



SCID and Combined Immunodeficiency Disease (CID) 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 ____/____/____ Gender: male [] female []
(mm/dd/yyyy)

Home

Address: _____

State: _____

Zip Code: _____

Phone: _____

Email: _____

State or Province of birth: _____

Country of birth: _____

Data transferred from earlier IDF Registry? Yes [] no []

Submitting Physician Information:

Name: _____

Address: _____

State: _____

Zip Code: _____

Phone: _____

Email: _____

Fax: _____

Diagnostic Criteria: SCID

Definitive []

Male or female patient **with:**

Absolute lymphocyte count of less than 3000/mm³, less than 20% CD3+ T cells, proliferative responses to mitogens less than 10% of control (**or** the presence of transplacentally acquired maternal T cells); and defect in one of the known SCID disease genes listed below.

Probable []

Male or female patient less than 4 years of age with less than 20% CD3+ T cells, an absolute lymphocyte count of less than 3000/mm³ and proliferative responses to mitogens less than 10% of control [] **or** the presence of maternal lymphocytes in the circulation [].

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Spectrum of disease

Patients with SCID usually develop failure to thrive and persistent diarrhea, respiratory symptoms and/or thrush in the first 2 to 7 months of life. Pneumocystis pneumonia, serious viral infections, significant bacterial infections and disseminated BCG infection are common presenting illnesses. Occasional patients do not have failure to thrive and are not recognized to have immunodeficiency until the second year of life, or rarely even later. SCID is usually fatal in the first 2 years of life unless the patient is treated with extremely restrictive isolation, hematopoietic stem cell transplantation or therapy that replaces the abnormal gene or gene product. Atypical cases of SCID may present with T cells because of hypomorphic mutations or somatic corrective mutations in T-cell precursors.

Differential diagnosis

- 1) HIV infection
- 2) Congenital rubella
- 3) Complete DiGeorge syndrome
- 4) Zap70 deficiency
- 5) Cartilage hair hypoplasia
- 6) MHC class II deficiency
- 7) PNP deficiency
- 8) FOXP1 deficiency

Diagnostic Criteria: Combined Immunodeficiency (CID)

There are a number of different genetic disorders of the immune system that result in combined immunodeficiency that generally does not reach a level of clinical and/or immunological severity to qualify as severe combined immunodeficiency. A listing of several of these disorders follows, although there may be many more syndromes that qualify for inclusion as CID that are not listed. Please use the "other" or "unknown" designations when registering patients in this later category.

Genetic Information

Pattern of inheritance:

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

Documented autosomal recessive by mutation analysis or affected relative(s) [],

or consistent with X-linked or autosomal recessive (e.g. male with male affected sibling) []

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Pedigree Analysis

Family history unknown []

Probable or documented consanguinity-Parents related? (explain) _____

Relation	Tested Normal	Carrier	Not Tested	S/CID-alive	S/CID-dead	Unknown	Undiagnosed with suggestive symptoms
Mother							
Father							
Brothers (fill in # in each category)							
Sisters (fill in # in each category)							
For below: fill in # in each category, indicate (M)aternal or (P)aternal							
Uncles							
Aunts							
Grandmothers							
Grandfathers							
Cousins							
Other affected family members							

Information on Other Affected Kindred Members listed above (e.g., sibling, cousin, &/or maternal uncle):

Relation	Initials	DOB	Gender

Gene Mutation

Molecular Type of SCID: γ c [], ADA [], Jak3 [], RAG1 [], RAG2 [], IL7R α [], Artemis [], CD3 δ [], CD3 ϵ [], CD3 ζ [], CD45 [], other [] _____, none found []

Molecular Type of CID: PNP [], ZAP70 [], CD25 [] Omenn (list abnormal gene if known) [], Bare lymphocyte (MHC class-II) [], Cartilage-Hair Hypoplasia [], other [] _____, none found []
 (Number nucleotides using Human Mutation 11:1-3, 1998)

Nucleotides affected, using "NM" reference RNA sequence (GenBank) and HUGO Nomenclature (e.g., allele 1: 289C>T; allele 2: 154delG) _____

Predicted Amino Acid Changes (e.g., allele 1: W140R; allele 2: M1I) _____

Complex mutation (please explain) _____

Gross deletion or duplication (explain and state how documented) _____

Mutation tested for, but not found _____

Publications or mutation database where this family's genotype information has been included (please give citation - if published) _____

Protein/Enzyme expressed? Yes_____, No_____, Not tested_____

Blood ADA level _____ Blood deoxyadenosine level _____
 (State assay used, whether in erythrocytes or leukocytes, control values)

Additional comments _____

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Clinical and Laboratory Features of Special Note

	Not seen	Observed	Prominent in this patient
Failure to thrive	_____	_____	_____
Growth retardation	_____	_____	_____
Pneumonia	_____	_____	_____
Ventilation required?	_____	_____	_____
Severe or protracted diarrhea	_____	_____	_____
Autoimmune hemolytic anemia	_____	_____	_____
Thrombocytopenia	_____	_____	_____
Eosinophilia	_____	_____	_____
Edema	_____	_____	_____
Hypoproteinemia	_____	_____	_____
Hepatosplenomegaly	_____	_____	_____
Graft versus Host Disease	_____	_____	_____
due to maternal engraftment	_____	_____	_____
due to blood transfusion	_____	_____	_____
Neutropenia	_____	_____	_____
Hepatitis	_____	_____	_____
Skin rash	_____	_____	_____
Lymphoproliferative disease	_____	_____	_____
Other features (specify) _____			
Comments _____			

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

	Yes	No	Age begun	Date begun (mm/dd/yyyy)	Duration	Still needed?	
						Yes	No
Oxygen supplementation							
Parenteral nutrition							

Other? Please explain _____

*Comments:

PEG-ADA Immune reconstitution; full [], partial [], Rx failure []
Dose schedule _____

*Comments:

Additional comments:
