

Kidney stone and urinary bladder telangiectasia in a patient with TAR syndrome

İpek Akil¹, Salih Gözmen², Ömer Yılmaz², Can Taneli²

Departments of ¹Pediatric Nephrology and ²Pediatric Surgery, Celal Bayar University Faculty of Medicine, Manisa, Turkey

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TAR syndrome is a congenital malformation syndrome characterized by bilateral absence of the radius and thrombocytopenia. The known urinary anomalies are duplex ureter, dilatation of renal pelvis, horseshoe kidney and functional problems like vesicoureteral reflux and pyelonephritis. In this report of a case with TAR syndrome, a kidney stone and bladder telangiectasia were found coincidentally during the investigation of hematuria. TAR syndrome is discussed in the light of the medical literature. To our knowledge, no case has been reported demonstrating nephrolithiasis and bladder telangiectasia in TAR patients.

Key words: kidney stone, bladder telangiectasia, TAR syndrome.

Thrombocytopenia and absence of the radii (TAR syndrome) was first described in 1951¹. It consists of gastrointestinal, hematological and cardiac anomalies². The incidence of TAR syndrome is 0.42/100,000. The basic reason for skeletal and hematological anomalies is an unknown etiology occurring during 6-8 weeks of gestational age. The main problem for patients with TAR syndrome is hemorrhage due to thrombocytopenia, which can appear as intracranial hemorrhages, hematuria, and mucosal bleeding, especially from the gastrointestinal system. Intracranial hemorrhage is important because of its lethal potential³.

In this report, a case of TAR syndrome accompanied with hematuria, kidney stone and bladder telangiectasia is discussed.

Case Report

A nine-year-old male patient with TAR syndrome was admitted with complaints of thrice-repeated bright red urine voids within a period of three years. He had no complaints of upper respiratory tract infection symptoms, pain in abdomen or dysuria. He had no family history of nephrolithiasis or renal disease. His weight was 22 kg (3-10 percentile), and height was 114 cm (<3 percentile). The blood pressure was 100/70 mmHg. He had no radii as a component

of TAR syndrome (Fig. 1). His hemoglobin was 14.2 g/dl, hematocrit 41.5%, and white blood cell and platelet counts 4350 and 62000/mm³, respectively. The renal function tests were within normal ranges. Microscopic hematurias with no accompanying erythrocyte dysmorphism or proteinuria were seen in repeated microscopic urine examinations. IgA, ANA, anti DNA serology was negative. Urine culture was clean. Stone formation (6x7 mm) in the upper pole of the right kidney was established by abdominal ultrasonography. Calcium, uric acid, oxalate, citrate, and cystine excretion in 24-



Fig. 1. The appearance of no radius as a component of TAR syndrome.

hour urine samples were as follows: Calcium: 2.95 mg/kg/day (N: <4 mg/kg/day), uric acid: 19.7 mg/kg/day (N: <15 mg/kg/day), oxalate: 0.4 mmol/m²/day (N: <0.24 mmol/0.84 m²/day), citrate 208 mg/g creatinine (N: >180 mg/g creatinine), and cystine 22 mg/m²/day (N: <60 mg/1.73 m²/day).

Since high levels of oxalate and uric acid were being excreted, an oxalate-restricted diet and hydration were recommended. Afterwards, the patient discontinued his follow-ups for a year. In that period, he had one attack of macroscopic hematuria and a renal calculus was said to pass spontaneously. When he presented for the second time, he reported macroscopic hematuria. On abdominal ultrasonography, there was no sign of the above-mentioned stone in the kidneys. Cystoscopy was done for etiology. Many tortuous and engorged vessels were evaluated in the bladder mucosa (Fig. 2). There was no active bleeding point.

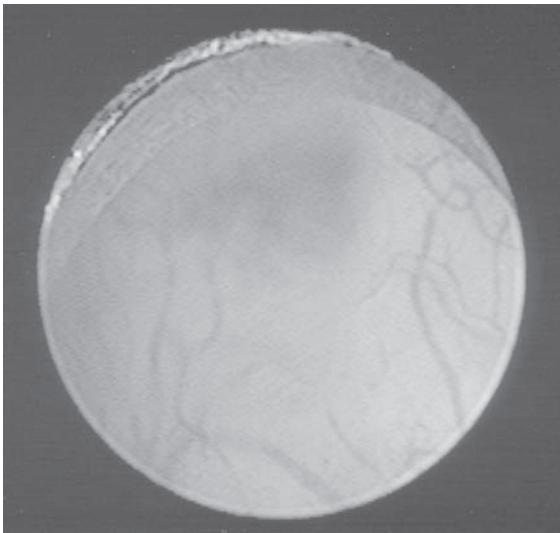


Fig. 2. Many tortuous and engorged vessels in the cystoscopy.

No stone was detected in ultrasonography; however, due to the history of a kidney stone, an abdominal computed tomography without contrast was performed and this imaging disclosed a stone formation in the right ureter. Postoperative chemical analysis of the stone revealed that it consisted of oxalate monohydrate. Our case was under oxalate-restricted diet; however, we concluded that reduced oxalate excretion could have ensued due to too little water intake.

Discussion

TAR syndrome is a congenital malformation characterized by bilateral absence of the radius and thrombocytopenia. The lower limbs, and gastrointestinal, cardiovascular, and other systems may also be involved⁴⁻⁶. The genetic basis of TAR syndrome is uncertain. Evidence for both autosomal recessive and autosomal dominant inheritance has been reported. Cases with consanguinity are rare in TAR syndrome^{1,2}. Some families are thought to have autosomal dominant condition with variable penetrance due to the proband having typical TAR syndrome, but the other family members having subtle signs such as limited pronation and supination or radial shortening⁷.

In Greenhalgh's² study, all cases had documented thrombocytopenia and bilateral radial aplasia, 47% had lower limb anomalies, 47% cow's milk intolerance, 23% renal anomalies (7 cases), and 15% cardiac anomalies^{2,8,9}. Renal abnormalities and dysfunction are not frequent in patients with TAR syndrome. In Greenhalgh's² study, one case had duplex ureter, one had mild dilatation of renal pelvis, two had horseshoe kidney, one had Wilms tumor, one had functional problems like vesicoureteral reflux, and one had pyelonephritis². Another case report presented a patient with TAR syndrome accompanied with horseshoe kidney¹⁰.

Hematuria is a common problem in pediatric nephrology. Hematuria can be gross or microscopic, transient or persistent, and associated with or without proteinuria. The prevalence of gross hematuria is accepted as 0.13%. There are many causes of hematuria appearing in widely varying clinical presentations. The cause could be established easily in 50% of patients presenting with hematuria¹¹, but for the remainder, further investigations must be done to determine etiology. Hematuria can be renal or non-renal in origin. Nevertheless, in such patients, the first steps are to ascertain history, physical examination and erythrocyte morphology in the urine sample. Furthermore, when the etiology of hematuria is being investigated, trauma, instituted medication, symptoms of upper respiratory tract infection as a sign of post-infectious glomerulonephritis, collagen vascular diseases, and family history of Alport syndrome, urolithiasis and polycystic kidney should be kept in mind¹².

Stone formation and hypercalciuria are the most frequent causes of hematuria in childhood¹². Urinary tract stone disease is common in the Mediterranean region. Turkey is accepted as an endemic area of urolithiasis in the world¹³. Those stones are usually composed of ammonium acid urate and oxalate. Dietary factors in the stone pathogenesis particularly show dependence on cereal or rice diets¹⁴. However, in the present case, the level of oxalate excretion was found to be high and a stone was visualized in abdominal ultrasonography, but hypercalciuria was not determined. In the chemical analysis of the stone, it was found to consist of oxalate monohydrate. Our case was under oxalate-restricted diet; however, we consider that reduced oxalate excretion could have ensued due to dehydration.

Telangiectasia in the bladder is a rare problem. There is a case report describing telangiectasia as a cause of gross hematuria in a patient with hereditary hemorrhagic telangiectasia¹⁵. Another rare condition related with telangiectasia in the bladder is cyclophosphamide usage^{16,17}. In our patient, cystoscopic examination revealed many tortuous and engorged vessels in the bladder mucosa. To our knowledge, there is no case report demonstrating nephrolithiasis and telangiectasia in patients with TAR.

In conclusion, TAR is a rare condition, and urolithiasis and bladder telangiectasia can be found as a cause of gross hematuria in TAR patients.

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