"IDIOPATHIC"- THE TRULY UNKNOWN OR YET ANOTHER HIDING PLACE FOR AUTOIMMUNITY?

A CASE OF CENTRAL DIABETES INSIPIDUS IN A YOUNG WOMAN WITH HASHIMOTO THYROIDITIS

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Introduction

Central diabetes insipidus is a rare, chronic disease characterized by polyuria and polydipsia due to a partial or total vasopressin deficiency (hypothalamic-neurohypophysial system damage). The ethiology can be familial (autosomal dominant inheritance, X-linked recessive traits; mutation of the vasopresin-neurophysin II genes), secondary (tumors, infections, infiltrative diseases, trauma, vascular lesions) and idiopathic (10-30%).

Case presentation

We present the case of a 24 year-old female who presented in our clinic for a polydipsia- polyuria syndrome (10 liters/day) with a sudden onset 3 months prior her admission. She had a familiy history of Hashimoto thyroiditis and Diabetes Mellitus type I, and a personal history of Hashimoto thyroiditis with hypothyroidism for which she had been taking treatment with L-thyroxine for almost one year.

Physical examination: overweight (BMI 28 kg/m2), blood pressure 120/ 95 mmHg, heart rate 108 bpm, no signs or symptoms of pulmonary disease. She had no modifications in circadian rhythm or appetite, normal menstrual cycle, no history of head trauma, pregnancy or childbirth.

<u>Lab tests:</u> routine investigations and inflammatory markers were normal, endocrinological testing revealed a normal anterior pituitary function and a balanced hypothyroidism sustitution (TSH=3.01microIU/ml, FT4=1.47ng/dl). The water deprivation test and ADH value when the test ended (<1 ng/L) were suggestive for central diabetes insipidus.

Hypothalamic- pituitary MRI with contrast showed no pathological modifications except for the absence of the physiological posterior pituitary bright spot.

The patient received long- term oral desmopressin treatment and her fluid intake and output normalised.

Conclusions

Central Diabetes Insipidus is rare chronic disease, even rarer in the case of young adults. When no familial or secondary cause of vasopressin deficiency is feasible, vascular lesions and autoimmune processes must be taken in consideration before labeling it as "idiopathic". In this particular case, given the personal and familial history of autoimmune diseases, despite the non-suggestive MRI findings, anti- pituitary and vasopressin-cell autoantibodies should be determined as an attempt to look even further and truly define a diagnosis.





