**BACKGROUND**

PHA1 is a rare disorder of congenital salt loss, resulting from resistance of kidney &/or other tissues to mineralocorticoids, arising from mutations in genes encoding Mineralocorticoid Receptor (MR: NR3C2, autosomal dominant), or Ephiallum Sodium Channel (ENaC) genes (SCNN1A/B/G: autosomal recessive) (see Riepe FG, 2009 for review). A milder clinical phenotype associates with renal PHA1 (NR3C2) and shows variable penetrance. Genetic PHA1 may be distinguished from PHA2 by urinary tract abnormalities by USP, with high levels of aldosterone and mineralocorticoid precursor metabolites characteristic in both conditions (see Steroid anabolic / catabolic pathways Fig.1 adjacent), with additional presence of raised cholesterol peak in PHA2.

**OBJECTIVES / METHODS**

Case reports: 2 families/3 patients.

Urine steroid profiles (USPs) represent Total Ion Current chromatograms of steroids as MO-TMS derivatized, obtained on a Perkin Elmer Clarus 500 single quadrupole GC-MS system.

**RESULTS**

**CONCLUSIONS**

- USP has major role in diagnosis of neonatal salt-wasting disorders
- Results may be available before full diagnostic panel of serum parameters
- Empirical treatment (Fludro + NaCl) in PHA can successfully manage patients before a specific diagnosis is confirmed
- In potential familial cases, USP can provide a non-invasive diagnosis in advance of apparent clinical deterioration
- The appearance of a significant cholesterol peak in USPs should raise suspicion of PHA2 but full clinical and endocrine assessment is required to support the diagnosis

**Reference**