of chromosome 14, and 1 sporadic case with epimutation at *DLK1* locus.
Conclusion: Pathogenic genetic defects were identified in 11.8% of patients with apparently idiopathic CPP involving four distinct genes. Altogether, these genetic findings indicate a context of changing in the distribution of the etiological diagnosis of CPP in both sexes, highlighting the genetic causes.

Bone and Mineral Metabolism

BONE AND MINERAL CASE REPORTS II

Presumed Parathyroid Infarction Leading to Remission of Primary Hyperparathyroidism: A Rare Clinical Occurrence

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Background: Infarction of a parathyroid adenoma, also known as 'parathyroid auto-infarction' or 'parathyroid apoplexy,' is a rare condition that can present and lead to acute and dramatic reduction of calcium (Ca2+) and parathyroid hormone (PTH) levels and spontaneous remission of primary hyperparathyroidism (PHPT). Some patients may experience neck pain, dysphagia, hoarseness, anterior neck swelling or ecchymoses, while others are asymptomatic. Ultrasound can show the lesion getting larger due to hemorrhage, but weeks to months later, there can be a size decrease of the adenoma due to loss of blood supply and necrosis. Sestamibi scans can also show non-localization on serial exams. Few case reports have evidence of infarction on pathology.
Clinical Case: A 38 year old man with no significant past medical history presented after a pedestrian-motor vehicle accident with polytrauma, including a cervical spine injury requiring tracheostomy and immobilization of his neck. Over the first week of his hospitalization, his Ca2+ rose as high as 14.0 mg/dL (8.9-10.2 mg/dL). Concurrent PTH level was 233 pg/mL (12-88 pg/mL) and 25-OH vitamin D level was 14.8 ng/mL (20.1-50.0 ng/mL). A neck ultrasound showed a hypoechoic nodule measuring 1.4 x 1.2 x 1.6 cm posterior to the superior aspect right thyroid lobe. A sestamibi scan with SPECT-CT showed a persistent focal activity in the region of right thyroid bed. He was treated with aggressive intravenous (IV) hydration, 7 doses of intranasal calcitonin 500 units, multiple doses of furosemide 40-80 mg IV, pamidronic acid 90 mg IV, and eventually transitioned to cinacalcet 30 mg twice daily. Due to his C-spine injury, parathyroid surgery was deferred.
Four months later, the patient developed acute muscle spasms. He denied anterior neck pain, dysphagia, bruising, or swelling. Ca2+ level was checked and found to be 7.0 mg/dL. Cinacalcet was decreased, and eventually had to be discontinued. His serial Ca2+ and PTH levels normalized to 9.8 mg/dL and 55 pg/mL, respectively. A repeat 25-OH vitamin D level was replete at 31.1 ng/mL. A follow up ultrasound redemonstrated a slightly ill-defined hypoechoic nodule, now only measuring 0.9 x 0.9 x 1.4 cm along the the right thyroid. A sestamibi scan was also obtained and did not localize any lesion.
The biochemical and imaging findings were most consistent with a parathyroid infarction resulting in spontaneous remission of PHPT. Now one year following his initial presentation, he remains normocalcemic.
Clinical Lesson: The differential diagnosis for a sudden remission of PTH-dependent hypercalcemia is limited. Parathyroid infarction is a rare condition with paucity of data regarding follow up, but these patients likely need to remain under close long term clinical and biochemical surveillance as recurrence has been documented in the literature.

Adrenal

ADRENAL PHYSIOLOGY AND DISEASE

LC-MS-Based Profiling of Adrenal Steroids Reveals Metabolic Signatures of 17á-hydroxylase Deficiency

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LC-MS-based profiling of adrenal steroids reveals metabolic signatures of 17α-hydroxylase deficiency
The comprehensive metabolic signatures of adrenal steroids are necessary to understand their pathophysiological functions in adrenal diseases, such as Cushing's syndrome (CS) and congenital adrenal hyperplasia (CAH). The 17α-hydroxylase deficiency (17α-OHD) CAH, accounted for <1% of CAH cases, is caused by mutations of CYP17A1 gene leading to impaired production of cortisol and adrenal androgens. It may be under-diagnosed in patients in whom routine screening for the early detection of CAH subtypes. A validated liquid chromatography-mass spectrometry (LC-MS)-based quantitative profiling of 27 adrenal steroids in human serum, therefore, has been developed and employed in patients with CS (n = 7) and 17α-OHD CAH (n = 1). In a patient with 17α-OHD, adrenal androgen levels were significantly decreased, especially DHEA sulfate (~1/1,000 times), while pregnenolone sulfate was increased against both healthy (n =43) and CS subjects ($p < 0.001$). In addition, increased mineralocorticoids and decreased glucocorticoids as well as DHEA-S/Preg-S were observed in a 17α-OHD patient, which mean DHEA-S, Preg-S, and these metabolic ratios could be good biomarkers for detecting 17α-OHD CAH in part of an overall plan of medical care. The developed LC-MS method can quantitatively profile biologically active adrenal steroids and sulfate conjugates in a single run to be a comprehensive diagnostic tool in adrenal diseases.