

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 yo 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-secreting 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-secreting 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.

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 scintigraphy 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 scintigraphy 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, 3 (15.8%) were easily identifiable lesions with atypical US features, 5 (26.3%) were lesions adherent to the thyroid gland and 9 (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 I 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 I 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 myotonic dystrophy in both maternal aunt and uncle as well as 2 cousins. EMG confirmed myotonia however genetic testing was not obtained due to cost. Due to her cognitive dysfunction and depression, she had difficulty to control diabetes with HbA1c of 9.9%, and multiple previous admissions for DKA. She was status post total thyroidectomy in 2008 for follicular variant of papillary carcinoma and Hashimoto's thyroiditis followed by I-131 therapy in 2009 and maintained on levothyroxine suppression therapy. Most recent Tg and Tg Ab were undetectable. On physical exam, the patient had a narrow, sallow face with temporal muscle atrophy, percussion myoclonus involving the thenar eminence of the hands, but no frontal balding. Work up showed LVEF of 20-24% with regional hypokinesis that led to catheterization and PCI to LAD. The patient had recurrent NSTEMI which eventually resulted in CABG 1 year after presentation. The association of autoimmune endocrinopathies, thyroid carcinoma and MD suggests a possible cause and effect relationship between these disorders. In patients with diabetes and MD, previously described insulin resistance as well as cognitive dysfunction can hinder good glycemic control increasing risk for complications. Although patients with MD are typically treated by neurologists, evaluation and therapy of endocrine dysfunctions are also necessary.

Bone and Mineral Metabolism

BONE AND MINERAL CASE REPORTS I

A Case Series: Discovery of Parathyroid Carcinoma During Parathyroidectomy for Primary Hyperparathyroidism

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SAT-367

Introduction: Parathyroid carcinoma is rare and represents <1% of patients diagnosed with hyperparathyroidism (1).

Clinical Cases: We present two cases of incidentally diagnosed parathyroid carcinoma during parathyroidectomy for primary hyperparathyroidism.

A 69-year-old female was referred for hypercalcemia of 10.7 mg/dL (normal range 8.4-10.2). She had bone pain, fatigue, and mild depression. She was taking triamterene-hydrochlorothiazide, vitamin D 2000 international units daily, and 1 caltrate daily. Her thiazide diuretic was discontinued, and a repeat calcium was 9.5 mg/dL with a PTH of 79 pg/mL (normal range 14-64). Vitamin D and renal function were normal. A 24 hour urine calcium was elevated at 706 mg/24 hours (normal range 100-321). A bone density revealed osteopenia.

Based on the high urine calcium, a thyroid ultrasound was completed and showed an enlarged right parathyroid gland. Intraoperatively, the surgeon found a superior parathyroid gland adherent to the local soft tissues with recurrent laryngeal nerve entrapment. The right superior parathyroid and right thyroid lobe were resected. Pathology demonstrated an infiltrating parathyroid carcinoma. Postoperative monitoring has included: calcium, creatinine, PTH and neck ultrasound every six months without evidence of recurrence.

A 79-year-old man was referred for an approximate 18-month history of hypercalcemia. He had a prior kidney stone and constipation. He was not on calcium supplementations or thiazides. On lab testing calcium was 11.0 mg/dl with prior levels of 11.7 mg/dl. PTH was 246 pg/ml and vitamin D was 20.1 ng/ml (normal range 30-80). Imaging was obtained for parathyroid localization. A neck ultrasound and nuclear medicine study showed a right inferior parathyroid adenoma. A bone density revealed osteoporosis. Intraoperatively, the surgeon found a bilobed parathyroid extending intrathyroidal and adherent to the recurrent laryngeal nerve. The right inferior parathyroid, right thyroid lobe, and isthmus were resected. Pathology was consistent with parathyroid carcinoma. The patient had recurrent laryngeal nerve damage with persistent hoarseness postoperatively.

Clinical Lessons: Patients with parathyroid carcinoma typically present with symptomatic hypercalcemia with mean levels of 13.5-14 mg/dL and mean PTH values 8.7 times the upper limit of normal (1). Given the rarity of the condition, there are no guidelines for surveillance (1). These cases highlight atypical, mild, and early presentations of an unusual and typically aggressive disease and serve as an example of how to monitor for recurrence.

Reference: 1- Stack BC, Bodenner DL. *Medical and Surgical Treatment of Parathyroid Diseases An Evidence-Based Approach*. Switzerland: Springer, Cham; 2017. <http://link.springer.com/chapter/10.10>

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Thyroid

THYROID DISORDERS CASE REPORTS III

Remarkable Euthyroid Hyperthyroxinemia Mistaken for Thyrotoxicosis

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MON-481

TITLE

Remarkable Euthyroid Hyperthyroxinemia Mistaken for Thyrotoxicosis

CLINICAL CASE

A 46 year old caucasian female with past medical history of menorrhagia was referred from primary care for evaluation of thyrotoxicosis. Thyroid function was assessed in the context of menometrorrhagia. She did not have any history of thyroid disorder or abnormal thyroid function tests. Per outside records, recent labs demonstrated TSH 0.88 uIU/mL (0.36-3.74), Free T4 > 8.00 ng/dL (0.76-1.46), Free T3 2.9 pg/mL (2.18-3.98). All other labs were within normal limits. Thyroid ultrasound revealed normal parenchyma and volume. She did not take any medications or supplements including biotin. She denied heat intolerance, anxiety, palpitations, dyspnea, tremors, hyperdefecation, or change in hair, skin, or mood. No epiphora, diplopia, or eye irritation was reported. Her father had been diagnosed with hyperthyroidism, mother with hypothyroidism.

Repeat labs at our visit revealed normal TSH of 1.05 uIU/mL (0.358- 3.74), normal Free T3 2.58 pg/mL