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Congenital heart disease in an infant with pseudohypoaldosteronism type 2 (Gordon syndrome)

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Pseudohypoaldosteronism type 2 (PHA2), also called Gordon's syndrome, is a rare autosomal dominant disease characterized by familial hypertension and hyperkalaemia; distal renal tubular acidosis despite normal glomerular filtration; normal or low normal serum aldosteron and plasma renin activity; and hypercalciuria and the increased risk of urolithiasis. All findings may not be determined in the infancy period. The congenital heart disease in PHA2 has not been reported previously. We, here, report an infant with PHA2 associated with cyanotic congenital heart disease.
A one-month-old boy, ninth child of a non-consanguineous marriage, was referred to our pediatric clinic with a history of cyanosis. He was born at term after unremarkable pregnancy. There was no family history of renal or other medical diseases. His weight and height were 4890 g and 57 cm (both 25-50th percentile), respectively. Physical examination was unremarkable apart from cyanosis and a soft systolic ejection murmur at the upper left sternal border. Vital signs were normal. Serum electrolytes, hepatic and renal function tests were normal. Echocardiography revealed complete atrioventricular septal defect; pulmonary atresia; and hypoplasic left ventricle and pulmonary arteries. Angiography confirmed the diagnosis. After 15th day of the admission he developed persistent hyperpotassemia. He was not receiving any drug leading to hyperkalaemia. The laboratory findings were as follows: serum sodium 139 mmol/l (reference range 135-145), potassium 6.4 mmol/l (3.5–5.5), chloride 103 mmol/l (98-106), creatinine 0.3 mg/dl (0.3-0.7), blood urea nitrogen 4.2 mg/dl (5-18), calcium 10 mg/dl (8.8-10.8), phosphorous 4.4 mg/dl (3.7-5.6), aldosteron 74 ng/dl (5-90), plasma renin activity 9.5 ng/ml/h (<16.6), trans-tubular potassium gradient 4.2 (>8), and spot urine calcium/creatinine 0.2 (<0.8). Venous blood gases were as follows: pH: 7.41 (7.35-7.45), PCO2 33 mmHg (35-45) and HCO3 21 mmol/l (22-29). Renal ultrasound was normal. He diagnosed as having PHA2. Hydrochlorothiazide (1 mg/kg/d) was given. On third day of hydrochlorothiazide, serum potassium level returned to normal (4.2 mmol/l).
PHA2 should be considered in patients with an isolated hyperkalaemia. This case suggests PHA2 may be associated with congenital heart diseases. This is an unreported constellation.