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.
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 _____

Practice Specialty _____ Physician Fax _____

Genetic Counselor _____ Counselor Phone _____

Patient's Ethnicity (check all that apply)

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

Does the patient have symptoms? ☐ No ☐ Yes (check all that apply)

☐ Age of onset: _____
☐ Recurrent infection
☐ Pneumonia
☐ Lymphadenitis
☐ Skin abscess
☐ Nausea/vomiting
☐ Diarrhea
☐ Colitis
☐ Urinary stricture/ obstruction
☐ Gastric obstruction
☐ Bone infection
☐ Poor wound healing
☐ Visceral abscess (location: _____)
☐ Other symptom(s): _____

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

Has the patient undergone previous DNA testing? ☐ No ☐ Yes ☐ Unknown
If yes, describe the test(s) and results: _____

_____

Is there any relevant family history? ☐ No ☐ Yes ☐ Unknown
If yes, attach a pedigree or specify the relative's relationship to the patient. List their symptoms and age of onset:

_____

_____

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

Has DNA testing been performed for the family member(s)? ☐ No ☐ Yes ☐ Unknown
If yes, attach a copy of the relative's DNA laboratory result. (REQUIRED for familial mutation testing)

Check the 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 relative's lab result is REQUIRED.

Master Label

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