

**Working Group for Endocrine Nurses
ESPE 2019**

WHAT'S NEW IN CAH?

**Leuan Hughes, University of Cambridge
and**

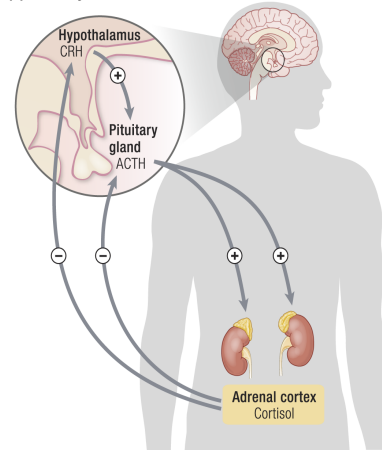
Kate Davies, London South Bank University

OUTLINE OF PRESENTATION

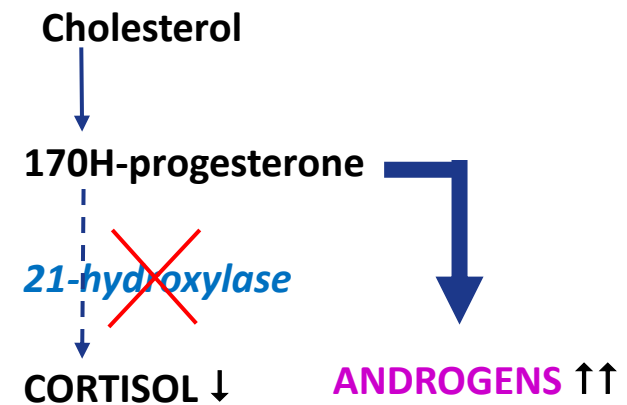
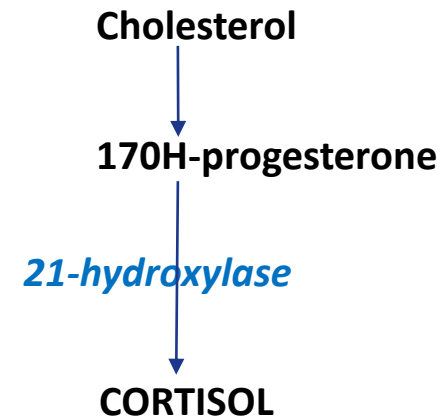
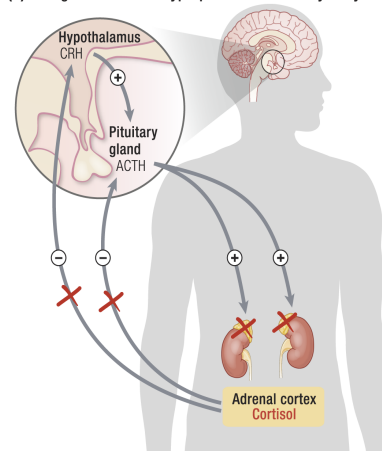
- **Pathophysiology of CAH**
 - **Medical Aspects of Management**
 - **Surgical Aspects**
 - **Outcomes**
 - **Key Role of Endocrine Specialist Nurse**
- * No conflicts of interest to declare**

PATHOPHYSIOLOGY OF CAH

(a) Healthy individual



(b) Congenital adrenal hyperplasia due to 21-hydroxylase deficiency

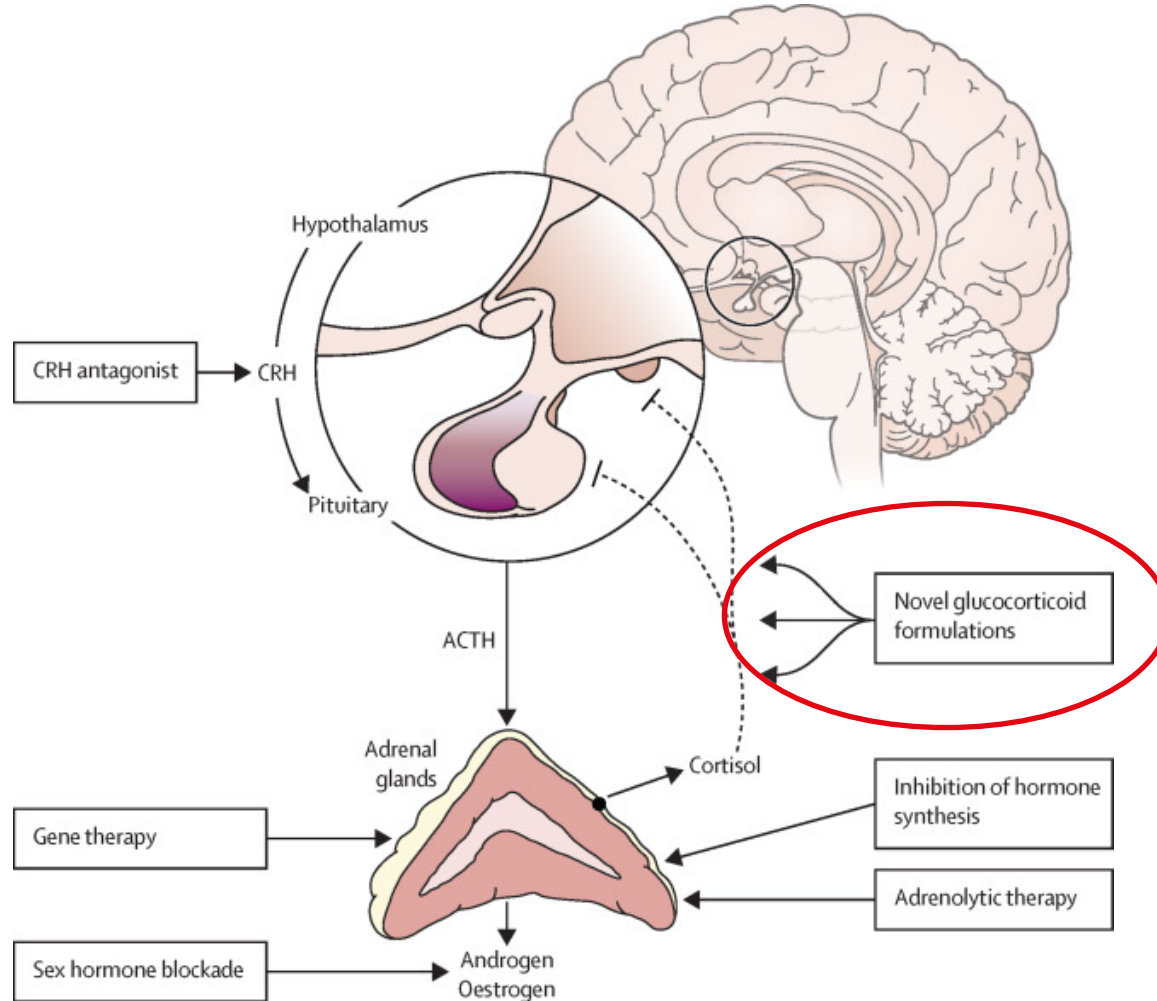


JUST REPLACE THE CORTISOL

Not so simple

- **Cortisol has a diurnal rhythm**
- **Peak 0700 – 0900; trough 2200 - 0300**
- **Hydrocortisone short half-life: 60 min**
- **Require tds dosing; provides < 24hr cover**

NEWER TREATMENTS FOR CAH



USE OF MODIFIED RELEASE HYDROCORTISONE

- **Plenadren**

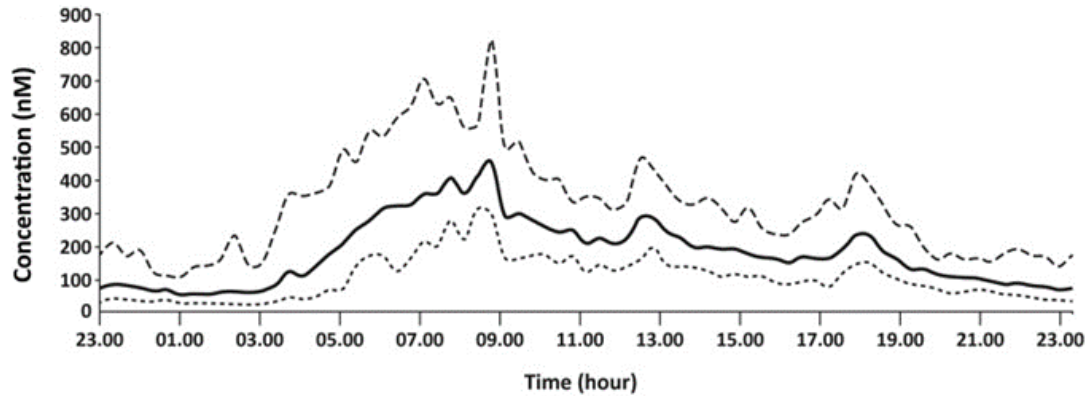
**immediate and delayed-release HC
combined once daily, on wakening**

- **Chronocort**

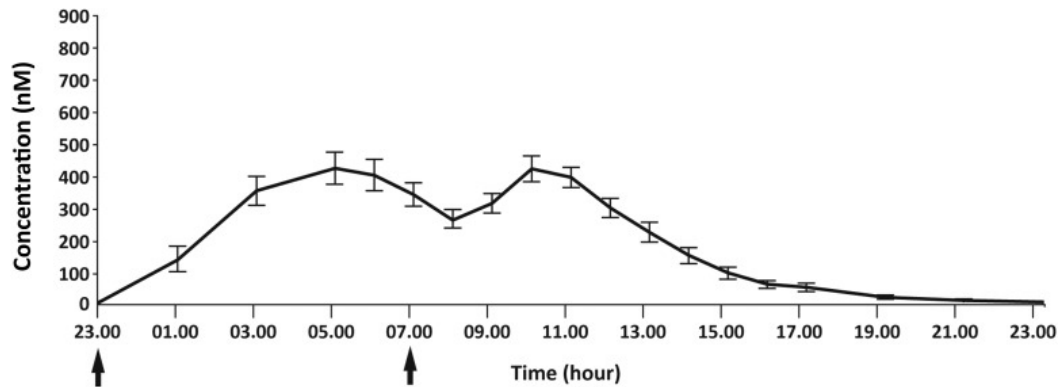
**modified-release formulation twice daily,
at night and on wakening**

REPLICATING THE CORTISOL DIURNAL RHYTHM

Normal cortisol profile



Chronocort in CAH



SURGICAL ISSUES



↑
Surgery ?



↑
Surgery ?

When ?

OUTCOMES IN CAH

- **Growth and puberty**

 - considerable improvements*

 - final height within population standards*

- **Reproduction**

 - high success rate in females, BUT low maternalism*

 - non-compliance in males; testicular adrenal rests*

- **Adult morbidity**

 - cardiovascular*

 - bone health*

 - decreased QoL*

 - increased mortality*

CONGENITAL ADRENAL HYPERPLASIA

Management

- **Fetal**
- **neonatal**
newborn screening
clinical diagnosis
early treatment
- **childhood/adolescence**
growth
puberty
- **adulthood**
metabolic aspects
fertility
pregnancy
genetic counselling

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Gynaecologist

Urologist

Obstetrician

Support groups

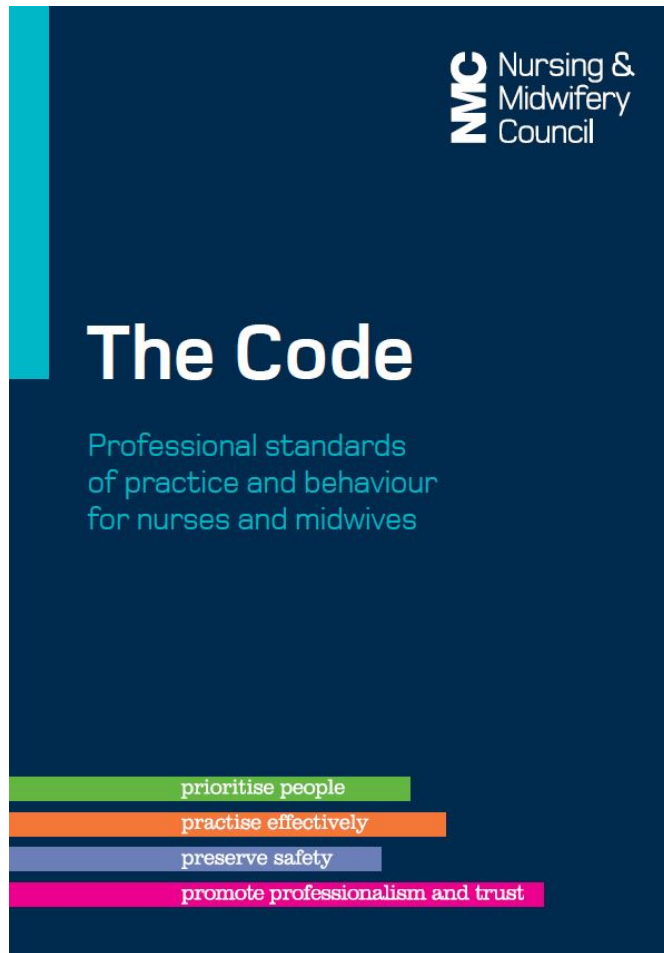
Psychologist

Endocrinologist

Geneticist

Specialist Nurse

Ethicist



- **3.1 pay special attention to promoting wellbeing, preventing ill health and meeting the changing health and care needs of people during all life stages**
- **3.4 act as an advocate for the vulnerable**
- **6.1 make sure that any information or advice given is evidence-based, including information relating to using any health and care products or services**

Literature search: CAH 2019



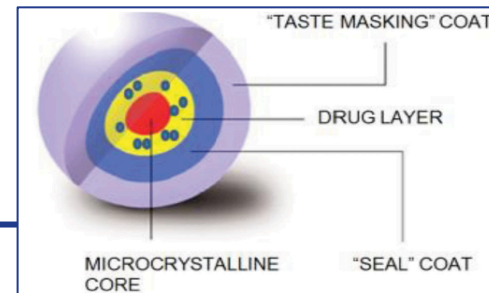


Cardiovascular Health in Children and Adolescents With Congenital Adrenal Hyperplasia Due to 21-Hydroxylase Deficiency

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- Children with CAH now at risk of cardiovascular risk factors
- Treatment regimens?
- PENS role



- **Alkindi Hydrocortisone granules well tolerated in children less than 6 years of age**

Neumann et al Clin Endocrinol 2018; 88: 21 – 9

1. Hold capsule so that the text is at the top and tap the capsule to make sure the granules are at the bottom



2. Gently squeeze the bottom of the capsule



3. Twist off the top of the capsule



4. Pour all granules out of capsule

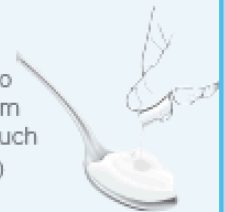


Either pour all the granules directly onto the child's tongue



OR pour all the granules directly onto a spoon and place them in the mouth

OR for children who are able to take soft food, sprinkle the granules onto a spoonful of cold or room temperature soft food (such as yoghurt or fruit puree) and give immediately





Hydrocortisone Granules Are Bioequivalent When Sprinkled Onto Food or Given Directly on the Tongue

Eleni Daniel,¹ Dena Digweed,² Jo Quirke,² Bernard Voet,² Richard J. Ross,¹
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Ethical issues with early genitoplasty in children with disorders of sex development

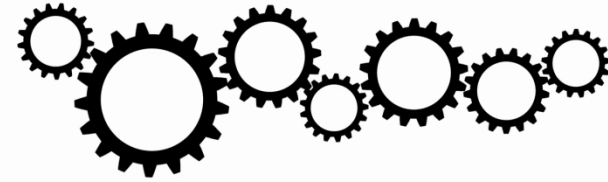
Rebecca M. Harris and Yee-Ming Chan

Curr Opin Endocrinol Diabetes Obes 2019 Feb;26(1):49-53

- When the child can make the decision or earlier?
- Patient advocacy groups
- Reduce stigma



@dsdfamilies



- Important to be aware of current research
- Evidence based practice
- Work in partnership with multi-disciplinary team

The NEW ENGLAND JOURNAL of MEDICINE

REVIEW ARTICLE

Julie R. Ingelfinger, M.D., *Editor*

Adrenal Crisis

R. Louise Rushworth, M.B., B.S., Ph.D., David J. Torpy, M.B., B.S., Ph.D.,
and Henrik Falhammar, M.D., Ph.D.

NEJM Aug 29 2019