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A giant adrenal tumor as a manifestation of congenital adrenal hyperplasia

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Congenital adrenal hyperplasia (CAH) comprises a group of genetic disorders requiring lifelong glucocorticoid replacement therapy to suppress excessive adrenocorticotropic hormone (ACTH) secretion. Untreated disease causes chronic cortisol deficiency and secondary ACTH overproduction.[1] Chronic ACTH elevation drives adrenal hyperplasia and, in some cases, masses including myelolipomas. Imaging findings may mimic adrenal incidentalomas or malignant tumors. Myelolipomas are benign tumors composed of fat and hematopoietic tissue.[2]

This case demonstrates a diagnostic discordance in untreated CAH, where an exceptionally large adrenal mass suggestive of myelolipoma or adrenocortical carcinoma proved on histology to be an adenoma with fatty metaplasia.

A 37-year-old woman with salt-wasting CAH diagnosed in the neonatal period, off treatment since age 18, was admitted after trauma. Trauma computed tomography revealed a cystic-solid

left adrenal mass (119 mm × 92 mm × 67 mm) with nodular right adrenal enlargement, initially suggestive of cancer. She was referred to the Department of Endocrinology, Diabetology and Internal Medicine at the Medical University of Bialystok, Poland for further evaluation.

Examination showed generalized hirsutism and virilization (Figure 1A and 1B). Laboratory tests showed elevated 17-hydroxyprogesterone, testosterone, and ACTH, with decreased cortisol and estradiol, a dehydroepiandrosterone sulfate (DHEA-S) within the reference range, and increased sex hormone-binding globulin (SHBG; Supplementary material, *Table S1*). Graves disease was also diagnosed. The diagnosis of salt-wasting CAH due to 21-hydroxylase deficiency, established in the neonatal period, was reconfirmed biochemically.[3]

Although DHEA-S was within the reference range, this does not exclude sustained ACTH-driven adrenal steroidogenesis, as androgen production in 21-hydroxylase deficiency also occurs through backdoor and 11-oxygenated pathways that are not adequately reflected by DHEA-S alone.[4]The elevated SHBG, atypical for hyperandrogenism, is better explained by the coexisting thyrotoxicosis, which increases hepatic SHBG, and argues against an autonomous androgen-secreting tumor.[5]

Unenhanced attenuation and washout differentiate these tumors.[1,2] These were unavailable (scan performed externally, documentation inaccessible), so magnetic resonance imaging was used, showing a large polycyclic left adrenal mass with an opposed-phase signal drop indicating lipid and contralateral lipid-containing nodules (Figure 1C and 1D). The appearances were most consistent with myelolipoma but did not exclude a lipid-rich adenoma or carcinoma.

Inability to exclude carcinoma and the large lesion size indicated adrenalectomy rather than surveillance.[1,2] An open approach was chosen over laparoscopy for a large, potentially malignant tumor to allow intact removal without capsular rupture.[2] Histopathology showed an adrenocortical adenoma (inhibin and pan-cytokeratin positive), with fatty metaplasia and lymphocytic infiltration of uncertain significance. Adrenocortical adenomas may rarely exhibit

myelolipomatous change with accumulation of mature adipose tissue within the tumor. The resulting macroscopic fat can mimic the imaging appearance of myelolipoma, whereas intracellular lipid may still produce opposed-phase signal loss characteristic of a lipid-rich adenoma, thereby explaining the discordant imaging features.[2]

Parameters normalized on fludrocortisone, dexamethasone, and methimazole, except cortisol, which remains low under dexamethasone and does not indicate undertreatment. Adrenal masses in untreated CAH require individualized assessment.[1,2] Uncomplicated hyperplasia needs no surgery, whereas a large indeterminate mass does, followed by hormone replacement, monitoring, treatment of Graves disease, and surveillance of the remaining adrenal.[1,3]

Supplementary material

Supplementary material is available at www.mp.pl/paim.

Article information

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References

1 Nermoen I, Falhammar H. Prevalence and characteristics of adrenal tumors and myelolipomas in congenital adrenal hyperplasia: a systematic review and meta-analysis. Endocr Pract. 2020; 26: 1351-1365.

2 Calissendorff J, Juhlin CC, Sundin A, et al. Adrenal myelolipomas. Lancet Diabetes Endocrinol. 2021; 9: 767-775.

3 Speiser PW, Arlt W, Auchus RJ, et al. congenital adrenal hyperplasia due to steroid 21-hydroxylase deficiency: an Endocrine Society* clinical practice guideline. J Clin Endocrinol Metab. 2018; 103: 4043-4088.

4 Yang M, White PC. Genetics and pathophysiology of classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency. J Clin Endocrinol Metab. 2025; 110: S1-S12.

5 Kjaergaard AD, Marouli E, Papadopoulou A, et al. Thyroid function, sex hormones and sexual function: a Mendelian randomization study. Eur J Epidemiol. 2021; 36: 335-344.

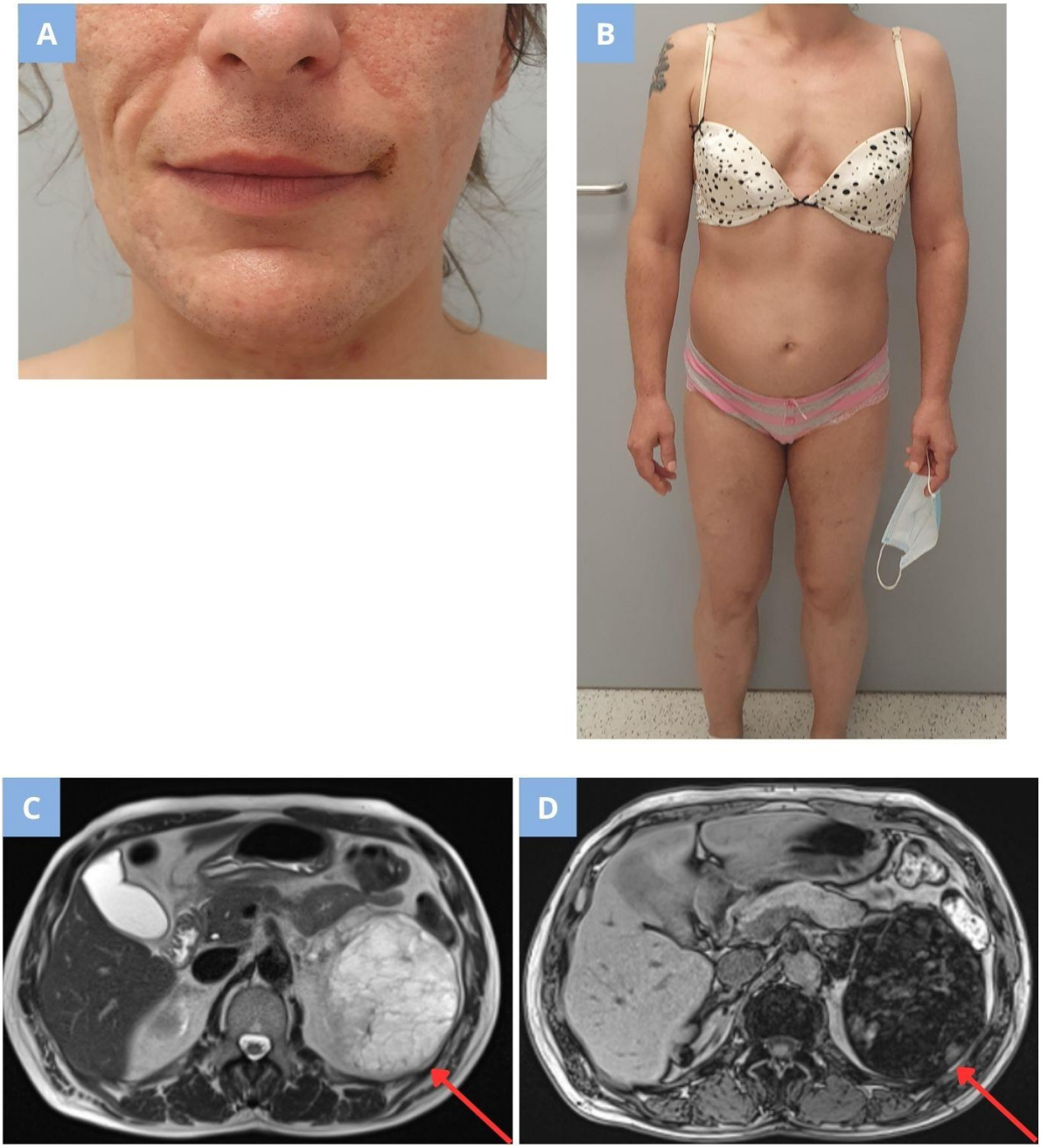

Figure 1 A, B – clinical features of hyperandrogenism in the patient, including visible hirsutism and masculinization of body habitus; **C, D** – contrast-enhanced magnetic resonance imaging (MRI) of the abdomen. A large, polycyclic retroperitoneal mass is visualized on the left side, located between the stomach, spleen, pancreas, and left kidney, causing varying degrees of displacement without evidence of invasion. The lesion measures up to 101 × 96 × 126 mm (AP × RL × HF). It demonstrates relatively high signal intensity on both T1- and T2-weighted images (Figure B – T1-weighted sequence; Figure A – T2-weighted sequence), with internal

septations and a solid component located paravertebrally, corresponding to the anatomical location of the adrenal gland. A signal drop on opposed-phase imaging is observed in the majority of the lesion, consistent with lipid content, along with a high apparent diffusion coefficient (ADC). The right adrenal gland is elongated and contains nodules (up to 16 mm in diameter) within the lateral limb, demonstrating fat content and peripheral contrast enhancement after gadolinium administration, with morphology similar to the lesion observed in the left adrenal gland.

Short title: Giant adrenal tumor in CAH