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NR0B1-RELATED ADRENAL HYPOPLASIA CONGENITA PRESENTING AS A NEONATAL SALT-WASTING CRISIS: A CASE REPORT

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Background. NR0B1-related X-linked adrenal hypoplasia congenita is a rare cause of primary adrenal insufficiency that may present in early infancy with a life-threatening salt-wasting crisis and clinically mimic congenital adrenal hyperplasia (CAH).

Case presentation. A male infant born at 39 weeks of gestation (birth weight, 3,630 g) developed lethargy and poor feeding on day 3 of life. Following an initial hospitalization for feeding difficulty and poor weight gain, he was readmitted approximately one week after discharge with hypovolemia, lethargy, and a depressed suck reflex. Laboratory investigations revealed metabolic acidosis, severe hyponatremia (112 mmol/L), and hyperkalemia (7.0 mmol/L), prompting evaluation for an adrenal salt-wasting disorder. Cortisol was below the laboratory reference interval, whereas serum 17-hydroxyprogesterone was not elevated (4.27 ng/mL; reference interval, 0.79–16.71). Aldosterone was 45.18 pg/mL (reference interval, 20–180). Electrolyte abnormalities persisted despite correction of acidosis and sodium supplementation. Hydrocortisone therapy was initiated and subsequently increased; after the addition of fludrocortisone, serum sodium and potassium normalized, feeding improved, and weight gain resumed. Genetic evaluation was initially directed toward CAH. Digital MLPA analysis of dried blood spot DNA did not identify a **CYP21A2** deletion; however, the available laboratory correspondence indicated that comprehensive **CYP21A2** testing was to be performed separately. Subsequent molecular laboratory correspondence reported a hemizygous frameshift variant in **NR0B1**, **NM_000475.5:c.595delT, p.(Ala194Argfs*70)**, providing molecular support for the diagnosis of X-linked adrenal hypoplasia congenita.

Conclusion. This case highlights NR0B1-related adrenal hypoplasia congenita as an important differential diagnosis in male neonates presenting with severe salt wasting. When 17-hydroxyprogesterone is not elevated, and the initial evaluation does not support classic 21-hydroxylase deficiency, consider alternative causes of primary adrenal insufficiency, with

molecular genetic evaluation helping define the underlying etiology.
Keywords: NR0B1; adrenal hypoplasia congenita; neonatal adrenal insufficiency; salt-wasting crisis; congenital adrenal hyperplasia; frameshift variant