

# Congenital Adrenal Hyperplasia (CAH)

## Case notification form

Please report any child from birth up to (but not including) 16 years of age with suspected or confirmed CAH seen for the first time in the last month. If diagnosis is awaiting confirmation, the child should still be reported.

### Case Definition:

A child will be considered to have a diagnosis of CAH if one or more of the criteria are met:

- Adrenal crisis (and/or salt wasting crisis)
  - Adrenal insufficiency (non-life threatening)
  - Virilisation of female genitalia
  - Incomplete masculinisation of male genitalia
  - Precocious puberty
  - Accelerated skeletal age
  - Short stature
  - Hypertension
  - Family History
- AND  
one or more of  
the following:
- Elevated 17-OHP in blood test
  - Positive synacthen stimulation test
  - Test diagnostic of rarer form of CAH (eg. 3 $\beta$  - HSD deficiency)

BPSU ID:   /     /

Date completed:   /   /

Consultant Name: \_\_\_\_\_

## Section 1: Child's details

- 1.1 Date of birth**   /   /
- 1.2 Initials**
- 1.3 Hospital Ref. No** \_\_\_\_\_
- 1.4 Home Post Code at Birth (first half only)**
- 1.5 Ethnic Origin** White  Asian/Asian British  Black/Black British   
Mixed  Other  *please specify:* \_\_\_\_\_ Not Known
- 1.6 Has there been difficulty with sex assignment?** Yes  No
- 1.7 If No: What sex is the child?**
- 1.8 If Yes: What sex was the child assumed to be at birth?**    
**What is the chromosomal sex?**    
**What sex has finally been assigned to the child?** Not Decided



## Section 2: Pregnancy/birth details (for infants)

2.1 Birth weight (grams) \_\_\_\_\_ Not Known

2.2 Was the child (circle your choice): Term / Preterm (less than 37 completed weeks)

If preterm, gestation \_\_\_\_\_ weeks + \_\_\_\_\_ days or Not Known

Was diagnosis made prenatally? Yes  No  Not Known

If so, when? Gestation \_\_\_\_\_ weeks + \_\_\_\_\_ days or Not Known

Was prenatal treatment given? Yes  No  Not Known

If yes, date   /   /   Treatment Details \_\_\_\_\_

## For children over 1 year of age

2.3 Most recent measured: weight (kgs) \_\_\_\_\_, height (cm) \_\_\_\_\_ on   /   /

## Section 3: Family history

3.1 Is/are there similarly affected sibling(s)? Yes  No  Not Known

If yes, details \_\_\_\_\_

3.2 Is/are there similarly affected relative(s)? Yes  No  Not Known

If yes, details \_\_\_\_\_

3.3 Have there been any unexplained neonatal deaths? Yes  No  Not Known

If yes, details \_\_\_\_\_

## Section 4: Mode of presentation

4.1 When CAH was first suspected:

What was the date?   /   /

What was the reason? Affected siblings/family history  Clinical symptoms

Other (please give details): \_\_\_\_\_

When was the diagnosis confirmed?   /   /

4.2 Have there been any previous episodes of illness that can be attributed to unrecognised CAH? Yes  No  Not Known

4.3 Which of the following symptoms were present at the time of diagnosis (or suspected diagnosis)? Please tick Yes/No/Not Known.

	Yes	No	NK	Date
Adrenal insufficiency (non-life threatening)	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="text"/> <input type="text"/> / <input type="text"/> <input type="text"/> / <input type="text"/> <input type="text"/>
Adrenal crisis (and/or salt wasting)	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="text"/> <input type="text"/> / <input type="text"/> <input type="text"/> / <input type="text"/> <input type="text"/>
Precocious puberty	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="text"/> <input type="text"/> / <input type="text"/> <input type="text"/> / <input type="text"/> <input type="text"/>
Advanced bone age	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="text"/> <input type="text"/> / <input type="text"/> <input type="text"/> / <input type="text"/> <input type="text"/>
Accelerated growth	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="text"/> <input type="text"/> / <input type="text"/> <input type="text"/> / <input type="text"/> <input type="text"/>
Hypertension	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="text"/> <input type="text"/> / <input type="text"/> <input type="text"/> / <input type="text"/> <input type="text"/>
Learning disability	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="text"/> <input type="text"/> / <input type="text"/> <input type="text"/> / <input type="text"/> <input type="text"/>

## Section 4: Mode of presentation continued

**4.5 Please answer the following as applicable for girls (\*see opposite):**

Virilisation of female genitalia

Yes  No

If yes, Prader score\*:

Date   /   /

**for boys:**

Incomplete masculinisation of male genitalia

Yes  No

Date   /   /

## Section 5: Initial Management

**5.1 At the first clinical episode (diagnosis) was the child:**

Referred (or admitted) for investigations? Yes  No  Not Known

Admitted (or transferred from maternity unit) to hospital for management of complications of CAH? Yes  No  Not Known

Admitted (or transferred from maternity unit) to intensive care unit for management of complications of CAH? Yes  No  Not Known

Details \_\_\_\_\_  
\_\_\_\_\_

**5.2 Treatment: Tick to indicate which of the following treatments were administered at first clinical episode (diagnosis), and please give brief details.**

Glucocorticoids: Hydrocortisone  Prednisolone  Cortisone acetate  Other

Details \_\_\_\_\_  
\_\_\_\_\_

Mineralocorticoids: Fludrocortisone  Other

Details \_\_\_\_\_  
\_\_\_\_\_

Genital Surgery: Yes  No

Date   /   /

Details \_\_\_\_\_  
\_\_\_\_\_

Other Treatments: Yes  No

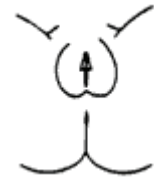
Details \_\_\_\_\_  
\_\_\_\_\_

Was the child referred to any other specialists?

Counselling  Psychiatrist  Psychologist  Geneticist  Other specialists

## Prader staging system for the degree of virilisation of the external genitalia

**Prader 0: Normal female external genitalia.**



**Prader 1: Female external genitalia with clitoromegaly.**

The clitoris (phallus) is enlarged



**Prader 2: Clitoromegaly with partial labial fusion forming a funnel-shaped urogenital sinus.**

The clitoris (phallus) is enlarged; the labia are partly fused; the vagina and urethra open into a funnel-shaped area



**Prader 3: Increased phallic enlargement. Complete labioscrotal fusion forming a urogenital sinus with a single opening.**

Increased enlargement of phallus; the labia are completely fused; the vagina and urethra share a single opening



**Prader 4: Complete scrotal fusion with urogenital opening at the base or on the shaft of the phallus.**

A large phallus; the labia are completely fused, like a scrotum; the vagina and urethra share a single opening near the base of the phallus



**Prader 5: Normal male external genitalia.**

The external genitals look like a typical boy's; the vagina and urethra share a single opening at the tip of the phallus.



Prader A (1954) Staging of ambiguous genitalia in patients with congenital adrenal hyperplasia. *Helv Paediatr Acta* 9: 231-248

## Section 6: Initial Outcome

- 6.1** Child admitted and currently in hospital Yes  No   
 Child admitted and discharged home on   /   /    
 Child never admitted (ie. remained at home) Yes  No
- 6.2** Child died Yes  No  If yes, date of death   /   /
- 6.3** If the child is alive, when was s/he last seen?   /   /    
 Were there obvious sequelae at that time? Yes  No  Too soon to tell   
 If yes, please give brief details \_\_\_\_\_

## Section 7: Diagnostic Test Results

- 7.1** 17-OHP serum/plasma  
 Result (nmol/l): \_\_\_\_\_ Date test taken   /   /    
 or Result Pending  Not Known  Not Done   
 Laboratory where analysis performed: \_\_\_\_\_
- 7.2** Synacthen stimulation test  
 Abnormal  Normal  Not Known  Not Done  Date test taken   /   /
- 7.3** Urine steroid profile  
 Result: \_\_\_\_\_ Date test taken   /   /    
 Not Known  Not Done
- 7.4** Was DNA mutation analysis undertaken? Yes  No   
 If yes, what mutations were found?  
 Allele 1 \_\_\_\_\_ or Normal  Result Pending  Not Known   
 Allele 2 \_\_\_\_\_ or Normal  Result Pending  Not Known   
 Genotype: Homozygote  Compound Heterozygote  Simple Heterozygote
- 7.5** Other tests (list): \_\_\_\_\_  
 Please give details: \_\_\_\_\_

## Section 8: Your Details

Your name (please print): \_\_\_\_\_  
 Address: \_\_\_\_\_  
 Telephone: \_\_\_\_\_ E-mail: \_\_\_\_\_  
 Date:   /   /