Management included IV calcium gluconate, targeted anti-failure therapy (captopril, furosemide, spironolactone), and mechanical ventilation. High-dose cholecalciferol replenished depleted stores, alongside an active vitamin D analog (alfacalcidol) to bypass impaired hepato-intestinal pathways, stimulating calcium absorption. Extubated after 2 weeks with stabilized serum calcium, he was discharged on anti-failure therapy, oral calcium carbonate (54 mg/kg/day elemental), D-cure 25,000 IU 4-weekly, Ursodeoxycholic acid (45 mg 12-hourly), and multivitamins. Repeat echocardiography is planned at 3 months.

CONCLUSION

This case underscores the vulnerability to nutritional rickets and late-onset hypocalcemia in SGA infants with inadequate maternal nutrition and impaired GI absorption. The calcium-phosphate axis is easily disrupted; high-phosphate substitutes (goat's milk) precipitously unmask hypocalcemia, overwhelming compensatory hyperparathyroidism. Proactive, dual-therapy metabolic supplementation is paramount to avert catastrophic, yet reversible, HDCM.

EP_P016

MATURITY-ONSET DIABETES OF THE YOUNG ASSOCIATED WITH A ABCC8 VARIANT

Jia Cheng Ong[1,2,3] and Suhaimi Hussain[1,2]
[1]*Department of Paediatrics, School of Medical Sciences, Universiti Sains Malaysia*
[2]*Hospital Pakar Universiti Sains Malaysia (HUSM)*
[3]*Faculty of Medicine, Universiti Sultan Zainal Abidin*

INTRODUCTION

Maturity-onset diabetes of the young (MODY) is a form of monogenic diabetes typically affecting young adults. There are 14 different genes that lead to pancreatic cell dysfunction causing MODY. Occurrence of MODY12 by ATP binding cassette subfamily C member 8 (ABCC8) gene is rare, comprising 1% of all the MODY subtypes.

CASE

A 13-year-old male was incidentally found to be hyperglycemic during medical examination. He denied any hyperosmolar symptoms, polyphagia, nocturia and recurrent skin infection. He did not have any history of neonatal diabetes mellitus. His father had been diagnosed with type 1 diabetes mellitus at the age of 22 years old. On examination, he is thinly built with no goiter or acanthosis nigricans. HbA1c was 8.8%. His insulin antibodies were negative. Whole exome sequencing revealed a heterozygous missense mutation in ABCC8 gene (c.2209G>A; p.Val737Ile), where there was a single nucleotide mutation of Valine to Isoleucine at the position 737 of the ABCC8 gene. This variant was reported in the ClinVar database as having uncertain significance. Considering the patient's clinical features, this mutation is responsible for the disease. He was initially treated with sulfonylurea, namely glibenclamide; however, his continuous glucose monitoring was not optimized and required change to insulin therapy.

CONCLUSION

This case explores the phenotypic spectrum of ABCC8-related MODY, showing that this mutation can present without neonatal diabetes and emphasizing the requirement of insulin as part of treatment. Genetic testing should be conducted in patients presenting with atypical clinical features of diabetes mellitus to initiate personalized treatment strategies.

EP_P017

RECOGNIZING RENI SYNDROME: A CASE OF ADRENAL INSUFFICIENCY, ICHTHYOSIS, AND PROTEINURIA

Noor Zakirah Noordin[1], Saw Shi Hui[1], Noor Arliena binti Mat Amin[2]
[1]*Paediatric Department, Hospital Raja Permaisuri Bainun*
[2]*Paediatric Department, Hospital Tunku Azizah*

INTRODUCTION

RENI syndrome (renal, endocrine, neurologic, and immune syndrome), also known as sphingosine-1-phosphate lyase insufficiency syndrome (SPLIS), is a rare autosomal recessive disorder caused by pathogenic variants in the SGPL1 gene. This gene encodes sphingosine-1-phosphate lyase, an enzyme responsible for the final step in sphingolipid degradation. Impaired enzyme activity results in the accumulation of sphingolipid intermediates, leading to multisystem involvement affecting the kidneys, adrenal glands, skin, immune system, and nervous system. Frequently reported manifestations include steroid-resistant nephrotic syndrome, primary adrenal insufficiency, ichthyosis, and neurological abnormalities.

CASE

We report a 15-year-old male with a history of primary adrenal insufficiency diagnosed at age three after presenting with recurrent vomiting and abdominal pain. He was started on steroid replacement therapy and remained stable without episodes of adrenal crisis. He also had ichthyosis requiring dermatological follow-up. During routine surveillance, persistent proteinuria was detected, and urinary protein excretion increased over

time. Ultrasound showed renal parenchymal changes despite preserved renal function. Whole-exome sequencing identified a homozygous variant of uncertain significance in the SGPL1 gene, consistent with RENI syndrome. He was started on enalapril for his proteinuria and continues to undergo multidisciplinary follow-ups. Both parents are consanguineous, but were not screened.

Mutations in SGPL1 have increasingly been recognized as a monogenic cause of syndromic steroid-resistant nephrotic syndrome, associated with endocrine and dermatological features. Renal histopathology in cases reported often shows focal segmental glomerulosclerosis, reflecting podocyte injury due to disruption of sphingolipid signaling pathways. Early recognition is crucial, as patients need multidisciplinary care.

CONCLUSION

This case highlights the importance of considering RENI syndrome in patients presenting with early-onset primary adrenal insufficiency accompanied by renal and dermatological symptoms. Increased clinical awareness and prompt genetic testing can enable earlier diagnosis, guide long-term monitoring, and support appropriate genetic counseling.

EP_P019

46XX, TESTICULAR DSD WITH A NEGATIVE SRY: WHAT YOU SEE IS NOT ALWAYS THE TRUTH

Chia Ying Kang[1] and Sze Teik Teoh[1]
[1]*Paediatric Department Hospital Sultanah Bahiyah*

INTRODUCTION

46,XX testicular DSD is rare with incidence ~1 in 20,000 live births. Over 80–90% of cases are caused by translocation of SRY gene to X-chromosome or an autosome. Most present later in life with small testes, infertility and azoospermia. In SRY-negative cases, mutations in other genes (SOX9, NR5A1, etc) or duplications of SOX9 are implicated. Our reported case is SRY negative, presenting at birth.

CASE

We report a case of 1-year 4-month-old term baby with atypical genitalia at birth. External genitalia examination showed penoscrotal malformation, genital tubercle (2.5 × 1.5 cm) with chordae, single opening at perineum, and two palpable gonads within both upper labio-scrotal folds. External genital score is 8.5. Ultrasound of inguinal region and pelvis showed bilateral hypoechoic ovoid structure at bilateral inguinal region suggestive of testes, with the right testis measuring 0.4 × 0.8 cm and left testis measuring 0.4 × 0.7 cm. No mullerian structures or ovaries were visualized. Initial chromosome results (10 cells analyzed, 30 counted) reported 46, XX. Consultation with genetic pathologist and 2nd karyotype result still revealed 46, XX with FISH analysis confirming the absence of SRY. Mini-puberty screen (at 72 hours of life) showed follicle-stimulating hormone 3.5 IU/L, LH 1.72 IU/L, and testosterone 2.29 nmol/L. HCG stimulation test indicated adequate testosterone rise from 13 nmol/L (Day 1) to 29.38 nmol/L (Day 3), confirming the presence of functional Leydig cells. Serum AMH was within normal male range for age (421.3 pmol/L, 29–364 days: 235.5–1125.9). Gender is assigned as male and he recently underwent 1st stage hypospadias repair. Whole exome sequencing result is pending.

CONCLUSION

46,XX testicular DSD, with a negative SRY, is rare, and early diagnosis at birth enables counseling for future concerns such as male infertility and hypergonadotropic hypogonadism. A multidisciplinary management (involving endocrinology, surgery/urology, and genetics) is needed to ensure understanding and emotional support.

EP_P020

THE SYSTEMIC COST OF INTRALESIONAL TRIAMCINOLONE FOR PAEDIATRIC KELOIDS

Naveen Nair Gangadaran[1], Mastura Ibrahim[1], Meenal Mavinkurve[2]
[1]*Department of Paediatrics, Hospital Tuanku Ja'afar*
[2]*Department of Paediatrics, International Medical University*

INTRODUCTION

Intralesional triamcinolone acetonide (TAC) is an accepted management option for paediatric keloid. TAC is a corticosteroid with five times the potency of hydrocortisone and acts by inhibiting fibroblast growth and thus keloid size. Dermatological adverse effects of TAC are common, but repeated high-dose intralesional TAC can cause the rare complication of Cushing syndrome and suppression of the hypothalamic–pituitary–adrenal (HPA) axis.

CASE

We report the case of an 11-year-old female referred for worsening lower limb striae and low serum cortisol. Prior to referral, intralesional TAC was administered by a general practitioner to a left lower limb keloid which had developed following an orthopedic intervention. A total of five injections were administered over 4 months: a cumulative dose of 150 mg (equivalent to 700 mg hydrocortisone). The last dose was administered 6 months prior to referral. The morning serum cortisol was low (<13.8 nmol/L) with a concomitantly low adrenocorticotropic hormone (ACTH 0.41 pmol/L) level, consistent with suppression of the