Genetic studies revealed a mutation in the parafibromin gene - *CDC73* (also called *HRPT2*), a tumor suppressor gene, which is on chromosome 1q25. The patient currently has 6 children ranging from age 5 months to 6 years. He was advised to have his children tested any time from age 7 years for the gene mutation.
The patient has remained stable 4yrs post operatively, with normal calcium and PTH levels. He does not have any history of jaw tumor. He never had an ultrasound kidney done. He is being monitored with yearly lab tests.
Conclusion: *CDC73* gene mutation-associated disorders are inherited as an autosomal dominant fashion, with variable penetrance. This gene mutation can be found in conditions such as hyperparathyroidism jaw tumor, familial hyperparathyroidism and parathyroid cancer.
Reference:
1.
CDC73-Related Disorders: Clinical Manifestations and Case Detection in Primary Hyperparathyroidism. J Clin Endocrinol Metab. 2017 Dec 1;102(12):4534–4540.

Thyroid

THYROID DISORDERS CASE REPORTS II

NIFTP: A Painstaking Diagnosis Through the Pathologist's Eyes

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SAT-461

Non-invasive encapsulated follicular variant of papillary thyroid cancer (EFVPTC) was recently reclassified as non-invasive follicular thyroid neoplasm with papillary-like nuclear features (NIFT-P).[1] In 2018, revised and stricter criteria were proposed for a lesion to qualify as NIFT-P including no well-formed papilla or BRAF-V600E mutation.[2] We are presenting an interesting case to highlight the importance of scrutinizing pathology slides to diagnose NIFTP with these more strict criteria.
35-year-old female from Puerto-Rico was diagnosed with Graves' disease. After 2 years of methimazole treatment, total thyroidectomy was planned for definitive treatment of Graves' disease. During the work up, she was noted to have a cystic nodule in isthmus, a 1.1 cm hypoechoic nodule in left mid-lobe and a 1.1 cm isoechoic rounded mass in left level III neck, which was initially thought to be a lateral aberrant thyroid remnant. Her thyroid uptake scan was consistent with a multinodular goiter with no uptake in the extrathyroidal mass. The mass was biopsied and showed Atypia of Undetermined Significance (AUS) Bethesda III with washout positive for thyroglobulin (Tg). Total thyroidectomy with bilateral central and left lateral neck dissection was performed. The pathology showed an intrathyroidal 1.2 cm EFVPTC with predominant follicular features and <1% papillae, without tumor capsular invasion. The initial diagnosis was NIFT-P with a background of chronic thyroiditis. However, on pathology, the level III neck mass was a 2 cm metastatic node with classical PTC. ThyroSeq mutational analysis of tissue blocks for both the thyroid nodule and lymph node were positive for NCOA4-RET (RET-PTC3) gene fusion, a BRAF-V600E-like mutation found in classical PTC. On review of her pathology, the thyroid lesion was noted to have more than one papilla, though <1% papillae and was >30% solid, hence not qualifying as NIFT-P and her histological diagnosis was changed to EFVPTC. She was staged as AJCC 8th edition stage 1 with intermediate ATA risk for which she received adjuvant therapy of 101 mCi ^{131}I.
Although classification into NIFT-P has been shown to reduce overtreatment of low risk encapsulated PTC, pathology slides should be closely scrutinized to ensure fulfillment of all criteria in order for a lesion to qualify as NIFT-P. This will minimize failure to recognize PTCs, that would warrant closer follow up and surveillance for recurrence.
1. Rossi, Esther D, et al. Noninvasive Follicular Thyroid Neoplasm with Papillary-Like Nuclear Features: Update and Diagnostic Considerations—a Review. *Endocrine Pathology* 30.2 (2019)
2. Nikiforov, Yuri E et al. Change in Diagnostic Criteria for Noninvasive Follicular Thyroid Neoplasm With Papillary-like Nuclear Features. *JAMA oncology* vol. 4,8 (2018)

Pediatric Endocrinology

PEDIATRIC ENDOCRINE CASE REPORTS II

A Perplexing Case of Hyponatremia and Abdominal Pain

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MON-067

Previously healthy 20-year-old female presented with diffuse lower abdominal pain, cramping in nature, multiple episodes of emesis as well as urinary frequency. On day 2 of symptoms, she was treated for a urinary tract infection with antibiotics, as well as NSAIDs and opiates for pain relief. Her serum sodium was 133 mmol/L at this time. On day 3 of symptoms, a CT scan of the abdomen was performed however did not reveal pathology. Her serum sodium was 129 mmol/L at this time. She presented to our ED, on day 5 of symptoms, where serum sodium was down to 122 mmol/L. Despite IV fluids, her sodium continued to decrease to a nadir of 117 mmol/L. Further testing, including a serum osmolality of 242 mOsm/kg, urine osmolality of 540 mOsm/kg, and urine sodium of 207 mmol/L, was consistent with a diagnosis of SIADH. Given persistence of abdominal symptoms along with SIADH further imaging studies, including US abdomen, CT brain and Chest XR, were ordered and returned unremarkable. The constellation of SIADH along with persistent abdominal pain, with negative imaging, lead to consideration of acute intermittent porphyria as a diagnosis. Random urine porphobilinogen was found to be elevated to 147.2 mcmol/L (≤ 2.4) leading to the presumptive diagnosis of acute intermittent porphyria presenting as a neurovisceral attack. Biochemical and genetic testing is being pursued to confirm her diagnosis.
Acute intermittent porphyria is an autosomal dominant hematologic disorder characterized by deficiency in porphobilinogen deaminase, an enzyme in the heme synthesis cascade. Acute attacks are caused by accumulation of porphyrin resulting in autonomic and peripheral

neuropathy which can present as abdominal pain, urinary retention, polyneuropathy, dark urine and psychiatric disturbance. Hyponatremia is present in 25–60% of cases which is caused by SIADH or sometimes renal and gastrointestinal sodium loss. Triggers for acute attacks include medications, starvation, infections, hormonal changes and alcohol. Treatment includes avoidance of triggers, IV dextrose and high carbohydrate diet. In severe attacks, IV hemin is used.

Our patient's urinary tract infection likely triggered her acute symptoms, which was further exacerbated by treatment with NSAIDS and opiates. She developed SIADH which improved with hypertonic saline and fluid restriction.

This case illustrates the need to consider acute intermittent porphyria in the differential diagnosis of SIADH presenting with abdominal pain of unknown etiology.

Genetics and Development (including Gene Regulation)

GENETICS AND DEVELOPMENT AND NON-STEROID HORMONE SIGNALING I

The Impact of FOXA3 on Testicular Steroidogenesis

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SUN-719

The Forkhead box(Fox) transcription factors are evolutionarily conserved in organisms and regulate diverse biological processes during development as well as adult life. Among the Fox family, FoxA subfamily members *Foxa1-3* have been termed `pioneer' transcription factors as they bind both nucleosome-bound and nucleosome-free DNA targets with the same recognition site. *Foxa3* is the only member of FoxA subfamily that is expressed in both male and female gonads. In the testis, *Foxa3* is expressed in spermatids and interstitial Leydig cells. We focused our study to elucidate the role of FOXA3 in Leydig cells and its impact on testicular steroidogenesis. Expression of FOXA3 dramatically decreased in mouse Leydig cells during testicular development. In addition, the time-dependent expression of FOXA3 showed an opposite pattern to that of steroidogenic genes in cAMP-induced primary Leydig cells. Meanwhile, Nur77 is among the prime regulators of steroidogenesis in the testicular Leydig cells. Overexpression of FOXA3 in MA-10 cells (mouse Leydig tumor cell line) repressed the cAMP-induced Nur77 promoter activity, which further resulted in the reduced activity of Nur77-target steroidogenic gene promoters (*StAR, CYP17A1*and *3*β*-HSD*). Similar to above results, the expression of Nur77 and its target genes,*StAR*, *3*β*-HSD* and *CYP11A1,* were repressed by adenovirus-mediated overexpression of FOXA3 in mouse primary Leydig cells, although the expression of CYP17A1, another steroidogenic gene, was differentially affected. These results suggest that FOXA3 locally regulates the expression of steroidogenic genes through Nur77 during testicular development.

Pediatric Endocrinology

PEDIATRIC ENDOCRINE CASE REPORTS I

Nutritional Deficiency of Calcium Mimicking P Seudohypoparathyroidism

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SAT-070

Nutritional Deficiency of Calcium Mimicking Pseudohypoparathyroidism

Background: Childhood hypocalcemia in general is caused by problems associated with calcium absorption and excretion, parathyroid hormone (PTH) secretion, and vitamin D metabolism. Clinical manifestations can vary from asymptomatic hypocalcemia to life-threatening conditions including convulsions, tetany and laryngeal spasm. As many symptoms are nonspecific, laboratory tests are essential for diagnosis. Nevertheless, the causes of hypocalcemia may not be determined by simple interpretation of baseline calcium, phosphorus, alkaline phosphatase (ALP), PTH and calcidiol (25OHD).

Case presentation: We report a case of 11-month-old female with a generalized tonic type seizure with low serum calcium level (5.7 mg/dl), 25OHD (23.2 ng/mL) and calcitriol ($1,25OH_2D$) (12.83 pg/mL). Serum phosphorus (5.9 mg/dL), ALP (209 mg/dL) were above normal range and PTH (484 pg/mL) was markedly elevated. She had a problem with weaning process after age of 5 months and milk powder was her main staple diet. Pseudohypoparathyroidism (PHP) was suspected due to slightly increased serum phosphorus, however Albright's hereditary osteodystrophy manifestation was absent and no GNAS methylation defect was identified. Serum calcium was normalized by intravenous calcium-gluconate followed by oral calcium carbonate and vitamin D supplement. Two months of oral oral calcium carbonate and vitamin D supplementation alone normalized all laboratory results.

Conclusions: Severe nutritional deficiency of calcium could mimic laboratory findings of PHP, therefore clinical judgement should not be made solely on biochemical markers.

Keywords: Hypocalcemia, pseudohypoparathyroidism, ri ckets

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Cardiovascular Endocrinology

ENDOCRINE HYPERTENSION AND ALDOSTERONE EXCESS

Difference in Aldosterone Dependency Between Cardiovascular Diseases and Renal Impairments in Patients with Primary Aldosteronism.

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