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

Nicole Legro, BS[1], Deborah Kees-Folts, MD[2], Roger Ladda, MD[2], Lina Huerta-Saenz, MD[2].

[1]Penn State College of Medicine, Hershey, PA, USA, [2]Penn State Children's Hospital/Penn State College of Medicine, Hershey, PA, USA.

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.

Pediatric Endocrinology

PEDIATRIC ENDOCRINOLOGY CASE REPORT

Paternalism in DSD Management: A Real and Present Threat

Kristen Moryan-Blanchard, MD[1], Lefkothea P. Karaviti, PhD,MD[2], Marni Axelrad, PhD[1], Paul Austin, MD[1], David Mann, MD, DBe[1].

[1]BAYLOR COLLEGE OF MEDICINE, Houston, TX, USA, [2]Baylor College of Medicine, Houston, TX, USA.

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