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Visual Vignette

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Case Presentation: A 25-year-old-male with simple-virilizing congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency reported doing well except for salt craving. He disclosed that he had stopped taking his medications for 6 years because of weight gain and moodiness, which he perceived to be medication related. Physical examination revealed generalized hyperpigmentation and testicular volume of 12 cc bilaterally (previously 25 cc). Laboratory evaluation (8 AM) showed adrenocorticotrophic hormone (ACTH) 889 pg/mL (normal range, 5 to 46 pg/mL), cortisol 3.6 µg/dL (normal range, 5 to 25 µg/dL), 17-hydroxyprogesterone 22,404 ng/dL (normal range, 13 to 120 ng/dL), androstenedione 1,762 ng/dL (normal range, 26 to 125 ng/dL), plasma renin activity 29 ng/mL/hour (normal range, 0.6 to 4.3 ng/mL/hour), total testosterone 275 ng/dL (normal range, 240 to 950 ng/dL), luteinizing hormone 0.2 U/L (normal range, 1 to 8 U/L), and follicle-stimulating hormone 0.2 U/L (normal range, 1 to 11 U/L). An abdominal computed tomography (CT) scan obtained at age 19 had shown diffuse enlargement of both adrenals (Fig. 1). Because of his noncompliance, a repeat abdominal CT was performed (Fig. 2).

What is the diagnosis?

Answer: Bilateral adrenal myelolipomas (AMLs) and hypogonadotropic hypogonadism. The challenge in treating CAH patients is to replace cortisol and control adrenal steroid production without making the patient Cushingoid. Prior glucocorticoid overtreatment may explain our patient's noncompliance. AMLs are benign hormonally inactive tumors composed of mature adipose tissue and myeloid components in varying proportions (hence the name "myelo + lipoma"). These uncommon tumors are usually identified incidentally on routine imaging; however, patients with large AMLs (>6 cm) can present with symptoms secondary to compression of surrounding structures and are at risk for rupture and hemorrhage (1). AMLs have been reported in patients with CAH and other states of elevated ACTH (1). CT features of AML vary from well-delineated, heterogeneous masses with areas of negative attenuation (due to high fat content) to higher attenuations due to the marrow elements (1). Adrenal carcinomas appear as large irregularly shaped masses with central areas of necrosis and hemorrhage, resulting in variable enhancement, which was not seen in our patient. Chronically elevated ACTH associated with longstanding medication noncompliance likely contributed to the large size of the AMLs at this young age (left, 12.5 cm; right, 12.1 cm). Aromatization of elevated adrenal androgens to estrogens led to the

development of hypogonadotropic hypogonadism with testicular shrinkage, a recognized cause of male infertility in CAH (2). We re-introduced glucocorticoid and mineralocorticoid therapy and reinforced the importance of medication compliance to reduce risk of adrenal crisis, treat hypogonadism, and halt further growth of the adrenal masses. Lifetime treatment is indicated for all patients with classic CAH, including the simple-virilizing form (3). Adrenal imaging may be useful in the patient with poor disease control or extended noncompliance.

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DISCLOSURE

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Fig. 1.

Fig. 2.