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INCREASED RISKS OF ATHEROSCLEROTIC HEART DISEASE, MALIGNANCY AND SLEEP APNEA WITH PRIMARY ALDOSTERONISM

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BACKGROUND
Primary aldosteronism (PA) is associated with increased risks of atherosclerosis, malignancy, and sleep apnea. We report a case of untreated PA complicated by coronary artery disease (CAD), renal cell carcinoma (RCC) and sleep apnea during the course of the illness.

CASE
Laboratory and radiological studies were performed at our hospital.

A 51-year-old male had a 12-year history hypertension and hypokalemia. His laboratory tests then showed plasma renin activity (PRA) 0.06 ng/mL/hr, aldosterone 6 ng/dL, normal plasma metanephrines and dexamethasone suppression test. Despite using several anti-hypertensive medications, his hypertension and hypokalemia remained poorly controlled. He was also diagnosed with sleep apnea and was placed on CPAP. At the age of 47 years, a diagnosis of Liddle syndrome was considered, and he was placed on amiloride. The following year, he was diagnosed with coronary artery disease. Repeat laboratory showed PRA <0.167 ng/ml/hr and plasma aldosterone 111.4 ng/dL. An abdominal CT revealed a left adrenal adenoma and a 1.8 cm left renal lesion suggestive of RCC. Adrenal venous sampling localized the left adrenal adenoma as the cause of PA. The patient underwent left adrenalectomy and left partial nephrectomy. Histopathology confirmed the diagnosis of adrenal adenoma and RCC. One week later, plasma aldosterone was 1.9 ng/dL and his hypertension improved with fewer medications without requiring potassium supplementation.

CONCLUSION
Our case illustrates that delayed diagnosis of PA may be associated with an increased risk of CAD, malignancy and sleep apnea. Early diagnosis and surgical intervention are recommended for aldosterone-producing adenoma.

PP-A-13

A NOVEL MUTATION ASSOCIATED WITH NON-CLASSICAL CONGENITAL ADRENAL HYPERPLASIA IN AN EMIRATI FEMALE

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BACKGROUND
Deficiency of the steroid 21-hydroxylase, encoded by CYP21A2 gene, accounts for 95-99% of all congenital adrenal hyperplasia (CAH) cases. We report a case of non-classical CAH (NCAH) due to a novel CYP21A2 mutation with unusual biochemical features.

CASE
A 24-year-old Emirati female with a BMI of 25.72 kg/m^2 presented with a history of repeated pregnancy losses (RPL) of spontaneous and in-vitro fertilization (IVF) pregnancies, hair fall and irregular menstrual cycles. She had acne, hirsutism, autoimmune thyroiditis and a thyroid nodule which were duly addressed. Biochemical tests showed very high levels of 17-OH progesterone (1071 ng/dL); however, testosterone, DHEAS and androstenedione levels were normal. Ovarian ultrasound showed no abnormalities.

In view of her known thyroid immunity and history of fatigue, screening of adrenal insufficiency using short synacthen test showed normal baseline, 30- and 60-minute values (239 nmol/L, 717 nmol/L and 850 nmol/L, respectively); unfortunately, paired 17-OH progesterone was not checked at the time. Genetic testing identified a novel heterozygous mutation c.337T>C p.(Tyr113His) in the CYP21A2 gene. This mutation is in a highly conserved region. Bioinformatic analysis suggests that it is highly likely to be pathogenic as a deletion of 8 codons (c332-339), including codon 337, is pathogenic.

CONCLUSION
We report an Emirati female patient with uncommon features of NCAH, including absence of classical hyperandrogenism due to a novel heterozygous variant. Genetic tests amongst relatives and additional molecular and clinical studies are planned to understand the role of this variant in CAH pathology.