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

https://doi.org/10.15605/jafes.040.S1.254

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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

https://doi.org/10.15605/jafes.040.S1.255

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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,

complicated by thyroid storm during the current pregnancy. Her TRAb levels prior to conception were over 23-fold above normal. The infant was born with suppressed cord TSH and markedly elevated TRAb level which is 13-fold above normal. She developed symptoms of neonatal hyperthyroidism in the second week of life and was started on carbimazole and propranolol, which were weaned off by the third week. Subsequent TFTs showed a phase of subclinical hyperthyroidism followed by hypothyroidism by two months of age requiring thyroxine replacement.

CONCLUSION

These cases highlight the diverse presentation of neonatal thyroid dysfunction associated with maternal GD, ranging from transient hypothyroidism to biphasic thyroid disturbances following neonatal hyperthyroidism. High maternal TRAb levels, as seen in Case 2, may serve as a predictor of a more severe case of evolving neonatal thyroid disease. Continuous postnatal monitoring is essential, as thyroid dysfunction may not be evident at birth and can evolve over time. Timely diagnosis and appropriate management are key to prevent complications and supporting optimal neurodevelopmental outcomes.

EP_P026

FAMILIAL DYSALBUMINEMIC HYPERTHYROXINEMIA: A RARE CAUSE OF EUTHYROID HYPERTHYROXINEMIA

https://doi.org/10.15605/jafes.040.S1.256

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INTRODUCTION

Euthyroid hyperthyroxinemia is a common clinical conundrum. It requires careful assessment to establish an accurate diagnosis. Differential diagnosis of euthyroid hyperthyroxinemia include assay interference, thyroid hormone resistance syndrome, familial dysalbuminemic hyperthyroxinemia (FDH) and TSH-oma.

CASE

A 7-month-old male was referred for incidental finding of persistent euthyroid hyperthyroxinemia. His birth history was unremarkable. Antenatally, his mother did not have any thyroid disorder. His paternal grandmother has been undergoing treatment for hyperthyroidism. Thyroid stimulating hormone (TSH) was elevated at 16.89 mIU/L. Routine prolonged jaundice investigations revealed free thyroxine (FT4) of 33.7 pmol/L and TSH of 7.4 mIU/L. Other investigations were normal. Clinically, he was euthyroid, not dysmorphic, no goitre and thriving well with normal developmental milestones. Repeated thyroid function test (TFT) via standard immunoassay at 2, 3 and 5 months of age showed similar results of high FT4 with unsuppressed TSH. FT3 was not available. TFT using a different assay was not done. Thyroid antibody screening was normal. He was initially suspected of having thyroid hormone resistance syndrome.

Family screening showed similar TFT pattern for his father and sister who were clinically euthyroid. His mother's TFT was normal. His family was referred for confirmatory genetic testing. Whole exome sequencing (WES) for his father identified a pathogenic missense mutation in albumin gene, resulting in the replacement of an arginine with a histidine (p.Arg242His) that is associated with FDH. No genetic testing was done for the children.

CONCLUSION

FDH is a rare cause of euthyroid hyperthyroxinemia. It is an autosomal dominant disorder characterized by an abnormally increased affinity of a mutant albumin molecule to serum thyroxine causing elevated total thyroxine (T4) and elevated or normal FT4 with normal TSH level. Genetic analysis is important to establish diagnosis, to avoid further unnecessary laboratory testing and even inappropriate treatment in FDH.

EP_P027

ATYPICAL GENITALIA IN SILVER-RUSSELL SYNDROME

https://doi.org/10.15605/jafes.040.S1.257

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INTRODUCTION

Silver-Russell Syndrome (SRS) is a clinically heterogeneous disorder which is often associated with growth restriction.