Congenital Adrenal Hyperplasia Questionnaire
Australian Paediatric Surveillance Unit

Survey form A – for patients diagnosed ≤ 6 months age

PAEDIATRICIAN

1. APSU Dr Code/Name: ☐☐☐☐/........................................
2. Report code ☐☐/☐☐
3. Address:..............................................................................................................................................
4. Telephone:...........................................................................................................................................
5. Fax:......................................................................................................................................................

PATIENT DETAILS

6. Surname (first two letters only): ☐☐ 7. First name (first two letters only): ☐☐
8. Date of birth (day, month, year): ☐☐/☐☐/☐☐ 9. Sex ☐ Male ☐ Female
10. Post code of family: ☐☐☐☐
11. Ethnic origin of mother ☐ Caucasian ☐ Other (please specify)
12. Ethnic origin of father ☐ Caucasian ☐ Other (please specify)

Features at diagnosis

13. How was the diagnosis of CAH made in this child ☐ Clinical
☐ Newborn screening (trial period in NSW)
☐ Prenatal screening

14. If diagnosed prenatally: Gestation at diagnosis ☐☐ weeks
Was prenatal treatment undertaken? ☐ Yes ☐ No
If treated give details ................................................................................................................................

15. Age at diagnosis postnatally (clinical or screening) ☐☐ months ☐☐ days ☐☐ hours
16. Clinical features of presentation (please indicate which of these were present at initial presentation, more than 1 may apply):

☐ Poor feeding, failure to thrive, lethargy, or vomiting
☐ Ambiguous genitalia
☐ Virilization without genital ambiguity
☐ Hyponatremia
☐ Adrenal crisis / hypotension / shock
☐ Other (please specify)................................................................................................................................

17. Was the child treated for an alternative diagnosis prior to confirmation of CAH eg sepsis, gastrointestinal disorder, renal disorder ☐ Yes ☐ No
If yes, please specify................................................................................................................................

18. History of CAH in a sibling ☐ Yes ☐ No
19. No of affected siblings ☐☐
Laboratory features

20. Biochemical diagnosis in this patient
   □ 21-hydroxylase deficiency
   □ Other adrenal enzyme deficiency (please specify) .............................................................
   □ Not yet available

*21 – 30. Biochemistry at presentation (if performed)

21. Was the infant hypoglycemic at presentation (blood glucose < 2.6 mmol/L) □ Yes □ No □ Unknown
   Please specify blood glucose level ........... mmol/L

22. Serum sodium ................ mmol/L

23. Serum potassium ................ mmol/L

24. Serum cortisol ................ mmol/L

25. Blood pH ...................arterial / venous / capillary (please circle)

26. Plasma renin activity ................ mmol/L Lab normal range and units ......................

27. ACTH ................... pmol/L Lab normal range and units ......................

28. Serum testosterone ................ nmol/L Lab normal range and units ......................

29. 17-hydroxyprogesterone (17-OHP) (if multiple determination in the first 30 days of life, please tick

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<thead>
<tr>
<th>Date</th>
<th>17 OHP (nmol/L)</th>
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17-OHP lab normal range:............................

30. If a synacthen test was performed, please indicate steroids measured and results:

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Selected management aspects

31. Age at commencement of treatment □ □ / □ □ / □ □ (days / months / years)

32. Initial therapy
   Were intravenous fluids given? □ Yes □ No
   Mode of initial glucocorticoid therapy □ Intravenous □ Intramuscular □ Oral

33. Continuing therapy
   Glucocorticoid □ Yes □ No If Yes, please specify type ...........................................................
   Mineralcorticoid □ Yes □ No
   NaCl supplements □ Yes □ No

If more convenient, a deindentified laboratory printout of results could be provided

Please return the questionnaire in the reply-paid envelope to Dr Geoff Ambler, Institute of Endocrinology, The Children’s Hospital, Camperdown, 2050. Phone 02 692 6464, Fax 02 516 4781.

Thank you for your assistance