CASE REPORT

# Truncal Varicosities and Bilateral Port-wine Stain with Spinal Deformities Associated with Klippel-Trenaunay-Weber Syndrome

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## Abstract

Klippel–Trenaunay–Weber syndrome (KTWS) is a rare congenital disorder characterized by asymmetric limb hypertrophy, usually of the lower limbs, as well as vascular anomalies and capillary malformations under the skin, termed as a port-wine stain. KTWS is prevalent in all parts of the world. It has a high degree of diversity of the associated malformations. In the present case, vascular/lymphatic malformations were evident by the presence of bilateral port-wine stain and lymphangioma. More interestingly, prominent aberrant veins (truncal varicosities) were found in the anterior chest wall, together with the presence of multiple angiolipomatosis. Bone deformities were more than limb hypertrophy and macrodactyl and extended to spinal deformities in the form of scoliotic changes.

**Key Words:** *Klippel–Trenaunay–Weber syndrome, lymphangioma, port-wine stain*

**What was known?**
Klippel–Trenaunay–Weber syndrome is a rare congenital disorder characterized by asymmetric limb hypertrophy, vascular anomalies, and capillary malformations under the skin.

## Introduction

Klippel–Trenaunay–Weber syndrome (KTWS) is a rare congenital disorder characterized by asymmetric limb hypertrophy, usually of the lower limbs, as well as vascular anomalies and capillary malformations under the skin, termed as a port-wine stain.[1] KTWS could be associated with other anomalies such as lymphatic obstruction, distal limb lipodermatosclerosis, affectation of the abdominopelvic vasculature leading to varying degrees of vascular malformations involving the gastrointestinal system, spleen, genitourinary, and central nervous system.[2]

## Case Report

We report a case of a 7-year old boy from Upper Egypt. The condition started at the age of 2 years old with a vesicular eruption on the right side of the abdomen, back, and thigh, with insidious onset and stationary course. He had a deformity on the left foot only since birth. There was no degree of consanguinity between parents. He was delivered vaginally, full term with no maternal history of medical problems during pregnancy. He had no history of other body system affection, with no family history of similar conditions.

On examination, there were grouped vesicles and papules on the right side of the abdomen, back, and upper thigh with superimposed secondary bacterial infections. These lesions were clinically correlated with lymphangioma circumscriptium. This was associated with port-wine stains on the right side of the abdomen (10 cm × 8 cm) and on the left side of the trunk (4 cm × 5 cm). Furthermore, there were vascular malformations on the left side of the trunk in the form of multiple elongated, tortious, and dilated veins (truncal varicosities) [Figures 1 and 2]. In addition, there were multiple subcutaneous swellings on the back (5 in number), with one at the right upper thigh and one at the lower chest. They were soft, freely mobile with normal underlying skin. There were enlarged axillary lymph nodes (discrete, firm, nontender,

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and mobile) on the right side (1 cm × 1 cm) and on the left side (0.5 cm × 0.5 cm). In addition, bone deformities were noted in the left feet (macrodactyl) with the back slightly curved to the left [Figure 3].

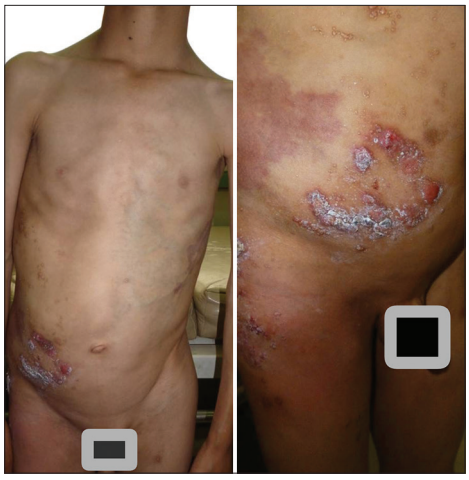

**Figure 1:** A 7-year-old boy with lymphangioma circumscriptum and port-wine stain on the right side of the abdomen

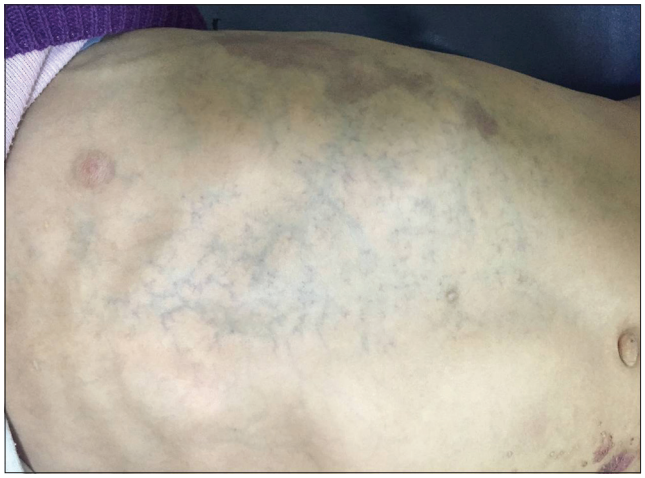

**Figure 2:** Truncal varicosities and port-wine stains on the left side of the trunk

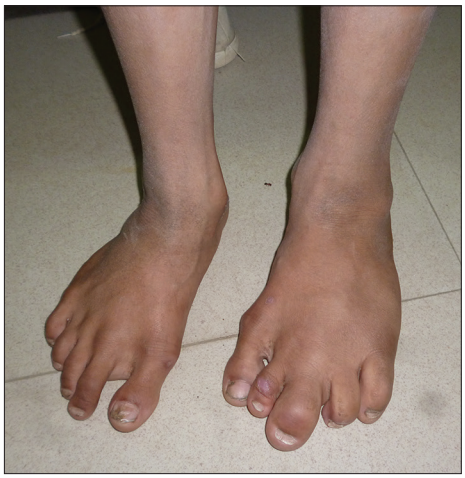

**Figure 3:** Feet deformities

Differential diagnosis included KTWS and Parkes–Weber syndrome, and the patient was investigated for proper evaluation as followed:

Pediatric evaluation: Weight was 24 kg (above the 50$^{th}$ percentile). Stature was 125 cm (above the 50$^{th}$ percentile). Ophthalmological evaluation was completely normal.

Laboratory investigations are shown in Table 1. Imaging investigations are shown in Table 2.

## Discussion

In our case, vascular/lymphatic malformations were evident by the presence of bilateral port-wine stain and lymphangioma. More interestingly, prominent aberrant veins (truncal varicosities) were found in the anterior chest wall, together with the presence of multiple angiolipomatosis. Bone deformities were more than limb hypertrophy and macrodactyl and extended to spinal deformities in the form of scoliotic changes.

It is a rare congenital mesodermal phakomatosis, affecting 1 in 100,000 with no gender or racial preference.[3] KTWS was first described in 1900 by two French physicians Maurice Klippel and Paul Trénaunay who described two patients who had a triad of port-wine stain, varicosities of an extremity, and hypertrophy of the affected limb bones and soft tissues.[4]

The cause of KTWS is unknown; however, a few theories have been postulated. The most popular one among them is that of Baskerville *et al.*,[5] who stated that a mesodermal defect during embryogenesis causes maintenance of microscopic arteriovenous communications resulting in KTWS.

The diagnosis of KTWS can be made when any two of the triad features is present. It is usually unilateral and

**Table 1: Laboratory investigations**

| Investigations | Result | Normal reference range |
|---|---|---|
| Erythrocyte sedimentation rate after the 1$^{st}$ h (mm) | 75 | 7-12 |
| Red blood cells (M/μL) | 4.15 | 4.0-5.2 |
| Hemoglobin (g/dL) | 11.1 | 11.5-15.5 |
| White blood cells (K/μL) | 8.06 | 4.5-14.5 |
| Platelets (K/μL) | 427 | 150-400 |
| Random blood sugar (mg/dl) | 79 | 70-110 |
| Creatinine (mg/dl) | 0.3 | 0.6-1.3 |
| ALT (U/L) | 2 | 0-41 |
| AST (U/L) | 15 | 0-41 |
| Total bilirubin (mg/dl) | 0.3 | 0-1 |
| Albumin (g/dl) | 3.9 | 3.5-5.2 |
| Total plasma proteins (g/dl) | 7.5 | 6.6-8.3 |
| Albumin/globulin ratio | 1.1 | 1.1-2.5 |

ALT: Alanine aminotransferase, AST: Aspartate aminotransferase

**Table 2: Imaging investigations**

| Imaging | Findings |
|---|---|
| Left feet X-ray | Showing deformity of the 3rd toe and shortening of the 1st big toe. Orthopedic recommendation was followed up for the overgrowth of the 3rd toe for 1 year and then for arthrodesis |
| Weight-bearing total spine X-ray | Showing slight scoliotic changes in dorsal spine with a curve toward the left side |
| Echocardiography | It was normal |
| Soft-tissue swelling ultrasonography | Subcutaneous fat deposition was noted on the right side of the back and right anterior abdominal wall regions. Associated dysplastic vessels were noted within these areas. A picture suggestive of multiple angiolipomas. Furthermore, there was a cystic lesion with turbid fluid contents at the lower anterior extent of the chest wall. It was about 3 cm × 3 cm. It might be related to small hematoma [Figures 4 and 5] |
| Color Doppler ultrasonography examination of both innominate and subclavian veins on both sides | It was revealing normal patency and compressibility<br>Color Doppler ultrasonography examination of both lower limbs: Revealed intact venous system [Figure 6] |
| Abdominal ultrasonography | The liver was mildly enlarged. There were no any other abnormal findings [Figure 7] |

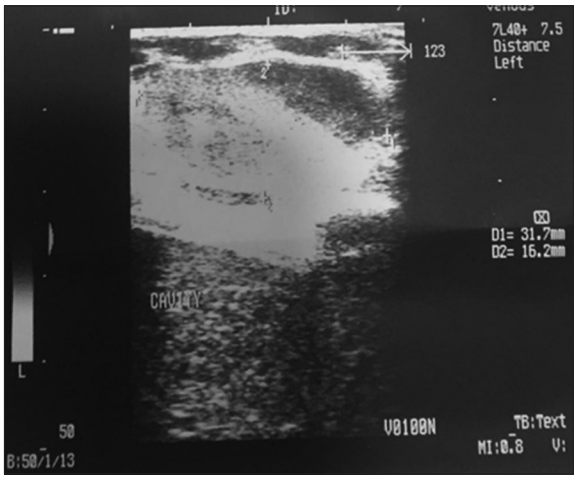


**Figure 4:** Soft-tissue swelling ultrasonography showing cyst at anterior chest wall

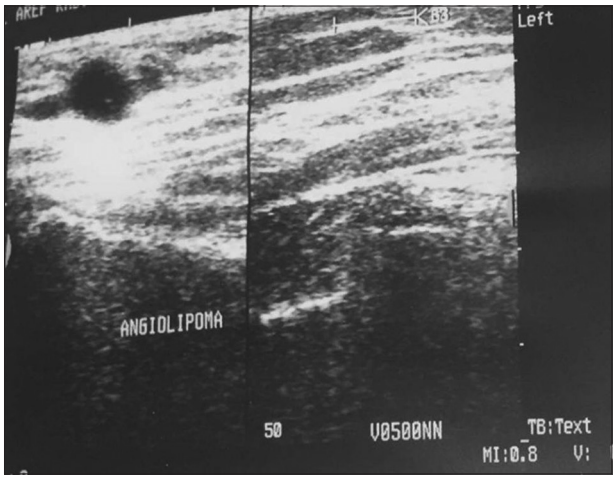


**Figure 5:** Soft-tissue swelling ultrasonography showing angiolipoma

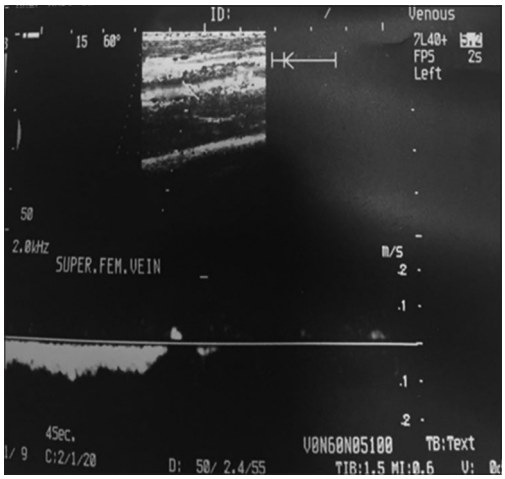


**Figure 6:** Color Doppler ultrasonography showing normal lower limb venous system

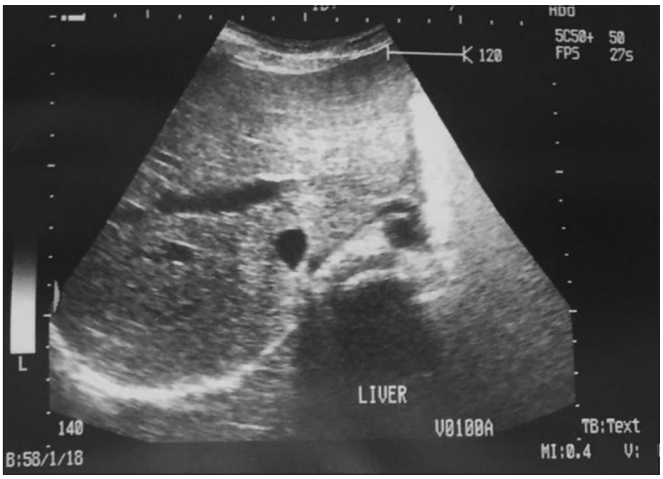


**Figure 7:** Abdominal ultrasonography showing mild hepatomegaly

almost exclusively involves lower extremities, buttocks, abdomen, and lower trunk. It is rarely bilateral and involves upper extremities. The varicosities appear mostly by the age of 12 years.[6]

Oduber *et al.*[7] expanded the definition to cover more anatomic variations, so that the vascular malformations and disturbed growth (hypertrophy or hypotrophy) coexist on the same or opposite sides involving part

of a limb, a whole limb, a hemibody, or a limb girdle. Hypertrophy of soft tissues may be prominent in small body parts such as toes (macrodactyly). Limb dystrophic disorders are nonessential, but still support the diagnosis of KTWS, including polydactyly, syndactyly, and clinodactyly.

Capillary malformations (port-wine stains) and venous malformations are both considered as diagnostic features of KTWS, whereas small congenital lymphatic malformations simply support the diagnosis of KTWS. Abnormal development (dysplasia) of the venous system involves mostly the deep veins of the lower limbs with vascular defects, phlebectasia, and hypoplasia. The common superficial venous system anomalies in KTWS are the persistence of the embryonic lateral marginal vein and varicose veins. The associated central nervous system abnormalities include microcephaly, macrocephaly, hemimegalencephaly, cerebral and spinal arteriovenous malformations or multiple aneurysms, and orbitofrontal varices.[6]

Complications that may also support the diagnosis of KTWS include thrombosis, thrombophlebitis, emboli, cellulitis, edema, hemorrhage from the involved epithelia, and autonomic dysfunction as evidenced by skin atrophy or hyperhidrosis.[8]

KTWS is a rare condition, but appears to be seen in all parts of the world. It has a high degree of diversity of the associated malformations.

## Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient's parents have given their consent for the patient's images and other clinical information to be reported in the journal. The patient's parents understand that the patient's name and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

## Financial support and sponsorship

Nil.

## Conflicts of interest

There are no conflicts of interest.

**What is new?**

Vascular/lymphatic malformations were presented as bilateral port-wine stain and lymphangioma. Truncal varicosities were found in the anterior chest wall, with multiple angiolipomatosis. Bone deformities were more than limb hypertrophy and macrodactyl and extended to spinal deformities in the form of scoliotic changes

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