

Paediatrics E-Poster

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 dysmorphism 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,