New Test: 3000544 Chronic Granulomatous Disease Panel (CYBB Sequencing and NCF1 Exon 2 GT Deletion) CGD PAN

Methodology: Polymerase Chain Reaction/ Sequencing/ High Resolution Melt Analysis
Performined: Sun-Sat
Reported: Within 2 weeks

Specimen Required: Collect: Lavender (EDTA), Pink (K$_2$EDTA), Yellow (ACD).
Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)
Storage/Transport Temperature: Refrigerated.
Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 6 months

Interpretive Data:

Background Information for Chronic Granulomatous Disease Panel (CYBB Sequencing and NCF1 Exon 2 GT Deletion):

Characteristics of Chronic Granulomatous Disease (CGD): A primary immunodeficiency disorder characterized by recurrent, severe bacterial and fungal infections of the skin, lymph nodes, liver, lungs, bones, or visceral organs. Dysregulated inflammatory responses result in granulomas.

Incidence: Approximately 1 in 250,000 births.

Inheritance: X-linked recessive for CYBB; de novo variants in 10-20 percent of affected males. Autosomal recessive for NCF1.

Cause: Pathogenic variants in the X-linked CYBB gene (60-70 percent), pathogenic variants in autosomal recessive genes NCF1 (25 percent), CYBA (Less than 5 percent), NCF2 (Less than 5 percent) and NCF4 (very rare).

Clinical Sensitivity: Up to 78 percent for CGD
Methodology: Bidirectional sequencing of the CYBB coding region and intron-exon boundaries. Polymerase Chain Reaction/High-Resolution Melt Analysis to assess for the common NCF1 c.75_76delGT variant.
Analytical Sensitivity: 99 percent for CYBB and homozygous NCF1 c.75_76delGT deletion, 90 percent for heterozygous NCF1 c.75_76delGT deletion.
Analytical Specificity: 99 percent.

Limitations: Diagnostic errors can occur due to rare sequence variations. Regulatory region variants, deep intronic variants, and large duplications will not be detected in patients of either sex; large deletions will not be detected in females. Variants in NCF1 other than c.75_76delGT are not evaluated. Because of potential recombination between NCF1 and its pseudogenes, the lack of detection of the c.75_76delGT variant does not rule out carrier status for autosomal recessive CGD.

CPT Code(s): 81479

New York DOH approval pending. Call for status update.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.