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