

Antonia Almeida¹ • Vinicius Aniceto¹ • Samara Reis¹ • Marcos Okido¹ • Weverton Luchi² • Conrado M. Coutinho¹

¹Hospital das Clínicas, Ribeirão Preto Medical School, University of São Paulo (HCFMRP-USP) • ²Hospital Universitário Cassiano Antonio Moraes, Federal University of Espírito Santo (HUCAM-UFES)

BACKGROUND

Polyhydramnios may have maternal, fetal or idiopathic causes. Among fetal etiologies, structural anomalies and genetic disorders are notable, including renal tubulopathies. We report a rare case of polyhydramnios associated with a hereditary salt-losing tubulopathy (SLT).

CASE REPORT

A 28-year-old G3P1A1 woman at 27+5 weeks of gestation presented with severe polyhydramnios, requiring four amniodrainages. Infectious screening was negative, and no maternal disease was identified. Amniotic fluid electrolytes, fetal karyotype (46,XX) and fetal MRI were normal, and the parents were non-consanguineous. Given the suspicion of a SLT, empiric indomethacin was initiated with partial response. Cesarean occurred at 35+3 weeks. The newborn presented metabolic alkalosis, hypokalemia, hyperchloruria, hypercalciuria and hyperreninemic hyperaldosteronism with normal blood pressure, suggesting a hereditary SLT. Postnatal ultrasonography revealed nephrocalcinosis and genetic testing identified a homozygous KCNJ1 missense variant, confirming neonatal BS.

DISCUSSION & CONCLUSION

This case of severe polyhydramnios without maternal or infectious causes, followed by neonatal electrolyte disturbances and nephrocalcinosis, raised suspicion for antenatal BS. It involves impaired electrolyte reabsorption in the thick ascending limb of Henle’s loop, leading to fetal polyuria, polyhydramnios and prematurity. Prenatal ultrasound findings, such as hyperechoic medullary pyramids or diffuse renal echogenicity, may support the diagnosis. When suspected, the Bartter index (BI) can be calculated by multiplying amniotic fluid total protein and alpha-fetoprotein levels; a BI < 0.5 suggests Bartter syndrome with high sensitivity and specificity. In cases where common causes of polyhydramnios are excluded, genetic tubulopathies should be considered. The presence of prenatal nephrocalcinosis further strengthens it and the BI may support prenatal diagnosis. Early recognition is essential for appropriate perinatal management and genetic counseling.

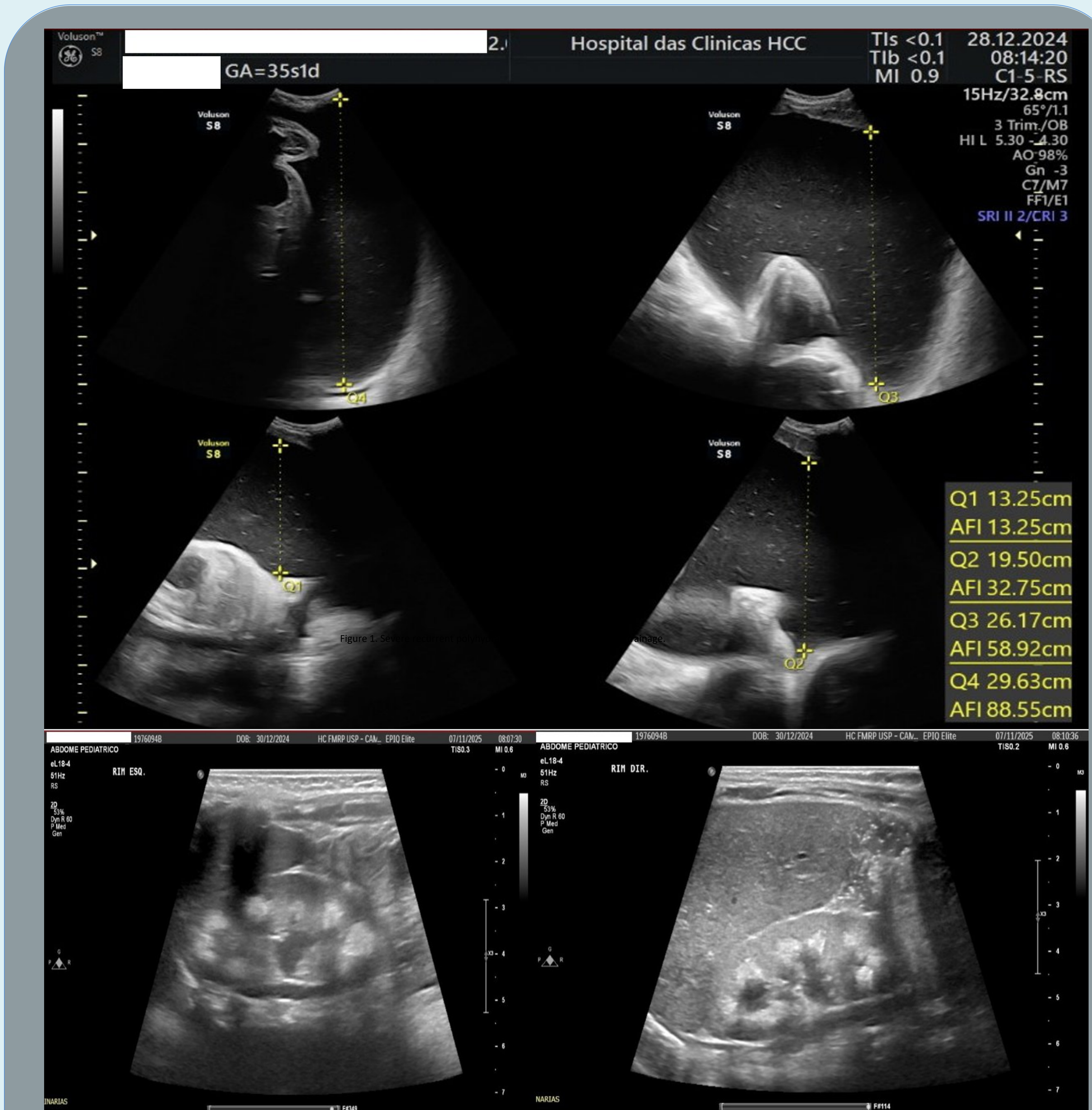


Figure 1. Prenatal obstetric ultrasound demonstrating massive polyhydramnios (above). Postnatal renal ultrasound demonstrating nephrocalcinosis (below).