WHAT’S NEW IN CAH?

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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

Cholesterol → 170H-progesterone → 21-hydroxylase → CORTISOL ↓ ANDROGENS ↑↑

Engels, 2019 (adapted)
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

CRH antagonist → CRH → Hypothalamus → ACTH → Adrenal glands → Cortisol

Gene therapy

Sex hormone blockade

Androgen Oestrogen

Inhibition of hormone synthesis

Adrenolytic therapy

Novel glucocorticoid formulations

El-Maouche, 2017
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
Normal cortisol profile

Chronocort in CAH

Porter et al, 2017
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

- Fetal
- Neonatal
  - Newborn screening
  - Clinical diagnosis
  - Early treatment
- Childhood/adolescence
  - Growth
  - Puberty
- Adulthood
  - Metabolic aspects
  - Fertility
  - Pregnancy
  - Genetic counselling

Management

- Obstetrician
- Endocrinologist
- Urologist
- Gynaecologist
- Geneticist
- Psychologist
- Support groups
- Ethicist
- Specialist Nurse

Professionals
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

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

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Surgery in girls

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
Nurses’ key roles

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

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