

Congenital Adrenal Hyperplasia Questionnaire

Australian Paediatric Surveillance Unit

Survey form A – for patients diagnosed \leq 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 cortisolmmol/L

25. Blood pHarterial / 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

Date					
17 OHP (nmol/L)					

17-OHP lab normal range:.....

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

.....
.....

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 deidentified 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