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)
↑ ACTH production in CAH

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

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<th>11-DOC</th>
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• **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/m2/day
  – Total dose spread 3 – 4 times throughout the day

• Fludrocortisone 100 mcg tablets
  – 150 mcg / m2/ 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
Why tablets?

- Hydrocortisone suspension not bioequivalent to Hydrocortisone tablets (Merke, 2001)
- Instructions given:
  - Cutting and crushing tablets
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 ¼'s using a tablet cutter

2. Crush ¼ of a tablet (2.5mg) using tablet crusher

3. Draw up 2mls of cooled, boiled water into a 2ml syringe

4. Mix the crushed ¼ 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
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 – 5mms/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:
Efocortesol 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

- 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

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

London Ambulance Service NHS Trust
Patient Specific Protocol
PSP Paediatric Steroid Dependent Crisis

Patient’s Name:
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 (Effertol) 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. Effertol 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 (Hypogly) 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

London Ambulance Service NHS Trust
Patient Specific Protocol
PSP Paediatric Steroid Dependent Crisis

The symptoms of a Steroid Dependent Crisis
- Weakness
- Mental confusion
- Coldness, in advanced cases slipping towards a coma
- Diarrhoea
- Nausea and vomiting
- Headaches
- Abnormal heart rate - either too fast or too slow
- Abnormally low blood pressure
- Possibly a fever
- Abdominal tenderness

The cause of a Steroid Dependent Crisis
- Physical shock, e.g. 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

Fienna Moore FRCS, FCSEM, FIMC RCS Ed
Medical Director
London Ambulance Service NHS Trust

Issue Date:
Great Ormond Street Hospital for Children
NHS Trust

Re:
Diagnosis: 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; > 5 yrs – 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
How to give an emergency Efctesol® injection

1. Equipment
2. Break the vial
3. Attach needle & syringe
4. Draw up

How to use Glucogel®

DO NOT USE IF THE PATIENT IS UNCONSCIOUS.

Gradually squirt the Glucogel® into the side of your child’s mouth, between the gums and the cheek. Alternatively, squirt the Glucogel® onto your fingertip and apply it between your child’s gums and cheek. Up to one-third of a 25g tube may be needed.

Massage your child’s cheek to allow the gel to be absorbed.

This should raise your child’s blood sugar level within 10 minutes.

Hold at angle and tap the vial

Attach needle and syringe

Once you have given your child the medication please tap below to call 999 and state your child is having an ADRENAL CRISIS

CALL 999
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.

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.

CORTISOL DEFICIENCY

THE OWNER OF THIS CARD IS ON CORTISOL REPLACEMENT THERAPY

Name __________________________
Address _________________________
Date of Birth / / __________________
Hospital No _______________________
Consultant _______________________
Hospital __________________________
Address __________________________
Tel __________________ Fax _________
General Practitioner ________________
Address __________________________
Tel __________________ Fax _________

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

NHS
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
Primary care – blood levels

- 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