NON-CLASSIC CONGENITAL ADRENAL HYPERPLASIA AND POLYCISTIC OVARY SYNDROME DIFFERENTIATION USING COMMERCIALLY AVAILABLE LC-MS/MS STEROID PROFILE KIT: A CASE REPORT

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INTRODUCTION: Hyperandrogenism and oligomenorrhea are common traits of patients with polycistic ovary syndrome (POCS) and non-classic congenital andrenal hyperplasia (NCAH). As these disorders have a similar presentation, their distinction using solely clinical symptoms is difficult. Of steroid hormones, 17-hydroxyprogesterone concentration is commonly used as discriminator between POCS and NCAH, although the universal cut off value is not yet established.

OBJECTIVE: To evaluate steroid hormone profile for differentiation of NCAH from PCOS in a patient.

METHODS: A 24 years old female patient visited endocrinologist because of acne, mild hirsutism and sporadically excessive sweating accompanied with hot flashes and tachycardia. Her menstrual cycles were regular, as it was ovary ultrasound. Patient also did an ergometry and heart ultrasound due to the sporadically high blood pressure and dyspnea, but these findings were not remarkable. As a part of laboratory work-up, steroid hormone profile which included cortisol, 11-deoxycortisol, 21-deoxycortisol, 11-deoxycorticosterone (11-DOC), dehydroepiandrosteronesulphate (DHEAS), androstendione, testosterone and 17-hydroxyprogesterone (17-OHP), was measured in morning serum obtained during early follicular phase using Recipe ClinMass Steroid in Serum/Plasma kit and Shimdzu Nexera X2 liquid chromatograph coupled with Shimadzu LCMS-8050 mass detector.
FSH and LH were measured with chemiluminescent microparticle immunoassay (CMIA) on Abbott Alinity i analyser.

RESULTS: Steroid hormone concentrations are shown in Table 1.

Table 1. Steroid hormone profile of the observed patient

Analyte (unit)	Value	Ref. interval
Cortisol (nmol/L)	377	127 - 568
11-deoxycortisol (nmol/L)	9.8	< 3.1
21-deoxycortisol (nmol/L)	< 0.02	0.06 – 0.44
11-DOC (nmol/L)	1.4	< 0.3
DHEAS (µmol/L)	0.6	1.2 – 8.7
Androstendione (nmol/L)	12.3	1.2 – 8.7
Testosterone (nmol/L)	2.52	0.07 – 1.56
17-OHP (nmol/L)	8.1	< 5.6
FSH (IU/L)	2.99	Follicular phase: 3.0 – 8.1
LH (IU/L)	5.33	Follicular phase: 2.4 – 6.6

CONCLUSION: Although clinical symptoms were mild, steroid profile and gonadotropin hormone measurement pointed to the NCAH. 11-DOC has mineralocorticoid effect and its elevated concentration is responsible for the elevated blood pressure, while increased testosterone and androstendione contribute to the acne and hirsutism occurrence. Elevated 17-OHP, 11-deoxycortisol and 11-DOC, decreased DHEAS together with elevated concentrations of other hormones from the profile, suggest 11β-hydroxylase deficiency. Steroid profile measurement is useful in PCOS and NCAH differentiation, as well as in distinguishing subtypes of CAH.