FSH- 0.2(prepubertal range). U/S-adrenals normal, right ovary was enlarged in size, 4x3cm cystic lesion. A CECT ABDOMEN showed Rt ovary -homogenously enhancing fairly well-defined solid lesion. Differential diagnosis was: NC/SVCAH with ovarian rest tumours vs ovarian virilising tumour. Since there was no non-invasive way of confirming between both the diagnosis, we gave a trial with hydrocortisone to see if her 17OHP reduces. After 1 month of therapy with hydrocortisone,17 OHP was still elevated -17.08ng/ml. Since there was no response, she underwent ovarian biopsy/excision. Right ovarian cystectomy was done. HPE showed -consider1. Steroid cell tumour NOS 2. Adrenal rest tumour. Note that these 2 tumours have identical morphology & IHC features. Followup:2 mon post-surgery: B5 P4 A3, mild facial hair,17 OHP - 1.53 ng/ml, Testosterone 2.5 ng/dl.12 months post-surgery child attained menarche had regular menstrual cycles, Serum 17OHP- 0.41 ng/ml, testosterone-11ng/dl.

Conclusion: The usual differential diagnosis in 46 XX female children presenting with signs of androgen excess is CAH, it is not uncommon to find SVCAH presenting in childhood with symptoms & signs of virilisation. Clinically both SV CAH and Steroid cell tumours can present with hyperandrogenic features in the childhood. Amongst steroid cell tumours, 3 subtypes are seen, they include 1. stromal luteoma 2. Leydig cell tumour 3. steroid cell tumour NOS. The steroid cell tumours of the ovary secrete androstenedione, alpha hydroxy progesterone & testosterone .17OHP is raised in both the conditions. The HPE of both adrenal rest tumours & steroid cell NOS is similar & a clear-cut differentiation cannot be made. They are similar in clinical presentation, biochemistry & HPE. It is important to recognise that steroid cell tumour NOS is a differential to SVCAH in childhood. Since these tumours can occur at any age, clues to differentiate both conditions are-raised 17OHP but normal DHEAS & on imaging- normal adrenals with enlarged ovaries with or without solid cystic lesions in steroid cell NOS of the ovary.

Pediatric Endocrinology

PEDIATRIC ENDOCRINOLOGY CASE REPORT

Successful Use of Intragastric Dextrose in a Unique Presentation of Congenital Hyperinsulinism

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Introduction: Congenital hyperinsulinism (HI) is the most common cause of persistent hypoglycemia in infants and can pose challenges if unresponsive to diazoxide. HI can be caused by monogenic mutations or can be associated with genetic syndromes. Macrocephaly Capillary malformation (MCAP) is a rare overgrowth syndrome, caused by heterozygous variants in the PIK3CA gene. A small number of pathologic variants in this gene have been reported to cause HI.

Clinical Case: A 4-month-old boy presented with a hypoglycemic seizure while fasting for an MRI. His history was notable for being born LGA and having macrocephaly, segmental infantile hemangioma, and ventriculomegaly requiring VP shunt. Critical labs were consistent with HI: plasma glucose (PG) of 23 mg/dL (54-117), inappropriately detectable insulin (2.7 mIU/mL) and c-peptide (1.6 ng/mL), low beta hydroxybutyrate (0.1 mmol/L) and low free fatty acids (0.16 mmol/L), and a positive glucagon stimulation test (increase in PG from 48 to 101 mg/dL in 30 minutes). Diazoxide was started at 5 mg/kg/day and titrated to 15 mg/kg/day, but he was unable to maintain PG >70 mg/dL. He was deemed unresponsive and the diazoxide was discontinued. His intravenous glucose infusion rate (GIR) was 14.4 mg/kg/min. An octreotide trial (8 mcg/kg/day) revealed a robust response: PG 64 mg/dL before initial dose, 105 mg/dL 3 hours later. However, he developed tachyphylaxis to the octreotide and it was discontinued. To further evaluate the etiology of his HI, he underwent an ^{18}F-DOPA PET scan, which showed diffuse uptake. Genetic sequencing for the 9 known HI genes was negative. At 6-months-old, he was evaluated by genetics who based on his clinical features diagnosed him with MCAP.

After failure of diazoxide and octreotide therapies, he was slowly transitioned from IV dextrose to continuous intragastric dextrose (IGD) using D20W. He was managed with a GIR of 10 mg/kg/min during the day (while receiving bolus feeds) and 5 mg/kg/min while on continuous feeds overnight. The continuous IGD allowed him to maintain euglycemia and develop his oral feeding skills. By 17-months-old, feeds by mouth improved and GIR had decreased to 6.5 mg/kg/min during the day and 2.5 mg/kg/min overnight. Genetic analysis eventually revealed a heterozygous p.Glu365Lys (c.1093 G>A) variant in the PIK3CA gene as the likely cause of the HI.

Conclusion: Genetic syndromes should be considered in infants with persistent hyperinsulinism and multiple congenital anomalies. Clinical work-up may provide important clues to the diagnosis. Diazoxide unresponsive HI can be treated with continuous IGD to prevent hypoglycemia-induced brain damage. Continuous IGD likely leads to better oral skills and decreased oral aversion compared to using continuous formula feeds.

Pediatric Endocrinology

PEDIATRIC ENDOCRINOLOGY CASE REPORT

Symptomatic Rebound Hypercalcemia After Denosumab Discontinuation in a Pediatric Patient With Cherubism

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Background: Pediatric bone diseases due to osteoclast overactivity have limited therapeutic options. Denosumab, a human monoclonal antibody against RANK-ligand, has been used in the treatment of these conditions despite limited pediatric safety data. Rebound hypercalcemia after denosumab cessation is a rare and potentially serious complication in children with an unpredictable course (1). We present the first report of a child with cherubism who experienced acute symptomatic hypercalcemia five months after denosumab cessation.