2019 the patient skipped levothyroxine replacement due to lapse in prescription refill and TSH increased to 28 ulU/mL. Around that time, she noticed an enlarging right sided neck mass. MRI of the neck showed an enhancing mass posterior to the submandibular gland measuring 1.6 x 1.4 cm. Fine needle aspiration in August 2019, showed a mixed population of lymphocytes and few benign appearing glandular cells. Pathology obtained after resection of the right neck mass in September 2019 revealed benign thyroid tissue with no lymphoid tissue, staining positive for TTF-1 and PAX-8.

Conclusion: TG deficiency has an incidence of 1:100,000 live births and is more common in cases of genetic consanguinity. There have been over fifty mutations of varying penetrance identified leading to differing consequences in TG production and processing. These mutations can cause structural changes in the TG protein, decreased tyrosine residue on TG leading to poor iodination and ineffective TG transport within the follicular cell. We propose that the right sided neck mass was regrowth of residual thyroid tissue due to the stimulatory effect of TSH after missed doses of levothyroxine caused TSH elevation. Our case is the first of TG deficiency wherein the patient developed ectopic thyroid tissue proliferation after a period of thyroid hormone deficiency.

Reproductive Endocrinology

SEX, GENDER, AND HORMONES

Duration of Testosterone-Induced Acyclicity Influences Corpora Lutea Formation and Stromal Changes in a Transgender Mouse Model

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OR27-03

The impact and reversibility of long-term gender-affirming testosterone (T) therapy on the reproductive axis of transgender men has not been well-established. Little is known about outcomes for transgender men interested in pausing T therapy to harvest oocytes or get pregnant. We previously established a translational mouse model to investigate T-induced acyclicity and ovarian perturbations. We hypothesized that the duration of T-induced acyclicity would impact the reversibility of cyclic and ovarian changes. To test this hypothesis, T-treated mice were assigned to two groups: (SHORT) 6 weeks of T therapy with immediate reversibility (1.5 mg T propionate pellet implant/ removal, n = 5) and (LONG) 6 weeks of T therapy with a prolonged T washout phase (subcutaneous oil injections of T enanthate at 0.9 mg once weekly, n = 5). Control groups (placebo pellets n = 5, sesame oil vehicle injections n = 5) were run in parallel. Estrous cycles were monitored using daily vaginal cytology. Following cessation of T therapy, mice were sacrificed in diestrus after resumption of cyclicity for 4 cycles and ovarian histology examined. Data were analyzed in GraphPad Prism using Welch’s t-test or Mann-Whitney where appropriate. T therapy led to persistent diestrus within a week after T administration for all T-treated mice and none of the controls. The total duration of acyclicity was 6±1 weeks for the SHORT group, which was significantly shorter than the 11±2 weeks for the LONG group (mean ± s.d., p = 0.0079). With resumption of cyclicity, both the SHORT and LONG groups had a significantly lower percentage of days in estrus and higher percentage of days in metestrus as compared to their parallel age-matched controls. Ovarian histology for the SHORT group all showed regular corpora lutea and minimal stromal changes, however, 3/5 mice in the LONG group lacked corpora lutea and 4/5 revealed marked stromal cell hypertrophy. Similar stromal cell changes were not seen in control mice. In conclusion, the length of time of T-induced acyclicity appears to impact the development of stromal cell hypertrophy and formation of corpora lutea even after resumption of cyclicity with similar alterations to the estrous cycles. These findings may have clinical relevance for transgender men interested in fertility, based on duration of gender-affirming T therapy. Future work will aim to separate out the respective contributions of T exposure and acyclicity to the stromal phenotype.

Adrenal

ADRENAL - HYPERTENSION

Identification of a New Heterozygous Germline ARMC5 Deletion in a Familial Case of Primary Bilateral Macronodular Adrenal Hyperplasia Co-Secreting Cortisol and Aldosterone

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MON-209

Context. Approximately 50% of familial cases of primary bilateral macronodular adrenal hyperplasia (PBMAH) are caused by mutations in the ARMC5 gene.

Case report. We report the case of a 37-year-old patient of Haitian origin, who presented with resistant hypertension. His workup showed high aldosterone (410 pmol/L) with suppressed renin levels (0.2 ng/mL/h) with an aldosterone to renin ratio (ARR) of 2050. Patient also had suppressed ACTH levels (<0.5 pmol/L (N: 2-12)) and high UFC (1103 nmol/d (N: <330)). He had an aberrant cortisol and aldosterone response to catecholamines and vasopressin (V,R). An abdominal CT scan showed bilateral enlargement of adrenal glands and a 3.3cm dominant nodule on the right gland. Moreover, a 2.8cm mass on the pancreatic tail was present. Patient underwent left laparoscopic adrenalectomy and distal pancreatectomy; the pathology confirmed PBMAH and a pancreatic neuroendocrine tumor (NET). Following surgery, ARR and UFC remained high and patient was treated successfully with B-blockers and MR antagonists. A head MRI showed no sign of intracranial meningiomas.

Genetic analyses. Following genetic counselling, MEN1 gene analysis was performed using sequencing/MLPA techniques but did not reveal a mutation. Initial genetic testing included ARMC5 gene analysis using direct Sanger sequencing which was negative. However, using Next-Generation Sequencing (NGS) and MLPA analysis, a
Bone and Mineral Metabolism
PARATHYROID HORMONE TRANSLATIONAL AND CLINICAL ASPECTS

Preoperative Parathyroid Ultrasound Imaging - Pitfalls and Ways to Improve Diagnostic Accuracy
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SAT-390
Introduction: Parathyroid ultrasound (US) is commonly used for pre-operative imaging to facilitate focused parathyroid surgery. It provides point-of-care imaging without ionizing radiation and is less expensive compared to nuclear sцинтigraphy or computed tomography (CT). Parathyroid US is, however, operator skill and experience dependent. Methods: The charts of all patient who underwent parathyroid surgery between 2016 and 2018 were reviewed. Investigators reviewed the pre-operative US images and correlated these findings with pathology reports, operative notes and with results of CT and nuclear sцинтigraphy imaging. The US characteristics of parathyroid lesions were described. Results: In total 146 patients underwent parathyroid surgery during the three-year study period. The average age of the cohort was 55.1 +/- 15.1 years and the male to female ratio was 1:2.6. The average pre-operative serum calcium and PTH levels were 11.6 +/- 0.9 mg/dL and 310.9 +/- 305 pg/ml, respectively. 134 out of 138 patients with preoperative PTH US had images available for review by investigators. Compared to the pre-operative read that identified 106 lesions, 19 additional parathyroid lesions were identified: seven (36.8%) were easily identifiable lesions with typical US features, three (15.8%) were easily identifiable lesions with atypical US features, five (26.3%) were lesions adherent to the thyroid gland and nine (47.4%) were small lesions that were likely only identified by the investigators due to knowledge of the final pathology and intraoperative findings. Forty-seven parathyroid lesions could not be identified by investigators and one or more of the following reasons were determined as possible explanations: lesion was small in size (14.9%), presence of a large thyroid gland (27.7%), location of the lesion deep in the neck or at an ectopic location (21.3%). The quality of stored images was inadequate in 31.9% of these cases. After review, 67.1% (compared to the actual detection rate of 61.3%) of parathyroid lesions should have been identified on preoperative US. Sixty percent of parathyroid lesions were left sided and 66.9% were inferior in location. The shapes observed were oval (48.8%), conforming (50.4%) or elongated (<1%) and echogenicity was hypoechoic (86.8%) or isoechoic (13.2%). Only a small percentage had a target sign (6.6%) or were partially cystic (7.4%). 74.8% demonstrated a feeding vessel and 33.6% had vascular arborization or scattered vascularity. Conclusions: While parathyroid lesions with typical US feature and locations are easily identified, the common reasons for failure to identify a lesion include atypical features, small size, slender configuration and adherence to the thyroid gland. Being aware of these possibilities can improve detection rate. Parathyroid lesions are also less likely to be identified when present deep in the neck or at an ectopic location outside of the neck.

Thyroid
THYROID DISORDERS CASE REPORTS II

Association of Myotonic Dystrophy with Autoimmune Endocrinopathies and Thyroid Carcinoma
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SAT-510
Myotonic dystrophy (MD) is a multisystemic, autosomal dominant disorder associated with progressive muscle weakness, premature cataracts, frontal baldness, and cardiac disturbances. MD has been associated with several endocrinopathies including primary testicular failure, autoimmune endocrinopathies (hypothyroidism, hyperthyroidism, multinodular goiter, and Addison’s disease), thyroid carcinoma (primarily papillary), insulin resistance, and type 2 DM. Development of diabetes is thought to be related to formation of an insulin-resistant receptor because of aberrant regulation of mRNA. We describe the first reported case of a patient with MD associated with type 1 diabetes mellitus, Hashimoto’s thyroiditis with hypothyroidism, and follicular variant of papillary thyroid cancer. A 49-year-old female presented with acute congestive heart failure. The patient had history of type 1 DM diagnosed at the age of 26, complicated by mild background retinopathy, peripheral neuropathy, and nephropathy with microalbuminuria. The patient first noticed proximal muscle weakness 1 year ago that gradually progressed resulting in multiple falls. She had history of bilateral cataracts status post cataract extraction at age 26. She also had progressive dysphagia requiring PEG placement, and cognitive dysfunction with mood disorder and depression. Family history was significant for heterozygous germline ARMC5 deletion of exons 5-8 was identified. The deletion is predicted to prematurely truncate the protein product and cause loss of function. The ARMC5 deletion segregated with the disease in his 24 year old son who had bilateral adrenal adenomas that appeared to be non-functional. The patient’s father was also known for having bilateral adrenal masses and hypertension. To our knowledge we report the second case of ARMC5 deletion in familial PBMAH. Suzuki et al. reported two patients, a mother and her son, carrying ARMC5 deletion of exons 1-5 and interestingly they were also affected by PBMAH co-secretting cortisol and aldosterone (1). As in this case report, the ARMC5 deletion was missed using Sanger sequencing initially.

Conclusion. These cases demonstrate that large deletions may be missed by Sanger sequencing and that the real prevalence of ARMC5 mutations may have been underestimated. The link between deletion of ARMC5 and correlation with PBMAH co-secretting aldosterone and cortisol remains to be determined but may be a step forward for genotype-phenotype correlation.

1.Suzuki S, et al. Endocrine practice: official journal of the American College of Endocrinology and the American Association of Clinical Endocrinologists. 2015;21 (10):1152-60.

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