A NEWBORN WITH PERSISTENT HYPERKALEMIA.

Marjan Klok-van Ledden and Ihor V. Yosypiv.

Department of Pediatrics, Tulane University Health Sciences Center, New Orleans, LA.

A 19 day old female born at 38 weeks of gestation after an uncomplicated pregnancy presented to nephrology clinic with serum potassium of 7.0 mmol/L in a non-hemolyzed sample. Family history, review of systems and physical examination were unremarkable with normal blood pressure of 88/52 mm/Hg. Notably, patient’s newborn screen for congenital adrenal hyperplasia was negative. Initial laboratory assessment showed normal renal function and serum sodium, absence of acidosis or urinary tract infection (UTI), presence of microcytic anemia with hemoglobin (Hb) of 10.8, hematocrit of 32.9 and mean corpuscular volume of 86. Renal ultrasonography ruled out presence of congenital structural abnormalities of the urinary tract. Hb electrophoresis revealed a fast moving Hb suggestive of α-thalassemia. Genotype was positive for 2 copies of 3.7kb α-Hb deletion confirming presence of a carrier state of α-thalassemia. Given that this condition in itself is unlikely to account for hyperkaliemia in the absence of documented hemolysis, further workup was pursued and revealed normal plasma renin (10 ng/ml/hr), aldosterone (17.8 ng/dl) and cortisol (10 µg/dl) levels. The calculated transtubular potassium gradient (TTKG) in the presence of serum potassium of 5.8 was low at 4.0. A low TTKG coupled with normal plasma aldosterone level suggested presence of pseudohypoaldosteronism (PHA). Absence of systemic hypertension or extrarenal manifestations was not consistent with the presence of PHA2 (due to inactivating mutations in WNK1 and WNK4) or PHA1 due to inactivating mutations in the epithelial sodium channel, respectively. A diagnosis of PHA1, which involves the mineralocorticoid receptor defect, was made. Patient was treated with kayexalate, a potassium-depleting resin, and sodium chloride to increase distal delivery of sodium and maximize potassium secretion into the urine. These therapeutic interventions resulted in normalization of serum potassium levels. In view of worsening of anemia, the patient was also referred to hematology for the evaluation of α-thalassemia. It is important to note that transient PHA may also occur in premature infants or due to UTI. Renal dysplasia or small bowel resection may also lead to PHA.