based on a pathogenic variant of the *SALL4* gene where an isolated growth hormone deficiency (GHD) was detected and has been successfully treated with growth hormone. **Acknowledgements:** Genetic testing was funded by AZV grant NV18-07-00283.

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PEDIATRIC ENDOCRINOLOGY CASE REPORT

Isolated Growth Hormone Deficiency as a Cause of Hypoglycemia Past Infancy

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Introduction: Hypoglycemia is a common manifestation of Growth Hormone (GH) deficiency in infancy, but is rarely seen beyond 1 year of age. Here, we describe the case of a 5 year 6- month- old child with recurrent episodes of hypoglycemia due to GH deficiency in the setting of malnutrition. **Experimental Methods / Case Presentation:** Case report and literature review **Results:** A 5y6m girl with history of atypical teratoid rhabdoid tumor, status-post surgical resection and adjuvant therapy with CNS irradiation (50.4 gray), presented with recurrent hypoglycemia despite continuous G-tube feedings. Child had undergone surgery and irradiation approximately 3 years prior to presentation. She was initially followed at the cancer survivor clinic but had been lost to follow up for 2 years. At presentation, whole blood glucose was 51mg/dL. Height was 93.5cm (-4 SDS) and BMI 10.6 kg/m2 (- 7.75 SDS); she appeared malnourished on exam with minimal subcutaneous fat. She was admitted to the hospital where blood glucoses ranged from 59-68 mg/dL, despite continuous enteral feeds. On evaluation, blood ketones were mildly elevated at 0.67 mmol/L (ref range < 0.3mmol/L), growth factors were low: IGF1 15 ng/mL (ref range 37 - 272 ng/mL); IGF-BP3 1.1 mg/L (ref range 1.1 - 5.2 mg/L), and other pituitary hormones were within normal range [stimulation test cortisol peak 31.9 mcg/dL, TSH 2.4 mIU/mL(Ref range: 0.700 - 4.100 uIU/ML), Free T4 1.2 ng/dL (Ref range 1.0-2.4 ng/dL)]. Hypoglycemia resolved within 48 hours of initiating empiric treatment with GH (0.2mg/kg/week) and patient's feeds were successfully compressed to 16 hours. Child was discharged home after passing an overnight 8-hour safety fast. **Conclusion:** Growth hormone deficiency can present as recurrent hypoglycemia outside the infantile period in the setting of malnutrition and needs to be considered in the differential diagnosis and evaluation of childhood hypoglycemia.

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PEDIATRIC ENDOCRINOLOGY CASE REPORT

Maffucci Syndrome, Calcium Homeostasis, and Endocrine Challenges in Management

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Maffucci syndrome is a rare disorder characterized by enchondromatosis and hemangiomata. It can occur due to sporadic, de novo, mosaic pathogenic variants in the gene encoding isocitrate dehydrogenase 1 (*IDH1*) or isocitrate dehydrogenase 2 (*IDH2*). *IDH1* variants are associated with endocrine manifestations, such as pituitary adenomas. However, literature is limited in describing other clinical features and available treatments in severe phenotypes. We report a pediatric patient with uniquely complex and severe Maffucci syndrome. Case: A 5-year-old boy was evaluated by pediatric endocrinology for chronic hypercalcemia as part of a multidisciplinary evaluation of his severe Maffucci syndrome. Past medical history included prematurity, restrictive lung disease, developmental delay, seizures, 2-OH glutaric aciduria, angiomas, and bicytopenia. Physical exam revealed angiomas, scoliosis, and severe bony deformities throughout the entire skeleton. During admission, laboratory assays revealed normal parathyroid hormone, phosphorus, 1,25-OH2D, and C-telopeptide; elevated serum calcium and PTH-related peptide; and low 25-OHD, alkaline phosphatase, and osteocalcin. Low-dose ACTH stimulation test yielded a peak cortisol level of 16.8. A 24-hour urine study confirmed hypercalciuria. Renal ultrasound demonstrated nephrocalcinosis. Skeletal survey revealed diffuse and chondromatous changes of nearly every bone. Whole-exome sequencing detected a presumed, mosaic de novo *IDH1* variant. DEXA scan revealed total body BMD z-score of -3.8. **Discussion:** Hypercalcemia in Maffucci syndrome is a rare phenomenon. The most likely etiology was due to the severe and chronic bony breakdown from the underlying progressive enchondromatosis. Subsequently, the body attempted to adapt to these chronic processes with abnormal mineral homeostasis, as seen in his laboratory assays. Chronic primary hyperparathyroidism was not likely, as his PTH, phosphorus, and 1,25 OH2D levels were not congruent with that diagnosis. Familial hypocalciuric hypercalcemia was not likely, as his urine calcium clearance ratio was >0.01. Finally, his slightly elevated PTHrP level was not due to PTHrP-mediated hypercalcemia of malignancy, as his bone marrow biopsy was negative. The options for short-term hypercalcemia management had their own inherent risks and were not suitable for long-term management. Although there is a lack of pediatric data to guide therapy in Maffucci syndrome, decision was made to proceed with bisphosphonate infusion given the benefits in the setting of his nephrocalcinosis, chronic hypercalcemia, and results of his DEXA scan. Given the rarity of Maffucci syndrome, few characteristics are well-described in the pediatric population. A multidisciplinary approach is necessary to review the severity of the disease and to determine the best treatment approach based on this information.

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PEDIATRIC ENDOCRINOLOGY CASE REPORT

Metreleptin and Metformin Use in an Infant With Congenital Generalized Lipodystrophy Secondary to AGPAT2 Mutation

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Background: Congenital generalized lipodystrophy (CGL) is a rare inherited disease characterized by widespread loss

of subcutaneous fat and severe metabolic abnormalities. Metreleptin, a synthetic analog of leptin, is a treatment modality that has been shown to decrease fasting triglycerides, fasting glucose, and HbA1c. Metformin use in infants has only been described in a few case reports of CGL and Donohue syndrome (insulin receptor mutation), and there is no established dosing for this age group. We report metreleptin, insulin, and metformin use in an infant with type 1 CGL who presented with marked hypertriglyceridemia, hypoleptinemia, hyperglycemia, and transaminitis.

Clinical Case: A 2-month-old African American female born SGA at term presented to her primary care physician for a well child check where she was noted to have poor weight gain, hyperphagia, and abdominal distension. She was subsequently admitted for failure to thrive with weight z-score of -2.17 and length z-score of -0.15. Initial labs were notable for triglycerides of 5,167 mg/dL, HDL 10 mg/dL, blood glucose 324 mg/dL, ALT 212 units/L, AST 215 units/L, and bicarbonate of 17 mmol/L. A random insulin level was elevated at 257 mcIU/mL. Adiponectin was undetectable and leptin was low at 0.3 ng/mL. Hemoglobin A1c was in diabetes mellitus range at 8.9%. She was started on detemir 1.0 units/kg/day on day 1 (titrated to a maximum dose of 4.4 units/kg/day) and metformin 50 mg/kg/day on day 3 of hospitalization. By day 4, triglycerides decreased to 758 mg/dL, AST to 119 units/L and ALT to 124 units/L. Pre-prandial glucoses improved ranging from 113 to 138 mg/dL. As her insurance denied coverage for detemir, she was discharged home on glargine 0.7 units/kg/day and metformin suspension. One month later, she was started on subcutaneous metreleptin, and glargine was discontinued. At 5 months of age, she had triglycerides 229 mg/dl, normal liver enzymes, and normal blood glucoses while on 0.056 mg/kg/day of subcutaneous metreleptin and metformin. Medications were well tolerated without side effects. She had improved growth and met all developmental milestones. Genetic evaluation revealed that she was homozygous for a pathogenic variant, c.589-2A>G; p.Gln196fs*228 (rs116807569), in the AGPAT2 gene.

Conclusion: Leptin is important in regulation of lipid and glucose metabolism, and patients with CGL are deficient due to lack of adipose tissue. Metabolic abnormalities, including stabilization of glucoses and improved hypertriglyceridemia, in our patient markedly improved with initiation of metreleptin, metformin, and insulin. We present successful dosing of these treatment modalities without adverse reactions in an infant with CGL.

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PEDIATRIC ENDOCRINOLOGY CASE REPORT

Mitotane in the Treatment of Adrenocortical Carcinoma: Metabolic and Endocrine Disruption

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Background: Mitotane is a well-known adrenocytolitic agent resulting in adrenal insufficiency. However, little is known about its multiple and complex metabolic and endocrine effects.

Clinical Case: Patient is a 6 year 9 month old female who presented with acne, pubic hair and rapid weight gain. Initial evaluation showed elevated testosterone (64 ng/dL), 17-OH-progesterone (236 ng/dL), DHEA-S (922 mcg/dL), and random cortisol (30.9 mcg/dL) with suppressed ACTH (<2.0 pg/mL). She had an inappropriate lack of suppression after a 1 mg overnight dexamethasone suppression test (cortisol 49 mcg/dL, nl <2 mcg/dL). Abdominal CT showed large necrotic-appearing right adrenal mass (10.7x8.8x15 cm) with no metastasis. Surgical excision was complicated by intraoperative rupture. Pathology confirmed stage III right adrenal cortical carcinoma. Patient was started on etoposide, cisplatin, doxorubicin, and mitotane as adjuvant therapy. 6 days after surgery, her DHEA-S and cortisol levels were undetectable. She was started on glucocorticoid replacement therapy with hydrocortisone at 18 mg/m^2/day. Due to severe nausea, she was switched to dexamethasone 5 mg/m2/day (245 mg/m^2/day dose equivalent to hydrocortisone). However, the patient developed hypotension, increased nausea and emesis and was switched back to hydrocortisone. The patient's clinical course was complicated by hyperlipidemia with total cholesterol 215 mg/dL, HDL 48 mg/dL and LDL 150 mg/dL, as well as central hypothyroidism with low FT4 (0.8 ng/dL) and an inappropriately low normal TSH (0.31 mcIU/mL). She was started on levothyroxine with a final dose of 2.6mcg/kg/day to achieve euthyroid state. Mineralocorticoid deficiency has been reported in a small number of case reports of mitotane use. Our patient continues to demonstrate adequate mineralocorticoid function based on her normal electrolytes, aldosterone and plasma renin activity level.

Conclusion: Mitotane exerts multiple clinically relevant metabolic and endocrine effects. Patients treated with mitotane should be monitored for complications including mineralocorticoid deficiency, central hypothyroidism, and hyperlipidemia. Dexamethasone must be avoided because of the rapid inactivation by CYP4503A4 leading to adrenal crisis. Typical glucocorticoid replacement dose must be doubled due to induction of CYP3A4 activity that leads to glucocorticoid inactivation.

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PEDIATRIC ENDOCRINOLOGY CASE REPORT

Multiple Concomitant Episodes of Diabetic Ketoacidosis and Acute Pancreatitis in a Pediatric Patient With Type 1 Diabetes

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Introduction: Abdominal pain is a common presenting symptom in diabetic ketoacidosis (DKA). Correction of the acidosis usually leads to resolution of the abdominal pain. In some instances, the pain may persist due to additional etiologies presenting alongside DKA. Though uncommon, there has been shown to be an association between DKA and acute pancreatitis (AP). In these rare cases, AP was secondary to the hypertriglyceridemia (HTG) state induced by DKA. We report a 13-year-old female known with type