



CGD Registry Data Collection Form

Patient Identification:

Patient Name (first, middle, last) _____

Date of this Record (mm/dd/yyyy): ____/____/____

Patient's Number Assigned by Registry _____

Is this the initial registration of this patient[] or follow-up?[]

Patient Initials ____/____/____ Date of Birth ____/____/____(mm/dd/yyyy) Gender: male [], female []

Home Address:

Address: _____

State: _____

Zip Code: _____

Phone: _____

Email: _____

State or Province of birth: _____

Country of birth: _____

Submitting Physician Information:

Name: _____

Address: _____

State: _____

Zip Code: _____

Phone: _____

Email: _____

Fax: _____

Diagnostic Criteria

Definitive []

Male or female patient with abnormal nitroblue tetrazolium (NBT), dihydrorhodamine (DHR), dichlorofluorescein (DCF) or respiratory burst in activated neutrophils (less than 5% of control) who has **one** of the following:

Mutation in gp91, p22, p47 or p67 phox []

Absent gp91, p22, p47 or p67 phox by immunoblot []

Maternal cousins, uncles, nephews or sibs with an abnormal respiratory burst []

Probable []

Male or female patient with abnormal respiratory burst in activated neutrophils (NBT, DHR, or DCF less than 5% of control) who has **one** of the following:

Deep seated infection (liver, perirectal or lung abscess; adenitis; or osteomyelitis) due to Staphylococcus, Serratia marcescens, Burkholderia, Nocardia or aspergillus []

Granulomatous obstruction of gastrointestinal or urogenital tracts []

Failure to thrive and hepatosplenomegaly or lymphadenopathy []

Maternal cousins, uncles, nephews or sibs with an abnormal respiratory burst []

Spectrum of Disease

Patients with the X-linked form of CGD (60-70% of patients) tend to present earlier and have more severe disease than patients with autosomal recessive forms. Many patients with X-CGD develop failure to thrive, bacterial adenitis, abscesses or osteomyelitis within the first 3 years of life. Pneumonia and lymphadenitis due to catalase-positive organisms - Staphylococcus, Serratia, Burkholderia, Nocardia, Chromobacterium and fungi are common. Symptoms of intestinal or urinary tract obstruction can be caused by granuloma formation. In both the X-linked and autosomal recessive forms, the first severe symptoms may not be recognized until adulthood.

Differential diagnosis: LAD, Sarcoidosis, Hyper IgE syndrome

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Patient Initials: ___/___/___

Tests Performed (check all that apply)

Indicate those tests/data used to establish/confirm the diagnosis by checking the indicated box []

- Dihydrorhodamine oxidation (DHR) [] **Stimulation index if known** []
- Nitroblue tetrazolium test (NBT) [] slide [], quantitative [], unknown [] data from IDF Reg
- Superoxide production []
- Hydrogen peroxide production []
- Oxygen consumption []
- Bacterial killing []
- Chemiluminescence []
- Dichlorofluorescein (DCF) []
- None of the above []
- Other (specify) _____

Genetic Information []

Pattern of inheritance:

Sporadic [] (no family history), X-linked [], autosomal recessive [], unknown []

Pedigree Analysis []

Family history unknown []

Consanguinity-Parents related? (explain) _____

Relation	Tested Normal	Carrier	Not Tested	CGD – alive	CGD – deceased	Unknown	Undiagnosed with suggestive symptoms
Mother							
Father							
Brothers (fill in # in each category)							
Sisters (fill in # in each category)							
Maternal uncles (fill in # in each category)							
Other affected family members							

Other affected family members? (explain): _____

Information on other affected kindred members listed above (e.g., sibling, cousin, &/or maternal uncle)

Relationship	Initials	Gender	DOB

Gene Mutation []

X-linked (gp91) []

Autosomal recessive: p22phox [], p47phox [], p67phox []

(Number nucleotides using Human Mutation 11:1-3, 1998)

Nucleotides affected (e.g., 361C>T) _____

Predicted Amino Acid Change (e.g., W140R) _____

Insertion / Deletion / Frameshift / Splice Site (please explain) _____

Mutation tested for but not found _____

Publications or mutation database (please give citation - if published) _____

Protein/Enzyme expressed? Yes _____, No _____, Not tested _____

CGD Registry Data Collection Form

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Clinical Features of Special Note List all complications at the end of this section *.

	Observed	Prominent	Organisms
Pneumonia			
Suppurative adenitis			
Osteomyelitis			
Septic arthritis			
Sepsis			
Lung abscess			
Liver abscess			
Brain abscess			
Subcutaneous abscess			
Meningitis			
Gastric outlet obstruction			
Colitis/enteritis			
Seborrheic dermatitis			
Other (explain)			
None of the above			
Comments:			

*Complications:

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Other Treatments/Procedures Used: List all complications at the end of this section *.

Anti-infectives List all complications at the end of this section*.

Prophylactic antibiotic	Continuous		Intermittent		Outcome	Currently used	
	Yes	No	Yes	No		Yes	No
Trimethoprim-sulfa							
Dicloxacillin/Keflex							
Other chronic antibiotics (list):							

Anti-fungals (systemic) (list):	Indication	Continuous		Intermittent		Outcome	Currently used	
		Yes	No	Yes	No		Yes	No

*Complications from anti-infectives::

Cytokines List all complications at the end of this section *.

	Yes	No	Age or Date	Dose	Response
Gamma-interferon					
G-CSF					
GM-CSF					
IL-2					
Other (list):					

*Complication from anti-inflammatory and immunosuppressive treatments: :

*Complications:

