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.

His weight remained persistently above the 95th centile, and height around the 90th centile, slightly exceeding mid-parental height expectations. Buried penis was identified at birth. Polydactyly is absent. He also demonstrates learning difficulties. Early diagnostic considerations include Klinefelter syndrome due to buried penis, and Prader-Willi Syndrome due to central obesity with co-existence of learning difficulties. Further evaluation excluded Prader-Willi Syndrome, and conventional karyotyping ruled out Klinefelter syndrome. Clinical assessment revealed pre-pubertal Tanner stage, bilaterally palpable testes (3 mL), and a stretched penile length <2 cm. Endocrine evaluation demonstrated a prepubertal response to LHRH stimulation, and poor testosterone response to hCG, suggesting hypogonadotropic hypogonadism with poor Leydig cell function. Whole exome sequencing confirmed a BBS2 gene mutation, establishing the diagnosis of Bardet-Biedl Syndrome. This mutation disrupts hypothalamic signaling, leading to the multisystem involvement observed. Over the follow-up period, he reported difficulty with night vision.

CONCLUSION

Not all syndromic obesity is immediately apparent—Bardet-Biedl Syndrome should be considered even in the absence of classical features. Marked phenotypic variability often leads to delayed diagnosis, highlighting the need for vigilant clinical suspicion and prompt genetic study to ensure timely recognition. Early recognition is critical, as endocrine and other associated complications can significantly impact long-term health.

EP_P025

ABCD SYNDROME: A RARE BUT UNDERRECOGNIZED CAUSE OF HYPERCALCEMIA IN DOWN SYNDROME

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INTRODUCTION

ABCD syndrome (ABnormal Calcium-Creatinine-Calcinosis in Down syndrome) is a rare tetrad of hypercalcemia, hypercalciuria, nephrocalcinosis, and renal impairment in children with Down syndrome, often with delayed diagnosis resulting in irreversible renal damage.

CASE

We report a 7-year-old male with Down syndrome, who previously had a stormy neonatal period due to large atrial-septal-defect and pulmonary hypertension, and required surgery at 3-years-old with prior prolonged ventilation. He was since bedbound with severe spastic diplegia, complicated by GERD and food aversion, requiring nasogastric tube feeding. He was discovered to have hypercalcemia and renal impairment during admission in district hospital for febrile illness with gastrointestinal symptoms. Initial serum calcium was 2.78 mmol/L, phosphate 1.54 mmol/L, magnesium 0.96 mmol/L, ALP 210 IU/L, urea 18.6 mmol/L, and creatinine 266 umol/L. Renal impairment did not improve and ultrasound KUB revealed bilateral small kidneys with medullary nephrocalcinosis. Consultation with the paediatric nephrologist concluded as CKD-stage-4. He was transferred to our centre. He demonstrated severe hypercalcemia (3.44 mmol/L), hypercalciuria (urine-calcium-to-creatinine-ratio of 1.17 mmol/mmol, >95 th%), and suppressed iPTH (0.9 pmol/L,1.6–6.9), suggesting PTH-independent process. 25-OH-Vit-D3 was 134 nmol/L (74–250, sufficient). He was more irritable and moody, but no seizures. ECG was normal. Skeletal assessment did not reveal osteolytic changes or increased resorption. He was investigated by paediatric hemato-oncologists, with negative findings, despite extensive search for hematological or bone malignancy and granulomatous disease. His ESR and immunoglobulin level was high for unknown reasons. He was treated with intravenous and oral hydration, dietary calcium restriction with modified low-calcium formula, assisted by dietitian, and IV pamidronate infusion (0.125 mg/kg) stat dose, resulting in stabilization of serum calcium level (2.5 mmol/L), which persisted for about 8 weeks during follow-up.

CONCLUSION

ABCD syndrome should be considered in Down syndrome children presenting with unexplained hypercalcemia. Early recognition and intervention are vital.

EP_P026

BEYOND GRAVES' DISEASE AND TYPE 1 DIABETES MELLITUS: THYROTOXIC HEART FAILURE AND EMERGING POLYGLANDULAR SYNDROME TYPE 2

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INTRODUCTION

The coexistence of autoimmune thyroid disease and type 1 diabetes mellitus is well established; however, their sequential or concurrent presentation in very young children remains uncommon. We demonstrate two paediatric cases with concurrent Graves' disease and type 1 diabetes mellitus, each illustrating distinct complications and clinical courses.