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

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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

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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

HPA axis secondary to exogenous steroids. Renin (84.80 mU/L), aldosterone (280.40 pmol/L), testosterone (0.31 nmol/L), estradiol (85.9 pmol/L) and thyroid function were normal. Physiological oral hydrocortisone replacement was promptly commenced, with education on adrenal crisis and sick days while awaiting recovery of the HPA axis.

CONCLUSION

This case illustrates the serious complication of Cushing syndrome with secondary HPA axis suppression due to repeated intralesional TAC therapy for paediatric keloid scars. Factors such as appropriate paediatric doses, size of the keloid, site of injection (dermal, not subcutaneous), and frequency of injections should all be given due consideration before using TAC. Clinicians prescribing steroids to children should be aware that suppression of the HPA axis is a very real and life-threatening complication and that steroids should be prescribed judiciously.

EP_P021

GROWTH HORMONE THERAPY IN PAEDIATRIC ONCOLOGY SURVIVORS: CASE SERIES FROM A MALAYSIAN TERTIARY CENTRE

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INTRODUCTION

Growth hormone deficiency (GHD) is a recognized endocrine complication among childhood cancer survivors resulting from disruption of hypothalamic–pituitary axis due to tumors, neurosurgery, or cranial irradiation. While recombinant human growth hormone (rhGH) therapy improves growth and metabolic outcome, concerns remain regarding its long-term safety with risk of tumor recurrence and secondary neoplasm. We present a case series of five paediatric oncology survivors with confirmed GHD receiving rhGH in Hospital Tunku Azizah, highlighting our clinical experience in comparison with international practice.

CASE

Five paediatric oncology survivors (acute lymphoblastic leukemia, craniopharyngioma, medulloblastoma, and supratentorial PNET) with GHD were commenced on rhGH therapy 2.5–5.5 years after completion of cancer treatment. All patients had significant short stature with a mean height SDS of −3.22 prior to the initiation of therapy. GHD was confirmed biochemically with low IGF-1 level and dynamic testing (peak GH 0.35–4.61 ng/mL). Among the four patients with brain tumors, two had stable residual disease while the remaining were tumor-free. rhGH therapy was temporarily ceased in two patients due to minor increment in tumor size but was successfully resumed following stabilization without further complications to date. Two patients are concurrently on pubertal induction. Most patients demonstrated catch-up growth with increased height velocity, supporting the efficacy of rhGH in this population. No secondary malignancies or significant adverse events were observed during follow-up.

CONCLUSION

Paediatric oncology survivors with GHD in this series demonstrated a favorable response to rhGH therapy, evidenced by improvements in height SDS and growth velocity, while maintaining oncological stability in the majority of cases. Our findings are consistent with international data, supporting cautious but appropriate use of rhGH in this population. Careful patient selection and close multidisciplinary monitoring remain essential.

EP_P022

WHEN PUBERTY COMES TOO SOON: CENTRAL PRECOCIOUS PUBERTY WITH PITUITARY MICROADENOMA AND PINEAL CYST

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INTRODUCTION

Central precocious puberty (CPP) is defined as premature activation of the hypothalamic-pituitary-gonadal axis, resulting in the development of secondary sexual characteristics before 8 years of age in girls. While most cases are idiopathic, intracranial pathologies such as pituitary lesions may cause CPP. Early identification is essential to optimize outcomes and minimize psychosocial impact. We report a case of CPP associated with pituitary microadenoma and pineal cyst from the Paediatric Department, Hospital Sultanah Nur Zahirah.

CASE

A 4-year-old female presented with bilateral breast enlargement, with ultrasonography showing normal fibroglandular tissue. Breast development was noted since 3 months of age, with whitish vaginal discharge from 2 years. She demonstrated accelerated growth, with height and weight above the 95th centile. There were no features suggestive of peripheral causes or raised intracranial pressure. Examination revealed Tanner stage A1B3P2. Bone age was markedly advanced (11 years vs. chronological age 4 years).