

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 ____/____/____ Gender F M
 Physician _____ Physician Phone (____) _____ Practice Specialty _____
 Genetic Counselor _____ Counselor Phone (____) _____

Patient's Ethnicity (check all that apply)

- | | | | |
|---|---|--|--------------------------------------|
| <input type="checkbox"/> African American | <input type="checkbox"/> Ashkenazi Jewish | <input type="checkbox"/> Asian | <input type="checkbox"/> Caucasian |
| <input type="checkbox"/> Hispanic | <input type="checkbox"/> Middle Eastern | <input type="checkbox"/> Native American | <input type="checkbox"/> Other _____ |

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

- Age at first presentation: _____
- | | | |
|--|---|--|
| <input type="checkbox"/> Recurrent infection | <input type="checkbox"/> Nausea/ vomiting | <input type="checkbox"/> Gastric obstruction |
| <input type="checkbox"/> Pneumonia | <input type="checkbox"/> Diarrhea | <input type="checkbox"/> Bone infection |
| <input type="checkbox"/> Lymphadenitis | <input type="checkbox"/> Colitis | <input type="checkbox"/> Poor wound healing |
| <input type="checkbox"/> Skin abscess | <input type="checkbox"/> Urinary stricture/ obstruction | <input type="checkbox"/> Other _____ |
| <input type="checkbox"/> 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 | <input type="checkbox"/> Abnormal | <input type="checkbox"/> Normal | <input type="checkbox"/> Not performed |
| Chest x-ray | <input type="checkbox"/> Abnormal | <input type="checkbox"/> Normal | <input type="checkbox"/> 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.

2006356 Chronic Granulomatous Disease (CYBB Gene Scanning and NCF1 Exon 2 GT Deletion) with Reflex to CYBB Sequencing
 Gene scanning of the *CYBB* coding region and intron/exon boundaries with identified variants confirmed by sequencing. Targeted testing for the common GT deletion in exon 2 of *NCF1*. Clinical sensitivity is 86% for CGD.

2006361 Chronic Granulomatous Disease, X-Linked (CYBB) Gene Scanning with Reflex to Sequencing
 Gene scanning of the *CYBB* coding region and intron/exon boundaries with identified variants confirmed by sequencing. Clinical sensitivity is 68% 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.

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

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