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.

CASES

A 3-year-old female presented with anterior neck swelling, weight loss, and persistent tachycardia. Biochemical evaluation confirmed severe thyrotoxicosis (T4 152.3 pmol/L, TSH <0.008 mIU/L) with markedly elevated anti-thyroglobulin and anti–thyroid peroxidase antibodies. Thyroid ultrasonography demonstrated a diffusely enlarged, hypervascular gland with nodular changes, consistent with Graves' disease. During longitudinal follow-up, she developed polyuria, polydipsia, and hyperglycemia, with positive pancreatic autoantibodies confirming type 1 diabetes mellitus. Notably, serial early morning cortisol measurements were persistently low, raising concern for evolving adrenal insufficiency and suspicion of autoimmune polyglandular syndrome type 2. Comprehensive endocrine evaluation is ongoing.

A 4-year-old male presented with a 1-year history of recurrent self-limiting fever followed by significant fatigability for 1 month. Salient examination on admission revealed tachycardia, anterior neck swelling, and an incidental pansystolic murmur. Thyroid function tests confirmed thyrotoxicosis (T4 38.6 pmol/L, TSH <0.008 mIU/L). Serial serum glucose was markedly elevated on admission, prompting further testing that confirmed type 1 diabetes mellitus with positive pancreatic autoantibodies. Echocardiography demonstrated moderate-to-severe mitral regurgitation with heart failure secondary to prolonged uncontrolled hyperthyroidism state. Cardiac function improved with restoration of a euthyroid state, highlighting the reversible nature of thyrotoxic heart failure.

CONCLUSION

These cases highlight the progressive nature of childhood autoimmunity, where Graves' disease may precede the development of type 1 diabetes mellitus and evolving features of autoimmune polyglandular syndrome type 2. They further emphasize that thyrotoxic heart failure is potentially reversible with restoration of a euthyroid state, underscoring the importance of early recognition and vigilant long-term surveillance.

EP_P027

NOT JUST DEHYDRATION: A CASE OF EARLY ONSET PERSISTENT HYPERNATREMIA IN A PRETERM BABY

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INTRODUCTION

Hypernatremia in preterm babies is often due to insensible water loss, inadequate fluid intake or sodium imbalance due to immature kidneys. Congenital nephrogenic diabetes insipidus (CNDI) is an uncommon cause of hypernatremia in neonates and presents shortly after birth. Early recognition is important to prevent severe dehydration, electrolytes imbalance and neurological complications.

CASE

We report a case of a preterm 32-week female infant, with a birth weight of 1.14 kg with persistent hypernatremia (146–156 mmol/L) from day 4 of life. She received total parenteral nutrition since birth and was started on breastmilk since day 5 of life. The baby was mildly dehydrated with weight loss and high urea (6.5 mmol/L) at day 7 of life. Total fluids were increased to 160 mL/kg/day but serum sodium remained elevated even though the urea had normalized. The infant was also noted to have high urine output (5–6 mL/kg/hour) since day 3 of life and suboptimal weight gain.

Further evaluation at age 1 month revealed urine osmolality 71 mOsm/kg, serum osmolality 308 mOsm/kg and urine sodium <20 mmol/L when serum sodium was 150 mmol/L, suggestive of DI. A trial of desmopressin showed unchanged serum sodium (Na), suggesting nephrogenic DI. Administration of intravenous fluids of 1/5NSD10% resulted in further increase of both sodium (159 mmol/L) and serum osmolality (326 mOsm/kg) with low urine osmolality (111 mOsm/kg). Intravenous fluids were discontinued and she was started on hydrochlorothiazide (1 mg/kg/dose bd), resulting in gradual normalization of sodium 138 mmol/L. Post discharge, she had good weight gain and normal developmental milestones at corrected age of 1.5-month-old. Due to early onset hypernatremia, the infant was referred for genetic testing to rule out CNDI.

CONCLUSION

Early onset persistent hypernatremia and high urine output despite adequate fluid management should prompt evaluation of DI. Early diagnosis is crucial to prevent a chronic state of hypernatremia that can lead to growth and developmental delay.