Hormonal evaluation showed elevated LH (3.28 IU/L) and follicle-stimulating hormone (4.79 IU/L), consistent with CPP. MRI demonstrated focal delayed enhancement in the posterolateral pituitary suggestive of microadenoma, and an incidental pineal cyst without complex features. Ophthalmological assessment was normal. Neurosurgical evaluation advised conservative management with annual imaging surveillance. Intramuscular Triptorelin was initiated at 4 years 1 month. Follow-up showed regression of pubertal progression, resolution of vaginal discharge, and reduced growth velocity to 6 cm/year.

CONCLUSION

This case highlights the importance of thorough evaluation in CPP to identify underlying intracranial pathology. Early initiation of gonadotropin-releasing hormone analog therapy effectively halts pubertal progression and preserves growth potential.

EP_P023

BECKWITH-WIEDEMANN SYNDROME PRESENTING AS PERSISTENT NON-KETOTIC HYPOGLYCEMIA IN A PRETERM INFANT

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INTRODUCTION

Beckwith–Wiedemann syndrome (BWS) is a congenital overgrowth disorder associated with dysregulation of genes on chromosome 11p15.5. Infants with BWS can present with hyperinsulinemic hypoglycemia. There are also distinctive clinical features including macrosomia, macroglossia and visceromegaly that may raise suspicion of this diagnosis.

CASE

A preterm female infant of 30 weeks gestation with a birth weight of 1.87 kg (97th centile) had recurrent hypoglycemia in the neonatal period, requiring escalation to a maximum glucose infusion rate of 17.6 mg/kg/min. Hypoglycemia only resolved after starting intravenous glucagon infusion. Critical sampling during hypoglycemia revealed serum insulin 3.9 µU/mL (3–25 µU/mL), serum ketone 0.2 mmol/L, cortisol 3,053 nmol/L and growth hormone 16.2 ng/mL. These findings supported the diagnosis of non-ketotic hyperinsulinemic hypoglycemia. She was commenced on oral diazoxide with resolution of hypoglycemia and discontinuation of glucagon.

Examination at birth revealed macroglossia, hepatomegaly and ballotable kidneys. An initial US abdomen at birth revealed bilateral enlarged kidneys but normal liver. An initial chromosomal study revealed karyotype 46, XX. Over time, additional clinical features became evident fulfilling the diagnostic criteria for BWS, that is, polyhydramnios, large for gestational age, transient hypoglycemia, hyperinsulinism, macroglossia, hemihypertrophy, facial nevi, bilateral ear creases, hepatomegaly and ballotable kidney.

A repeat US abdomen surveillance at 4 months old revealed a heterogeneous liver mass with marked vascularity, prompting a diagnosis of hepatic hemangioma. She was commenced on oral propranolol, with reduction in the size and vascularity of the liver hemangioma and decline in alpha-fetoprotein levels.

CONCLUSION

The clinical features of BWS may not be recognizable in a preterm baby in early neonatal period. Careful clinical examination should be done in a baby with persistent hyperinsulinemic hypoglycemia for underlying syndromal causes. US surveillance should also be carried out due to increased risk of hepatoblastoma or liver hemangioma in BWS, as was seen in this case.

EP_P024

BEYOND OBESITY: DIAGNOSTIC CHALLENGES OF BARDET-BIEDL SYNDROME

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INTRODUCTION

Bardet-Biedl Syndrome is a rare autosomal recessive disorder characterized by multisystem manifestations, such as early-onset obesity, hypogonadotropic hypogonadism, retinal dystrophy, polydactyly, renal anomalies and intellectual disability. Phenotypic variability makes diagnosis challenging. Central obesity and hypogonadotropic hypogonadism are frequently observed, but often overlooked.

CASE

We report a 12-year-old male who was referred at the age of 10 years for evaluation of central obesity and buried penis. He was born at term with birth weight of 3,200 g. Both parents were non-consanguineous, and antenatal history was unremarkable. Early-onset excessive weight gain was noted from infancy, with hyperphagia during early childhood.