

CGD Registry Data Collection Form

Patient Initials: ___/___/___

Patient Identification:

Patient Name (first, middle, last) _____

Patient's USIDNET Registry Number assigned after online enrollment _____

Date of Birth ___/___/___ (mm/dd/yyyy) or Year of Birth _____ Male [], Female []

Home Address:

Address: _____

State: _____

Zip Code: _____

Phone: _____

Email: _____

State or Province of birth: _____

Country of birth: _____

Date of this Record Completion (mm/dd/yyyy): ___/___/___

Date of Visit (mm/dd/yyyy): ___/___/___

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

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), dichlorofluoroscein (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 []

CGD Registry Data Collection Form

Patient Initials: ___/___/___

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

Tests Performed (check all that apply)

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

Test	Yes	No	Unknown
Dihydrorhodamine oxidation (DHR)			
Stimulation index if known _____			
Nitroblue tetrazolium test (NBT)			
slide [] OR quantitative []			
Superoxide production			
Hydrogen peroxide production			
Oxygen consumption			
Bacterial killing			
Chemiluminescence			
Dichlorofluorescein (DCF)			
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) _____

Please list additional relationships. If more space is needed, please use the Memo section at the end of this form.

Relation	CGD – Alive	CGD – Deceased	Tested Normal	Carrier	Not Tested	Unknown	Undiagnosed with suggestive symptoms
Mother							
Father							

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

Relationship	Initials	Gender	Year of Birth	Listed in Registry? Yes/No/Unknown

CGD Registry Data Collection Form

Patient Initials: ___/___/___

Gene Mutation

X-linked (gp91) [] OR 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 [] which gene(s) _____

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

Protein/Enzyme expressed? Yes _____, No _____, Not tested _____ [] Western [] FACS

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

	Not Seen	Observed	Prominent	Unknown	Organism(s)
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:

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::

Cytokine Treatment List all complications at the end of this section *.

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

*Complications:

CGD Registry Data Collection Form

Patient Initials: ___/___/___

Other treatments used List all complications at the end of this section *.

	Yes	No	Unknown	Age begun		OR Date begun (mm/yyyy)	Duration	Still needed?	
				Mos.	Yrs.			Yes	No
Chronic oxygen									
Parenteral nutrition									

	Yes	No	Unknown	Age begun		OR Date begun (mm/yyyy)	Cells	Vector	Outcome
				Mos	Yrs				
Gene therapy									
Publication?									

Other? Please explain _____

Was steroid / immunosuppressive therapy needed for management? Yes [], No [], Unknown []

What kind? _____ dose _____ duration _____

What kind? _____ dose _____ duration _____

What reason?

ITP
 Hemolytic Anemia
 Lung Disease

IBD or other gastrointestinal disease
 Gastric outlet obstruction
 Bladder outlet obstruction
 Lymphoma
 Vasculitis

Other (specify): _____

Did any of these treatments lead to significant complications? Yes [], no [], unknown [], explain:

*Complications:

Additional comments:
