

Specimen Collected: 25-Oct-21 09:19

Biotinidase Deficiency by NGS	Received: 26-Oct-21 08:40	Report/Verified: 26-Oct-21 08:44
Procedure	Result	Units
Reference Interval		

Biotinidase Deficiency Whole Blood

Specimen

Biotinidase Deficiency See Note ¹¹

Interp

Test Information

11: Biotinidase Deficiency Interp

BACKGROUND INFORMATION: Biotinidase Deficiency (BTD) Sequencing

CHARACTERISTICS: Deficiency in biotinidase enzymatic activity impairs the body's ability to recycle and reuse the vitamin biotin, resulting primarily in neurologic and dermatologic symptoms. Manifestations of profound biotinidase deficiency (BTD) include ataxia, hypotonia, developmental delay, seizures, vision problems, hearing loss, alopecia, metabolic ketolactic acidosis, organic aciduria, and hyperammonemia.

EPIDEMIOLOGY: The incidence of profound and partial biotinidase deficiency is approximately 1:60,000

CAUSE: Pathogenic germline variants in the BTD gene

INHERITANCE: Autosomal recessive

CLINICAL SENSITIVITY: 99 percent

GENE TESTED: BTD (NM_000060)

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 BTD. This test only detects variants within the coding regions and intron-exon boundaries of the BTD gene. Deletions/duplications/insertions of any size may not be detected by massively parallel sequencing. Regulatory region variants and deep intronic variants will not be identified. Diagnostic errors can occur due to rare sequence variations. In some

*=Abnormal, #=Corrected, C=Critical, f=Result Footnote, H-High, i-Test Information, L-Low, t-Interpretive Text, @=Performing lab

Unless otherwise indicated, testing performed at:

ARUP Laboratories

500 Chipeta Way, Salt Lake City, UT 84108

Laboratory Director: Tracy I. George, MD

ARUP Accession: n/a

Report Request ID: 15055207

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Test Information

i1: Biotinidase Deficiency Interp

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.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

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