

Hyperinsulinaemic hypoglycaemia of infancy Questionnaire Australian Paediatric Surveillance Unit

Please telephone Dr Ristan Greer (07) 3365-5338 if you have wish to discuss this questionnaire.

REPORTING CLINICIAN

1. APSU Dr Code/Name /.....2. Month/Year of Report /.....
3. Date questionnaire completed / /

PATIENT

4. First 2 letters of first name 5. First 2 letters of surname
6. Date of Birth / / 7. Sex M F 8. Postcode
9. Date of diagnosis: / /
10. Country of Birth: Australia Other specify Don't know
11. Mother's country of birth Australia Other specify Don't know
12. Father's country of birth Australia Other specify Don't know
13. Is the child of Aboriginal or Torres Strait Islander origin Yes No Don't know

If this patient is primarily cared for by another physician who you believe will report the case, please complete the questionnaire details above this line and return to APSU. Please keep the patient's name and other details in your records. If no other report is received for this child we will contact you for information requested in the remainder of the questionnaire.

The primary clinician caring for this child is: **Name:**

Hospital:

Instructions: Please answer each question by ticking the appropriate box or writing your response in the space provided. DK= Don't Know, NA = Not applicable

Presentation & diagnosis

14. Date of presentation: / /
15. Place of initial presentation:
- | | | | | |
|---|------------------------------|-----------------------------|-----------------------------|--|
| a. general practitioner | Yes <input type="checkbox"/> | No <input type="checkbox"/> | DK <input type="checkbox"/> | |
| b. hospital accident and emergency | Yes <input type="checkbox"/> | No <input type="checkbox"/> | DK <input type="checkbox"/> | |
| c. maternity hospital/neonatal nursery/birth centre | Yes <input type="checkbox"/> | No <input type="checkbox"/> | DK <input type="checkbox"/> | |
| d. other hospital | Yes <input type="checkbox"/> | No <input type="checkbox"/> | DK <input type="checkbox"/> | |
| e. other, please specify | | | | |
16. Signs and symptoms at presentation:
- | | | | | | | | |
|---|------------------------------|-----------------------------|-----------------------------|------------|------------------------------|-----------------------------|-----------------------------|
| a. seizure | Yes <input type="checkbox"/> | No <input type="checkbox"/> | DK <input type="checkbox"/> | b. floppy | Yes <input type="checkbox"/> | No <input type="checkbox"/> | DK <input type="checkbox"/> |
| c. 'funny turn' | Yes <input type="checkbox"/> | No <input type="checkbox"/> | DK <input type="checkbox"/> | d. sweaty | Yes <input type="checkbox"/> | No <input type="checkbox"/> | DK <input type="checkbox"/> |
| e. staring episode | Yes <input type="checkbox"/> | No <input type="checkbox"/> | DK <input type="checkbox"/> | f. jittery | Yes <input type="checkbox"/> | No <input type="checkbox"/> | DK <input type="checkbox"/> |
| g. poor feeding | Yes <input type="checkbox"/> | No <input type="checkbox"/> | DK <input type="checkbox"/> | | | | |
| h. asymptomatic at diagnosis Yes <input type="checkbox"/> No <input type="checkbox"/> DK <input type="checkbox"/> | | | | | | | |
| i. other, please specify | | | | | | | |
17. Was the child diagnosed through:
- | | | | |
|---|------------------------------|-----------------------------|-----------------------------|
| a. routine neonatal blood sugar surveillance | Yes <input type="checkbox"/> | No <input type="checkbox"/> | DK <input type="checkbox"/> |
| b. asymptomatic but high index of suspicion e.g. Family Hx) | Yes <input type="checkbox"/> | No <input type="checkbox"/> | DK <input type="checkbox"/> |
| c. investigation of child with suggestive signs or symptoms | Yes <input type="checkbox"/> | No <input type="checkbox"/> | DK <input type="checkbox"/> |
18. What was the specific diagnosis, if available:
- | | | | |
|--|------------------------------|-----------------------------|-----------------------------|
| a. defect of SUR/Kir6.2 gene | Yes <input type="checkbox"/> | No <input type="checkbox"/> | DK <input type="checkbox"/> |
| b. defect of glutamate dehydrogenase gene (hyperammonaemic hypoglycaemia) | Yes <input type="checkbox"/> | No <input type="checkbox"/> | DK <input type="checkbox"/> |
| c. defect of glucokinase gene | Yes <input type="checkbox"/> | No <input type="checkbox"/> | DK <input type="checkbox"/> |
| d. Congenital disorder of glucosylation | Yes <input type="checkbox"/> | No <input type="checkbox"/> | DK <input type="checkbox"/> |
| e. SCHAD1 (SCHAD is a defect in the gene encoding short 3-hydroxylacyl-CoA dehydrogenase) | Yes <input type="checkbox"/> | No <input type="checkbox"/> | DK <input type="checkbox"/> |
| f. Beckwith-Weidemann syndrome | Yes <input type="checkbox"/> | No <input type="checkbox"/> | DK <input type="checkbox"/> |
| g. Other, please specify | | | |

Child & family history

19. Child's gestation at birth: weeks DK 20. Child's birth weight: grams DK
21. Child's birth length: cm DK
22. Child's delivery: normal vaginal Forceps assisted Caesarian section Other DK
23. Did the child suffer any birth trauma? Yes No DK
- IF YES, please specify
24. What is the ethnic origin of mother: DK
25. What is the ethnic origin of father: DK
26. Are the parents consanguineous? Yes No DK
27. If there is consanguinity, please specify relationship (if known)
28. Are any siblings affected? Yes No DK NA, no siblings

Management & short-term outcome

29. How were the initial episode/episodes of hypoglycaemia treated? (tick all that apply)
- a. intravenous glucose Yes No DK
 - b. frequent feeds Yes No DK
 - c. Diazoxide Yes No DK
 - d. referral to endocrinologist Yes No DK
 - e. referral to surgeon Yes No DK
 - f. other, please specify
30. Was surgical therapy (pancreatectomy) undertaken/ or is it planned? Yes No DK
31. If pancreatectomy was performed, what was the extent of resection?
- a. total (all possible tissue removed) Yes No DK
 - b. 95% (to the level of the bile duct) Yes No DK
 - c. subtotal (e.g. tail or adenoma) (specify)
 - d. date of surgery (or child's age (days or weeks) at surgery) ____/____/____ DK
32. What was the short term outcome of the HI episode/s?
- a. Any apparent ill-effects of HI episode/s? Yes No DK
 - b. Medically unstable eg. recurrent episodes hypoglycaemia? Yes No DK
 - c. Is the infant hyperglycaemic or on insulin? Yes No DK
 - d. Is the child neurologically normal? Yes No DK
 - e. Is neurological damage established? Yes No DK
 - f. death Yes No DK
 - g. other, please specify
33. a. Is the child currently alive? Yes No DK
- b. If not, what was the date of death (estimate if necessary) ? ____/____/____ DK
- c. If not, what was the cause of death? DK
34. What is the ongoing treatment?
- a. none Yes No DK
 - b. medical therapy Yes No DK
- If yes, please specify
- c. other, please specify

Please return this questionnaire in the addressed reply-paid envelope to Dr Ristan Greer, Department of Endocrinology and Diabetes, Mater Children's Hospital, South Brisbane, QLD 4101.

Thank you for your assistance with this study, which has been approved by a Human Ethics Committee. The APSU is a Unit of the Royal Australasian College of Physicians (Division of Paediatrics and Child Health) and is funded by the NHMCR (Enabling Grant No. 40284), the Department of Health and Ageing, and the Faculty of Medicine at the University of Sydney.