1 diabetes (T1D) who presented with multiple concomitant episodes of DKA and AP and normal triglyceride levels. **Case Presentation:** The patient is a 13-year-old female with T1D who presented with two days of hyperglycemia, nausea, and diffuse abdominal pain. Initial laboratory evaluation was remarkable for point-of-care glucose of >500 mg/dL (60-99), venous pH of 7.006 (7.330-7.430), bicarbonate of < 5 mmol/L (20-28), beta-hydroxybutyrate of 5.6 mmol/L (0.0-0.8); consistent with severe DKA. She received normal saline bolus fluids and then started on the DKA protocol with improvement of acidosis, though with the persistence of abdominal pain. Due to concern for other causes of her abdominal pain, additional workup was done, notable for elevated lipase of 624 U/L (10-52), amylase of 434 U/L (25-100), and triglyceride of 121 mg/dL (30-149). An abdominal ultrasound showed findings consistent with AP, lipase levels peaked at 1753 U/L before down-trending to 959 U/L, and amylase decreased to 389 U/L. After several days abdominal pain resolved, and the patient was discharged home. The patient was readmitted six weeks and again one year later for laboratory and symptoms, including abdominal pain consistent with DKA. Both lipase and amylase were elevated during both admissions with normal triglyceride levels. Magnetic resonance cholangiopancreatography was significant for findings compatible with acute pancreatitis with no evidence of cholelithiasis or choledocholithiasis. The patient underwent genetic testing, including normal PRSS1, SPINK1, CFTR, CPA1, and CTRC. A variant of unknown clinical significance was identified in the CTRC gene (c.550G>A), which was not thought to be the cause of her recurrent pancreatitis. Interestingly, since her hemoglobin A1c has been in a better range for the past year, she did not have any recurrent episodes of pancreatitis. **Conclusion:** The insulin-deficient state associated with DKA can lead to moderate to severe HTG, which in turn can cause AP. Even though abdominal pain is a common symptom in patients presenting in DKA, one should think about other causes when the abdominal discomfort is out of proportion or not improving as acidosis resolves. Our patient had recurrent pancreatitis for unknown etiology; however, she has not had any pancreatitis episodes in the last year since her diabetes has been under better control.

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PEDIATRIC ENDOCRINOLOGY CASE REPORT

Novel Heterozygous Calcium Sensing Receptor (CASR) Genetic Variant in Child with Unique Phenotype: Hypocalcemia, Mandibular Hypoplasia, Renal Cysts and Type E Brachydactyly

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Background: There are over 230 disease-causing variants in the calcium-sensing receptor gene (*CaSR*). Gain-of-function missense mutations in *CaSR* cause Autosomal Dominant Hypocalcemia (ADH) characterized by hypocalcemia (hCa), hypoparathyroidism (hPTH), and hypercalciuria. Patients with ADH are sensitive to fluctuations in serum calcium (Ca); and supplementation with Ca and vitamin D can cause inappropriate renal calcium retention leading to hypercalcemic events and long-term renal complications. **Clinical Case**: A 15-year-old adopted (at age 18 months) Korean female was initially diagnosed with hPTH and chronic hCa after presenting with hCa seizures. Laboratory values showed hCa (7.7 mg/dL), hyperphosphatemia (7.6 mg/dL) and hPTH (< 3 pg/mL.) Initially, she was treated with Ca supplementation (20 mg/kg/day elemental Ca), and calcitriol (0.01 mcg/kg/day). She presented at age 4 with hematuria and was found to have obstructive nephrolithiasis requiring operative intervention. Renal ultrasound (US) showed bilateral medullary nephrocalcinosis. She continued treatment with Ca and calcitriol. At age 6, a thiazide diuretic and potassium citrate supplement were added due to hypercalciuria. She had recurrent nephrolithiasis and persistent nephrocalcinosis. Follow-up renal US also showed bilateral renal cysts. Biweekly laboratory evaluation demonstrated an exuberant response to calcium supplementation. Serum Ca levels oscillated between 7.0 -10 mg/dL, but she showed minimal symptoms of hCa. At age 14, she was also recognized to have submandibular hypoplasia and brachydactyly of the 4^{th} and 5^{th} metacarpals and metatarsals bilaterally and genetic testing for CaSR gene mutation was requested. Sshe developed acute kidney injury and hypercalcemia, possibly precipitated by viral illness. However, 3 weeks before, calcitriol dose was increased to 1.25 mcg twice a day (0.07 mcg/kg/day). At admission, serum Ca was 12.7 mg/dL, iPTH 5.2 mg/dL, phosphorus 4.5 mg/dL, BUN 36 mg/dL, creatinine 1.85 mg/dL. Symptoms included headache, muscle spasm and throat spasm. She received intravenous fluids and recovered, but had an extended hospital stay. Targeted genetic analysis of the *CaSR* gene was completed, and identified a heterozygous variant (c.2506G>T, p.V836L) which is predicted to be likely pathogenic and cause ADH. After CaSR gene mutation identification, the calcitriol and also elemental Ca dosing were decreased to achieve a low Ca level (~7 mg/dL) with normal urine Ca/creatinine ratio. Patient remains asymptomatic. **Conclusion**: This is the first case of a novel mutation in the *CaSR* (c.2506G>T, p.V836L) associated with ADH, brachydactyly, renal cysts, and mandibular hypoplasia. Timely genetic testing for ADH in patients with newly diagnosed hPTH can lead to changes in therapy and improved prognosis.

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PEDIATRIC ENDOCRINOLOGY CASE REPORT

Paternalism in DSD Management: A Real and Present Threat

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In 1965, a botched circumcision left Bruce Reimer, a healthy, 8-month old XY male, with a disfigured penis. At the recommendation of Dr. John Money and physicians at Johns Hopkins, the infant was reassigned to female sex and underwent an orchiectomy and vaginoplasty. The

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family renamed the child "Brenda." Unaware of her history, Brenda struggled with significant gender identity, psychological, and behavioral issues throughout her childhood and adolescence. When made aware of this history, she transitioned to male gender and assumed the name "David." After years of psychological distress, David Reimer committed suicide in 2004. Despite the myriad lessons gleaned from this tragic story, medical and surgical management of children with atypical genitalia still remains often misguided, as providers continue to assume paternalistic roles in determining sex assignment and surgical interventions. A fifteen year old XY male with Robinow Syndrome presented for evaluation of hypogonadism and urinary incontinence. At birth, the patient was discovered to have a micropenis and perineal hypospadias and was diagnosed with hypogonadotropic hypogonadism. At the recommendation of the medical team, the infant underwent bilateral orchiectomy at eight months of age followed by urethroplasty and vaginoplasty at six years of age. The child was then given a female sex assignment. At twelve years of age, the child felt discordant from the sex of rearing and wished to be identified as male—his natal, genetic sex. He transitioned to male gender and began testosterone injections. He had history of recurrent UTIs and severe incontinence requiring diaper use. He strongly desired neophallus and urethral reconstruction for improved quality of life. The patient endorsed prior depression and desires to self-harm. He had significant concerns regarding his gender presentation and transition. He shared his difficulties in continuing in the same school system with peers who knew him as a female prior to transition and was concerned about peers knowing his medical history. In the years since the famous David Reimer case, the medical system has made tremendous strides in recognizing the need for patient autonomy and shared decision-making in patients with Differences of Sex Development and genital atypia. However, the paternalistic history of this field continues to leave its indelible mark more than 20 years since David Reimer's case made headlines, as physicians continue to recommend definitive sex assignments and surgical interventions. As with the David Reimer case, the bodily integrity of this XY infant was altered in a permanent fashion with inadequate education of his family and little to no credence given to the autonomy of the child himself. We, as physicians, cannot continue to paternalistically apply John Money's concept of gender neutrality and rigidly mandate sex assignments and early surgical interventions.

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PEDIATRIC ENDOCRINOLOGY CASE REPORT

Pediatric Giant Prolactinoma Presenting With Acute Obstructive Hydrocephalus and Intracranial Hypertension

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Background: Pediatric prolactinomas (PP) are rare but represent 50% of all pediatric pituitary adenomas. Girls are affected more frequently than boys, although PP tend to be larger and more aggressive (earlier age, larger mass, and higher prolactin levels) in boys. Thus, microadenomas (tumors < 10 mm in diameter) are typical in females and macroadenomas (10–40 mm in diameter) are typical in males. Giant prolactinomas (> 40 mm in maximum diameter), an unusual subset of macroprolactinomas, are also commonly found in boys. In a large case series, the largest tumor volume reported was 93.5 cm^3. Here we report a giant prolactinoma in a female requiring V/P shunt for decompression. **Clinical Case:** A 16-year old female presented with 2 weeks of intractable headache, nausea and vomiting, vision impairment, and changes in balance described as running into stationary household objects. Historical review revealed primary amenorrhea and short stature. On initial exam, the patient had a right eye afferent pupillary defect, concern for loss of color vision, and bilateral optic nerve edema with blurred disc margins. Brain MRI showed a large lobulated mass centered in the suprasellar cistern, measuring approximately 6.4 x 5.8 x 5.7 cm with a tumor volume of 105 cm^3. There was extension superiorly, anteriorly, and laterally, with homogeneously enhancing and cystic components, and mass effect resulting in obstructive hydrocephalus. Differential diagnoses included craniopharyngioma, germinoma, and adenoma. Initial tests demonstrated prolactin of >2,000 ng/mL, with diluted result of 17,811.16 ng/mL. Morning fasting labs confirmed multiple anterior pituitary hormone deficiencies including central hypothyroidism, ACTH deficiency, GH deficiency, and hypogonadotropic hypogonadism. The patient was started on hydrocortisone and levothyroxine. Due to obstructive hydrocephalus and vision impairment, she underwent VP shunt placement for decompression. She was started on cabergoline for medical treatment of the tumor and did not require surgical resection. Repeat prolactin measurements have shown striking improvement (to 2,350 ng/ml, 824 ng/ml, and 152 ng/ml at 1 week, 1-month, and 2-month-follow-up, respectively) with central vision improved in both eyes, papilledema resolved, and resolution of headaches. **Conclusion:** Giant prolactinomas presenting with hydrocephalus and intracranial hypertension are very rare in pediatrics, especially in girls, and can vary greatly in mass characteristics and resulting hormone deficiencies. Our patient is unique with her large tumor volume and the extent of pituitary hormone deficiencies. Prolactin levels should be measured with all sellar masses, as this may prevent unnecessary invasive intervention and possibly provide prompt response to medical management.

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PEDIATRIC ENDOCRINOLOGY CASE REPORT

Pituitary Macroprolactinoma Apoplexy in a Prepubertal Girl

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