Congenital Nephrogenic Diabetes Insipidus: A case report in a Hispanic male

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Background: Diabetes Insipidus (DI) is a rare condition that manifests with large volumes of diluted urine and plasma hyperosmolarity secondary to decreased production or response to antidiuretic hormone (vasopressin). Congenital forms represent <10% of cases. We present a case of familial nephrogenic DI in a male with multiple family members with the same clinical findings.

Case presentation: A 32 year-old-male with a history of long-standing polydipsia (10 L/day) and polyuria, with similar symptoms in his son and nephew, was admitted to the intensive care unit following a motor vehicle accident. The patient was initially sedated and intubated and his sodium level at admission was 140 mmol/L. He developed severe hypernatremia 6 hours after arrival (Na+ 163 mmol/L), with a serum hyperosmolarity of 340 mOs/kg and urine hyperosmolality of 191 mOs/kg. Trauma imaging per protocol showed pneumocephalus within the pituitary fossa and fracture in the superior margin of the nasal bone. A high-dose desmopressin challenge test failed to improve the hypernatremia and urine output. This along with his family history established the diagnosis of congenital nephrogenic DI. We started treatment with indomethacin 50 mg BID and hydrochlorothiazide 25 mg BID, which improved the polyuria and decreased sodium levels to Na+ 138 mmol/L.

Conclusions: Increased serum sodium and failure to concentrate the urine in the presence of high plasma vasopressin and parenteral administration of desmopressin are diagnostic of NDI. Molecular genetic testing is also done to identify congenital etiologies. This rare condition is often overlooked due to the variable symptomatology among family members and socioeconomic status, especially among the South Texas Hispanic population. The intrinsic social determinants of health in our population limit the opportunity to seek genetic testing and to afford treatment. Treatment goals include reducing polyuria by up to 50%, thiazide diuretics in standard to high doses, often in combination with NSAIDs or potassium-sparing diuretics as well as dietary sodium restriction to 1mmol/kg/day. Diagnosis and management of these patients at an early age can improve their quality of life and empower them to have regular employment opportunities.

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