INDIANA UNIVERSITY
SCHOOL OF MEDICINE

LAB TESTS FOR CAH: IMPORTANT THINGS TO KNOW

2016 CONGENITAL ADRENAL HYPERPLASIA

PATIENT & FAMILY SUMMIT

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PROFESSOR OF CLINICAL PEDIATRICS

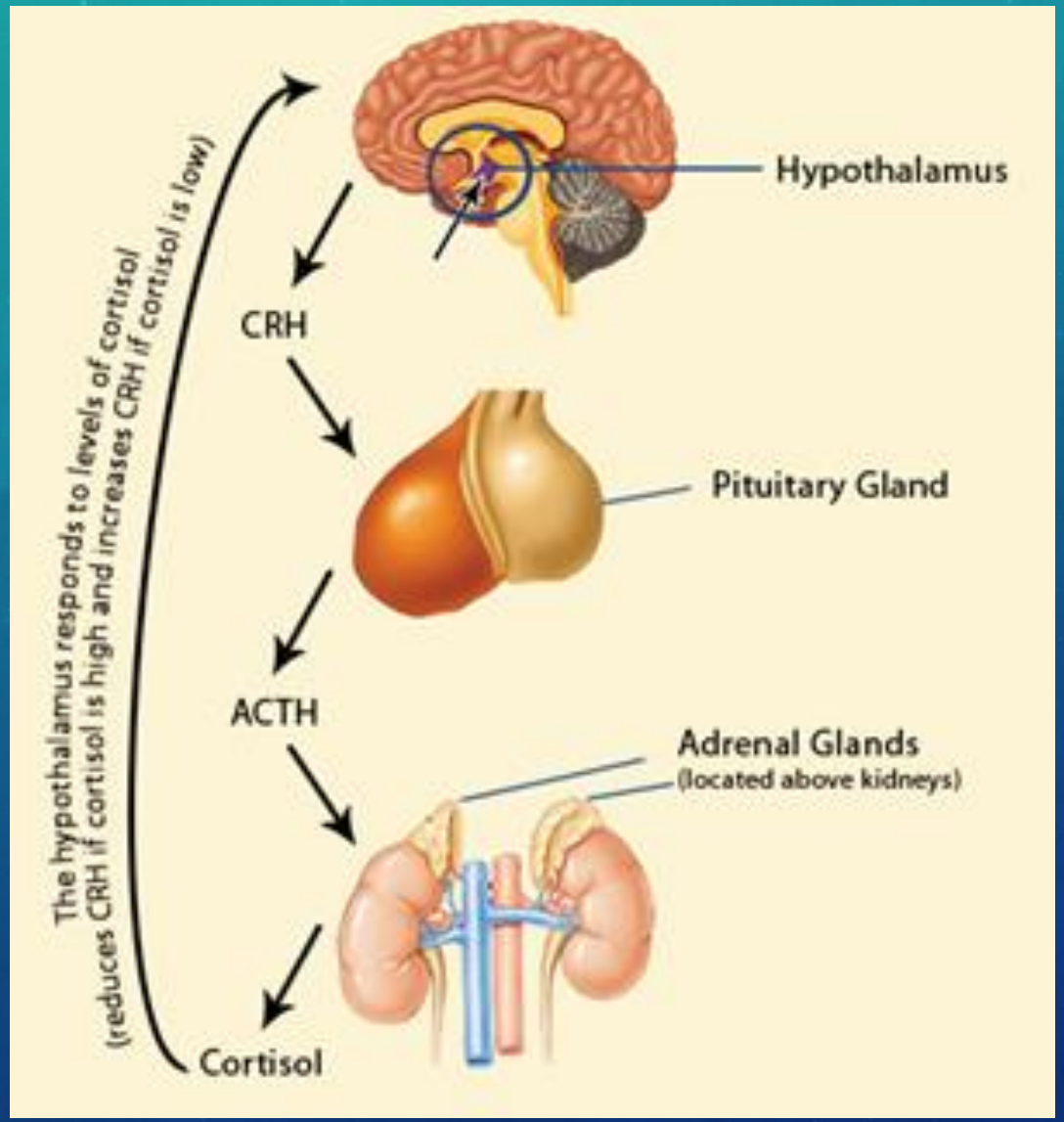
Riley Hospital for Children

WHY DO TESTS FOR CAH?

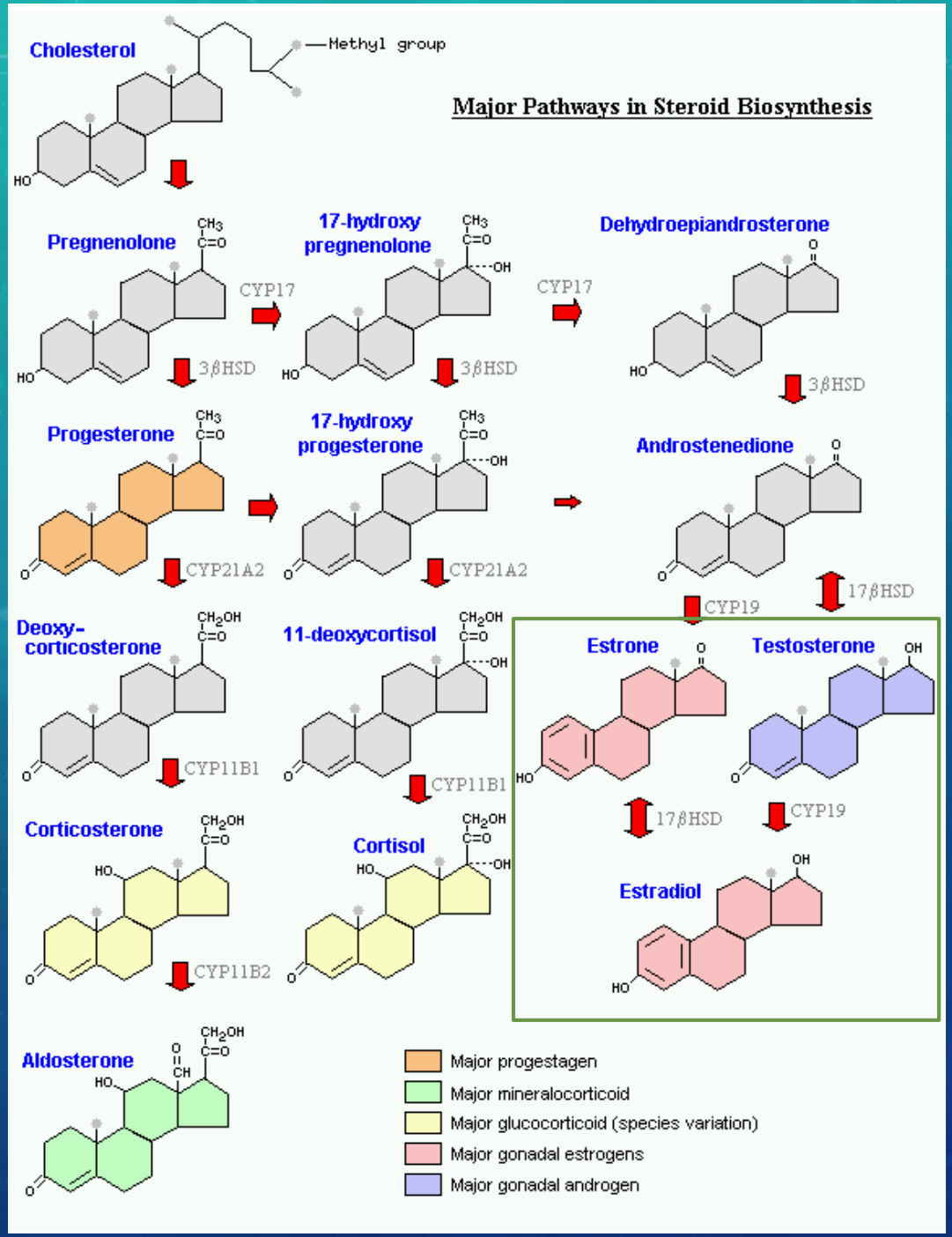
- Diagnosis
  - Newborn screening
  - Evaluation of suspected newborns
  - Precocious puberty
  - Abnormal reproductive function and signs of virilization in teens/women
- Monitoring of treatment
  - Ensure proper doses

CONTROL OF STEROID SECRETION

- HPA axis



NORMAL STEROID SYNTHESIS



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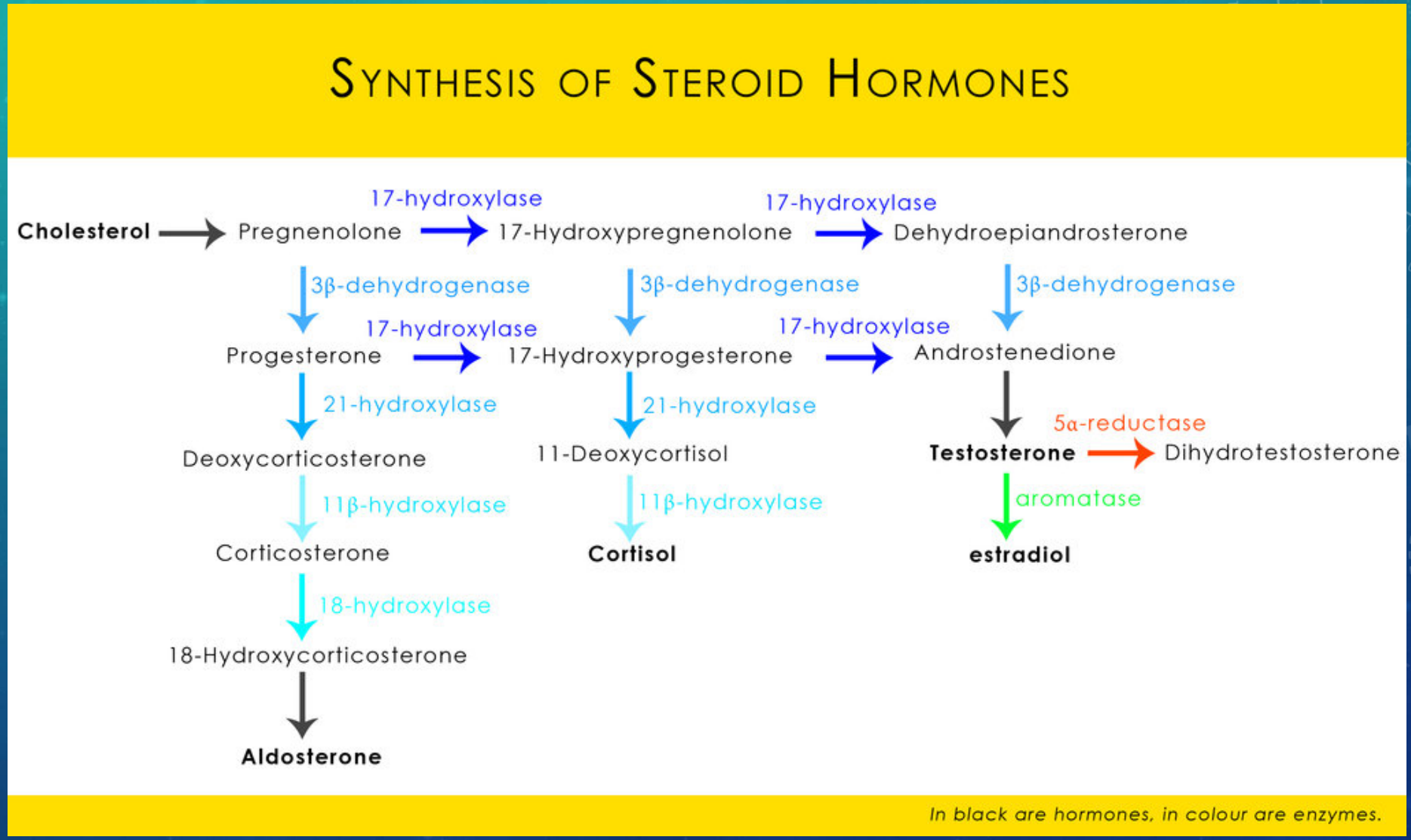
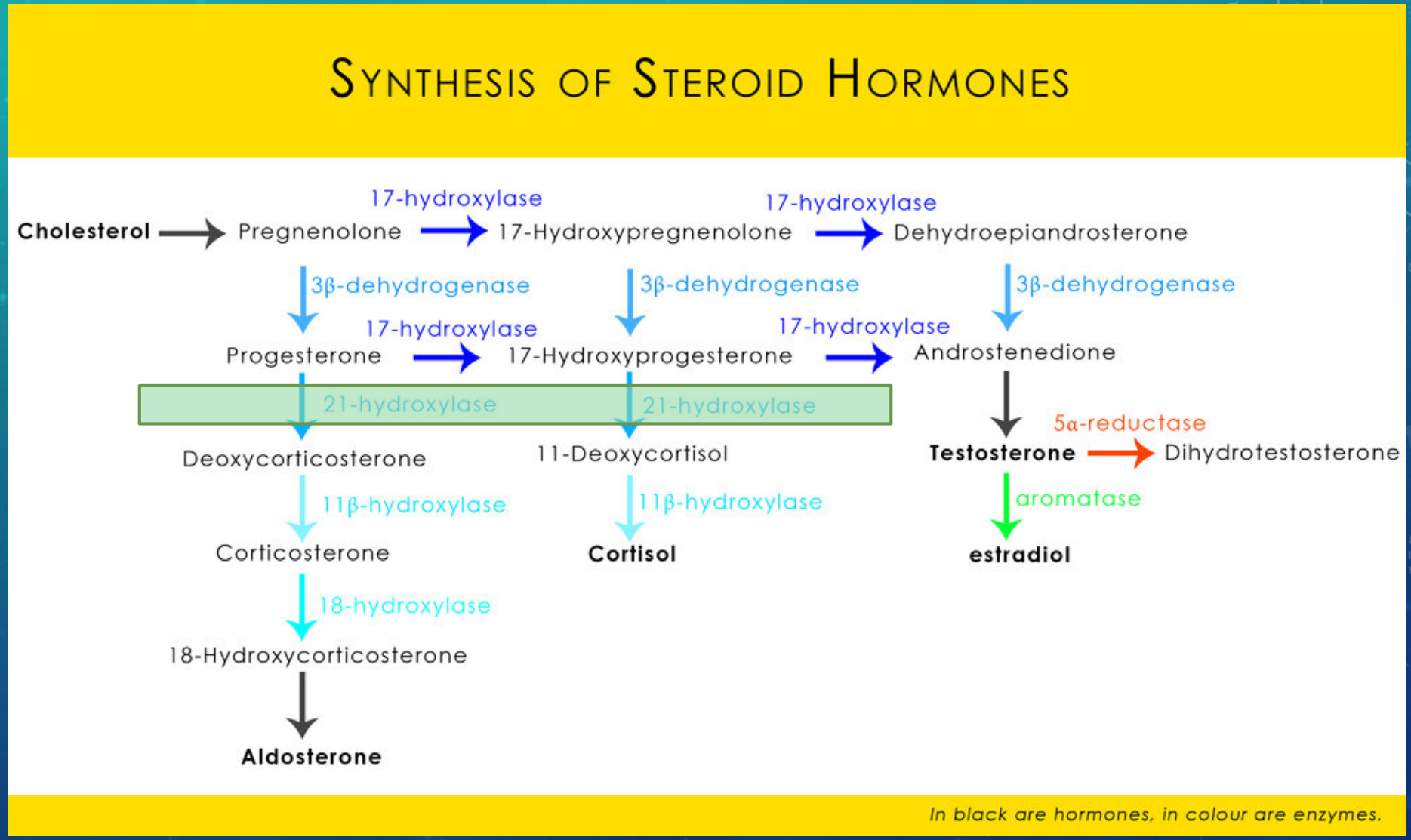


STEROID SYNTHESIS IN CAH



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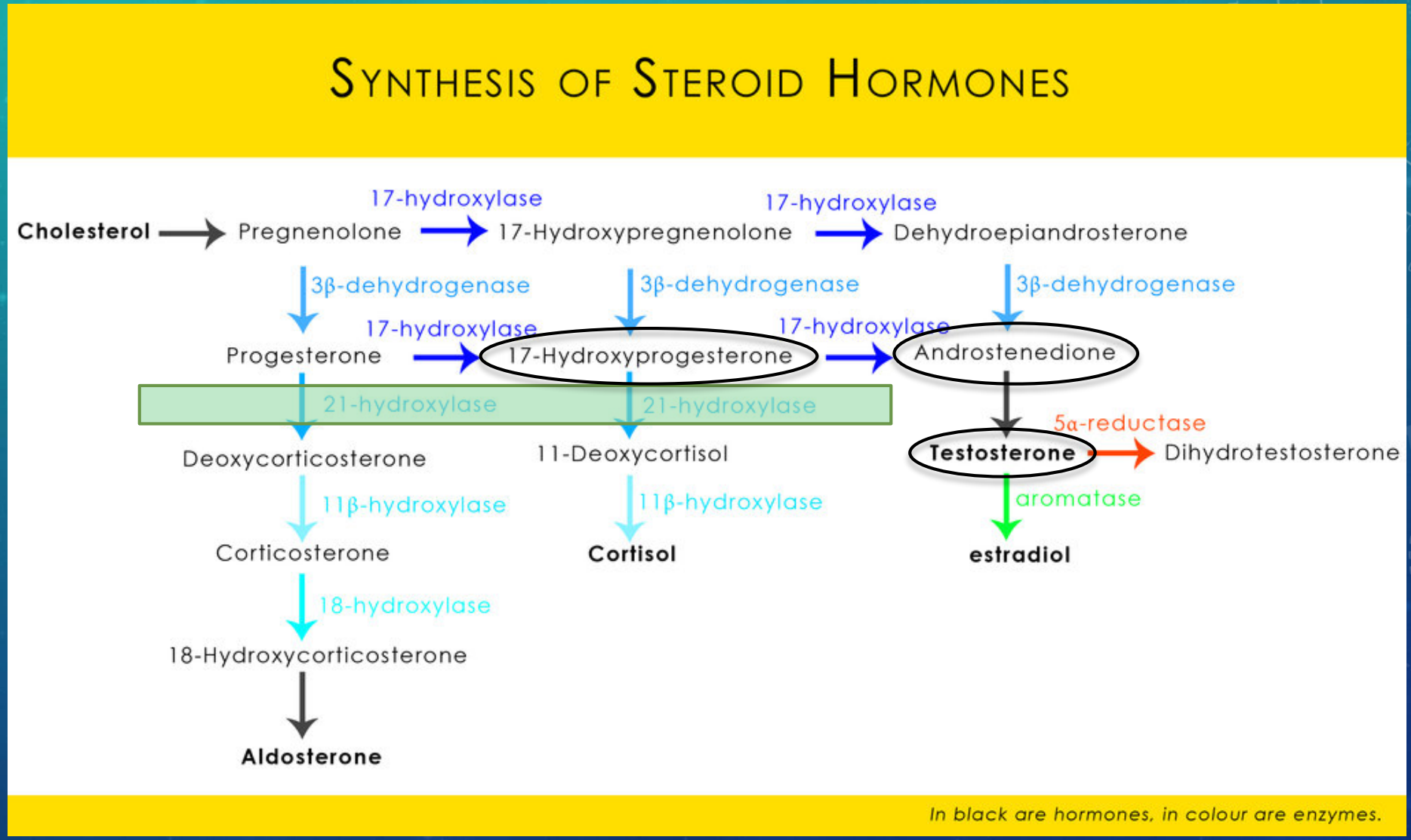
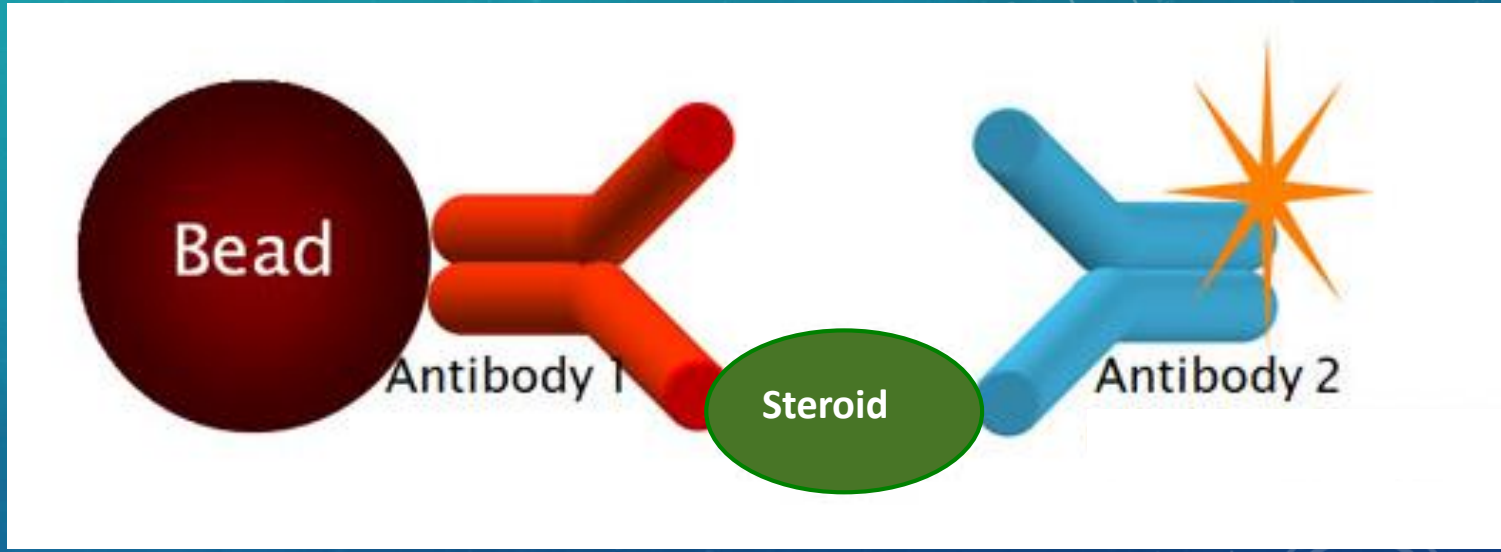
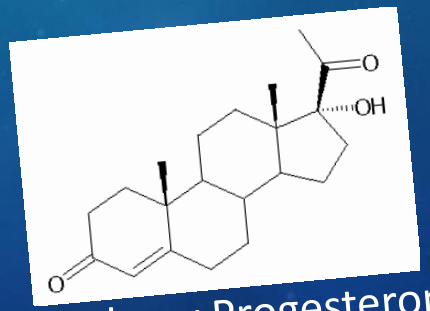


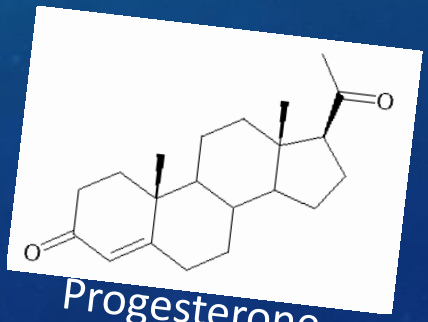
HOW ARE THESE SUBSTANCES MEASURED?

- Immunoassays
  - Routinely available at most labs
  - Can get results quickly
  - May be difficult to distinguish between similar substances





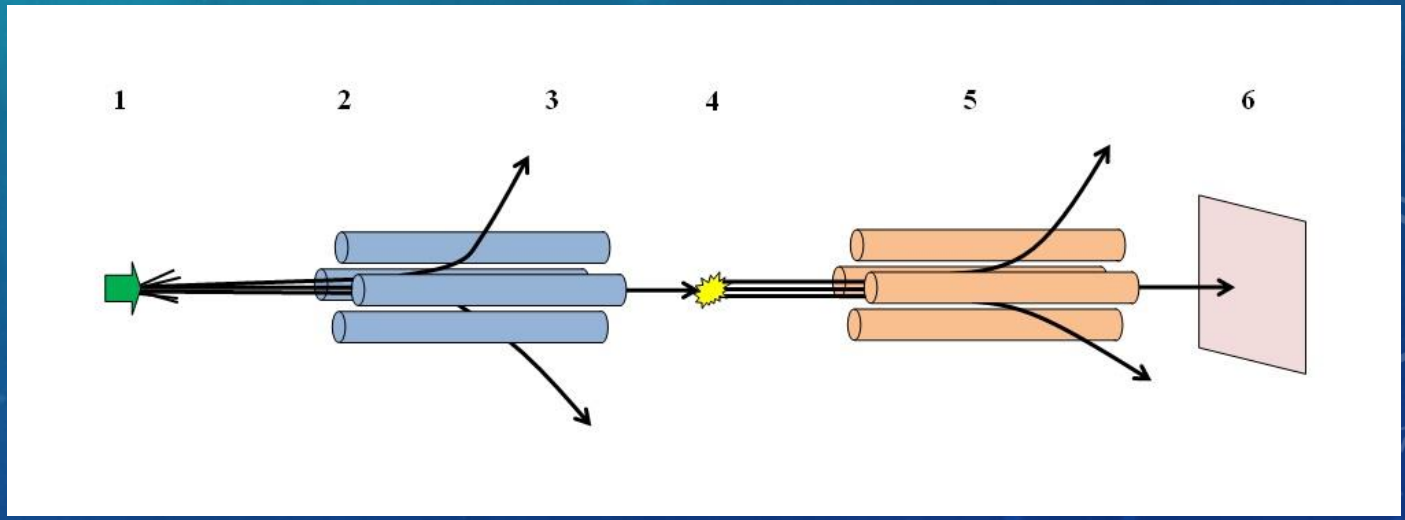
17-Hydroxy Progesterone



Progesterone

HOW ARE THESE SUBSTANCES MEASURED?

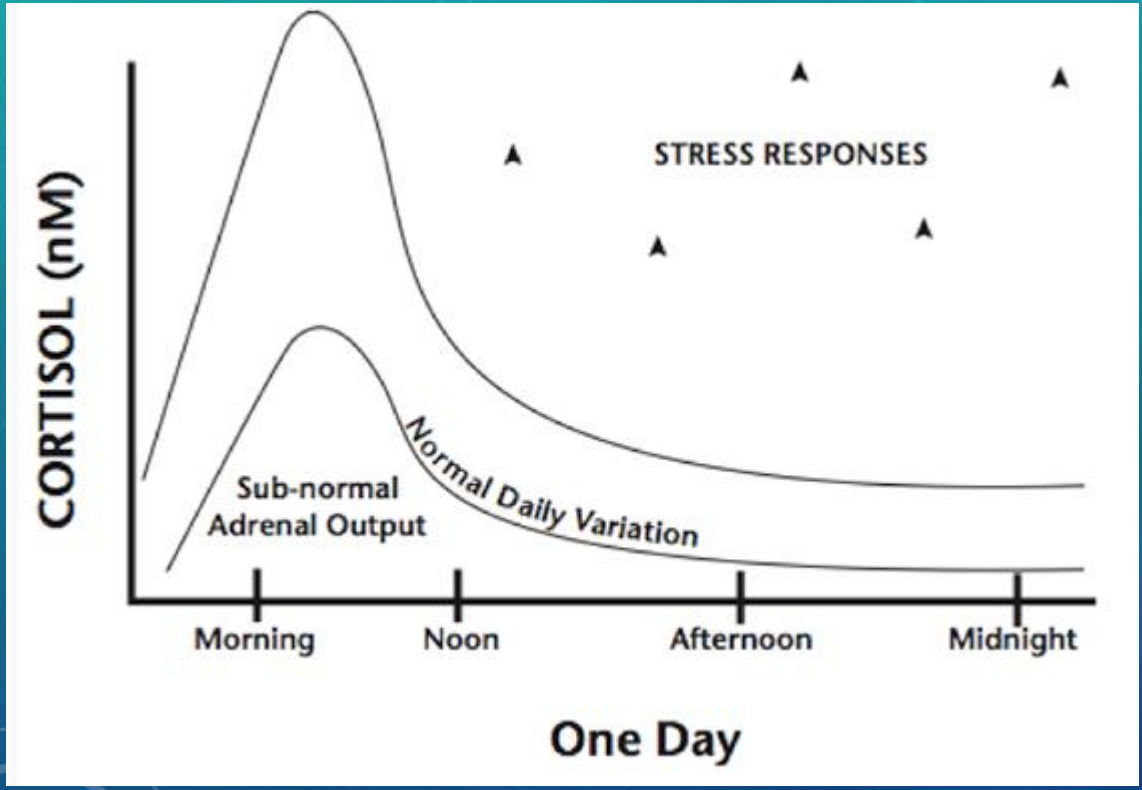
- Tandem mass spectrometry
  - Able to distinguish between similar substances
  - Not routinely available at hospital or community labs

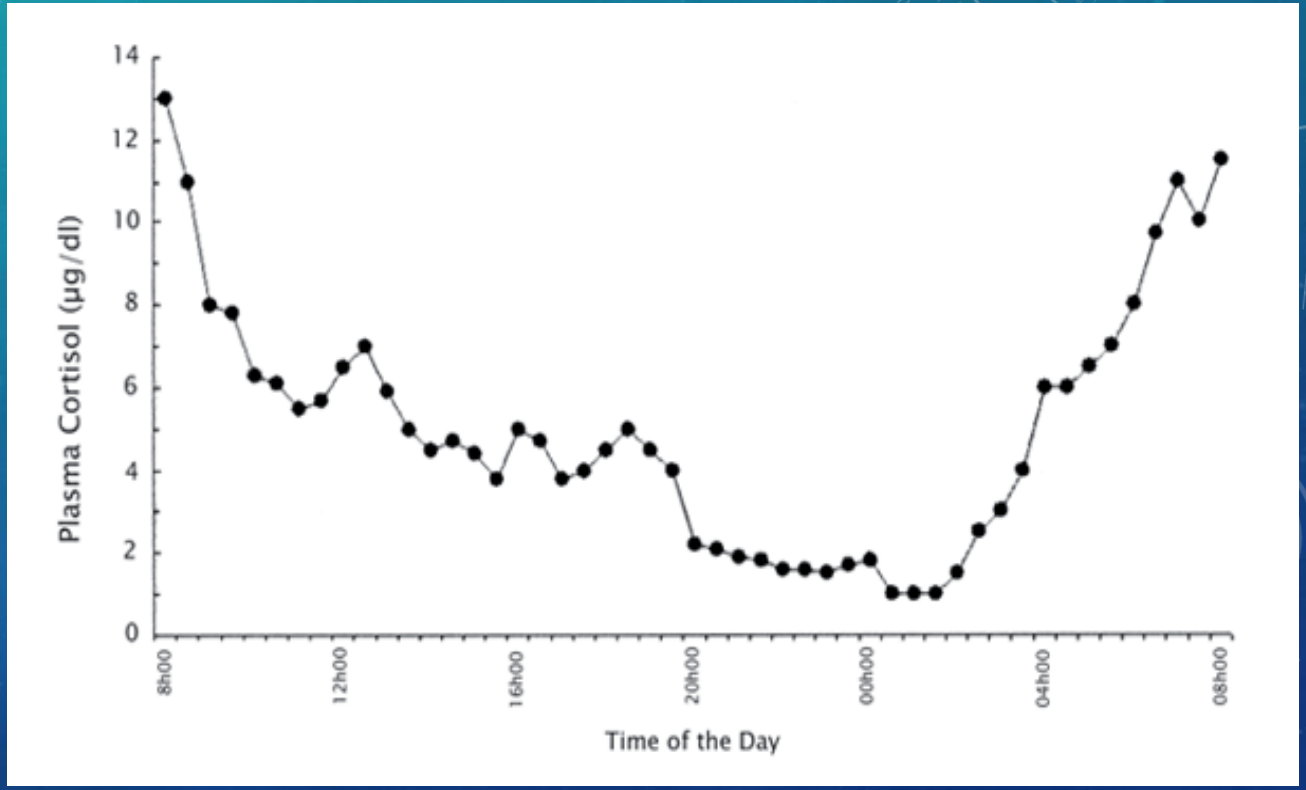


ISSUES IN CAH MONITORING

- Time of blood tests
- Time of last medication
- Recent stress dosing
- Stress of blood drawing

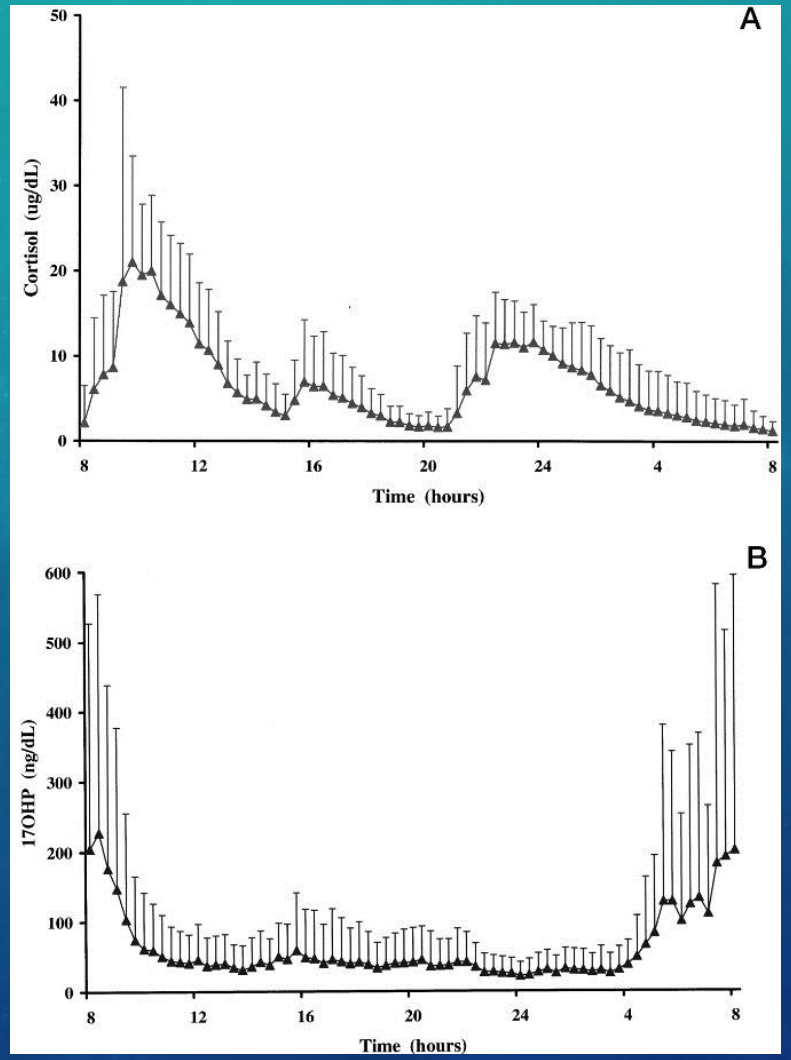
CONTROL OF STEROID SECRETION

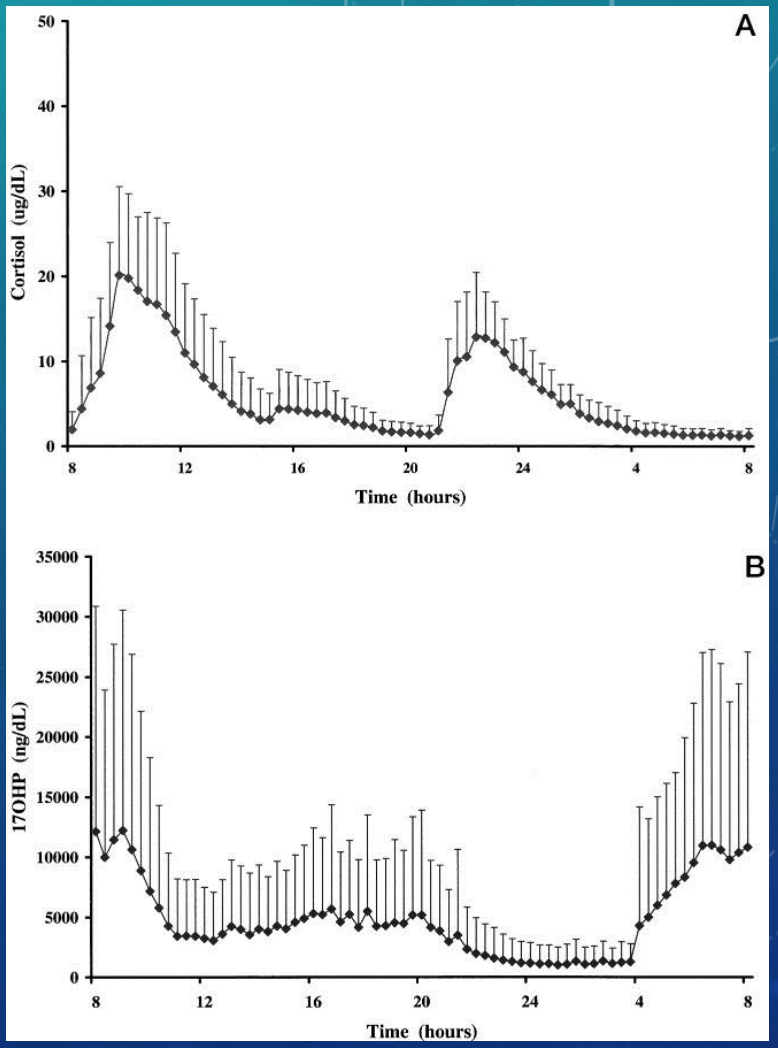




STEROID SECRETORY PATTERN IN CAH

- 36 children with CAH
- Every 20 minute measurement of cortisol and 17OHP
- Usual hydrocortisone doses

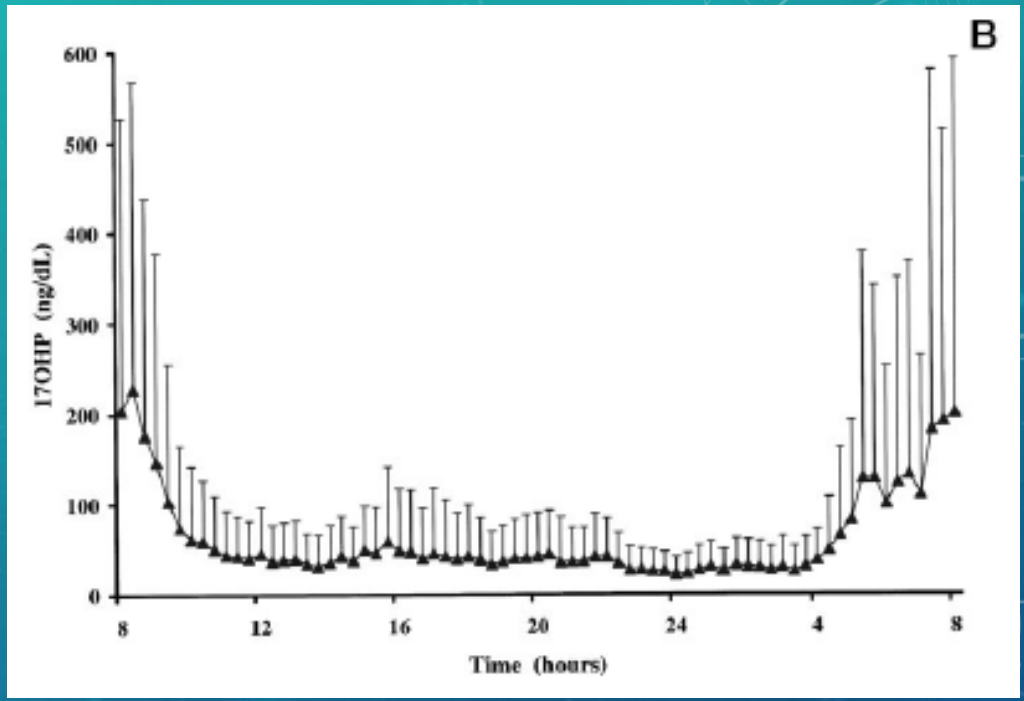


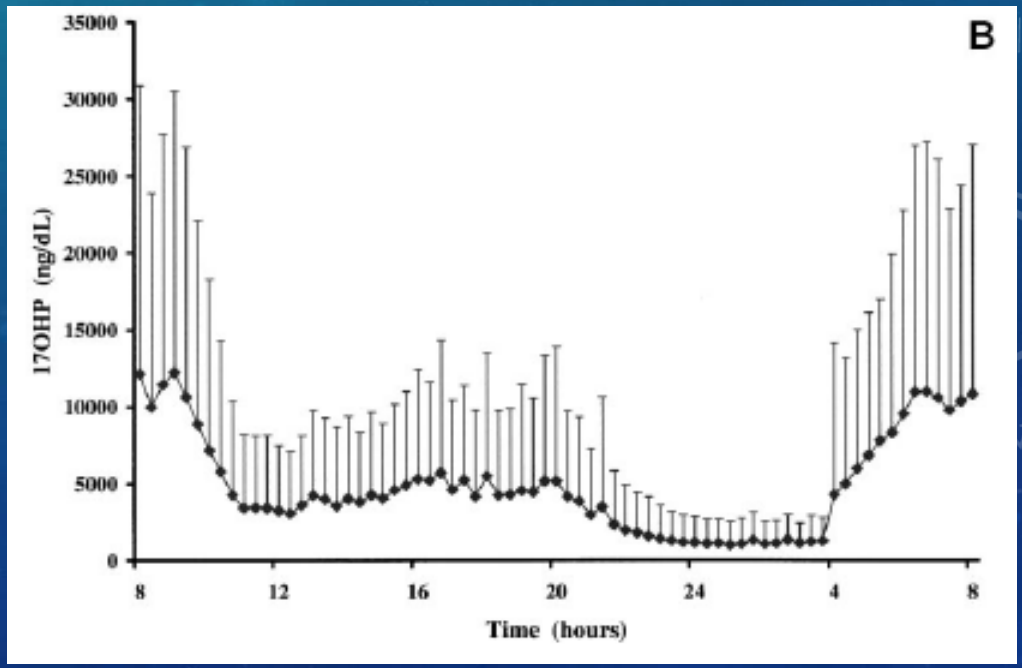


Charmandari, et al. JCEM 86:4679, 2001

TIME OF DAY EFFECTS

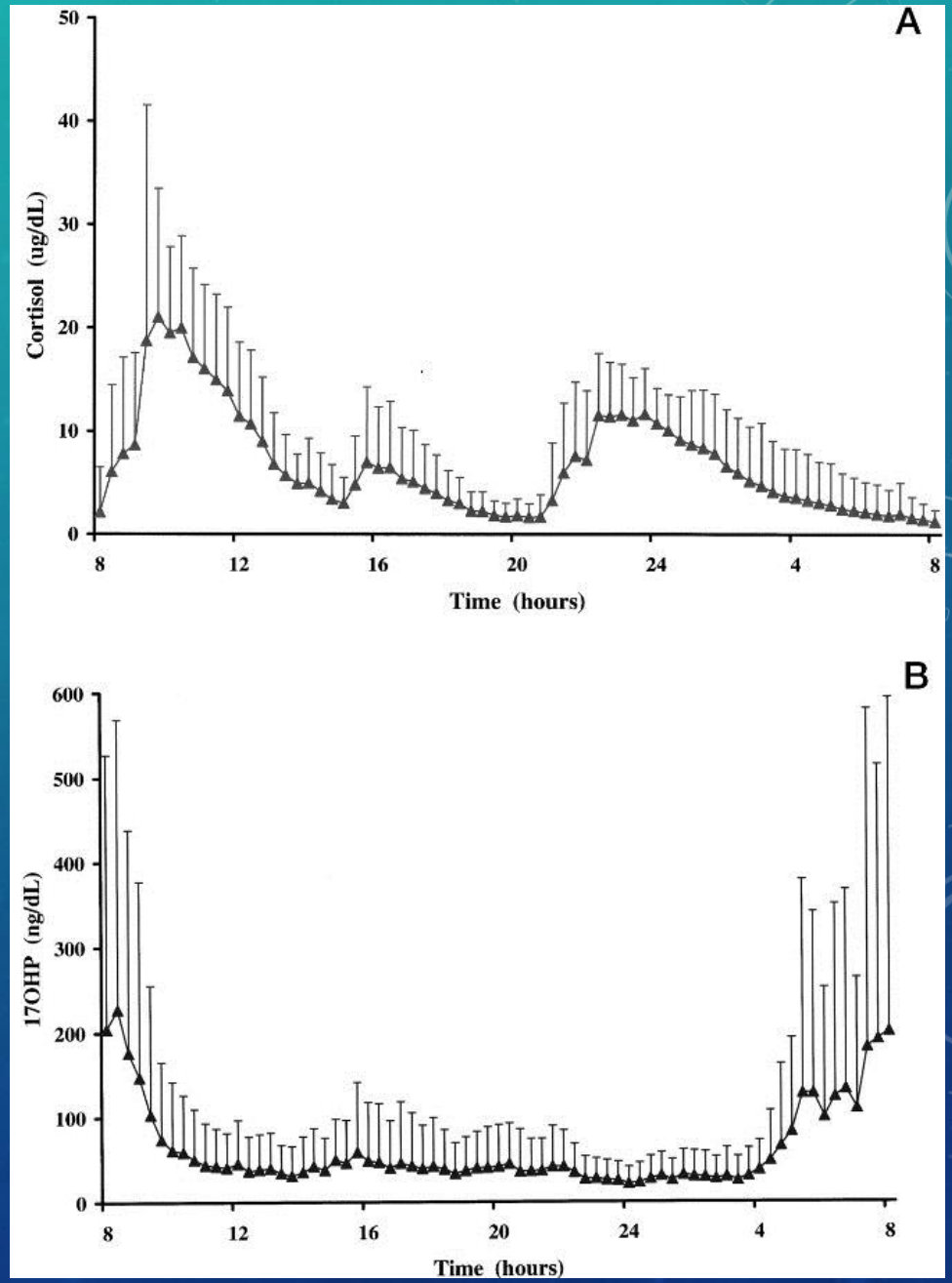
- Like cortisol, 17OHP is low overnight
- Both increase at about 4:00 AM
- Regardless of degree of control
  - Better control means lower overall levels
- Easier to suppress later in the day





PROXIMITY TO STEROID DOSING

- Rapid decline in 17OHP and androstenedione after hydrocortisone
- With the morning dose, the lowest levels are 3-4 hours afterwards
- Testosterone is more steady

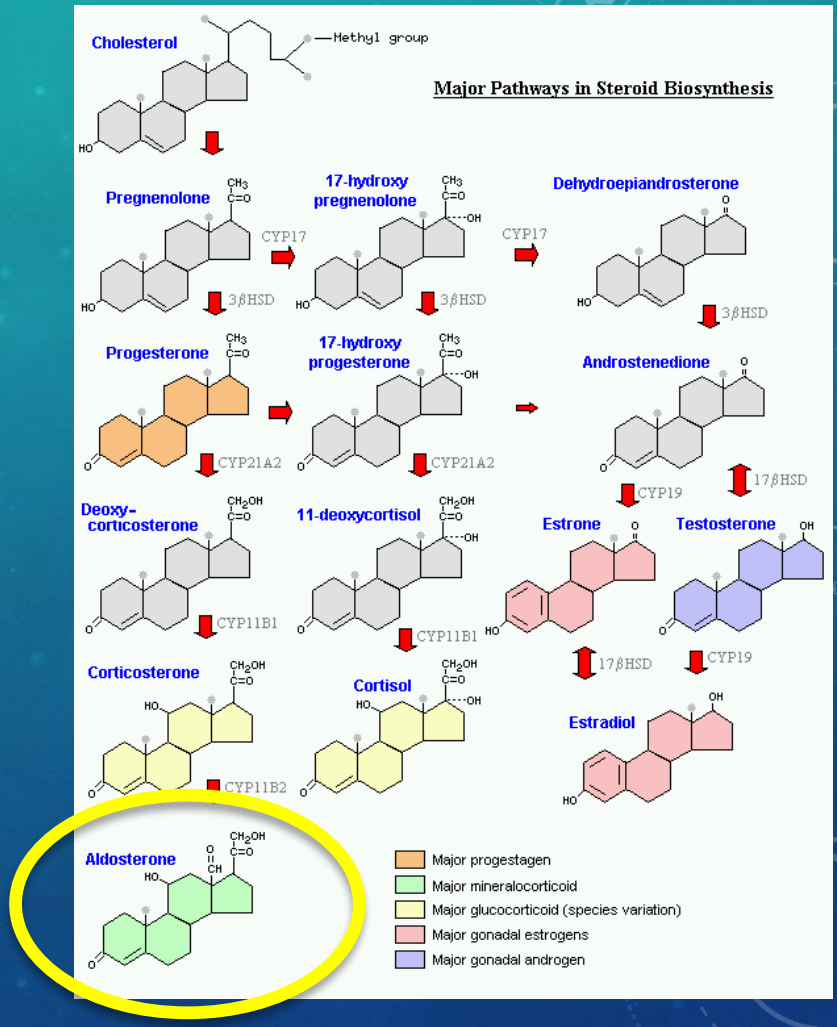


OTHER CONSIDERATIONS IN TESTING

- Effects of stress dosing
  - Suppression of 17OHP, androstenedione, and testosterone
  - May last for days after administration
  - Avoid lab tests soon after stress dosing
- Stress of blood drawing
  - Physical or emotional stress (pain, anxiety) leads to rapid increases in 17OHP
    - Androstenedione and testosterone are less responsive

PLASMA RENIN ACTIVITY

- Assesses mineralocorticoid effect
- High in untreated salt-wasting CAH
- Used to decide on fludrocortisone (Florinef) dose and salt intake

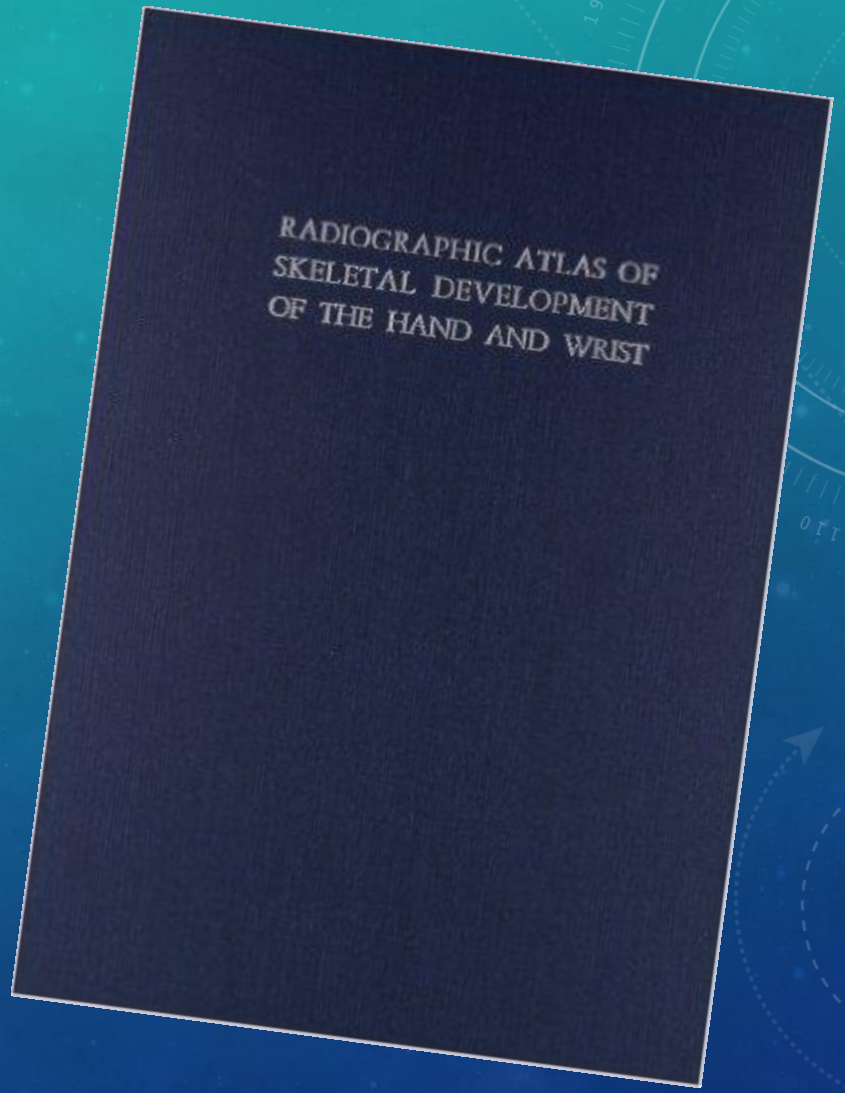


ALTERNATIVE TESTING APPROACHES

- Salivary testing
- 24 hour urine collection
- Filter paper blood spot

BONE AGE

- Done to measure skeletal maturity
- Greulich and Pyle atlas, ©1950
- Compares child's x-ray of hand/wrist to a series of x-rays from children of different ages



BONE AGE

- Advanced bone ages predict earlier attainment of adult height
- Example:
  An 8 year old girl with a bone age of 11 only has 3 years of growth remaining instead of 6 years

OTHER CONSIDERATIONS IN MONITORING CAH

- Growth chart
- Bone age
- Clinical factors