Monitoring of treatment in children with congenital adrenal hyperplasia (CAH) includes assessment of growth and bone maturation, and measurement of hormone concentrations. Hormone measurement is difficult, because steroid production follows a circadian rhythm and is influenced by short-term stress and steroid drugs. Steroid assessment in urine collected over 24h is most reliable, but for some children this is not feasible. Thus, urine spots might be an alternative. In healthy children, steroid concentrations in 24h and spot urine do not correlate well, but in children with CAH who have a unique urine steroid signature, this could be different.

**OBJECTIVE**

To investigate whether steroid metabolites in 24h urine collections correlated with those from urine spot samples in children with CAH.

**METHOD**

- **Study design:** Observational study including data from three European countries (Greece, The Netherlands, Switzerland).
- **Sample:** 40 Children and adolescents with CAH due to 21-hydroxylase deficiency.
- **Collection:** Both 24h urine and urine spots. Urine spots: 20/40 collected in the morning; 20/40 later during day (non-morning).
- **Biochemical analyses:** 40 steroid metabolites using GC-MS.
- **Statistical analyses:** Correlations between 24h and spot urine metabolites assessed using Kendall’s tau-b correlation. Correlation were considered significant if Kendall’s tau b >0.45 and p<0.002 to account for multiple testing.

**RESULTS**

- **Strong correlation between 24h and morning spot urine for 17 of the 40 measured metabolites and between 24h and non-morning spot urine for 11/40 metabolites** (Figure 1).

Figure 1: Steroid pathway depicting measured metabolites and their correlation between 24h and spot urine (modified from Black et al., JSBMB 2020).

**CONCLUSIONS**

- **Urinary steroid profiling in children with CAH revealed correlations between 24h urine specimens and spot urines.**
- **Morning spot urine might suffice to recognize the specific pattern of 21-hydroxylase deficiency for management of CAH children (e.g. through marker metabolites Pregnanetriolone, TH-11-deoxy-cortisol or substrate/product ratios).**
- **In which subgroups of CAH a spot urine can inform about metabolic control of treatment needs to be tested in a larger sample.**

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