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A RARE PAEDIATRIC CASE OF AVPR2-RELATED NEPHROGENIC SYNDROME OF INAPPROPRIATE ANTIDIURESIS IN PENANG

Wei Lian Lean[1], Raja Aimee Binti Raja Abdullah[1], Gaik Siew Ch'ng[2], Voon Lee Lim[3]
[1]Paediatric Endocrinology Unit, Department of Paediatric, Hospital Pulau Pinang
[2]Genetic Unit, Hospital Pulau Pinang
[3]General Paediatric, Department of Paediatric, Hospital Pulau Pinang

INTRODUCTION
Nephrogenic syndrome of inappropriate antidiuresis (NSIAD) is an uncommon X-linked genetic condition marked by euvolemic hyponatremia, which arises from a gain-of-function mutation in the arginine vasopressin receptor type 2 (AVPR2) gene. In contrast to the classic syndrome of inappropriate antidiuretic hormone secretion (SIADH), NSIAD is characterized by urine that is inappropriately concentrated even when arginine vasopressin (AVP) levels are low or undetectable. Failure to recognize NSIAD may result in recurrent hyponatremia and neurological morbidity.

CASE
A 2-year-old male was referred to us after recurrent vomiting and subsequently developed generalized tonic-clonic seizures due to hyponatremia (with a sodium level of 115 mmol/L). He needed sodium replacement through an intravenous drip of 3% saline and was stabilized with oral sodium chloride 20%. Urine osmolality was elevated, serum osmolality remained normal, and copeptin levels were low. He was discharged after a 10-day hospital stay. During follow-up, hyponatremia recurred despite his typical fluid intake of 1,200 mL/day. He was instructed to limit his fluid intake to 600–700 mL, which successfully normalized his serum sodium levels. Serum levels of adrenocorticotropic hormone, the aldosterone-to-renin ratio, thyroid function tests, and serum cortisol were all within normal ranges. Plasma AVP was unusually low (4.0 pmol/L). Whole exome sequencing (WES) confirmed a likely pathogenic hemizygous variant in AVPR2 gene located on chromosome Xq28, which was inherited from his mother. He is the only child, born from a non-consanguineous marriage, with no other significant family medical history.

CONCLUSION
Nephrogenic syndrome of inappropriate antidiuresis (NSIAD), is an uncommon, yet clinically significant condition. This case highlights the importance of considering a broad range of differential diagnoses when a patient presents with hyponatremic seizure. Furthermore, obtaining a detailed family history is essential to identify potential hereditary factors that could contribute to the condition and for genetic counseling.

EP_P013

PARTIAL ANDROGEN INSENSITIVITY SYNDROME PRESENTING AS AMBIGUOUS GENITALIA IN A 46, XY INFANT

Muhammad Shafiq Safwan bin Md Latip[1], Shi Chyn Lim[1], Yee Lin Lee[1]
[1]Department of Paediatrics, Faculty of Medicine and Health Science, Hospital Sultan Abdul Aziz Shah, Universiti Putra Malaysia

INTRODUCTION
Partial androgen insensitivity syndrome is a rare X-linked 46, XY disorder caused by androgen receptor dysfunction, producing variable genital ambiguity and complex diagnostic challenges in early infancy and childhood.

CASE
We report a term infant, currently aged 9 months, who was admitted to the Neonatal Intensive Care Unit at birth for glucose-6-phosphate dehydrogenase (G6PD) deficiency and noted to have ambiguous genitalia. The infant had no hypoglycemia, feeding intolerance, or electrolyte disturbances throughout admission. She is the second child of non-consanguineous parents, with no family history of disorders of sex development. Examination of the external genitalia revealed a prominent phallus without erectile tissue with mildly pigmented and rugated labioscrotal folds. There was no labioscrotal fusion. Bilateral gonads were palpable in both labioscrotal folds with one opening seen.

Pelvic ultrasonography showed bilateral small testes within the labioscrotal folds, with absence of Müllerian structures. Karyotype analysis confirmed 46, XY genotype. Human chorionic gonadotropin stimulation (HCG) testing at day 7 of life demonstrated normal baseline testosterone 4.9 nmol/L, but minimal testosterone rise 5.4 nmol/L at day 4 post HCG. Normal testosterone to dihydrotestosterone ratio and testosterone to androstenedione ratio excluded defects in androgen synthesis, while normal adrenal steroid profile ruled out congenital adrenal hyperplasia.

Gonadotropin-releasing hormone stimulation at 2 months old revealed a peak follicle-stimulating hormone of 8 IU/L and peak LH of 4 IU/L. Anti-Müllerian hormone level was markedly elevated >328 pmol/L (normal 5.5–103 pmol/L).