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 exons1-5 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