Paediatrics E-Poster

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.