**New Test** | 3000541 | Chronic Granulomatous Disease, X-Linked (CYBB) Sequencing | CYBB FGS

**Methodology:** Polymerase Chain Reaction/Sequencing  
**Performed:** Sun-Sat  
**Reported:** Within 2 weeks

**Specimen Required:**  
- **Collect:** Lavender (EDTA), Pink (K2EDTA), 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, X-Linked (CYBB) Sequencing:**

**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.

**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:** 51-60 percent for CGD.

**Methodology:** Bidirectional sequencing of the CYBB coding region and intron-exon boundaries.

**Analytical Sensitivity and 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 genes other than CYBB are not evaluated.

**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.