Bone and Mineral Metabolism
BONE AND MINERAL CASE REPORTS I

Severe Hypocalcemia Secondary to Pseudohypoparathyroidism
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SAT-371
Introduction: Pseudohypoparathyroidism (PHP) is a rare disorder characterized by PTH resistance due to a mutation in the GNAS gene causing decreased cyclic AMP generation. The 5 subtypes of PHP include type 1a, 1b, 1c, 2, and pseudo-PHP with type 1a being the most common. Patients with PHP present with hypocalcemia, hyperphosphatemia, appropriately elevated PTH, and suppressed calcitriol levels. PHP type 1a patients have characteristic features including obesity, short stature, round facies, and shortened metacarpals. PHP patients should be evaluated for other endocrinopathies as mutations in the GNAS gene may result in resistance to other hormones like TSH, GHRH, and gonadotropins.

Case Report: This patient is a 25 year old male who presented to clinic for evaluation of hypocalcemia. He denied any personal or family history of calcium disorders, thyroid disease, or parathyroid disease. He admitted to severe fatigue and muscle cramps for over one year leading to a car accident. He was sent to the emergency room with normal findings. He elected to terminate the high-risk pregnancy. Based upon her young age at presentation, family history of early onset hypertension, grossly elevated aldosterone: renin activity was 1.3 ng/ml/hr, with a ratio of 373, diagnostic of primary aldosteronism. Due to the markedly high ratio, a saline suppression test was deemed unnecessary for confirmation. Her serum potassium was normal at 3.6 mEq, likely due to poor renal clearance. Given renal failure, a CT non-contrast of the adrenal glands was performed with normal findings. She elected to terminate the high-risk pregnancy.

Adrenal
ADRENAL CASE REPORTS II

The Creatinine, the Crib and the Manometer - Navigating the Labyrinth of Primary Aldosteronism
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SUN-191
A 21-year-old Ethiopian female with a five-year history of hypertension presented to medicine clinic with headaches and fatigue for two weeks. She was hypertensive to 163/113 mmHg. She had recently moved to the US and no prior medical records were available. She had been taking an unknown antihypertensive until three weeks prior. She was found to have a creatinine of 3.49 mg/dL. Renal ultrasound revealed bilateral, small echogenic kidneys without any evidence of renal artery stenosis. An intrauterine pregnancy was also incidentally discovered. Her aldosterone level was elevated to 486 ng/dL and her renin activity was 1.3 ng/ml/hr, with a ratio of 373, diagnostic of primary aldosteronism. Due to the markedly high ratio, a saline suppression test was deemed unnecessary for confirmation. Her serum potassium was normal at 3.6 mEq, likely due to poor renal clearance. Given renal failure, a CT non-contrast of the adrenal glands was performed with normal findings. She elected to terminate the high-risk pregnancy.
Primary aldosteronism (PA) usually presents with recall-citrant hypertension, hypokalemia and an elevated aldosterone: renin ratio. It is commonly attributed to adrenal adenomas or hyperplasia with familial hyperaldosteronism (FH) remaining a rare etiology. FH is sub-divided into glucocorticoid remediability, type I, and non-glucocorticoid remediability, types II – IV. The initial diagnosis of such a condition during pregnancy and in the setting of worsening kidney disease presents a diagnostic and management challenge as this precludes adrenal vein sampling and contrast imaging. Our case highlights the importance of early screening for PA and illustrates the need for updated guidelines on aldosteronism workup in the setting of ESRD and pregnancy.

Tumor Biology
ENDOCRINE NEOPLASIA CASE REPORTS I

Carney Complex: A Case of a Rare Multiple Endocrine Neoplasia Misdiagnosed as Peutz-Jeghers Syndrome
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SUN-940

Carney Complex (CNC) is an extremely rare multiple endocrine neoplasia caused by germline inactivating mutation in protein kinase A type I-alpha regulatory subunit (PRKAR1A gene). Mode of inheritance is mostly autosomal dominant; 25% of cases are due to de novo mutations. Only 750 world-wide cases have been reported. Most patients are diagnosed in the second or third decade. Clinical features include cutaneous myxomas, angiomyxoid nodules, lentiginous skin pigmentation, cardiac myxomas, and benign and rare malignant endocrine tumors. These endocrine tumors include and are not limited to prolactinomas, thyroid tumors, primary pigmented nodular adrenocortical disease (PPNAD), and large cell-calcifying Sertoli cell tumors (LCCSCT). Diagnosis is often challenging as disease manifestations can occur sporadically over a large span of time, and patients may present with various conditions such as Cushing syndrome, like our case. We present a case that demonstrates the importance of early recognition of this rare disorder.

A 28-year-old Caucasian male with PMH of HFrEF, HTN, Sertoli cell tumor status post orchiectomy, vertebral fractures, and surgical removal of lip angiomyxoma presented to clinic for hypogonadal. Physical examination revealed marked Cushingoid features and facial lentigines above his eyes and on his lips. His ecletic medical history and unique exam findings lead to finding of a unifying diagnosis. His labs revealed severe Cushing syndrome, and computed tomography (CT) of his abdomen was performed due to ACTH independent hypercortisolism, demonstrating a bilateral lobular appearance of the adrenal glands. Combination of labs and physical exam findings of lentigines, skin myxomas, cushingoid features, rare angiomyxoma, LCCSCT and hypercortisolism lead to diagnosis of Carney Complex. He was misdiagnosed with Peutz-Jeghers in his adolescence due to LCCSCT and mucosal lentigines; therefore, hormonal screening was not routinely performed. Untreated Cushings led to severe osteoporosis with vertebral fractures and heart failure. Treatment included bilateral adrenalectomy. Pathology report confirmed rare PPNAD. PPNAD and LCCSCT are extremely rare tumors almost exclusively linked to Carney Complex. Interestingly, family history did not reveal endocrine disorders, cancers, or severe illnesses. Genetic testing returned positive for the PRKAR1A gene mutation. Given the consequences of untreated hormonal aberrations seen in this disorder, an early and accurate diagnosis is imperative.

Thyroid
THYROID CANCER CASE REPORTS II

Papillary Thyroid Carcinoma Arising in a Thyroglossal Duct Cyst: A Case Report
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Introduction: Thyroglossal duct cysts (TGDCs) are uncommon benign congenital entities. Rarely, thyroid carcinoma can arise from a TGDC; the most common being papillary thyroid carcinoma (PTC). Similar to TGDC, carcinomas originating within them can present as an asymptomatic midline neck mass. Signs of malignancy include dysphagia, dysphonia, weight loss, and rapid growth. Given the rarity of TGDC carcinomas, clinical management remains controversial, particularly regarding the requirement for total thyroidectomy.

Case: A 52-year-old female with history of an anterior central neck mass initially noted in 2017. A 0.3-cm left lobe mid-segment cyst and a complex thyroglossal avascular simple cyst measuring 2.4 × 1.1 × 1.8 cm was observed during ultrasound (US). She presented to the endocrinology clinic in April 2019 due to progressive enlargement of the mass. Repeat thyroid US revealed that the cystic structure had become complex with a peripheral solid component and measured 3.3 × 2.1 × 2.2 cm. FNA was performed and found to be suspicious for PTC (Bethesda category V) and positive for the BRAF V600E mutation. Patient was referred for surgical evaluation. Physical examination revealed a midline anterior 10-cm, painless, and fixed mass above the thyroid that moved with deglutition and tongue protrusion. Contrast computed tomography scan showed a large multicellular cystic structure measuring 4.1 × 4.4 × 5.9 cm. A lobulated soft tissue mass measuring 2.2 × 2.4 × 3.0 cm was noted internally along the inferior margin of the cyst. She underwent en-block resection of the TGDC in addition to a total thyroidectomy. Histopathological examination identified a 7.5 × 5.5 × 5.0 cm cystic mass with fluctuation and a firm, solid area measuring 4.1 × 4.4 × 5.9 cm. Thyroid gland examination was otherwise unremarkable. No areas of extension of the mass into the thyroid tissue were clearly identified and no other gross lesions were observed. The solid area within TGDC contained a tumor with findings characteristic of PTC. Postoperatively, she was placed on thyroid hormone replacement therapy.