A Case with Congenital Adrenal Hyperplasia Diagnosed by Malnutrition

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Introduction:

Congenital adrenal hyperplasia is an autosomal recessive disorder characterized by enzyme deficiencies in the adrenal steroidogenesis pathway. The most common type is 21 hydroxylase deficiency and is divided into two groups as classical and nonclassical type. 75% of the classical type of cases is salt-losing type, and cortisol and aldosterone deficiency symptoms occur in patients. Female cases presented with ambiguus genitalia due to hyperandrogenemia at birth. In male cases, scrotal hyperpigmentation and macropenis are present.

Case:

A 7-month-old male patient was referred to us because of the high level of TSH in his laboratory examination. His past medical history revealed that he was born with 3000 grams and he did not have any complaints such as hypoglycemia, concomitant vomiting or diarrhea. It has been learned that the child has been followed up by the department of gastroenterology for developmental delay since 3 months.His parents are first degree cousins.

On his physical examination weigth: 4.7 kg (-3.92SDS), height: 62 cm (-2.61SDS), he was malnourished, thyroid gland was nonpalpable, axillary hair was negative, pubic hair was Tanner stage1, testes were 2/2 ml and
palpable bilateral in scrotum, penis length was 3 cm, there was no scrotal hyperpigmentation.

In the laboratory; the values: fT4 : 1.01 ng/dl, TSH: 10.5 uIU/ml, glucose: 75 mg/dl, sodium: 127.9 mmol/L, potassium:5.74 mmol/L were found. ACTH:41.8 pg/ml, cortisol: 4.42 ug/dl, 17 OH progesterone >19.2 ng / ml, renin 2.59 ng / ml / h(0,3-4,88), aldosterone 19.1 ng / dl (5-90) , the patient was hospitalized because of mineralicorticoid deficiency symptoms and the standard dose ACTH stimulation test was performed.
ACTH at baseline: 14.3 pg / ml, cortisol: 0.68 ,6g / dl, 17 OH progesterone: 2.26 ng/ml; peak cortisol: 1.47 :g / dl, peak 17 Oh progesterone: 92.3 ng / ml. Hydrocortisone 15 mg/m2/day, fludrocortisone 2x 0.1 mg and 3 gr/day oral salt treatment was started with diagnosis of adrenal insufficiency. In the genetic analysis, the CYP21A2 gene revealed a c.293-13C> G homozygous mutation previously reported in the literature. The patient's parents were also shown to be carriers of the same mutation.

Conclusion:

This case was diagnosed because of hyponatremia and hyperkalemia at 7 months of age due to malnutrition. There is no macropenis and scrotal hyperpigmentation in the clinic and there is no hypoglycemia or vomiting symptoms. Despite aldosterone deficiency findings, he did not show low cortisol and aldosterone levels at first application. Tuhan et al defined this condition in their case report ,high aldosterone level was suggested to be associated with cross-reactivity of adrenal steroid precursors (including 17-OH progesterone), and the radioimmunoassay (RIA) technique was defined as the responsible method for cross-reactivity. The normal cortisol level may reflect the still active steroid production in the fetocortex.
This case is presented because of its late diagnosis and presentation to a different clinic.

References:

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