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.