

METHODOLOGY: Probe hybridization-based capture of all coding exons and exon-intron junctions of the targeted genes, followed by massively parallel sequencing. Sanger sequencing was performed as necessary to fill in regions of low coverage and confirm reported variants. Human genome build 19 (Hg 19) was used for data analysis.

ANALYTICAL SENSITIVITY/SPECIFICITY: The analytical sensitivity is approximately 99 percent for single nucleotide variants (SNVs) and greater than 93 percent for insertions/duplications/deletions (indels) from 1-10 base pairs in size. Indels greater than 10 base pairs may be detected, but the analytical sensitivity may be reduced. Specificity is greater than 99.9 percent for all variant classes.

LIMITATIONS: A negative result does not exclude a diagnosis of Wilson disease. This test only detects variants within the coding regions and intron-exon boundaries of

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the ATP7B gene. Regulatory region variants and deep intronic variants will not be identified, including the Sardinian founder variant, c.-436_-422del15. Deletions/duplications/insertions of any size may not be detected by massively parallel sequencing. Diagnostic errors can occur due to rare sequence variations. In some cases, variants may not be identified due to technical limitations caused by the presence of pseudogenes, repetitive, or homologous regions. This test is not intended to detect low-level mosaic or somatic variants, gene conversion events, complex inversions, translocations, mitochondrial DNA (mtDNA) mutations, or repeat expansions. Interpretation of this test result may be impacted if this patient has had an allogeneic stem cell transplantation. Non-coding transcripts were not analyzed.

Unless otherwise indicated, testing performed at:
ARUP Laboratories
500 Chipeta Way, Salt Lake City, UT 84108
Laboratory Director: Tracy I. George, MD