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A case of hypodipsic hypernatremia in autosomal dominant polycystic kidney disease

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Introduction: Serum osmolarity is maintained by two major responses: thirst and antidiuretic hormone(ADH) secretion. Any defects in these defense mechanisms could result in osmolar & electrolyte imbalance. Autosomal dominant polycystic kidney disease(ADPKD) is an inherited disease characterized by progressive formation of multiple cysts in both kidneys, often accompanied by extra-renal manifestations such as cerebral aneurysms, hepatic & pancreatic cysts, hypertension. Here, we introduce a case of 58-year-old male with ADPKD presented with hypernatremia caused by hypodipsia. **Case:** A 58-year-old male with ADPKD visited our nephrology clinic with hypodipsia started several months ago. He underwent neck clipping of right anterior communicating & left posterior communicating artery aneurysm 13 years ago(Fig.1). From that time, his serum creatinine level maintained 1.2-1.4 mg/dL. His systolic/diastolic blood pressure was 112/51 mmHg and pulse rate was 45 /min. Physical examination revealed decreased skin turgor. Laboratory study showed serum blood urea nitrogen 43.2 mg/dL, creatinine 2.22 mg/dL, osmolarity 349 mOsm/kg, sodium 163 mmol/L and urine osmolarity 638 mOsm/kg, sodium 117mmol/L. He was admitted for intravenous hydration and we planned urine output & serum ADH level measurement, water deprivation test & pitressin test to make a differential diagnosis among diabetes insipidus, essential and hypodipsic hypernatremia. He was not polyuric. During water deprivation test, urine osmolarity decreased, so we stopped the test and urine concentration was revealed after desmopressin injection. His serum ADH level was 2.6-2.9 pg/mL which seems at lower margin of normal range and it did not change despite serum & urine osmolarity changed. After free water hydration, azotemia and hypernatremia improved. He was discharged after education of oral hydration of free water, 1-1.5 L/day. **Conclusion:** In this case, hypernatremia was due to hypodipsia occured by destruction of osmoreceptors in anterior hypothalamus that regulate thirst. It was caused by the anterior communicating aneurysm accompanied by ADPKD. Hypodipsic hypernatremia can be corrected by just adding water.

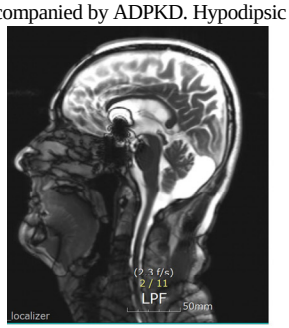

Figure 1.

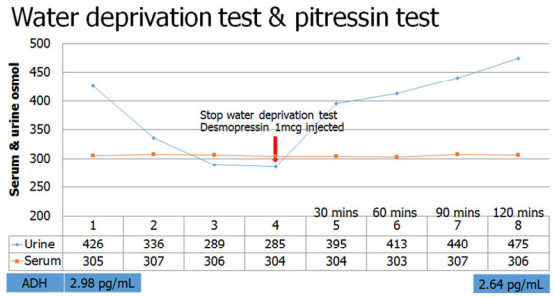

Figure 2.

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Monitoring globotriaosylsphingosine before and after enzyme replacement therapy in Fabry disease

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Fabry disease is an X-linked lysosomal storage disorder caused by deficiency of α-galactosidase A activity. It leads to dysfunctions in multiple organs, including the kidney, heart, and brain. Representative deposits are globotriaosylceramide (Gb3) and globotriaosylsphingosine (lyso-Gb3). Recent studies have proposed lyso-Gb3 to be a therapeutic biomarker. Herein, we report a case in which lyso-Gb3 was traced before and after enzyme replacement therapy in a patient with Fabry disease. A 29-year-old man was diagnosed with Fabry disease on the basis of reduced α-galactosidase A enzyme activity in leukocytes (3.3 nmol/h/mg protein) and α-galactosidase A gene mutation (Asp92Gly). At baseline, the patient had angiokeratoma, decreased kidney function (serum creatinine, 1.90 mg/dl; estimated glomerular filtration rate, 46.6 ml/min/1.73 m2), proteinuria (random urine protein-to-creatinine ratio, 3.1 g/g), cardiac basal septal wall hypertrophy (14 mm), and a suspected ischemic lesion in the right posterolateral putamen (6-mm-high signal intensity on a T2-weighted-Fluid-Attenuated Inversion Recovery image, and low intensity on a T1-weighted brain magnetic resonance image). A biopsied specimen from the kidney showed diffuse lipid deposits in podocytes, glomerular endothelial cells, and mesangial cells. The patient was treated biweekly with agalsidase beta (Fabrazyme®), and both plasma Gb3 and lyso-Gb3 levels were estimated every 3 months. Before enzyme replacement therapy, the plasma Gb3 and lyso-Gb3 were 10.9 μg/ml (reference range, 3.9–10.9 μg/ml) and 108.0 ng/ml (reference range, ≤1.74 ng/ml), respectively. After 6 months of treatment, the levels of Gb3 and lyso-Gb3 dropped to 6.4 μg/ml and 18.7 ng/ml, respectively. During the treatment period, kidney function did not decrease further (serum creatinine, 1.81 mg/dl; random urine protein-to-creatinine ratio, 2.2 g/g). The present case suggests that lyso-Gb3 can be useful in monitoring the response to enzyme replacement therapy.