SEVERE COMBINED IMMUNODEFICIENCY (SCID)

CANADIAN PAEDIATRIC SURVEILLANCE PROGRAM

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REPORTING INFORMATION
(To be completed by the CPSP Senior Coordinator)

Report number:	
Month of reporting:	
Province:	
Today's date:	

Please complete the following sections for the case identified above.
Confidentiality of information will be assured.

CASE DEFINITION FOR SEVERE COMBINED IMMUNODEFICIENCY

Report any child less than two years of age with:

- the clinical features of SCID (i.e., chronic diarrhea, recurrent pneumonia, failure to thrive, persistent thrush, opportunistic infections, etc.)

and at least one of the following:

- an absolute lymphocyte count of less than 3000/mm^3 or less than 20% CD3$^+$ T cells
- familial history of primary immunodeficiency.

Exclusion criteria

Infants with HIV infection or cystic fibrosis.

SECTION 1 – DEMOGRAPHIC INFORMATION

1.1 Date of birth: ____ /_____ /_____ 1.2 Sex: Male ___ Female ___
DD MM YYYY

1.3 Province/Territory of residence: _____

SECTION 2 – ETHNICITY

2.1 Born in Canada? Yes ___ No ___ Unknown ___
If no, specify country of birth: _____

2.2 Is the child Aboriginal? Yes ___ No ___ Unknown ___
If yes, First Nations (or North American Indian) ___ Inuit ___ Métis ___ Unknown ___
If the child is First Nations, does he/she live on reserve? Yes ___ No ___ Unknown ___

SECTION 3 – FAMILIAL HISTORY

3.1 Other child(ren) in the family with immunodeficiency? Yes ___ No ___ Unknown ___

3.2 Family history of infant death either unexplained or by infection? Yes ___ No ___ Unknown ___

3.3 Any family history suggestive of an X-linked condition, such as male maternal relative being affected?
Yes ____ No ____ Unknown ____

3.4 Are the parents consanguineous (descended from common ancestor)? Yes ___ No ___ Unknown ___

SECTION 4 – CLINICAL FEATURES

4.1 Date of diagnosis: ____ /_____ /_____
DD MM YYYY

SECTION 4 – CLINICAL FEATURES (cont'd)

4.2 Clinical presentation

4.2.1 Failure to thrive: Yes ___ No ___ Unknown ___ If yes, age (months) at onset: _____

4.2.2 Chronic diarrhea: Yes ___ No ___ Unknown ___ If yes, age (months) at onset: _____

4.2.3 Interstitial pneumonia: Yes ___ No ___ Unknown ___ If yes, age (months) at onset: _____

If yes, please specify:_____

4.2.4 Persistent bronchiolitic-like illness: Yes ___ No ___ Unknown ___

If yes, age (months) at onset: _____

4.2.5 Persistent or recurrent superficial candidiasis: Yes ___ No ___ Unknown ___

If yes, age (months) at onset: _____

4.2.6 Oral and/or genital ulcers: Yes ___ No ___ Unknown ___ If yes, age (months) at onset: _____

4.2.7 Rash: Yes ___ No ___ Unknown ___ If yes, age (months) at onset: _____

4.2.8 Absent lymph nodes and tonsils: Yes ___ No ___ Unknown ___

4.2.9 Lymphadenopathy +/– hepatosplenomegaly: Yes ___ No ___ Unknown ___

If yes, age (months) at onset: _____

4.2.10 Opportunistic infections: Yes ___ No ___ Unknown ___ If yes, age (months) at onset: _____

If yes, please specify: _____

4.2.11 Any other significant infections: Yes ___ No ___ Unknown ___

If yes, age (months) at onset: _____

If yes, please describe: _____

4.2.12 Any other immune-related problems (e.g., autoimmunity, blood cytopenias):

Yes ___ No ___ Unknown ___ If yes, age (months) at onset: _____

If yes, please specify: _____

4.3 Disseminated BCG infection

4.3.1 BCG immunization: Yes ___ No ___ Unknown ___

If yes, date of immunization: ____ /_____ /_____
DD MM YYYY

4.3.2 Disseminated BCG infection: Yes ___ No ___ Unknown ___

If yes, date of diagnosis: ____ /_____ /_____
DD MM YYYY

4.3.3 Treated successfully? Yes ___ No ___ Unknown ___

SECTION 5 – INVESTIGATIONS RELATED TO SCID DIAGNOSIS
(Please check all investigations completed and provide the earliest relevant results where available.)

5.1 Total lymphocyte count:_____

5.2 Immunoglobulin levels (mg/dl) along with the appropriate units and normal ranges for your laboratory:

	Results	Units	Normal range
IgM	_____	_____	_____
IgA	_____	_____	_____
IgG	_____	_____	_____
IgE	_____	_____	_____

5.3 Lymphocyte subset absolute numbers (percentage of total lymphocytes)

CD3 T cell:_____ (____%) CD4 Tcell:_____ (____%) CD8 Tcell:_____ (____%)

CD19 B cell:_____ (____%) CD16 NK cell:_____ (____%) CD56 NK cell:_____ (____%)

5.4 Chest X-ray:

Presence of thymic shadow: Yes ___ No ___ Unknown ___

SECTION 6 – REFERRAL AND SPECIALIZED INVESTIGATION

6.1 Referred to a pediatric immunologist or other specialist? Yes ___ No ___ Unknown ___
If yes, name of specialist: _____
Name of specialist's hospital: _____

If the information is available, please complete sections 6.2. to 6.6; otherwise proceed to section 7.

6.2 Lymphocyte proliferation following mitogen stimulation
Results: Low/Absent proliferation ____ Normal proliferation ____

6.3 Purine and pyrimidine studies
ADA level: normal ____ abnormal ____

6.4 X-inactivation pattern in T cells of mother:
Unilateral pattern? Yes ___ No ___ Unknown ___

6.5 Specific molecular testings? Yes ___ No ___ Unknown ___
If yes, specimen sent to National Institutes of Health? Yes ___ No ___ Unknown ___
Results:_____

6.6 SCID classification
6.6.1 Is this X-linked SCID? Yes ___ No ___ Unknown ___
6.6.2 Is this an ADA deficiency? Yes ___ No ___ Unknown ___
6.6.3 Is this due to another genetic mutation? Yes ___ No ___ Unknown ___
If yes, which one:_____

SECTION 7 – TREATMENT

7.1 Bone marrow transplantation data
7.1.1 Referred for hematopoietic stem cell transplant: Yes ___ No ___ Unknown ___
If yes, where? _____
If no, why? _____
7.1.2 Received hematopoietic stem cell transplant: Yes ___ No ___ Unknown ___
If yes: related, matched ____ related, haploidentical ____ matched, unrelated ____ stem cell ____
If no, why? _____

7.2 Septra prophylaxis: Yes ___ No ___ Unknown ___

7.3 Regular administration of intravenous immunoglobulins (IVIG): Yes ___ No ___ Unknown ___

7.4 Regular administration of enzyme replacement (PEG-ADA): Yes ___ No ___ Unknown ___

7.5 Referred for gene therapy: Yes ___ No ___ Unknown ___
If yes, where? _____
Results: _____

SECTION 8 – OUTCOME

8.1 Child still in hospital Yes ___ No ___ Unknown ___

8.2 Child discharged home Yes ___ No ___ Unknown ___

8.3 Child died _____ Date of death: ____ /_____ /_____
DD MM YYYY
Cause of death: _____

8.4 Lost to follow-up _____

SECTION 9 – REPORTING PHYSICIAN

First name_____ Surname_____

Address_____

City_____ Province_____ Postal code_____

Telephone number_____ Fax number_____

E-mail_____ Date completed_____

Thank you for completing this form.

(SCID 2004-04)