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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:80.4cm (SDS:-8.9), weight:10,2 kg (SDS:-7.1), head circumference:46.5cm (SDS:-3.94), prepubertal] after remission of ketoacidosis revealed epiphysial dysplasia (Figure 1).

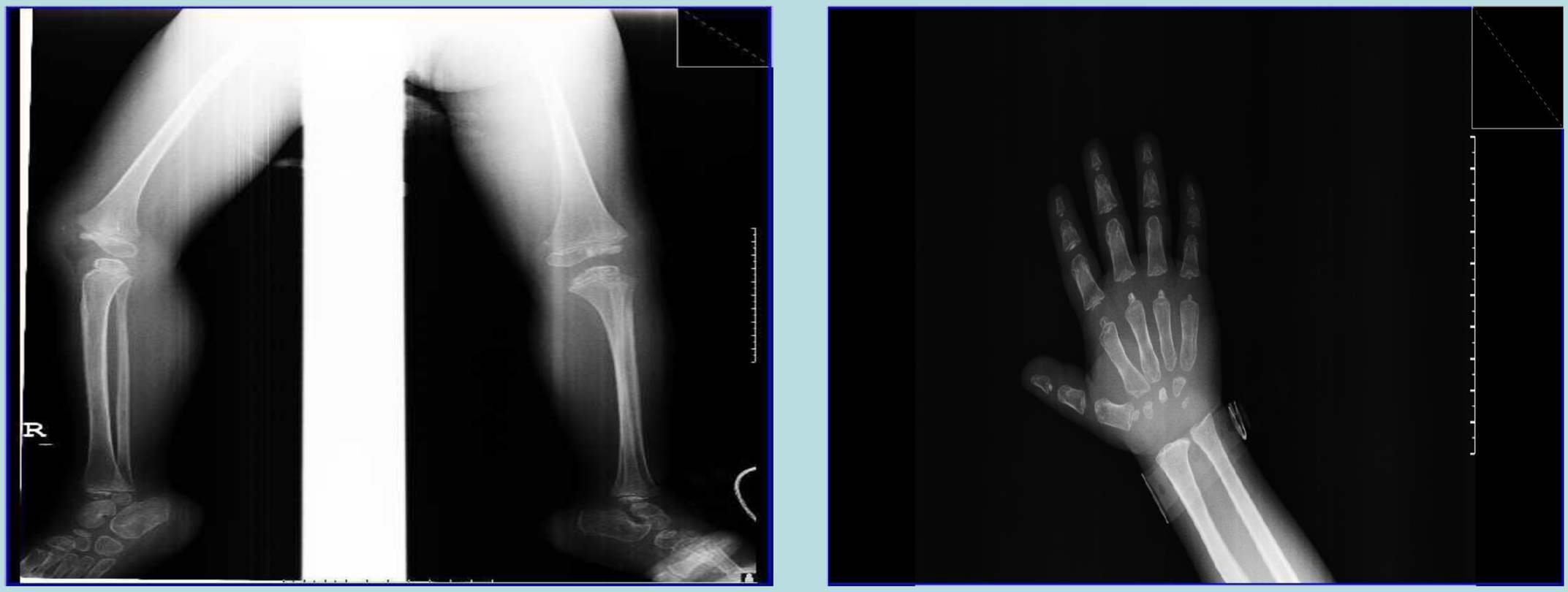

Figure 1. Bilateral knee and left hand and left wrist radiographs of the patient with Wolcott-Rallison syndrome indicating epiphyseal dysplasia

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, aldosteron (upright: 241.3 pmol/l), plasma renin level: 39 pg/ml (normal range: 5,41-34,53 pg/ml). Estimated glomerular filtration rate-Schwartz 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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