Case Report of Syndrome of Nephrogenic Inappropriate Antidiuretic Hormone Secretion (SIADH) Caused by Rare AVPR2 Gene Active Mutation

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Objective:
The AVPR2 gene mutation usually cause nephrogenic diabetes insipidus. We report a patient who carried an active gene mutation of AVPR2 presenting persistent hyponatremia, finally diagnosed the syndrome of nephrogenic inappropriate antidiuretic hormone secretion (NSIADH)

Method:
We describe a patient whose clinical and laboratory evaluation were consistent with hyponatremia, which hardly to be corrected to normal. After the AVPR2 gene mutation identified, the level of serum sodium was increased with furosemide orally.

Result:
The patient was 5 years old boy with several times generalized seizures caused by hyponatremia in 2 years. His sister was in good health. There was no family history of hyponatremia. He showed apathy with otherwise normal physical examinations. His blood pressure was normal, his height and weight were in normal range. Initial laboratory evaluations demonstrated hyponatremia of 120 mmol/L with inappropriately elevated urinary sodium levels of 181.7 mmol/L, and the serum levels of potassium and bicarbonate were normal. The serum osmolality was lower to 252 mOsm/L. He had normal blood urea nitrogen and low serum creatinine levels. Adrenal hormone and thyroid function tests were all normal. Imaging studies of the head and chest were unremarkable. Despite clinical and laboratory presentations consistent with the presence of NSIADH, we cannot find any pathogenic reasons. The hyponatremia was hardly to be corrected by high doses of sodium supplementation.

DNA sequencing of the patient's AVPR2 gene identified missense mutations of nucleotide 770 mutated from cytosine to thymine, which changed codon 137 from arginine to cysteine. This mutation was reported as constitutive activation of AVP receptor by other researchers.

The patient was initially treated with fluid restriction and high doses of sodium supplementation, but both were useless. He was treated with furosemide, resulting in increased urinary output and normalization of the serum sodium level.

Conclusion:
The gene mutation can cause nephrogenic diabetes insipidus and NSIADH, depending on the kinds of mutations. The administration of furosemide normalized the serum sodium level and increased the urinary output. The AVP antagonist of Tolvaptan could suppress AVP receptor activity, which could prescribe to NSIADH, but is more expensive than furosemide.

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