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.

Cousin B and C. Two siblings with 46, XY karyotype and ambiguous genitalia presented at different points following birth.

Sibling 1. The elder sibling, aged 4 years and 2 months, presented with ambiguous genitalia and a left inguinal hernia at 1 month old. Physical examination revealed a single opening, incomplete labioscrotal fusion and palpable gonads bilaterally at the inguinal region. Laboratory tests showed high testosterone and anti-Müllerian hormone levels, with absent uterus and ovaries.

Sibling 2. The younger sibling, aged 2 years and 6 months had ambiguous genitalia with bilateral palpable gonads in the inguinal region at birth. Despite having a male genotype, they exhibited predominantly female phenotypic traits and were raised as females. The family declined genetic testing due to financial constraints.

CONCLUSION

Three cousins with varying presentations of the AIS highlight the phenotypic diversity of the condition and the challenges in sex assignment and management, underscoring the need for genetic counselling and multidisciplinary care.

EP_P024

CHROMOSOME 9p DUPLICATION AND SHORT STATURE: A CASE REPORT

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INTRODUCTION

Chromosome 9p duplication, also referred to as partial duplication syndrome, is a rare chromosomal disorder with fewer than 200 cases reported worldwide. It is characterized by duplication of a segment of the short arm of chromosome 9 and is frequently associated with short stature, craniofacial dysmorphisms and intellectual disability.

CASE

We present the case of a 6-year-old male born at term via emergency lower segment cesarean section (EMLSCS) at 37 weeks due to intrauterine growth restriction (IUGR). Since infancy, he exhibited persistent failure to thrive and subtle dysmorphic features. His height and weight remained consistently below the 3rd percentile despite adequate nutritional intake. Physical and systemic examinations were otherwise unremarkable.

Initial workup, complete blood count, renal profile were normal, Hormonal work out showed the following: thyroid stimulating hormone: 5.36, free T4: 19.85, luteinizing hormone: <0.1 IU/L, follicular stimulating hormone: 1.23 IU/L, cortisol: 168.5 nmol/L growth hormone: 0.8 ug/L and IGF-1: 266.4 ng/mL. Gross karyotyping (46,XY) and radiological imaging yielded normal results. Chromosomal microarray analysis subsequently revealed a microduplication in the 9p12-p11.2 region, establishing the diagnosis of chromosome 9p duplication syndrome.

CONCLUSION

This case underscores the importance of considering chromosomal microduplication in children presenting with unexplained short stature and developmental concerns. The role of growth hormone therapy in this population remains uncertain due to limited evidence regarding its efficacy in this specific genetic condition.

EP_P025

THE VARIED FACES OF NEONATAL THYROID DYSFUNCTION IN THE CONTEXT OF MATERNAL GRAVES DISEASE: A CASE SERIES

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INTRODUCTION

Maternal hyperthyroidism, most commonly due to Graves' disease (GD), can cause a range of thyroid dysfunction in the fetus and neonate. Neonatal thyroid function is influenced by factors such as maternal disease activity, levels of TSH receptor antibodies (TRAb) and in-utero exposure to antithyroid drugs (ATDs). We report two neonatal cases illustrating this variability.

CASE

Case 1. A term male neonate was born to a mother diagnosed with GD during the first trimester, who was well-controlled on oral carbimazole. She had a positive TRAb with hyperaemic thyroiditis on ultrasound. The infant had a normal cord thyroid stimulating hormone (TSH) at birth but TRAb measured at one week was 2-fold above the upper limit of normal. A thyroid function test (TFT) on Day 5 showed subclinical hypothyroidism which resolved spontaneously by one month without treatment.

Case 2. A female neonate was delivered at 34 weeks' gestation to a mother with a six-year history of GD,