ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

A nonprofit enterprise of the University of Utah and its Department of Pathology

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THIS IS NOT A TEST REQUEST FORM.
The information below is required to perform CGD testing.
Please fill out this form and submit it with the test request form or electronic packing list.

PATIENT HISTORY FOR CHRONIC GRANULOMATOUS DISEASE (CGD) TESTING

Patient Name_____ **Date of Birth**_____ **Sex** ☐ **F** ☐ **M**

Physician_____ **Physician Phone** _____

Physician Specialty_____

Genetic Counselor_____ **Counselor Phone** _____

Patient's ethnicity (check all that apply)

☐ African American ☐ Ashkenazi Jewish ☐ Asian ☐ Caucasian
☐ Hispanic ☐ Middle Eastern ☐ Native American ☐ Other _____

Does the patient have SYMPTOMS of CGD? ☐ No ☐ Yes, check all that apply

Age at first presentation:_____

☐ Recurrent infection ☐ Nausea/vomiting ☐ Gastric obstruction
☐ Pneumonia ☐ Diarrhea ☐ Bone infection
☐ Lymphadenitis ☐ Colitis ☐ Poor wound healing
☐ Skin abscess ☐ Urinary stricture/ obstruction ☐ Other_____
☐ Visceral abscess (location:_____)

Has FUNCTIONAL TESTING (neutrophil oxidative burst assay; DHR) been performed? ☐ **No** ☐ **Yes** ☐ **Unknown**

If yes, what was result? ☐ Abnormal ☐ Normal ☐ Unknown

Radiological Findings

Bone scan	☐ Abnormal	☐ Normal	☐ Not performed
Chest x-ray	☐ Abnormal	☐ Normal	☐ Not performed

Does the patient have a FAMILY HISTORY of CGD? ☐ No ☐ Yes ☐ Unknown

If yes, attach a PEDIGREE or specify the RELATIONSHIP of the family member(s) to the patient and detail the symptoms/age of onset in each symptomatic relative.

_____

What is the disease severity in affected males in the family? ☐ Severe ☐ Mild ☐ Unknown

Was DNA testing performed for these family members? ☐ Yes ☐ No ☐ Unknown

If yes, please attach a copy of the relative's DNA laboratory result (REQUIRED for familial mutation testing).

Has the patient undergone previous DNA testing for CGD? ☐ No ☐ Yes

If yes, please describe test(s) and results_____

Check the CGD test you intend to order.

☐ **3000544 Chronic Granulomatous Disease Panel (*CYBB* Sequencing and *NCF1* Exon 2 GT Deletion)**
Sequencing of the *CYBB* coding region and intron/exon boundaries; targeted testing for the common pathogenic *NCF1* c.75_76delGT variant. Clinical sensitivity up to 78% for CGD.

☐ **3000541 Chronic Granulomatous Disease, X-Linked (*CYBB)* Sequencing**
Sequencing of the *CYBB* coding region and intron/exon boundaries. Clinical sensitivity 51-60% for CGD.

☐ **2006366 Chronic Granulomatous Disease (*NCF1*) Exon 2 GT Deletion**
Targeted testing for the common GT deletion in exon 2 of *NCF1* associated with autosomal recessive CGD. Clinical sensitivity is 18% for CGD.

☐ **2001961 Familial Mutation, Targeted Sequencing**
Tests for a *CYBB* sequence change previously identified in a family member. A copy of an affected relative's DNA laboratory result is REQUIRED.

Master Label

For questions, contact an ARUP genetic counselor at (800) 242-2787, ext. 2141