

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 Kang¹ and Sze Teik Teoh¹

¹Paediatric 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 müllerian 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 Gangadaran¹, Mastura Ibrahim¹, Meenal Mavinkurve²

¹Department of Paediatrics, Hospital Tuanku Ja'afar

²Department 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 adrenocorticotrophic hormone (ACTH 0.41 pmol/L) level, consistent with suppression of the