Nephrogenic syndrome of inappropriate antidiuresis in a 19 months old child with gross developmental delay

Sodium plays an important role in normal physiological function of cells, especially neurons. There are several conditions that could affect sodium maintenance in the body with potential severe neurological outcomes. We present a case of rare genetically predisposed cause of hyponatremia due to mutation in vasopressin receptor in kidneys.

19 months old African American boy with nonsignificant past medical history presented with inability to walk, hypotonia, chewing difficulties, and speech delay. Initial laboratory evaluations demonstrated serum hyponatremia of 120-129 mEq/dl, low serum osmolality 261 mosm/kg (normal serum osmolality 275-295 mosm/kg) with inappropriately high urine osmolality 445 mosm/kg (normal urine osmolality 300-1000 mosm/kg). Blood work for thyroid, adrenal, and pituitary function was unremarkable. Vasopressin level of 2.4 pg/ml (normal 0.0 - 4.7 pg/mL) did not support the diagnosis of Syndrome of Inappropriate Antidiuretic Hormone Secretion (SIADH). Fludrocortisone challenge test ruled out aldosterone deficiency with insufficient decrease in urine sodium and no response in serum sodium. Renal ultrasound and brain MRI were normal. Genetic testing revealed AVPR2 X-linked mutation c.409C>T p.R137C, consistent with nephrogenic syndrome of inappropriate antidiuresis (NSIAD). The patient was discharged on sodium chloride tablets 2g every 8 hours and remained stable, asymptomatic. One month after discharge patient showed significant neurological improvement.

Sodium is known to play a significant role in growth and maturation of the whole body, especially of the nervous system. Normal fluid and electrolyte balance is achieved by the thirst mechanism and free water excretion in kidneys. Hereditary nephrogenic syndrome of inappropriate antidiuresis is very rare mutation in vasopressin receptor, which causes gain of function and inappropriately high water retention.