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EP_P020

PAEDIATRIC PRIMARY HYPERPARATHYROIDISM PRESENTING WITH BILATERAL SLIPPED UPPER FEMORAL EPIPHYSES: A CASE REPORT

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INTRODUCTION
Primary hyperparathyroidism (PHPT) is a rare endocrine disorder in children and adolescents with a prevalence of 2-5 in 100,000. PHPT in this age group is often due to single parathyroid adenoma, whereby surgery remains the definitive treatment. Postoperative transient hypocalcemia is a common complication. Nevertheless, severe hungry bone syndrome (HBS) in paediatric is uncommon and is a challenge in the post-operative management of PHPT.

CASE
A 14-year-old Malay, male presented with trivial falls and subsequently developed bilateral lower limb pain for 2 months, which led to an abnormal, painful gait for 1 week. He also had nausea, intermittent vomiting, abdominal pain, loss of weight and appetite.

Biochemical investigations were consistent with primary hyperparathyroidism. He had severe hypercalcemia, hypophosphatemia with elevated alkaline phosphatase and intact parathyroid hormone (iPTH). Pelvic x-ray revealed bilateral slipped upper femoral epiphysis (SUFE) and periosteal bone resorption at the pelvic bones. A neck ultrasound showed a hypoechoic nodule located posterior-inferior to the right lobe of the thyroid gland. A Tc-99m sestamibi parathyroid scan detected an avid lesion inferior to the right thyroid lobe.

His severe hypercalcemia was managed by hydration and loop diuretics. For preoperative optimisation, he received intravenous zoledronate and subcutaneous calcitonin. He underwent a right-focused parathyroidectomy and histopathology confirmed the diagnosis of parathyroid adenoma. Post-operatively, he developed severe HBS. He had symptomatic hypocalcemia post-parathyroidectomy for which he required continuous calcium gluconate infusion, calcitriol, calcium carbonate and cholecalciferol. Continuous intravenous calcium and intravenous alfacalcidol were given for four weeks and stopped when serum phosphorus and alkaline phosphatase levels returned to normal limits.

CONCLUSION
PHPT in children and adolescents often presents with non-specific symptoms leading to a delay in diagnosis. A diagnosis of PHPT should be considered when they present with bone pain or skeletal deformity associated with radiological imaging of osteolytic lesions.

EP_P021

THE HIDDEN THREAT: DIABETES MELLITUS IN A CHILD WITH CONGENITAL RUBELLA SYNDROME

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INTRODUCTION
Congenital rubella syndrome arises from maternal infection with rubella virus, particularly during the first trimester of pregnancy. While rubella is primarily associated with ocular, cardiac and auditory defects, its effects on the endocrine system, particularly in relation to diabetes mellitus, are seldom reported. This case report underscores the necessity of close monitoring in children with a history of rubella exposure, given the potential risk for the subsequent development of diabetes mellitus.

CASE
A 1-year 9-month-old male had right corneal clouding and absent red reflex. He was born at term with a birth weight of 2.6 kg. His mother had a multinodular goitre with no history of fever and rashes during pregnancy. He was diagnosed with right eye glaucoma and left uveitic cataract. He underwent evisceration of the right eye at 4 months and left lens surgery at 6 months.

The patient presented recently with lethargy, excessive thirst, frequent urination and weight loss. He had global developmental delays and showed signs of dehydration during the examination. His growth was within percentile. Investigations revealed blood glucose of 46 mmol/L, positive serum ketone and metabolic acidosis (pH, 7.126; HCO_3, 10.1 mmol/L; base excess -22.7 mmol/L; $PaCO_2$, 20 mm Hg). He had no skin lesion and other systemic examinations were unremarkable.

He was diagnosed with diabetic ketoacidosis and was treated with intravenous fluids and insulin. Following metabolic stabilization, he was transitioned to subcutaneous insulin.

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There was an increased incidence of insulin-dependent diabetes mellitus with congenital rubella syndrome. Pathogenesis is multifactorial, potentially involving the viral destruction of pancreatic β-islet cells and autoimmunity. Rubella virus peptides mimic glutamic acid decarboxylase (GAD) peptides in the pancreas. This activates T-cell-mediated autoimmune destruction and progressive loss of insulin-producing pancreatic beta-cells due to cross reaction.

CONCLUSION

This case highlights a significant endocrine complication associated with congenital rubella syndrome and emphasizes the importance of early diagnosis and management.

EP_P022

PERICARDIAL EFFUSION SECONDARY TO SEVERE HYPOTHYROIDISM IN DOWN'S SYNDROME

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INTRODUCTION

Hypothyroidism is a recognized cause of pericardial effusion. Among children with Down's syndrome, hypothyroidism may be an associated feature.

METHODOLOGY

We report a case of a 4-year-old female with Down's syndrome and severe pericardial effusion secondary to hypothyroidism. She was born with no history of maternal thyroid disease. The diagnosis of Down's syndrome was made postnatally. She was diagnosed with congenital hypothyroidism and was started on treatment during her stormy neonatal period. She had a recurrent lung infection, developed chronic lung disease and worsening pulmonary hypertension. Due to multiple hospital admissions, she was non-compliant to her thyroid medications. She has been asymptomatic apart from failure to grow and mild constipation which was attributed to poor nutrition and presumed gastroesophageal reflux disease. At the age of 3 years and 6 months, she was noted to have muffled heart sounds. Her vitals were normal for age, but ECG showed a relative bradycardia with a rate of 65 bpm with low voltage and flattening of the T-wave. Her echocardiogram showed large pericardial effusion. Her thyroid-stimulating hormone (TSH) was 1085.52 mIU/L and free thyroxine (FT4) of <1.3 pmol/L, confirming severe hypothyroidism. She was started on intravenous levothyroxine for five days before changing to oral levothyroxine to a maximum dose of 100 mcg (8 mcg/kg/day) daily. She did not require pericardiocentesis and was discharged well. Three months later, her thyroid function test showed normalization of TSH and FT4. Repeated echocardiogram showed smaller pericardial effusion.

CONCLUSION

This case report highlights a rare presentation of significant pericardial effusion secondary to severe primary hypothyroidism in a young female with Down's syndrome. Furthermore, it emphasizes the need for vigilant monitoring of thyroid function in this population and timely intervention to prevent potentially serious complications.

EP_P023

ANDROGEN INSENSITIVITY SYNDROME: A FAMILY CASE SERIES

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INTRODUCTION

Androgen insensitivity syndrome (AIS) is a rare X-linked recessive disorder caused by mutations in the androgen receptor. In Malaysia, only four cases of complete androgen insensitivity syndrome (CAIS) have been reported.

CASE

We present three biological cousins born to two sisters from the same maternal lineage, presenting with varying degrees of genitalia ambiguity.

Cousin A. A 1-year-and-5-month-old child presented with ambiguous genitalia at 1 month old. Physical examination revealed a 3 cm genital tubercle, penoscrotal hypospadias and fused symmetrical scrotal labia, with both testes retractile in the inguinoscrotal region. Antimüllerian hormone level was elevated, and an HCG stimulation test showed an increase in testosterone response. Karyotyping confirmed a 46, XY karyotype and whole exome sequencing identified a hemizygous pathogenic variant in the AR gene: p. Arg841His. Gender was assigned as male.