

Case Report

Klippel Trenaunay Weber syndrome with unilateral polycystic kidney disease: a rare presentation

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ABSTRACT

Klippel Trenaunay Weber syndrome (KTWS) is a rare disease characterized by hemihypertrophy, variceal enlargement of the veins, and arteriovenous (AV) malformations. Renal involvement in KTWS is not known except in rare case reports. Herein, we present a case of KTWS with unilateral polycystic kidney. A 52-year-old male was admitted due to pain left lumbar region for the last three months. The physical findings were increased diameter and increased length of the left leg compared with the right one, diffuse variceal enlargements on left leg, portwine stain on left side on neck, thorax, abdomen, left upper limb and left lower limb and a few hemangiomas on the left leg. Radiographic findings were cystic lesions in the left kidney, varicose veins in left leg, and hypertrophy of the soft tissues of the proximal left leg. Color Doppler of left lower limb showed incompetence of the saphenofemoral junction. He was diagnosed to have KTWS with these findings. Renal function tests of the patient were in the normal range. Patient's only complaint was left lumbar region pain, mild in intensity. Patient was managed symptomatically.

Keywords: Hemihypertrophy, Polycystic kidney disease, Portwine stain

INTRODUCTION

Klippel Trenaunay Weber syndrome (KTWS) is a rare idiopathic disease characterized by hemihypertrophy of the bones and soft tissues, variceal enlargement of the veins in the involved extremity, and arteriovenous (AV) malformations. The disease was first described by Klippel and Trenaunay as Klippel Trenaunay syndrome (KTS) which included hemihypertrophy and varices in 1900 after which Weber called the disease Klippel Trenaunay Weber syndrome with the addition of AV malformations in 1907.^{1,2} The pathogenetic mechanism of the increased angiogenesis is thought to be mutations in the angiogenic factor (VG5Q) gene via transcription and increased activity.³ VG5Q gene has been identified in blood vessels, is secreted during angiogenesis, and increases endothelial cell proliferation. The involvement is unilateral typically, of the lower extremity in 95%, upper extremity in 5%, and both lower and upper

extremities in 15% of the cases.⁴ Capillary lesions are associated with soft tissue swelling and bone hypertrophy. Patients with this syndrome have a wide spectrum of presentation from asymptomatic disease to life-threatening bleeding and embolism.

The symptoms appear before the age of ten in about 75% of cases in this congenital disease.⁵ Although the treatment strategy is conservative unless complicated, patients need close orthopedic follow-up since the length of lower extremities differs frequently.⁶ The differential diagnosis of KTWS includes KTS, Maffucci syndrome, Proteus syndrome, and other capillary malformations not associated with any syndrome.⁷

Renal involvement in KTWS is not known except in rare case reports. Herein, we present a case of KTWS diagnosed at the age of 52 together with polycystic kidney

CASE REPORT

A 52-year-old male was admitted to the outpatient clinic of the department of internal medicine with the complaint of pain in left lumbar region for the last three months. The patient had variceal enlargements of the veins and a port wine stain on left side, extending upto midline, from the time of birth and his left leg was thicker than the right one, but he did not have a certain diagnosis. The family history was negative. The pathological findings on physical examination at the time of presentation were large hemangiomas lesions in form of patches on the left leg, left arm, left side of neck, thorax and abdomen shown in Figure 1 and 2. These skin lesions were limited strictly to left side with typical demarcation upto midline and sparing of the right side. Figure 3 showing increased diameter and increased length of the left leg compared with the right one, with diffuse variceal enlargements, increased diameter and length of the left arm as compared to right arm, left hand and digits larger than the right hand, similarly left foot and digits larger than the right foot. The laboratory findings were in the normal limits including the renal function tests. The abdominal ultrasonography revealed multiple cystic lesions in the left kidney which were further proved by CT scan too.



Figure 1: Portwine stain on left side of the body (chest, left arm).



Figure 2: Hemihypertrophy of left side of the body (left upper limb, left lower limb) varicose vein in left lower limb and portwine.



Figure 3: Hypertrophied left lower limb with varicose veins.

DISCUSSION

Klippel Trenaunay Weber syndrome is a rare vascular abnormality characterized by hemihypertrophy, variceal veins, and AV malformations. It is usually sporadic as in our case, although few familial cases have been reported.⁸ It is a congenital disease diagnosed usually in childhood, but it should be kept in mind that there may be cases undiagnosed until adulthood as in our case.

The disease involves usually the lower extremity like our case although the trunk or face may be affected unilaterally. Hemangiomas may be limited to the skin or may be seen in bones, muscles, and solid organs. The patient we present had hemangiomas in the liver, spleen, and rectum besides the lower extremity. Variceal veins appear in the first years of life and increase in dimensions until adolescence.⁷ They may cause pain, lymphedema, thrombophlebitis, and skin ulcerations. Hemihypertrophy presents either as increased length of the bones or as increased diameter due to soft tissue involvement. Hemihypertrophy is present at birth and progresses until adolescence at which time it ceases to progress. The presented case had hypertrophy of both bone and soft tissue. There may be eye abnormalities in KTWS including vascular pathologies of optic nerve, iris, choroid, retina, and orbit. The pathological findings detected at the examination of the presented case were thought to be related with KTWS.

Renal involvement in KTWS is so far presented as case reports only. A case with KTWS and renal failure was reported of which the renal biopsy showed abnormal accumulation in the mesangial tissue.⁹ One case with KTWS and proteinuria was reported and one case with KTWS and unilateral polycystic kidney of the same side was reported.^{10,11}

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