A CASE REPORT OF AN INFANT WITH IDIOPATHIC HYPERTENSIVE CRISIS IN A 16-YEAR OLD GIRL WITH NPHRIPATHY AND NEPHROLITHIASIS ASSOCIATED WITH CYP24A1 ENZYME POLYMORPHISM

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CYP24A1 is an enzyme that inactivates vitamin D and encodes vitamin D 24-hydroxylase. Mutations in this enzyme have been linked with idiopathic infantile hypercalcemia, nephrolithiasis, and nephrocalcinosis. The genetic testing for this mutation should be considered in the presence of hypercalcemia, elevated serum calcium, elevated 24- hour elevation vitamin D, and suppressed parathyroid hormone.

We present a previously healthy eight-month-old male infant with macroscopic hematuria. He was born full-term with no perinatal risks. Infant was breastfed for up to four months, and afterwards fed with milk formula, some mixed fruits and vegetables, supplemented with vitamin D according to the recommendations. The family history was negative for nephrolithiasis and urinary tract abnormalities. On the admission, he was in good general condition, afibrile, with normal vital parameters. His body weight was 8.05 kg (15th centile), length 76 cm (94th centile), and head circumference 46.1 cm (77th centile), without any deviations in his clinical examination. In laboratory findings, there were 90% dysmorphic erythrocytes in urine and elevated calcium/creatinine ratio (>1.5 mmol/mmol). 24- hour urine sample showed hypercalciuria (6 mg/kg), albuminuria (54 mg/24h). The values of alpha-1-microglobulin, parathyroid hormone, hypercalciuria (6 mg/kg/24h), and citrates in urine and coagulation tests were normal.

In conclusion, we omitted vitamin D supplementation. Initially we had a good therapy response, but considering relapse of hypercalciuria after lowering the dose of hydrochlorothiazide (0.5 mg/kg) and known risk of non-melanoma skin cancer in patients on hydrochlorothiazide, the therapy was changed to potassium citrate (2 mmol potassium ion/kg/day). During the follow-up, there was no relapse of macrohematuria, the infant was in good general condition with all tests within reference values. The ultrasound of the urinary tract remained unchanged.

Children presenting with hypercalcemia, hypercalciuria and nephrolithiasis should be tested because of the importance of recognition, genetic diagnosis and proper treatment of CYP24A1 mutations that can present with a wide range of phenotypic presentations, from asymptomatic to chronic renal disease.

HYPERTENSIVE CRISIS IN A 16-YEAR OLD GIRL WITH ACCESSORY RENAL ARTERY: A CASE REPORT

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Introduction The most common pattern of kidney vascularisation is a single renal artery originating from the abdominal aorta. However, in 20-30% of general population an accessory renal artery can be found being more frequently present (up to 80%) in patients with essential hypertension.

A possible pathomechanism of hypertension is the impaired renal perfusion since the diameter of a single renal artery is usually larger than when multiple arteries are present. Whether an accessory renal artery could be a cause of hypertension is still controversial.

Case Presentation We describe a case of a 16-year old girl who presented with hypertension crisis. She was previously healthy with no record of abnormal blood pressure (BP).

At admission she reported nausea and severe headache while her BP was 220/120 mmHg. Her body mass index was normal and physical examination unremarkable.

Initial workup showed normal renal function with normal serum electrolytes and plasma glucose. Urine dipstick and urine toxicology screen were also normal. She had hypercholesterolemia and mild proteinuria but no signs of other target organ damage (electrocardiogram, echocardiography, fundoscopy and computed tomography of the brain were normal). Ambulatory blood pressure monitoring confirmed severe ambulatory hypertension.

Further evaluation was aimed at determining the possible cause of secondary hypertension. Urine metanephrines, urinary free cortisol, plasma cortisol, ACTH and thyroid function tests were within reference ranges. High normal plasma renin with elevated plasma aldosterone led to a suspicion of renovascular hypertension.

Magnetic resonance imaging revealed no pathology of the adrenal glands. Although renal ultrasonography with Doppler was normal, magnetic resonance angiography and later CT angiography showed two nonstenotic right renal arteries.

Conclusion Although nonstenotic, an accessory renal artery should be considered as a possible cause of renovascular hypertension in children and adolescents.