Conclusion: Novel genetic defects were identified in 23% cases of CPP associated with complex phenotypes. Three chromosomal regions represented loci potentially implicated in CPP: Xp22.33, 7q11.23 and 1p31.3. Five genes were identified as candidate genes associated with CPP: NFIA, ARELI, TNRC6B, MKKS and UGT2B4.

Thyroid

ThYROID DISORDERS CASE REPORTS III

Evaluating Conflicting Thyroid Function Tests in New Admissions: Discordance of TSH, Free T4 and Clinical Status: A Clinical Challenge!

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Background: It is not uncommon to encounter patients whose thyroid function tests (TFTs) seem mutually inconsistent or inconsistent with a patient's clinical status, At times, the simplest reconciliation of the findings invokes a rare disorder that we are hesitant to accept. In this case, a clinically euthyroid patient presents with elevated TSH and Free T4 (FT4) suggestive of a TSH-producing tumor or of Thyroid Hormone Resistance.

Clinical Case: A 72 yr-old man with cardiomyopathy on amiodarone is admitted to the Medical Service for treatment of anasarca. He had no symptoms or signs of thyroid dysfunction and was not taking L-T4, amphetamines or propranolol. Findings on exam included normal VS, runs of atrial tachycardia, and edema from feet to scrotum. Thyroid exam was normal. Serum creatinine was 2.24 mg/dl (NL: 0.67-1.17). Bili was 2.7 mg/dl (NL: 0.67-1.17); AST and ALT were normal, Chest x-ray revealed cardiomegaly with clear lung fields. Thyroid ultrasound revealed a normal size gland containing a few sub-centimetric nodules. On Day 2 The serum FT4, by analog assay, was elevated at 1.66 ng/dl (NL: 0.76-1.46) and TSH was elevated at 9.06 mIU/L (NL: 0.35-3.74), Anti-peroxidase and anti-thyroglobulin antibodies were negative. The Medical Service diagnosis was “amiodarone-induced thyroiditis.” The amiodarone was discontinued and diuresis was induced with bumetanide, Endocrinology consultation was requested On Day 4 the FT4 and TSH were still elevated at 1.62 and 10.1, respectively. FT4 by dialysis was not elevated at 1.62 ng/dl (NL: 0.9-2.2). The FT3 was 2.34 pg/ml (NL: 2.18-3.98). On Day 5 Anti-thyroxine antibodies and Thyroid Stimulating Immunoglobulins (TSI) were negative. Paired TSH samples with and without neutralization of Human Anti-Mouse Antibodies (HAMA) were identical: both elevated at 8.30 mIU/L (NL: 0.4-4.50). Serum Iodine was markedly elevated at 2288 mcg/L (NL: 52-109).

The FT4 levels by analog assay therefore appear to have been falsely elevated (as indicated by the dialysis assay) though not by recognized factors such as thyroxine antibodies, amphetamines or propranolol. Continued
observation is necessary to further assess the transience of the post-admission TFTs.

**Conclusion:** In patients admitted to Acute Medical or Psychiatric Services, most combinations of high or low TSH and FT4 have been reported as well other aberrations of “non-thyroidal illnesses.” In patients with conflicting TFTs at admission, especially those who are clinically euthyroid, it is generally better to allocate a few weeks for observation and monitoring than to immediately launch into searches for rare disorders. This is especially important when multiple potentially thyro-active clinical states exist, such as renal and hepatic compromise, amiodarone use, and highly elevated iodine levels.

### Thyroid

**THYROID CANCER CASE REPORTS I**

**A Very Rare Case of Thyroid Cancer**

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**Background:** Cribiform-Morular variant of Papillary Thyroid Cancer (PTC-CMV) is an exceedingly rare subtype of thyroid cancer that predominantly affects younger females. As the name implies, it is a papillary thyroid carcinoma with predominantly cribriform and morular pattern of carcinoma cells on cytopathology. While completion thyroidectomy is usually recommended for larger and higher-risk Papillary Thyroid Cancer (PTC), surveillance may be acceptable with PTC-CMV, which tends to be a less aggressive malignancy. Clinical Case: A 46-year-old Guyanese woman presented with a three week history of an enlarging right-sided neck mass associated with a globus sensation while swallowing food. She denied any history of radiation exposure. Her exam findings were positive for a tender, right-sided neck mass. CT neck without contrast revealed a 4.1 x 4.0 x 5.9cm heterogeneous mass within the right thyroid lobe causing mild tracheal deviation to the left. Ultrasound of thyroid gland showed a solid heterogeneous hypoechoic 4.22 x 2.39 x 2.46cm right lobe nodule with no microcalcifications, border irregularity or taller-than-wider morphology. Fine Needle Aspiration of the nodule came back as Atypia of Undetermined Significance. The patient then underwent a core needle biopsy. The resultant pathology was negative for thyroid carcinoma or medullary thyroid carcinoma but was suggestive of a bronchial cleft cyst versus bronchogenic cyst with atypical glandular proliferation. She subsequently underwent a right hemithyroidectomy which revealed a final pathological diagnosis of a 3.5cm PTC-CMV. Such pathology warranted the patient to undergo a colonoscopy which was negative for Familial Adenomatous Polyposis (FAP). Given her negative GI workup and non-contributory family history for colonic polypsis or carcinoma the decision was made to continue surveillance rather than performing completion thyroidectomy as the disease was presumed to be sporadic.

**Discussion:** PTC-CMV accounts for 0.2% of all PTC. It is associated with FAP in more than 50% of cases but can also occur sporadically. This subtype of PTC generally follows a less aggressive course. Review of current literature revealed several case series of CMV-PTC patients. In the largest one, 32 cases were observed over a 19 year period and only two out of twelve patients with FAP-associated PTC-CMV initially treated with hemithyroidectomy developed recurrence to the contralateral lobe. Interestingly, none of the remaining patients with the sporadic type developed recurrence suggesting that completion thyroidectomy may not be mandatory in this group. It is, therefore, critical to identify these patients and screen them with a colonoscopy to avoid the potentially unnecessary resection of the contralateral lobe and the consequent need for thyroid hormone replacement.

### Adrenal

**ADRENAL CASE REPORTS I**

**Adrenal Myelolipoma in a Patient with Sickle Cell Disease**

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**Background:** Adrenal myelolipoma is a rare benign tumor of the adrenal gland, consisting of adipose tissue and hematopoetic elements. It has been reported to be associated with many chronic diseases such as haematological disorders. The majority of the adrenal myelolipomas are diagnosed in the fourth-sixth decades of life, usually as incidental findings. Despite its benign behavior, it may cause difficulties in the differential diagnosis.

**Clinical Case:** A 33-year-old woman was found to have a right adrenal incidentaloma in a kidney ultrasonography, during work-up for suspected kidney pathology ultimately not confirmed. Her past medical history was significant for sickle cell disease. There was no history of hypertension nor personal or family history of endocrine diseases. The physical examination was unremarkable. The ultrasonography revealed a mass above the right kidney, measuring 5.5 cm in diameter, markedly heterogeneous and hypoechoic in the center. On CT imaging, one month later, this mass measured 6.2x4.3cm. The absolute contrast washout of 52% was indeterminate for adrenal adenoma. The MRI, twelve months later, showed a well-demarcated and heterogeneous tumor with 8x4.7x4cm, with fat areas, suggesting adrenal myelolipoma, but not excluding the possibility of a malignant lesion, such as liposarcoma. The mass was in contact with the liver although not invaded it nor the kidney or inferior vena cava. There was no evidence of metastatic disease. Basal biochemical work-up did not disclose hormonal hypersecretion. The ACTH level was 19.6 pg/ml (N7.2-63.3); free urinary cortisol (24 hours) was 101 μg/24h (N36-137) and the overnight 1 mg dexamethasone suppression test was also normal (0.5 μg/dL). Serum metanephrines were within the normal range: metanephrine 7.8 pg/mL (N <65) and