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.