had PAC > 10 ng/dl were diagnosed as PA. The proportion of unilateral PA determined by adrenal vein sampling (AVS) was higher in patients who had PAC >30 ng/dl or those with spontaneous hypokalemia who had PAC between 20 and 30 ng/dl than those who did not meet the criteria (76% vs. 17%, P<0.001). **Conclusion**: Confirmatory tests in PA could be spared in patients who have typical features of PA and these patients had a high probability of unilateral PA on AVS.

Pediatric Endocrinology

PEDIATRIC ENDOCRINE CASE REPORTS I

Hidden in Plain Sight: Rethinking Our Approach to Allan-Herndon-Dudley Syndrome

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SAT-081

Background: Allan-Herndon-Dudley (AHD) is a rare X-linked disorder with neurological manifestations secondary to a mutation in monocarboxylate transporter 8, a protein that transports T3 into nerve cells in the brain. AHD is characterized by increased serum free T3, decreased serum free T4 and normal serum TSH levels as well as the severe neurological manifestations including global developmental delay, hypotonia, and joint contractures (1). A phase 2 trial using triodyothyroacetic acid has shown promise in treating this disorder (2). We report on three children who were diagnosed by whole exome sequencing after presenting with neurological manifestations.

Clinical Cases: Patient 1 presented at 4 months to the neurology clinic for seizures. He had a normal newborn screen. Worsening developmental delays and central hypotonia prompted a brain MRI that revealed delayed myelination for age. At 6 months a chromosomal microarray and metabolic work-up were performed and were nondiagnostic. Whole exome sequencing was obtained at the age of 4.5 years revealing a mutation in the SLC16A2 gene (p.Ser210Tyr). Thyroid studies were consistent with the diagnosis.

Patient 2 presented to neurology at 9 months for developmental delay. A brain MRI was obtained which was within normal limits. At 14 months an acylcarnitine profile was obtained which indicated a possible CPT1 deficiency, which did not fit his clinical picture. Chromosomal microarray as well as work-up for inborn errors of metabolism were performed and were nondiagnostic. Thyroid studies were obtained which showed low free T4 with normal TSH. Whole exome sequencing was obtained at the age of 2.5 years, which revealed a mutation in SLC16A2 (p.R371C).

Patient 3 presented as sibling of patient 2 with known AHD syndrome. Testing for SLC16A2 was performed at the age of 5 months and returned positive for same mutation as sibling (p.R371C).

Conclusion: Allan-Herndon-Dudley syndrome is a rare neurological disease secondary to a mutation in the T3 transporter protein to nervous tissue. A high index of suspicion as well as thyroid studies should be obtained in patients presenting with central hypotonia and global developmental delay with normal newborn screens, particularly in states that use TSH as a screening test. This is especially important as treatments are becoming available that may help prevent neurological devastation seen in these patients.

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Thyroid

HPT-AXIS AND THYROID HORMONE ACTION

Influence of Smoking on Thyroid Function in Japanese Subjects: Longitudinal Study for One Year of On-Off Smoking

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SAT-437

Recent studies showed that various factors, including age, gender, race, iodine intake, obesity, the thyroid peroxidase antibody (TPO-Ab), and/or smoking, influence the thyroid status. In the present study, we analyzed and investigated the effects of these factors, particularly smoking and the thyroid peroxidase antibody (TPO-Ab) in Japanese euthyroxinemia individuals with serum free T4 level within normal range. A total of 12,289 subjects who underwent health check-ups were analyzed in a cross-sectional and longitudinal study. The mean age of subjects was 50 ± 10 years (age range: 21–88 years). Serum TSH levels and the prevalence of positivity for TPO-Ab increased with age in Japanese euthyroxinemia subjects. Mean and median serum TSH levels increased with age in smokers and non-smokers, but were significantly lower in smokers than in non-smokers among men and women in most age groups; the median 97.5th percentiles of TSH levels were 1.2 mU/liter and 2.9 mU/liter in smokers, and 1.4 mU/liter and 3.9 mU/liter in non-smokers in 31- to 40-year-old men, p<0.01, and 1.4mU/liter and 4.3 mU/liter, and 1.8mU/liter and 6.2 mU/liter in 61- to 70-year-old men, p<0.01. However, smoking had a negligible effect on serum TSH levels in women older than 50 years; 1.3 mU/liter in smokers and 1.6 mU/liter in non-smokers in 31- to 40-year-old women, p<0.01, and 1.5 mU/liter and 1.8 mU/liter in 51- to 60-year-old women, p=0.3. Furthermore, the present study confirmed that serum free T4 levels in men progressively decreased with age, whereas no significant change was observed in women. Smoking did not affect the relationship

between age and serum free T4 levels in men or women, except for men in their 20s. Serum TSH levels were significantly higher in subjects with positivity for TPO-Ab than in those with negativity at all ages and in both genders; however, smoking did not affect free T4 levels or the positivity for TPO-Ab. The rate of smokers in men was significantly higher in patients with subclinical hyperthyroidism (25%) than in those with subclinical hypothyroidism (10%, $p<0.05$). Furthermore, the results of the longitudinal study revealed a significant decrease in serum TSH levels one year after the start of smoking in men ($p<0.05$). Since smoking appears to lower serum TSH levels in Japanese euthyroxinemia subjects careful consideration of the smoking status is needed when evaluating subclinical thyroid function.

Adrenal

ADRENAL CASE REPORTS II

Acnes Fulminans in a Very Young Patient with Adrenal Carcinoma After Surgical Treatment

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SUN-165

Introduction:
Acne *fulminans* is a rare disease, which most often affects teenagers and young adults, being described associated with exogenous testosterone use and with elevated adrenal androgens in congenital adrenal hyperplasia. We describe a case of acne *fulminans* in a toddler with adrenocortical carcinoma.
Clinical Case:
A 2.3 years-old male diagnosed with adrenocortical tumor presenting pubarche, increased penis and body hair, severe acne lesions in face, chest and back and also arterial hypertension. He was previously healthy, with normal development at his first year. The investigation showed adrenal hyperandrogenism and hypercortisolism. Serum values of Total Testosterone >1500 ng/dL (<7,00 - 25,91 ng/dL), ACTH: 7,2 pg/mL (<46 pg/mL), DHEAS: 682 µg/dL (<15 µg/dL), Aldosterone: 11 ng / dL (2.5-39.2 ng /dL), Serum Cortisol 8AM: 30.65µg/dL (5.27 to 22.45 µg/dL), Salivary midnight cortisol: 29.5nmol/L (11:30PM-00:30AM < 7.6 nmol/L). The abdominal computed tomography (CT) showed the left suprarenal gland with a 5,1 x 3,9 cm lesion. Left adrenalectomy was performed, whose histology confirmed adrenocortical carcinoma, without exceeding the capsule, but with a vascular invasion focus and Ki-67 rate of 20%. Hormonal levels in the early follow up were normal and prophylactic corticosteroid therapy was progressively removed. At the three months after surgery outpatient follow-up, worsening of his skin acneiform lesions was observed. He presented painful papules, pustules and crusts at face, chest and back, with purulent exudation and bleeding lesions, associated with axillary and inguinal adenomegaly and acne *fulminans* was diagnosed. Clinical and ultrasonographic examination also showed the presence of hepatosplenomegaly,. The treatment was initially made with oral corticosteroids and antibiotics. At this time, laboratory tests of androgens, DHEAS and salivary cortisol were normal, but inflammatory markers were high. A new postoperative abdominal CT showed, at the left adrenal topography, two lesions, 47HU attenuation, 1.5 x 1.2 x 1.1cm and 2.3 x 1.7 x 2.0cm, and also confirmed mild hepatosplenomegaly. Biopsy of inguinal lymph nodes was performed, and histology showed lymphoid follicles hyperplasia.
Conclusions:
This case report showing the association between acne *fulminans* and adrenocortical carcinoma, at postoperative time, when the child already had normal serum androgens and after hypercortisolism resolution, without oral corticosteroid therapy. Disordered immune response and/or hypersensitivity reaction to *Propionibacterium acnes* antigens, are also considered as possible triggering factors, although the etiology for this cases is not well established. The description of an acne *fulminans* case in such a young patient with adrenocortical carcinoma seems unusual and such association deserves further elucidation.

Thyroid

THYROID DISORDERS CASE REPORTS III

A Rare Case of Thyroglobulin Deficiency with Ectopic Thyroid Tissue

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MON-461

Background: The production of thyroglobulin (TG) is initiated by TSH binding to the TSH receptor leading to the transcription and production of TG protein. TG protein then gets transported to the lumen of the follicular cell where its tyrosine amino acids get iodinated and coupled to form T3 and T4. TG deficiency is a rare autosomal recessive genetic condition characterized by congenital thyroid goiter, increased thyroid uptake and primary hypothyroidism.
Clinical Case: A 21 year old South Asian female with a history of TG deficiency, hypothyroidism, and thyroid goiter status post thyroidectomy presented with a right sided neck mass. She was diagnosed hypothyroidism with a goiter at the age of five. Levothyroxine replacement was initiated and she had normal growth and development. At the age of ten upon transferring care to our health network, there was concern for enlargement of the goiter. Labs showed TSH of 2.85 uIU/mL (normal: 0.36-3.74 ulU/mL), thyroid binding globulin of 18.2 ug/mL (normal: 13-30 ug/mL), TG antibody less than 20 IU/mL (normal: less than 40 lU/mL) and TG level less than 0.2 ng/mL (normal: less than 55 ng/mL). Thyroid uptake and scan showed overall increased uptake. Genetic studies revealed homozygous mutation of thyroglobulin deficiency. Further history obtained confirmed patient's parents were first cousins with heterozygous thyroglobulin mutations. Due to enlarging multinodular goiter with thyroid gland measuring 6.2 cm x 4 cm on the right and 6.2 x 3.6 cm on the left, total thyroidectomy was completed in 2012. Pathology showed benign thyroid tissue. In May