



SCID and Combined Immunodeficiency Disease (CID) 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 _____

Gender: 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: 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 under Molecular Type of SCID on page 3.

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) _____

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

| Relation | S/CID-alive | S/CID-dead | 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):

| Relation | Initials | Gender | Year of Birth | Listed in Registry? Yes/No/Unknown |
|----------|----------|--------|---------------|------------------------------------|
| | | | | |
| | | | | |
| | | | | |

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 (which genes) _____, _____, _____, _____, _____, _____, _____, _____, _____

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 _____ Western [] FACS []

| | Level | Assay Used | Cells | | Control Values |
|---|-------|------------|--------------|------------|----------------|
| | | | Erythrocytes | Leukocytes | |
| Plasma ADA (umol/h/ml) | | | | | |
| RBC deoxyadenine nucleotide (umol/ml/RBC) | | | | | |

Additional comments _____



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Clinical and Laboratory Features of Special Note

| | Not seen | Observed | Unknown | Prominent |
|--------------------------------|----------|----------|---------|-----------|
| 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 | Unknown | Age begun | OR 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:
