A 14 Year Old Female with Primary Amenorrhea
Varshini Chakravarthy, MD1, Sehar Ejaz, MD2.
1Nassau University Medical Center, East Meadow, NY, USA,
2Nassau Univ Med Ctr, East Meadow, NY, USA.

Background: Swyer Syndrome is an extremely rare disorder of sexual development. These patients often present with primary amenorrhea during adolescence and are phenotypically female with 46 XY chromosomes. Given the association of invasive gonadal malignancies with this disorder, suspicion should be high in patients who present with a stagnant or decreased rate of pubertal progression. We present a case of Swyer Syndrome in a 14-year-old female with primary amenorrhea in the setting of decreased pubertal progression. Case: A 14-year-old female presents with a chief complaint of primary amenorrhea. She first noticed breast budding 2 years prior but reports no significant increase in breast tissue over the last 2 years. She does not appreciate any other signs of puberty. She denies any acne, body odor, hirsutism, hair loss, or abdominal/pelvic pain. She denies any changes in her diet or physical activity and is not on any medication. No history of cancer, surgeries, or radiation exposure. There is no family history of infertility or delayed puberty. Her vitals on presentation are within normal limits. Her growth parameters are the following: weight-69.9 kilos, height-163 cm, and BMI-26.3. Physical exam shows a well-appearing adolescent with grossly female external genitalia and the breast exam is SMR II. No pubic or axillary hair appreciated on the exam. Although our patient did not meet the traditional definition of primary amenorrhea, a workup was started due to the slow progression of puberty. Initial blood testing shows normal levels of AMH (0.52 ng/mL) and inhibin A (1pg/mL) confirms suspicion for ovarian insufficiency. Chromosomal analysis and pelvic ultrasound findings of a small uterus and ovaries led to our diagnosis of Swyer syndrome. Our patient had surgical resection of both ovaries and fallopian tubes and the ovarian pathology showed gonadoblastoma with invasive dysgerminoma in both gonads. She was started on hormone replacement after gonadectomy.

Conclusion: Although Swyer syndrome is uncommon with an incidence of 1 in 80,000, this case illustrates that suspicion for Swyer Syndrome should be high in patients with slow progression of puberty and primary amenorrhea (1). Early diagnosis is critical, as patients with gonadal dysgenesis are at great risk for germ cell cancers. Though most of these patients have an identifiable genetic mutation, we were unable to elicit the exact mutation in our patient despite whole-genome sequencing. References: Jaideep Khare, Prasun Deb, Prachi Srivastava & Babul H. Reddy (2017) Swyer syndrome: The gender swayer?, Alexandria Journal of Medicine, 53:2, 197–200, DOI: 10.1016/j.ajme.2016.05.006 Varshini Chakravarthy, Seher Ejaz. A 16-Year-Old With Amenorrhea and Delayed Breast Development - Medscape - Jan 14, 2020.
After surgery, he received flare-dosing PVO at 20 mg/day for 4 weeks, then 10 mg/day for 8 weeks. Post-surgical imaging 12 weeks after the surgery showed new bridging HO at the site of intramedullary rod insertion and around the distal screw. Nine months after the fracture the patient had a second fall resulting in a right intertrochanteric fracture. He underwent intramedullary nailing of the right hip, in a modified procedure which did not require distal screw placement. PVO was increased similarly to the above flare protocol, but, at the time of fracture occurrence rather than post-surgery. He had no skin or healing complications with either treatment regimen. After each fracture the patient had prolonged recurrent flare-ups at the injury sites, significantly increasing his number of flare-ups per year. After the fractures there was new Brooker class D HO at the left hip, originating at the insertion of the intramedullary rod, and new class B HO at the right greater trochanter, again near the insertion site of the intramedullary rod compared to his pre-surgery baseline. In contrast, there was no new HO at the right distal intramedullary rod whereas HO occurred around the screw placement site at the left distal rod.

Conclusion: This case suggests that PVO in the dosing regimen received by this patient can be tolerated in an individual with FOP following major surgery. HO still occurred in this patient, particularly along the rod insertion track, suggesting that the PVO regimen may need to be optimized for surgical cases or that poly-trauma events may not be adequately blocked by the dosing regimen received by this patient. However, PVO did not negatively impact fracture healing or osteointegration, and no major skin healing effects were identified. Further investigation is needed to assess whether PVO can lead to a dose-dependent reduction in HO in the setting of trauma and surgery.

Thyroid

THYROID DISORDERS CASE REPORTS I

Dissociation of Clinical Course of Coexisting Autoimmune Hepatitis and Graves Disease

Aesha Patel, MD, Camille Stanback, M.D., Priyathama Vellanki, MD

1Emory Univ Sch of Med, Atlanta, GA, USA, 2Emory University Hosp, Atlanta, GA, USA, 3Emory University, Atlanta, GA, USA.

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Title: Dissociation of Clinical Course of Coexisting Autoimmune Hepatitis and Graves Disease

Introduction: The association between Graves’ disease and coexisting autoimmune hepatitis is well known. Treatment of autoimmune hepatitis with glucocorticoids can concurrently lower thyroid hormone levels. Additionally, recurrence of hyperthyroidism has been shown to be associated with recurrence of autoimmune hepatitis. We present a case of a patient with autoimmune hepatitis and Graves’ thyrotoxicosis, which initially improved with prednisone therapy, but thyrotoxicosis recurred during the prednisone taper while hepatitis stayed in remission.

Clinical Case: A 47-year-old female initially presented with fatigue, nausea, and jaundice. Liver enzymes showed elevated AST (1634 U/L) and ALT (1956 U/L) with total bilirubin 15.1 mg/dL. Liver biopsy was consistent with autoimmune hepatitis. Treatment was initiated with ursodiol 300 mg TID and liver enzymes improved to ALT 579 IU/L and AST 544 IU/L with total bilirubin 1.9 mg/dL. Nine months later, she presented with worsening upper and lower extremity weakness, slurred speech, abdominal pain, and nausea and vomiting. AST and ALT were again elevated to >1,000 U/L. TFTs were checked due to symptoms of palpitations and heat intolerance. TSH was 0.024 uIU/mL with elevated free T4 of 3.74 ng/dL and TSI of 435%. She was treated with prednisone, cholestyramine, and propranolol and discharged with a one-month prednisone taper starting at 60 mg daily. LFTs and TFTs normalized after one month. Cholestyramine was discontinued and prednisone was tapered to 5 mg daily. Seven months later, she had symptoms of palpitations and heat intolerance. TFTs were consistent with hyperthyroidism (TSH 0.049 uIU/mL, free T4 3.96 ng/dL). LFTs remained in normal range. Since thionamides have relative contraindications in patients with liver disease, prednisone was increased from 5 mg to 20 mg daily and cholestyramine was resumed to treat hyperthyroidism. After five months, TSH remained suppressed to 0.019 uIU/mL however free T4 improved to 1.74 ng/dL. The patient was referred for total thyroidectomy.

Conclusion: This case illustrates an example of the rarely reported occurrence of thyrotoxicosis recrudescence despite initial improvement with treatment of underlying autoimmune hepatitis. Recognition of recurrence of hyperthyroidism independent of recurrence of autoimmune hepatitis indicates the need for early definitive therapy for hyperthyroidism.

Thyroid

BENIGN THYROID DISEASE AND HEALTH DISPARITIES IN THYROID II

Subclinical Hypothyroidism in Hospitalized Patients with Chronic Heart-Failure or Chronic Renal-Failure Is Most Probably Not Due to Thyroid Disease

Amir Bashkin, MD, Wagde Abu Saleh, MD, Ohad Ronen, MD.

Guliel Medical Center, Naharia, Israel.

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Introduction: Subclinical hypothyroidism is common in chronic diseases such as heart failure and advanced chronic renal failure. It is unclear whether this is a thyroid disease or an isolated TSH elevation. The goal of this study was to investigate the prevalence of worsening thyroid function in these patients with recurrent admissions.

Methods: We performed a retrospective review of medical records of hospitalized patients in non-surgical wards from 2013–2016. First, all patients with TSH levels above the normal range (4.95 mIU/L) and up to 12 mIU/L with FT4 levels in the normal range were identified. We then investigated which of these patients were re-hospitalized at least once within at least six months. According to data from the re-hospitalization, an increase in TSH level above 12 mIU/L or initiation of levothyroxine treatment was defined as worsening of thyroid function. Patients treated with a drug affecting thyroid function or with a known thyroid disease prior to first hospitalization were excluded from the

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