

Klippel-Trenaunay-Weber syndrome with hydronephrosis and vesicoureteral reflux: an unusual association

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SUMMARY: Yıldızdaş D, Antmen B, Bayram İ, Yapıcıoğlu H. Klippel-Trenaunay-Weber syndrome with hydronephrosis and vesicoureteral reflux. an unusual association. Turk J Pediatr 2002; 44: 180-182.

The Klippel-Trenaunay-Weber syndrome is a rare disorder characterized by congenital vascular hamartomas, limb hypertrophy, cutaneous manifestations, lymphangiomas and atresia of lymph vessels with non-pitting edema. A three-year-old boy was referred to our clinic for progressive hypertrophy of leg and feet with 32-month history. We diagnosed Klippel-Trenaunay-Weber syndrome, and determined vesicoureteral reflux in our patient. To our knowledge, hydronephrosis and vesicoureteral reflux have not been described previously in the KTWS.

Key words: Klippel-Trenaunay-Weber syndrome, hydronephrosis, vesicoureteral reflux.

The Klippel-Trenaunay-Weber syndrome (KTWS) is a rare disorder characterized by congenital vascular hamartomas, limb hypertrophy, cutaneous manifestations, lymphangiomas and atresia of lymph vessels with non-pitting edema. The basic defect is assumed to involve vascular embryogenesis and can result in malformations of almost any body area¹⁻³. Mental retardation and/or convulsion, short stature, anomalies in genitalia, aberrant big vessels and hemimegalencephaly are associated in some rare cases with KTWS^{4,5}. We report a child with KTWS and vesicoureteral reflux and hydronephrosis.

Case Report

A three-year-old boy was referred to our clinic for progressive hypertrophy of leg and feet with 32-month history. His height was at the 90th percentile weight at the 75th and head circumference at the 50th. The parents were not related, and his father was 47 years old and his mother 39 years old. Physical examination was remarkable for hyperpigmented vascular nevus in his left hypochondriac region, varicose veins and lymphedema in the legs and feet, polydactyly of feet, and bilateral diffuse hypertrophy of gluteal area (Fig. 1). There were no neurological disturbances. Laboratory data values at presentation were as follows: WBC: 9,600/mm³, Hct: 36%, Hb: 12 g/dl and platelet count: 277,000/mm³. Pyuria was detected in urine and



Fig. 1. General appearance of the patient.

cultured *P. aeruginosa*. X-rays showed only hypertrophy of soft tissue of the legs and feet. Voiding cystography revealed grade V reflux in the left collecting system (Fig. 2). Computerized

tomography (CT) of the abdomen showed a large, hyperdense, lobulated soft tissue mass in the mesentery of the small bowel (Fig. 3). Bowel loops were displaced anteriorly towards the abdominal wall. We did not detect any hemangiomatous involvement in the kidney. Dynamic DTPA scintigraphy of the right kidney revealed normal pattern of perfusion, uptake and excretion (Fig. 4); however, there was dilatation of the left collecting system and ureter with significant clearing following diuretic infection. DMSA scintigraphy revealed a normal right kidney but focal lesions in the upper and lower poles of the left kidney attributable to renal scarring (Fig. 5).

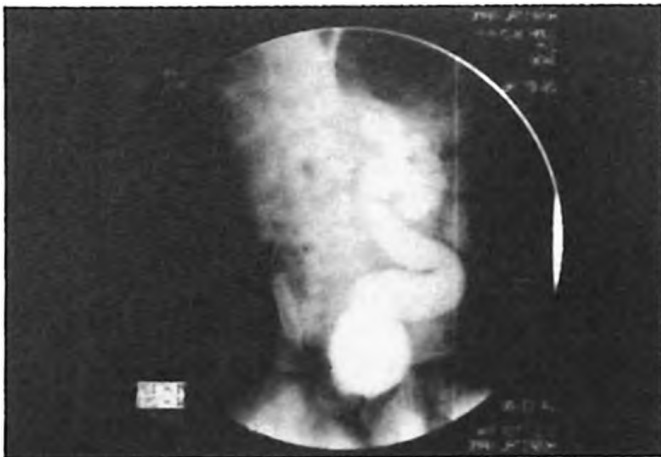


Fig. 2. Voiding cystography revealed grade V reflux in the left collecting systems.

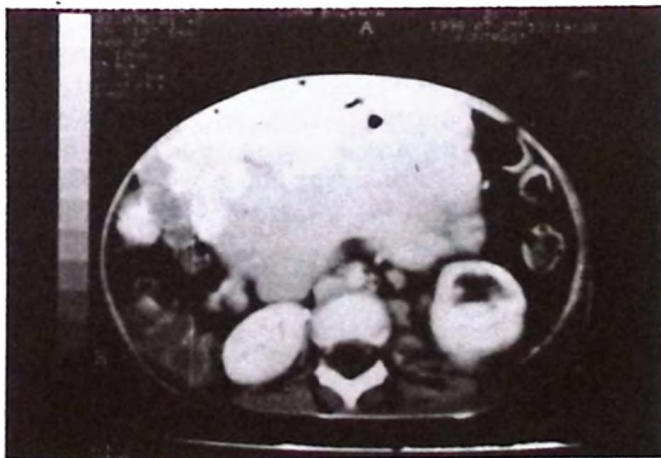


Fig. 3. CT of the abdomen showed a large hyperdense, lobulated soft tissue mass in the mesentery of the small bowel. Bowel loops are displaced anteriorly towards abdominal wall.

Discussion

The Klippel-Trenaunay-Weber syndrome is a rare sporadic congenital malformation of the vascular system that was first described in 1900⁶. The

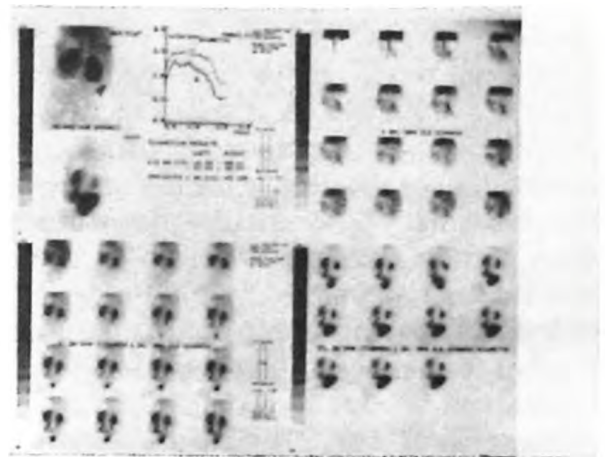


Fig. 4. Dynamic DTPA scintigraphy of right kidney revealed normal pattern of perfusion, uptake and excretion, but there was dilatation of the left collecting system and ureter with significantly clearing following diuretic infection.



Fig. 5. DMSA scintigraphy revealed a normal right kidney and focal lesions in the upper and lower poles of the left kidney attributable to renal scarring.

current review was not undertaken as a population study. This syndrome is characterized by: 1) a more or less extensive nevus on the involved limb, 2) varicose veins on the involved limb arising in childhood, and 3) hypertrophy of usually one, but occasionally of more than one limb. There are well documented reports in which other congenital anomalies have been found to be associated with this syndrome⁷. Hamartomas and vascular malformations described in the KTWS are separate from the flat nevus and are usually localized to the affected limb. Cutaneous nodular hemangiomas consisting of capillary and cavernous elements together with lymph vessels may then be connected to a malformed venous system with reduplications, agenesis, arteriovenous shunts or

fistula^{1,2,8}: Digital anomalies are well known and have been described in case reports or small series⁹⁻¹¹. KTWS is also associated with lymphatic malformations resulting from the obstruction of the deep veins and with common lymphedema. The vascular malformations have been seen in different systems. Varicose veins and vascular anomalies have been described in different sites including vascular proliferative patches in the mouth¹²; varicosities in the small bowel, colon¹³⁻¹⁴, and esophagus¹⁵; and ocular hemangiomas with glaucoma¹⁶.

Although complications may develop, KTWS is generally a nonprogressive disorder. The problems, which include edema, stasis dermatitis, skin ulceration, cellulites, anemia¹⁴, thrombosis, phlebitis, bone and joint abnormalities, scoliosis, and paresthesia, tend to be chronic in nature¹⁷. Reported life-threatening complications include disseminated intravascular coagulation¹⁸, gastrointestinal bleeding as a result of hemangiomas in the intestine¹⁸⁻²⁰, and systemic infection with Gram-negative bacteremia²¹. In our case, typical findings of KTWS are described. During the screen for gastrointestinal involvement by sonography, we detected hydronephrosis. The hydronephrosis was also confirmed with CT. We performed voiding cystography and detected grade V vesicoureteral reflux in our patient. DTPA and DMSA scintigraphies demonstrated dilatation of the left collecting system and scars on the left kidney. To our knowledge, hydronephrosis and vesicoureteral reflux have not been described previously in the KTWS.

Our patient was followed closely. He had infections in the urinary tract and cultured. *P. aeruginosa* was cultured in urine. He was treated with ceftriaxone along with nutritional support. We consulted with pediatric surgeons on this case, and planned a surgical approach.

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