CR-GE-25

A CASE OF CLASSICAL CONGENITAL HYPERPLASIA (CAH) PRESENTING WITH BILATERAL ADRENAL MASSES

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INTRODUCTION

Congenital Adrenal Hyperplasia (CAH) presenting with adrenal masses has been reported to be less than 0.4%. We report a 25-year-old diagnosed with simple virilizing Classical Congenital Adrenal Hyperplasia (CAH) with bilateral adrenal masses.

CASE

A 25-year-old phenotypic male came in due to right flank pain. He has an ambiguous genitalia. He has male secondary features and was raised as male since birth. Karyotype showed a female genotype.

CONCLUSION

It is recommended that all patients with ambiguous genitalia be tested for karyotyping and evaluation should be undertaken as soon as possible for immediate management and intervention.

KEY WORDS

congenital adrenal hyperplasia, bilateral adrenal mass, ambiguous genitalia

CR-GE-26

A CLINICAL FEATURE OF SEVERE HYPERTRIGLYCERIDEMIA IN REPRODUCTIVE WOMAN WITH PRIMARY POLYDIPSIA

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INTRODUCTION

Severe hypertriglyceridemia defined as triglyceride level above 1000 mg/dL may cause fatal cardiac event due to its atherogenic impact and also acute pancreatitis. This report described severe hypertriglyceridemia that occurred coincidentally with polyuria related to a psychogenic disorder.

CASE

A 35-year-old woman came to our clinic presenting with polyuria more than 6.5 L/day. She complained of nocturia more than 10 times/night with absence of weight loss and polyphagia. She denied consuming excessive water. She complained of dyspnea with absence of orthopnea. 2D-echocardiography showed concentric left ventricular hypertrophy with preserved EF (62.9%). She is obese (BMI 30,9 kg/m²) with absence of diabetes mellitus, acanthosis nigricans and xanthomas. Her morning urine osmolality was low (213 mOsm) with normal plasma osmolality (283 mOsm). Her triglyceride was high (2375 mg/dL), high total cholesterol (385 mOsm), low HDL (25 mg/dL) and low LDL (48 mg/dL) wih normal blood glucose (70 mg/dL) and normal HbA1C 5.4%. In the 5th hour of water deprivation test, her urine osmolality exceeds 600 mOsm (688 mOsm). Dynamic contrast pituitary MRI revealed no intracranial lesions. Treatment with fibrates and water restriction showed gradual improvement in triglyceride level with last result 305 mg/dl. She was diagnosed with mixed depression and anxiety due to her cervical carcinoma in situ.

Certain literature has stated that hypertriglyceridemia can be secondary to other diseases including diabetes insipidus. Water deprivation test can be applied to differentiate diabetes insipidus and primary polydipsia. Since pituitary MRI with contrast showed absence of abnormality, psychogenic polydipsia was considered in this patient. Anxiolitic treatment relieved polyuria because of reduction in water consumption. Fibrates were chosen to reduce the high triglyceride level.

CONCLUSION

Hypertriglyceridemia can be primary or secondary. After exclusion of diabetes mellitus, hypothyroidism, alcohol consumption and drug effect, the precipitant of severe hypertriglyceridemia in this patient was the primary polydipsia. Lipid lowering agent and avoidance of excess water consumption led to significant improvement in triglyceride level.

KEY WORDS

phepertriglyceridemia, desmopressin acetate, gemfibrozil