RELATIVE HYPOALDOSTERONISM IN A PATIENT WITH WOLCOTT-RALLISON SYNDROME

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INTRODUCTION

Wolcott-Rallison syndrome (WRS) is a multi-system disorder with autosomal recessive transmission and early onset of diabetes in infancy.

CASE REPORT

A 9 year old girl presented to our emergency department with ketoacidosis and multi-organ failure. She had been followed-up due to diabetes and growth failure from another centre. Evaluation for short stature [height: 80.4 cm (SDS: -0.9), weight: 10.2 kg (SDS: -7.1), head circumference: 46.5 cm (SDS: -3.94), prepubertal] after remission of ketoacidosis revealed epiphyseal dysplasia (Figure 1).

![Figure 1. Bilateral knee and left hand and left wrist radiographs of the patient with Wolcott-Rallison syndrome indicating epiphyseal dysplasia](image)

A homozygous nonsense p.W521* mutation in EIF2AK3 gene confirmed the clinical diagnosis of WRS (Exeter, UK, Prof Sian Ellard, Dr Jayne Houghton). On follow-up, serum sodium levels ranged between 126 meq/l to 130 meq/l, and serum potassium ranged between 5.8 to 6.3 meq/l. Venous blood glucose was between 135 to 210 mg/dl. Hormonal work-up while the patient was given no intravenous fluid (serum sodium: 129 meq/l, serum potassium: 6.2 meq/l) was as follows: ACTH: 31 pg/ml, cortisol: 18.7 μg/dl, aldosterone (upright: 241.3 pmol/l), plasma renin level: 39 pg/ml (normal range: 5.41-34.53 pg/ml). Estimated glomerular filtration rate-Schwarz was 36.9 ml/min/1.73 m², and the result was consistent with stage 3 chronic renal failure. Transtubular potassium gradient (TTKG) [(Urinary potassium+[Urinary osmolality/plasma osmolality])=plasma potassium] was 1.39 (urinary potassium: 20 meq/l, urinary osmolality: 649 mosmol/kg, plasma osmolality: 288 mosmol/kg). Relative hypoaldosteronism was diagnosed, and a diet poor in potassium and rich in sodium was started. Failure of response to dietary intervention prompted a trial of fludrocortisone (FC) at a dose of 0.05 mg/day. The TTKG calculated four hours after fludrocortisone was 5.3. Serum sodium was 137 meq/l and potassium was 4.1 meq/l on the third day of therapy.

CONCLUSIONS

Relative hypoaldosteronism should be suspected in metabolically stable patients with Wolcott-Rallison syndrome in case of persistently elevated potassium levels. An aldosterone agonist can be considered when conventional approaches fail.

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