Peds I – Inborn Errors of Metabolism
2/11/18

Don't be scared of the inborn errors of metabolism and endocrinology. They are really quite simple to screen for. Focus less on knowing the details of each one, and more on the general concept of what byproducts are building up and what substance is missing.

Inborn errors of metabolism

- Involves the buildup of toxic intermediates or lack of others vital substances
    - Buildup of: **ammonia** (ornithine transcarbamylase def)
    - Buildup of: **acid** (methylmalonic acidemia)
    - Lack of: **glucose** (glycogen storage disorder)
- Hundreds of diseases can be screened for with THREE lab tests
    - 1. Glucose
    - 2. Ammonia
    - 3. Acid (Electrolyte panel – CO2)
- Symptoms – nonspecific

Congenital Adrenal Hyperplasia

- Missing enzyme: **21-Hydroxylase**
- Missing substances:
    - Aldosterone – low Na+ / low BP
    - Cortisol – low glucose, hyperpigmentation
- Buildup of:
    - Sex hormones (androgens) – fused labia, partial male genitalia, etc