



# Diagnosis of CAH in infancy and management in a new nurse led adrenal clinic

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# Introduction

- Diagnosis – boys and girls
- Management in a tertiary referral centre
- Biochemical investigations
- Holistic management
  - Discharge
  - Sick day and emergency management
- Adrenal nurse led clinic
  - Short and long term support
- Professional issues

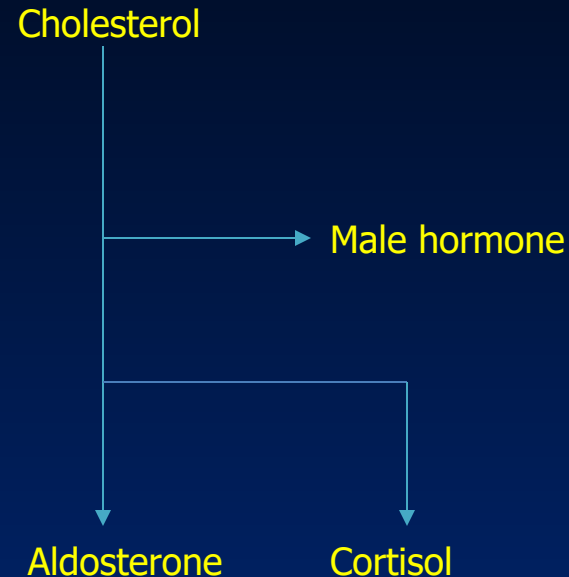
# What is CAH?

- An adrenal enzyme defect
- Classical 21-hydroxylase deficiency is the most common
  - 1 in 15,000 births in the UK
- Results in glucocorticoid and mineralocorticoid deficiency
  - ↑ ACTH secretion by the anterior pituitary
  - Accumulation of steroid precursors prior to the enzyme defect
  - ↑ androgens production

# Normal steroid production

- Steroids made in the adrenal cortex
- Made from cholesterol:
  - Cortisol
  - Aldosterone (salt retaining steroid)
  - Androgen (male hormone)

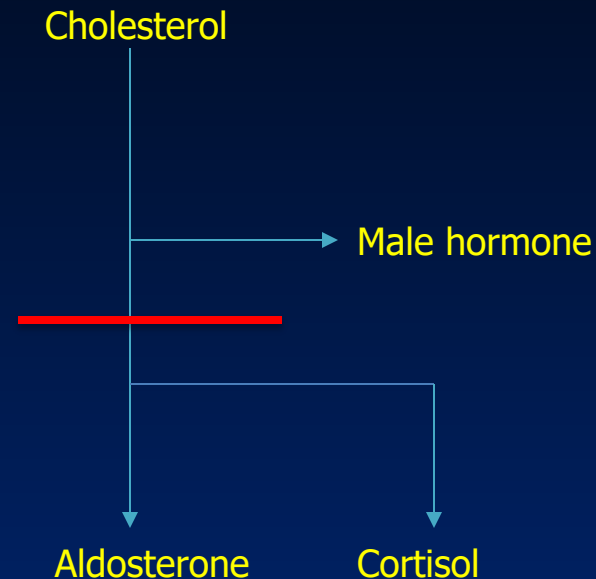
## Steroid Pathway



# ↑ ACTH production in CAH

- Causes the adrenal gland to go 'hyperplastic' (increase in amount of tissue)
- Deficiencies in aldosterone and cortisol

## Steroid Pathway



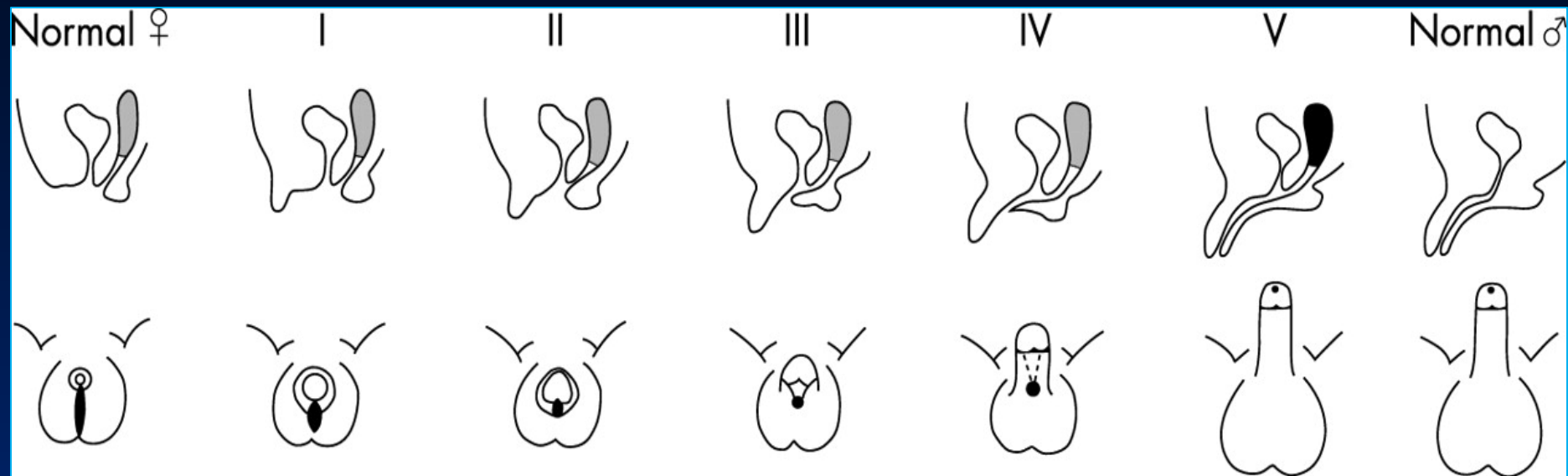
# Diagnosis in Boys

- Can have hyperpigmented scrotum and genitalia at birth, but usually look 'normal'
- Presentation
  - Day 5
  - Second week of life
    - Poor feeding, weight loss, failure to thrive
  - If CAH not recognised
    - Salt losing crisis
      - Due to the aldosterone loss

# Diagnosis in Girls

- Genitalia are usually virilized due to excess testosterone
  - Allows earlier diagnosis
- Mild clitoromegaly to full masculinisation
  - Prader staging
- DSD service

# Prader staging in girls



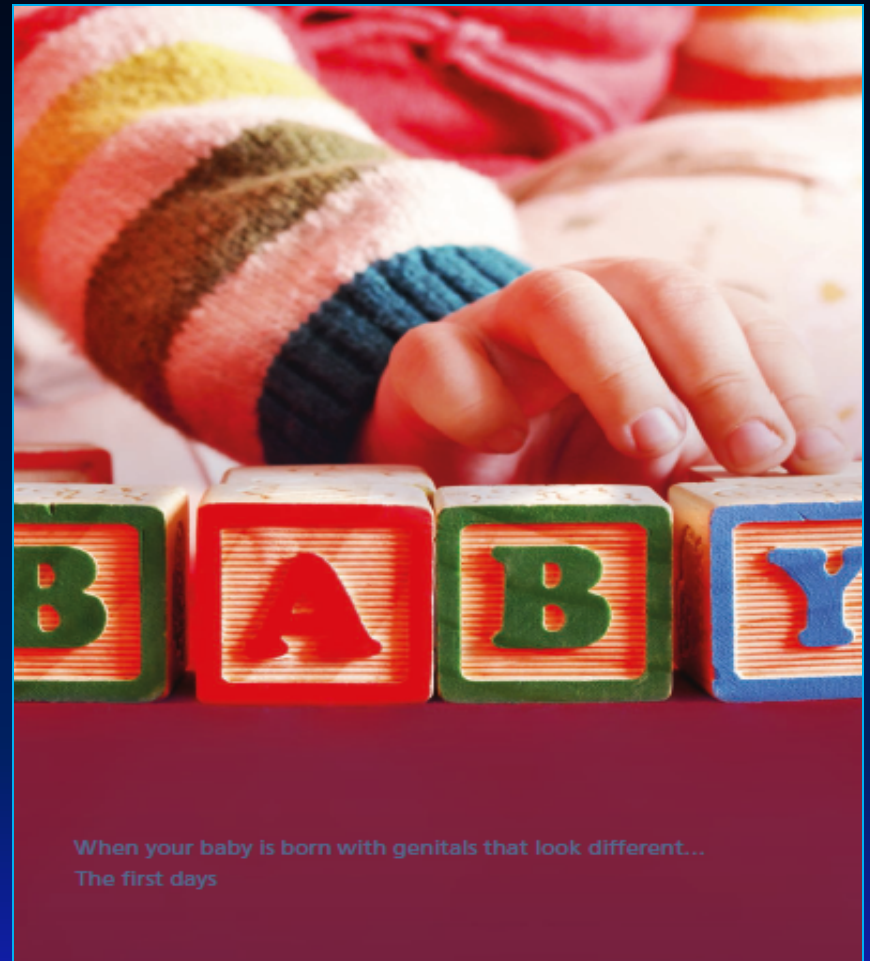


# Management at GOSH - Boys

- Referred from local for management of salt losing crisis
- Biochemical investigations for confirmation of diagnosis
- Support and education for family
- Enter into shared care

# Management at GOSH - Girls

- Usually brought in to be reviewed by the DSD team at GOSH within first week of life
  - Endocrinologist
  - Urologist
  - Psychologist
  - CNS
  - Gynaecologist



# Diagnosis

- Confirmed by a raised 17OHP level after day 3 of life
- Salt wasting confirmed by:
  - Low plasma sodium
  - High potassium
  - Increased urinary sodium excretion
  - Virilised girls
    - Chromosome analysis
    - Pelvic ultrasound

# Biochemical investigations

- Short synacthen test

Time	Cortisol	17-OHP	11-DOC	A4	ACTH	Renin
0	✓	✓	✓	✓	✓	✓
30	✓	✓	✓	✓		
60	✓	✓	✓	✓		

- Synacthen given IM or IV

- 0 – 6 months: 62.5mg

- Urine

- Steroid analysis to confirm the 21-hydroxylase deficiency defect

# Medical management

- Hydrocortisone 10mg tablets
  - 10 – 15 mg/m<sup>2</sup>/day
  - Total dose spread 3 – 4 times throughout the day
- Fludrocortisone 100 mcg tablets
  - 150 mcg / m<sup>2</sup>/ day
- Salt supplements
  - Oral salt supplements (until one year of age) in the 5mmol/ml 30% Sodium Chloride solution – 5mmols/kg/day, in 4 divided doses = .....mls per dose four times a day
  - Can stop when fully weaned

# Holistic management

Education

Resources

Safety at home

# Why tablets?

- Hydrocortisone suspension not bioequivalent to Hydrocortisone tablets (Merke, 2001)
- Instructions given:
  - Cutting and crushing tablets



# Great Ormond Street Hospital for Children



NHS Trust

Gastroenterology, Endocrinology, Metabolic & Adolescent Medicine (GEMA)

Great Ormond Street  
London WC1N 3JH

Tel: 020 7405 9200  
Direct Line: 0207-813-8214

## How to give Hydrocortisone 1.25 mg using 10 mg tablets

### Hydrocortisone

Dose: Hydrocortisone 1.25 milligrams (mg) three times a day (morning, early afternoon and evening approx every 8 hours)

1. Take 1 x 10mg hydrocortisone tablet and cut into  $\frac{1}{4}$ 's using a tablet cutter
2. Crush  $\frac{1}{4}$  of a tablet (2.5mg) using tablet crusher
3. Draw up 2mls of cooled, boiled water into a 2ml syringe
4. Mix the crushed  $\frac{1}{4}$  of a tablet with the 2mls of cooled boiled water
5. Then draw up 1ml of the mixture to give 1.25mg of hydrocortisone
6. Give by mouth as shown by ward nurses

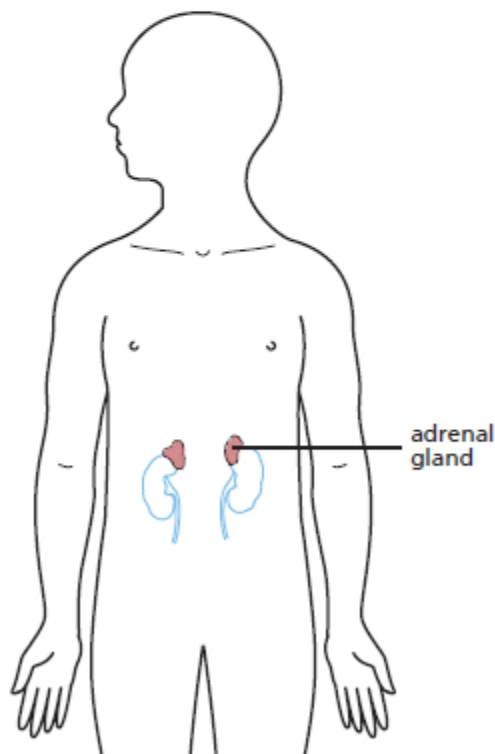




Great Ormond Street Hospital for Children NHS Foundation Trust: Information for Families

## Congenital adrenal hyperplasia (CAH)

This information sheet from Great Ormond Street Hospital (GOSH) explains about the medical condition congenital adrenal hyperplasia (CAH) and what to expect when your child comes to GOSH for assessment and treatment.



Congenital adrenal hyperplasia is a group of inherited conditions that are present at birth (congenital) where the adrenal gland is larger than usual (hyperplasia). In CAH, the body is missing an enzyme (chemical substance) that stimulates the adrenal glands to release the cortisol hormone. Lacking this hormone means that the body is less able to cope with stress, either emotionally or physically, which can be life threatening. It also makes the level of androgen (male hormone) increase, which causes male characteristics to appear early in boys or inappropriately in girls.

The adrenal glands rest on the tops of the kidneys. They are part of the endocrine system, which organises the release of hormones within the body. Hormones are chemical messengers that switch on and off processes within the body.

The adrenal glands consist of two parts:

- the medulla (inner section) which makes the hormone 'adrenaline' which is part of the 'fight or flight' response a person has when stressed. This is not usually affected in CAH.

# GP letter on discharge

## GP details

Date

Dear Dr

RE:

Diagnosis: Congenital Adrenal Hyperplasia

..... was referred to us on ..... from ..... Hospital, and a diagnosis of Congenital Adrenal Hyperplasia has been made, and s/he has cortisol deficiency.

She/he has been commenced on the following medication and we would be very grateful if you could commence a repeat prescription for:

Hydrocortisone ..... mg (as Hydrocortisone 10mg tablets, NOT suspension)

Fludrocortisone ..... mcg daily

Oral salt supplements (until one year of age) in the 5mmol/ml 30% Sodium Chloride solution – 5mmols/kg/day, in 4 divided doses = .....mls per dose four times a day

Also to be included on his/her prescription:

Hydrocortisone Emergency Pack to be renewed yearly:

Efcortisol 100mg vials, 25/50/100mg to be given IM in an emergency.


Glucose Gel 25g tube, 1/3 tube orally to be given in an emergency.

We have given .....’s parents a tablet cutter and a tablet crusher, and have educated them in how to prepare and administer their medication. ....’s parents have had education in his/her management during times of illness and they have been trained in giving IM hydrocortisone, 25/50/100mg, should the need arise, along with oral glucose gel. A steroid card has been given, and also information about Medic Alert jewellery to start wearing as soon as possible.

We have arranged with the local hospital and the local Paediatrician (.....) to have fast track access should he/she require emergency IM hydrocortisone. We have also set up an arrangement with .....Ambulance Service to ensure a red alert system is in place.

- Medication onto repeat prescription system
- Tablets not suspension
- Also highlights that the baby can have all of their usual childhood immunisations

# Sick day and emergency management



Great Ormond Street Hospital for Children NHS Foundation Trust: Information for Families

## Cortisol deficiency and steroid replacement therapy

This leaflet explains about cortisol deficiency and how it is treated. It also contains information about how to deal with illnesses, accidents and other stressful events in children on cortisol replacement.

### Where are the adrenal glands and what do they do?

The adrenal glands rest on the tops of the kidneys. They are part of the endocrine system, which organises the release of hormones within the body. Hormones are chemical messengers that switch on and off processes within the body.

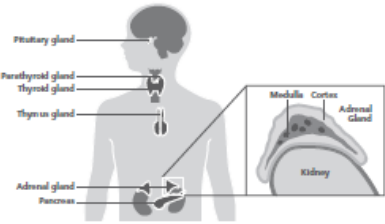
The adrenal glands consist of two parts:

- the medulla (inner section) which makes the hormone 'adrenaline' which is part of the 'fight or flight' response a person has when stressed.
- the cortex (outer section) which releases several hormones.

The two most important ones are:

- **Aldosterone** – this helps regulate the blood pressure by controlling how much salt is retained in the body. If a person is unable to make aldosterone themselves, they will need to take a tablet called 'fludrocortisone'.
- **Cortisol** – this is the body's natural steroid and has three main functions:
  - helping to control the blood sugar level
  - helping the body deal with stress
  - helping to control blood pressure and blood circulation.

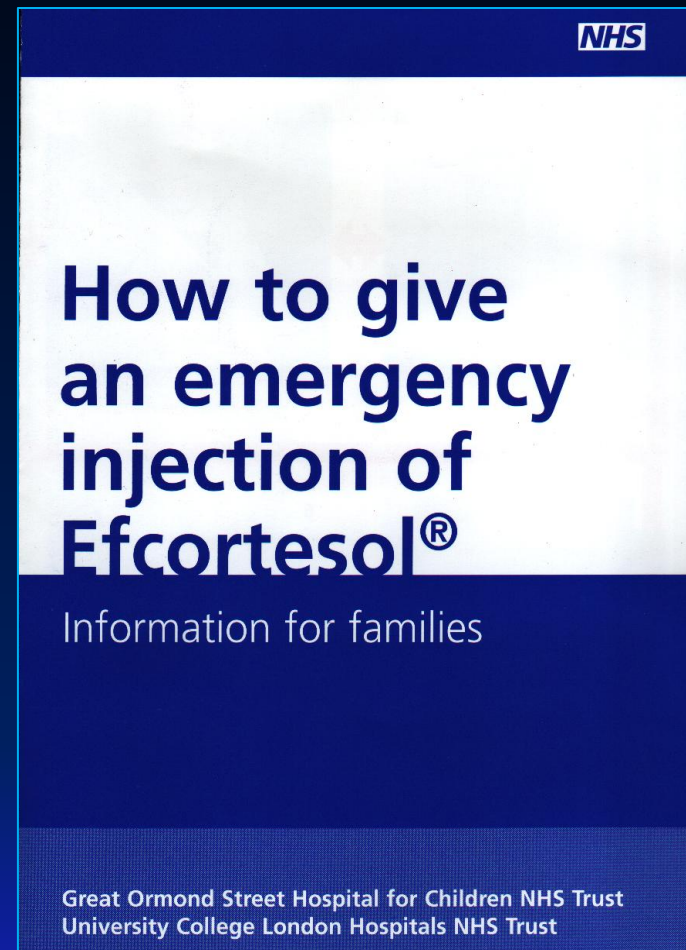
If a person is unable to make cortisol themselves, they will need to take a tablet to replace it. The most common form used is hydrocortisone, but other forms may be prescribed.



- Doubling up on hydrocortisone when unwell
- Additional 4am dose (same as morning dose)

# Sick day and emergency management

- Emergency injection of hydrocortisone and oral glucogel
- Liaise with nurseries
  - Schools when older
- Medic alert jewellery
- Usually dispense x2 emergency packs
  - Home
  - Bag
    - Another when older for nursery / school



# Emergency services



London Ambulance Service NHS Trust

Patient Specific Protocol  
PSP Paediatric Steroid Dependent Crisis

PSP

This protocol has been specifically prepared for **STERIOD DEPENDENT CRISIS** patients and details the treatment to be given in specified circumstances.

Patient's Name:

Date of Birth:

NHS Number:

Address:

School:-

Local hospital:

Reason for protocol: Administration of IM hydrocortisone in possible adrenal crisis

Specific Treatment / Instructions: Patient may have an adrenal crisis if IM hydrocortisone not administered in an emergency situation

In the event that this child is involved in an accident or develops diarrhoea or vomiting and presents with any symptoms of a steroid dependent crisis whilst at **Home** or at **School** they are to be administered IM hydrocortisone as detailed over leaf.

**Note:-** The IM hydrocortisone (Efcorlesol) is kept both by the parents and by the school in an emergency pack.

Please transport this child to the above local hospital if possible, otherwise to the nearest paediatric A&E unit.

**All other aspects of clinical care remain unchanged.**

**For further advice if necessary please contact the Endocrine Registrar on call via switchboard at Great Ormond Street Hospital on 020 7405 9200**

1. Efcorlesol 1ml ampoule (Hydrocortisone 100mg/ml - as sodium phosphate)

Dose: Age 0-1 years 25 mg IM  
Age 1-5 years 50mg IM  
Age 5+ years 100mg IM

2. Please also administer Glucogel (Hypostop) 25 gram tube, required dose in an emergency - up to 1/3 tube if not already previously administered by carers.

Following administration of the hydrocortisone remove to hospital with full monitoring and oxygen therapy as required.

**All other aspects of clinical care remain unchanged.**

**If required contact EOC and ask for the Clinical Support Desk**

**PTO for further general info on Steroid Dependent Crisis**

- Contact details for all UK ambulance services
- Red flag system



London Ambulance Service NHS Trust

Patient Specific Protocol  
PSP Paediatric Steroid Dependent Crisis

The symptoms of a Steroid Dependent Crisis

- Weakness
- Mental confusion
- Drowsiness, in advanced cases slipping towards a coma
- Dizziness
- Nausea and/or vomiting
- Headache
- Abnormal heart rate - either too fast or too slow
- Abnormally low blood pressure
- Possibly a fever
- Abdominal tenderness

The causes of a Steroid Dependent Crisis

- Physical shock, e.g. a car accident
- Infection, e.g. flu with a high temperature
- Dehydration, e.g. stomach bug with vomiting

**All other aspects of clinical care remain unchanged.**

**NOT SUITABLE FOR LAS CLINICAL TELEPHONE ADVICE**

**If required contact EOC and ask for the Clinical Support Desk**

Fiona Moore FRCS, FCEM, FIMC RCS Ed  
Medical Director  
London Ambulance Service NHS Trust

Issue Date:

# Emergency hospital letter

Great Ormond Street   
Hospital for Children  
NHS Trust

Great Ormond Street  
London WC1N 3JH

Tel: 020 7405 9200

Gastroenterology, Endocrinology, Metabolic & Adolescent Medicine (GEMA)  
Direct Line: 0207-813-8214

Re:

Diagnosis:

Medications: Hydrocortisone (oral)

Fludrocortisone (oral)

NaCl supplements 5mmol/ml 30% solution:

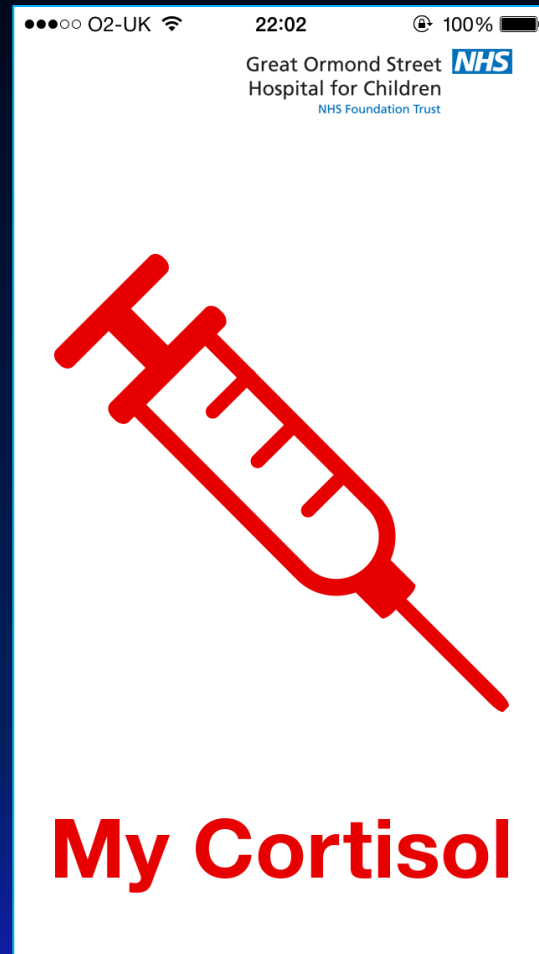
Instructions for Hospital Doctor

In view of this patient's cortisol deficiency, if this patient is brought to hospital as an emergency, the following management is advised:

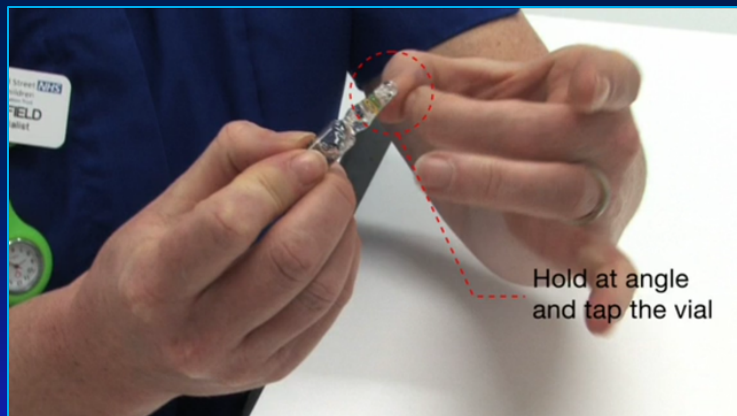
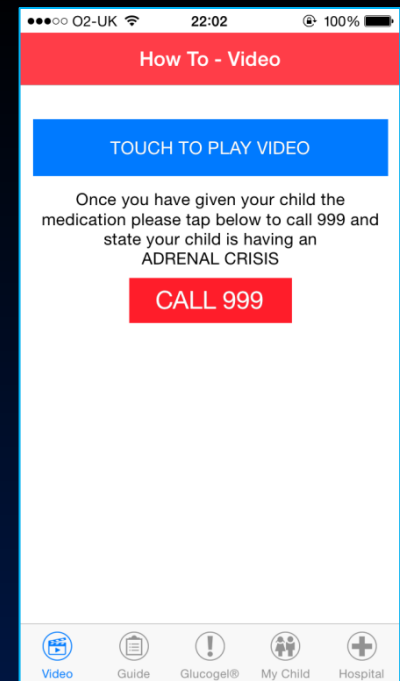
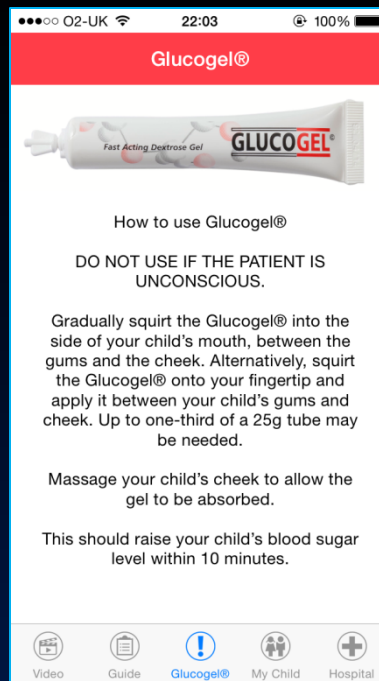
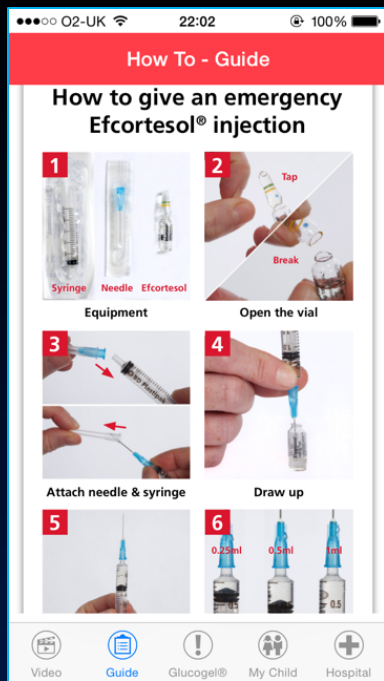
- If patient drowsy and unresponsive give IM hydrocortisone in the following doses immediately (0-1yr – 25mgs; 1-5 yrs – 50mgs; > 5yrs – 100mgs) if patient has not already had IM hydrocortisone administered by ambulance crew or parents.
- Take blood for U&Es, glucose and osmolality
- If blood glucose is < 2.5 mmol, give bolus of 2mg/kg 10% dextrose
- If patient is drowsy, hypotensive and peripherally shut down, give 20ml/kg of normal saline, insert an IV cannula and then continue with usual dextrose saline infusion
- Continue with bolus IV hydrocortisone at 2mg/kg every 4 hours until patient is tolerating oral fluids and then swap to double usual oral Hydrocortisone doses until patient fully recovered and back to normal self (usually 2-3 days on double usual hydrocortisone doses).
- **Important:** Please admit for a minimum of 12 hours

If there is any doubt about this patient's management, advice can be obtained via Great Ormond Street Hospital switchboard (0207 405 9200, asking for the Endocrine Registrar on Call).

# My Cortisol App









# Steroid card

## Instructions for Hospital Doctor

Dear Doctor,

If this patient is brought to hospital as an emergency the following management is advised:

- 1) Insert an IV cannula
- 2) Take blood for U&Es, glucose, and perform any other appropriate tests (e.g. urine culture)
- 3) Check capillary blood glucose level
- 4) Give 100 mg hydrocortisone intravenously as bolus (unnecessary if patient has already been given IM hydrocortisone)
- 5) Commence IV infusion of 0.45% sodium chloride and 5% glucose at maintenance rate (extra if patient is dehydrated). Add potassium depending on electrolyte
- 6) Commence hydrocortisone infusion (50 mg hydrocortisone in 50ml 0.9% sodium chloride via syringe pump)
- 7) Monitor for at least twelve hours before discharge

**IMPORTANT!** If blood glucose is  $< 2.5$  mmol/l, give bolus of 2 ml/kg of 10% glucose

If patient is drowsy, hypotensive and peripherally shut down with poor capillary return give 20ml/kg of 0.9% sodium chloride stat.

**If in any doubt about this patient's management, please contact the urgent advice numbers**

## Useful Contact Numbers:

**GOSH Switchboard**

Tel: 020 7405 9200

**For Urgent Advice:**

Tel: 020 7405 9200 and ask to be put through to the endocrine registrar on call

**University College  
Hospital Switchboard**

Tel: 0845 155 5000

**For Urgent Advice:**

Tel: 0845 155 5000 and ask to be put through to the endocrine registrar on call.

Great Ormond Street Hospital for Children NHS Trust and  
University College London Hospitals NHS Foundation Trust



## CORTISOL DEFICIENCY

**THE OWNER OF THIS CARD IS ON  
CORTISOL REPLACEMENT THERAPY**

Name \_\_\_\_\_

Address \_\_\_\_\_

Tel \_\_\_\_\_

Mobile: \_\_\_\_\_

Date of Birth \_\_\_\_/\_\_\_\_/\_\_\_\_

Hospital No \_\_\_\_\_

Consultant \_\_\_\_\_

Hospital \_\_\_\_\_

Address \_\_\_\_\_

Tel \_\_\_\_\_ Fax \_\_\_\_\_

General Practitioner \_\_\_\_\_

Address \_\_\_\_\_

Tel \_\_\_\_\_ Fax \_\_\_\_\_

Affix  
photo  
here

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# Primary Care

- Open access onto local paediatric ward
- Contact details for all UK childrens' community nursing teams
- Medical team to liaise with local medical team to arrange formal shared care plan

Great Ormond Street **NHS**  
Hospital for Children  
NHS Trust

Great Ormond Street  
London WC1N 3JH

Tel: 020 7405 9200

Gastroenterology, Endocrinology, Metabolic & Adolescent Medicine (GEMA)  
Direct Line: 0207-813-8214

Date:  
Reference:

Dr  
Paediatric Consultant

Dear Dr

RE:

..... is a .... year old ..... under the care of ..... at Great Ormond Street Hospital. He is a boy/girl with ....., he/she was referred with ..... and we have since found he also has cortisol deficiency.

He/She has been commenced on Hydrocortisone at a dose of 2.5mg mane, 2.5mg at lunchtime, and 2.5mg nocte. ....'s mum has had education in his/her management during times of illness and has been trained in giving IM hydrocortisone should the need arise.

I would be extremely grateful if you could arrange for ..... to have fast track access at the ..... should he/she require emergency IM hydrocortisone. Please let us know on the number below.

Please do not hesitate to contact me should you require more information on 0207 813 8214.

Many thanks,

Yours sincerely

.....  
Clinical Nurse Specialist

# Primary care – blood levels<sup>P</sup>

## Cortisol bloods plan for new CAH baby

Discharge following birth

Week 1

Week 2

Week 3

Week 4

Week 6

Week 8

Week 10

Week 12 / Month 3

Month 4

Month 5

Month 6

Month 7

Month 8

Month 9

Month 10

Month 11

Month 12

On discharge following birth: weekly bloods for 4 weeks.

2 weekly for the next 8 weeks.

4 weekly until fully weaned.

When weaned (around age 1yr) - check 4 weeks later, and then at annual reviews in clinic / 6 monthly.

- Liaise with local teams for community nurses to visit family and take regular bloods for U&E
- Ensure results are fed back to GOSH

# Adrenal nurse led clinic

- First appointment
  - One month after diagnosis / discharge from GOSH
  - Discuss
    - Compliance
    - Management of medication
    - Re-educate sick day and emergency management
      - Teach injection technique
  - Follow up on any queries the family have
  - Liaise with Urology if female
  - Liaise with local teams for recent blood results
  - Discuss patient support groups

# Adrenal nurse led clinic

- Subsequent appointments
  - Alternate clinic appointments with medical consultations
  - Compliance issues
  - Re-education
  - Any prescription problems
    - Hydrocortisone tablets
    - Salt supplements
    - Emergency hydrocortisone
  - Arranging annual reviews in readiness for medical appointment
    - 24 hr profile
    - Bone age
    - Clinical examination for under / over dosing of hydrocortisone
  - Transition discussions
    - → Transfer to UCLH for adolescence

# Nurse Led Clinics

- Managerial support
  - Increase revenue into the Trust
  - Room
  - Admin
- Team support
  - Smooth liaison between medical and nursing personnel
    - Consultants and junior Doctors

# Professional Issues

- Non medical prescribing course
  - Case study – Infant with CAH
  - Bioequivalence of hydrocortisone tablets and suspension (Merke, 2001)
- Led on to further modules:
  - Advanced assessment of the presenting child
    - Children's Advanced Nurse Practitioner

# Conclusion

- Diagnosis of CAH
  - Complexity of management
    - Boys – salt losing crisis
    - Girls – Ambiguous genitalia
- Medical management
- Holistic and practical management
  - Administration of medication
  - Sick day and emergency management
- Adrenal nurse led clinic
  - Ongoing support until adolescence



# Thank you

