**AUSTRALIAN PAEDIATRIC SURVEILLANCE UNIT (APSU) STUDY: PRIMARY IMMUNODEFICIENCY DISEASES (PID)**

**Abbreviation codes:**
- Autosomal recessive = AR
- X Linked = XL
- Immunoglobulin = Ig
- Purine nucleoside phosphorylase = PNP
- Adenosine deaminase = ADA
- Assoc = associated
- Def = deficiency
- Immunodef = Immunodeficiency

<table>
<thead>
<tr>
<th>PAEDIATRICIAN:</th>
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<tbody>
<tr>
<td>1. Dr. Code □□□□</td>
<td>2. Report Code ..............................................</td>
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<thead>
<tr>
<th>PATIENT:</th>
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<tbody>
<tr>
<td>3. First 2 letters of surname ..............</td>
<td>4. First 2 letters of first name ..............................................</td>
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<tr>
<td>5. Date of Birth (d, m, y) ....../...... /.......</td>
<td></td>
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<tr>
<td>6. Sex</td>
<td>□ M □ F</td>
</tr>
<tr>
<td>7. Postcode</td>
<td>□□□□</td>
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<tr>
<td>8. Child’s country of Birth:</td>
<td>□ Australia □ Other: ..............................................</td>
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<th>FAMILY:</th>
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<tbody>
<tr>
<td>9. Mother’s country of birth</td>
<td>□ Australia □ Other: ..............................................</td>
</tr>
<tr>
<td>10. If mother was born in Australia is she an</td>
<td>□ Aboriginal □ Torres Strait Islander</td>
</tr>
<tr>
<td>11. Parents 1st cousins?:</td>
<td>□ Yes □ No □ Don't know</td>
</tr>
<tr>
<td>12. Family history of PID:</td>
<td>□ Yes □ No □ Don't know</td>
</tr>
<tr>
<td>If Yes, briefly describe:</td>
<td>..........................................................................................</td>
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| 13. Have there been any unexplained deaths of male siblings? | □ Yes □ No □ Don't know |
| If yes: Age at death: ...................... | Year of death: .............................................. |

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<tr>
<th>CLINICAL DETAILS:</th>
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<tr>
<td>14. Has the patient been referred to an immunologist?</td>
<td>□ Yes □ No □ Don't know</td>
</tr>
<tr>
<td>If so please give the immunologist’s name:</td>
<td>..........................................................................................</td>
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</table>

If this patient has been referred to an immunologist whose name you have supplied there is no need to answer any further questions.

*Thank you for completing this questionnaire.*

Please return it in the reply-paid envelope to:
Dr Alyson Kakakios, Department of Immunology, The Royal Alexandra Hospital for Children, PO Box 3515, Parramatta NSW 2124

If this patient has not been referred to an immunologist please complete the following questions.
CLINICAL DETAILS (CONT):

15. Age at presentation: ..............................................

16. Age at definite diagnosis: ..............................................

16. DIAGNOSTIC CATEGORIES:

16.1 Antibody Deficiency: □ Yes □ No □ Don't know

If no go to 16.2, if yes please specify:

□ XL agammaglobulinaemia □ Sporadic
□ Common variable immunodef. □ Assoc. with thymoma □ Assoc. transcobalamin II def
□ Hyper IgM syndrome: □ XL □ AR □ Sporadic
□ Ig class deficiency □ Assoc with IgG subclass def (please specify)
□ Ig subclass deficiency
□ Ig light chain deficiency
□ Deficiency of specific antibodies (please specify) ..............................................................

16.2 T cell and combined deficiencies:
(T and B cell deficiencies)

□ Yes □ No □ Don't know

If no go to 16.3, if yes please specify:

□ DiGeorge anomaly
□ Ataxia telangiectasia
□ Other chromosomal breakage syndromes (please specify) ..................................................
□ Wiskott Aldrich syndrome
□ T cell receptor deficiency (please specify) ...........................................................................
□ ADA deficiency
□ PNP deficiency
□ XL severe combined immunodeficiency
□ Omenn’s syndrome
□ Sporadic B cell positive severe combined immunodeficiency
□ Other forms of SCID/CID (please specify) ...........................................................................
□ Def HLA expression □ def HLA class I + II expression
□ Short limbed dwarfism □ def HLA class II expression
□ Chronic mucocutaneous candidiasis
□ Other (please specify) ...........................................................................................................

16.3 Complement deficiencies □ Yes □ No □ Don't know

If no go to 16.4, if yes please specify:

□ C1q □ C1r □ C1s □ C2 □ C3 □ C4 □ C5 □ C6 □ C7 □ C8 □ C9
□ Factor B □ Factor D □ Properdin
□ C1-esterase inhibitor def □ functional □ quantitative
16.4 Phagocytic disorders

If no go to 16.5, if yes please specify:

- Chronic granulomatous disease
  - XL
  - Autosomal
  (please specify subcomponent)

- Primary neutropenia (Kostmann’s disease)
- LFA deficiency
  - LAD I
  - LAD II
- Chediak-Higashi syndrome
- Congenital asplenia
- Other (please specify)

16.5 Other

If no go to 17, if yes please specify:

- X linked lymphoproliferative syndrome
- Hyper IgE syndrome (Job’s syndrome)
- Interleukin deficiency (please specify)
- Interleukin receptor deficiency (please specify)
- Any other deficiency (please specify)

INITIAL INVESTIGATIONS:

17. Haemoglobin

18. Platelet count

19. White cell count

20. Absolute lymphocyte count

21. Lymphocyte subset analysis:
  - CD3
  - CD4
  - CD8
  - CD19

22. Lymphocyte response to mitogens:
  - specify mitogen
    - Absent
    - Low
    - Normal
    - Not done

23. Immunoglobulins:
  - IgG
  - IgG1
  - IgG2
  - IgG3
  - IgG4
  - IgA
  - IgM

24. PNP activity

25. ADA activity

26. HIV antibody:
  - Negative
  - Positive
  - Not done
MANAGEMENT:

27. Duration of care at your centre: ..........................................................................................................................

28. Is this patient receiving gammaglobulin: □ Yes □ No □ Don't know

29. **If no go to question 34**
   **if yes please specify:**
   Route of administration: □ IV □ IM □ SC
   Frequency of therapy: ................................................................................................. times/year
   Amount given on each occasion: .......................................................................................... grams
   Duration of therapy at your centre: ..........................................................................................
   Usual brand of immunoglobulin: ..........................................................................................
   Have they suffered serum sickness as a result of Ig therapy?: □ Yes □ No □ Don't know
   Have they contracted hepatitis as a result of Ig therapy?: □ Yes □ No □ Don't know
   Please specify type if known: ..........................................................................................
   Have they suffered any other complication of Ig therapy (please specify) ..........................

30. Bone marrow transplant performed: □ Yes □ No □ Don't know

31. If yes, Date(s): (1)........../......../........, (2)........../......../........, (3)........../......../...........
   Relationship of donor: ..........................................................................................
   Donor match ........................................ out of ........................................ antigens
   T-cell depleted marrow □ Yes □ No
   Other tissue (e.g., cord blood) ..........................................................................................
   Conditioning □ Yes □ No

32. Lymphokine therapy
   Gamma interferon □ Yes □ No
   Alpha interferon □ Yes □ No
   G-CSF □ Yes □ No
   GM-CSF □ Yes □ No

33. Other therapy
   C1-esterase inhibitor □ Yes □ No
   Other (please specify) ..........................................................................................

OUTCOME INFORMATION

34. □ Alive □ with disease □ disease free
    □ Died Cause of death if known: ..........................................................................................

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