

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 _____

Circle 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