Background
Controlling therapy of infants, especially from neonates onwards, with classic congenital adrenal hyperplasia (CAH) is challenging due to the lack of reference values.

Objective and hypotheses
Retrospective analysis of the urinary steroid metabolome obtained by gas chromatography-mass spectrometry (GC-MS) for treatment monitoring of infants with CAH.

Methods
We retrospectively analyzed repeated spot urinary steroid hormone metabolite profiles determined by gas chromatography–mass spectrometry (GC-MS) of 60 infants aged ≤ 4 years with classic 21-hydroxylase deficiency (21-OHD) on hydrocortisone and fludrocortisone treatment. Fig. 1 gives an overview of analyzed urinary steroid metabolites.

Results
Infants aged 1 year (N=14) demonstrated a reduction of their height (H-SDS: -1.0 ± 1.7) (Fig. 2).
H-SDS was significantly correlated with tetrahydrocortisone (THE) to tetrahydrocortisol (THF) ratio (R_s = 0.70; P < 0.01), demonstrating an impact of the individual metabolism of hydrocortisone on growth in infants (Fig. 3A).
Additionally, H-SDS was negatively correlated with the ratios of THF to the 17-hydroxyprogesterone (17-OHP) metabolite pregnanetriol (R_s = -0.64; P = 0.02) (Fig. 3B), THF to 11-hydroxyandrostenedione (11-OH-An) (R_s = -0.68; P = 0.01) (Fig. 3C), and THF to summed androgen metabolites (androsterone, etiocholanolone and 11-hydroxyandrostenedione) (R_s = -0.71; P < 0.01) (Fig. 3D).
In contrast, the hydrocortisone dosage was not related to H-SDS (not shown).

Conclusions
A substantial proportion of infants with CAH were over treated. The urinary steroid hormone metabolite profiles, but not the prescribed hydrocortisone dosage, were related to height at one year in infants with classic CAH. Additionally, the individual metabolism of hydrocortisone, as shown by the tetrahydrocortisone-to-tetrahydrocortisol ratio, influences the growth in infants treated with hydrocortisone.

Disclosure: The authors have nothing to disclose.