Whole-exome sequencing identified a hemizygous androgen receptor gene variant, p.(Pro818Ala), classified as a variant of uncertain significance with deleterious in silico predictions, consistent with X-linked partial androgen insensitivity syndrome.

CONCLUSION

This case highlights the necessity of early, systematic endocrine evaluation integrated with genomic testing in 46, XY DSD to achieve a definitive diagnosis, optimize sex assignment, and guide longitudinal, multidisciplinary management.

EP_P014

CONGENITAL TOXOPLASMOSIS WITH CRANIAL DIABETES INSIPIDUS AND HYDROCHLOROTHIAZIDE

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INTRODUCTION

Congenital toxoplasmosis (CTox) is common in Malaysia and classically presents with brain and eye involvement, causing hydrocephalus, intracranial calcifications, cataract, chorioretinitis, blindness, epilepsy, and psychomotor or mental impairment. Cranial diabetes insipidus (CDI) and panhypopituitarism rarely complicate its clinical course.

CASE

From 2024 to 2026, we had encountered three cases of CTox infants, of whom 2 (C1, C2) had a stormy neonatal period and passed away by 3 months old due to refractory seizures, while the 3rd (C3) survived. C1 (2.2 kg) was diagnosed antenatally from fetal ultrasound brain with severe ventriculomegaly, whereas C2 (2.47 kg) had multiple syndromic features, cleft lip, palate and anopthalmos. C3 (2.6 kg) was only diagnosed later by 1-month-old when she had afebrile seizures. C1 and C2 had severe hypernatremia (Na >150 mmol/L) within 1st week of life, but C3 had hypernatremia after surgical drainage of her hydrocephalus and administration of steroids. All were diagnosed with CDI and fulfilled the triad of polyuria, hypernatremia and inappropriate paired osmolality. During acute period, they were managed with IV vasopressin infusion with intensive monitoring. At low dose (0.1–0.3 mcg/kg/hour), all achieved eunatremia and euvolemia within 12 hours with no inadvertent hyponatremia. They also had central hypothyroidism and received L-thyroxine, with prior oral hydrocortisone, except C1. Their CDI persisted with highest Na 165 mmol/L in C1. They were started on tab hydrochlorothiazide (HCTZ) alongside low renal solute load formula (LRSL) and EBM. HCTZ dose was titrated gradually from 0.5 to 1.0 mg/kg/dose and further to 1.5 mg/kg/dose. In between, subcutaneous desmopressin (DDAVP) 0.02–0.04 mcg were given during breakthrough DI. All cases responded to HCTZ at 1.5 mg/kg/dose, with varying intervals (daily to TDS). C3 went home after 6 weeks with HCTZ, L-thyroxine and hydrocortisone, together with oral anti-toxoplasmosis and anticonvulsants.

CONCLUSION

CTox with CDI presents a challenge during infancy, and a combination of LRSL with thiazide diuretics is an acceptable alternative, prior to definitive DDAVP therapy later.

EP_P015

STARVED BONES, FAILING HEART: UNCOMMON YET LIFE THREATENING HYPOCALCEMIC CARDIOMYOPATHY IN NUTRITIONAL RICKETS

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INTRODUCTION

We report an insidious presentation of hypocalcemic dilated cardiomyopathy (HDCM) in an infant with nutritional rickets and severe hypocalcemia, highlighting the myocardium's strict reliance on calcium-phosphate homeostasis.

CASE

A 3-month-old, small-for-gestational-age male (birth weight: 1.93 kg), with congenital cataracts and persistent neonatal cholestasis (negative hepatopathy and congenital infection screens) presented acutely with post-feeding cyanosis, requiring intubation for aspiration pneumonia. The mother denied prior apneic episodes, seizures, feeding difficulties, or heart failure symptoms. Clinically, there was no murmur or hyperactive pericardium. Chest radiograph revealed globular cardiomegaly. Echocardiography confirmed HDCM (LVEF 41%).

Bloodwork revealed profound hypocalcemia (1.16 mmol/L; normal 2.1–2.6), hyperphosphatemia (2.97 mmol/L), and markedly elevated alkaline phosphatase (2,411 IU/L). 25(OH)D3 was deficient (34.08 nmol/L). Secondary hyperparathyroidism (iPTH 19.55 pmol/L) represented an appropriate response. The mother had introduced goat's milk instead of cow's milk due to inadequate lactation. She took no supplements during pregnancy and was also vitamin D deficient.

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

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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

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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