Klippel-Trenaunay Syndrome: A Clinical Case Report

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Abstract

KTS is a rare congenital disorder. We present a 14 year old boy with the characteristics corresponding to the same i.e. venous varicosities, capillary malformation and limb hypertrophy. The patient also had other bony deformities like pectus carinatum, thoracolumbar scoliosis and campylodactyly. Such lumbar and digit deformities have been reported earlier also in patients with KTS. Patient was managed by a multidisciplinary approach, although a good follow up was not assured by the parents.

Keywords : *KTS, varicosities, limb hypertrophy, scoliosis, campylodactyly, Pectus carinatum.*

Introduction

Klippel-Trenaunay Syndrome (KTS), a syndrome of capillary-lymphatic-venous malformation associated with soft tissue and skeletal hypertrophy. It is a rare congenital disorder with a very low incidence of about 1:100,000. It has no predilection for gender, race, or geographical area and occurs sporadically [(1)]. It manifests as a triad of cutaneous capillary malformations, venous varicosities, and hypertrophy of the osseous and/or soft tissue of the extremities [(2-6)]. Vascular malformations are always present and usually (but not always) affect only one extremity, particularly the lower extremities [(7)].

Case Report

A 14 years old male presented with complaint of intermittent, low-grade fever from past 1 month which was not associated with chills or rigors. History of left lower limb painful swelling which progresses while doing daily activities and regresses while lying down or after elevation of the legs and also abnormal shape of chest which is progressing with age. The developmental growth was normal at each age range. There was no history of similar kind of illness in the family. On examination, the vitals of the patient were stable. The blood pressure of upper limbs and lower limbs were compared which did not show any variation. In anthropometric examination; weight, height and body mass index of the patient were 29.1 kg (-3.53 z), 144 cm (-2.38 z) and 14.0 kg/m^2 (-3.34 z) respectively. The arm span was 163 cm (figure 1) and arm span to height ratio was 1.13 and the upper segment to lower segment ratio was 0.8. The deformities seen were Pectus Carinatum (figure 2), Thoracolumbar scoliosis and Camptodactyly in little finger of both hands (left > right) (figure 3). Also there was increase in size/ diameter/mass of the left lower limb. His right leg was 6 cm longer than the left leg. There were painful venous varicosities on left lower limbs with gross tortousity of veins (figure 4). Visible pulsations were seen over the entire pericardium. However, apex beat was heard in 5ht intercostal space in mid clavicular line. On auscultation, heart sound were normal with no murmur. Neurological examination was intact. There was no respiratory or abdominal abnormalities. On Radiological investigations, thoracolumbar scoliosis with cardiomegaly on anterio-posterior and lateral chest x-rays (figure 5 and 6). Ultrasound of left leg showed extensive varicosities with many of them thrombosed with ectasia of venous channels more so in mid and lower leg with ectasia of superficial femoral vein in distal thigh. A definitive diagnosis with genetic testing and extensive family history and examination was not possible due to financial constrains and a poor follow up by the patient.

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Discussion

KTWS is a combination of cutaneous angiomatosis, varicose veins, and enlargement of soft tissue first described in 1900. The KTWS occurs mostly sporadically, affecting males and females in equal numbers in about 1/100,000 people [8].

Limb hypertrophy is often present at birth or during infancy and continues to grow until the child stops growing, although it may continue to progress over time. The hypertrophy is a result of soft tissue and/or bony overgrowth or lymphatic and venous malformations [9, 10]. Although any part of the body can be involved, the most commonly affected sites are the lower extremities [11].

This patient has camptodactyly. Various other limb anomalies including macrodactyly, syndactyly, clinodactyly, ectrodactyly, and congenital hip dislocation have been reported in association with KTS [10].

The exact cause and mechanism of scoliosis in the patient in this study is unknown. It may be secondary to limb length discrepancy (about 6 cm), striking pelvic obliquity and long-term claudication.

A color Doppler ultrasound should be performed for prenatal diagnosis of limb hypertrophy and to assess the underlying cause of any cystic lesion. Most patients present with the complete clinical triad, with port wine stains and vascular malformations first appearing at birth and varicose veins usually appearing during infancy and progressing in adolescence [9]. Although such couldn't be assumed in our case.

KTS can cause significant morbidity from the vascular anomalies including deep venous thrombosis, bleeding, pulmonary embolism, stasis dermatitis, cellulitis, and limb enlargement that may require amputation. Patients also suffer from scoliosis and gait abnormalities related to limb hypertrophy. Therefore, KTS must be suspected, recognized, and appropriately managed in all infants with capillary malformations involving one or more extremity at birth [11].

In an adolescent, vitamin D deficiency and iron-deficiency anemia are quite uncommon. In a study by Jiliang Zhai et al,[12] serum ferritin level were found extremely low in a patient of Klippel-Trenaunay Syndrome and a severe form of iron deficiency anemia was noticed in the patient. Elevated D-Dimer were probably due to extensive venous malformations. Previous studies found that patients with large venous malformations has chronic low-grade consumptive coagulopathy [13]. Mazoyer [14] proposed that the coagulopathy among patients with venous malformations was a result of localized intravascular coagulation.

There is currently no cure or definitive treatment for KTS. However, management should be multidisciplinary and aim to reduce the symptoms and complications of the disease. For instance, compression stockings can be used for varicose veins, and heel inserts can be used and are usually sufficient for leg length discrepancies of 1.5 cm or less, although surgical closure of the growth plate at the knee is sometimes needed to equalize the leg length [15].

Conclusion

Here we present a case of KTS with a unique constellation of signs: unilateral limb involvement and campylodactyly. This case not only serves as a review of the atypical features of the syndrome, but also highlights that patients must be followed up on a regular basis by a multidisciplinary team and more research is required for proper diagnosis and treatment guidelines.

Figure 1: Arm span of the patient was 163 cms.

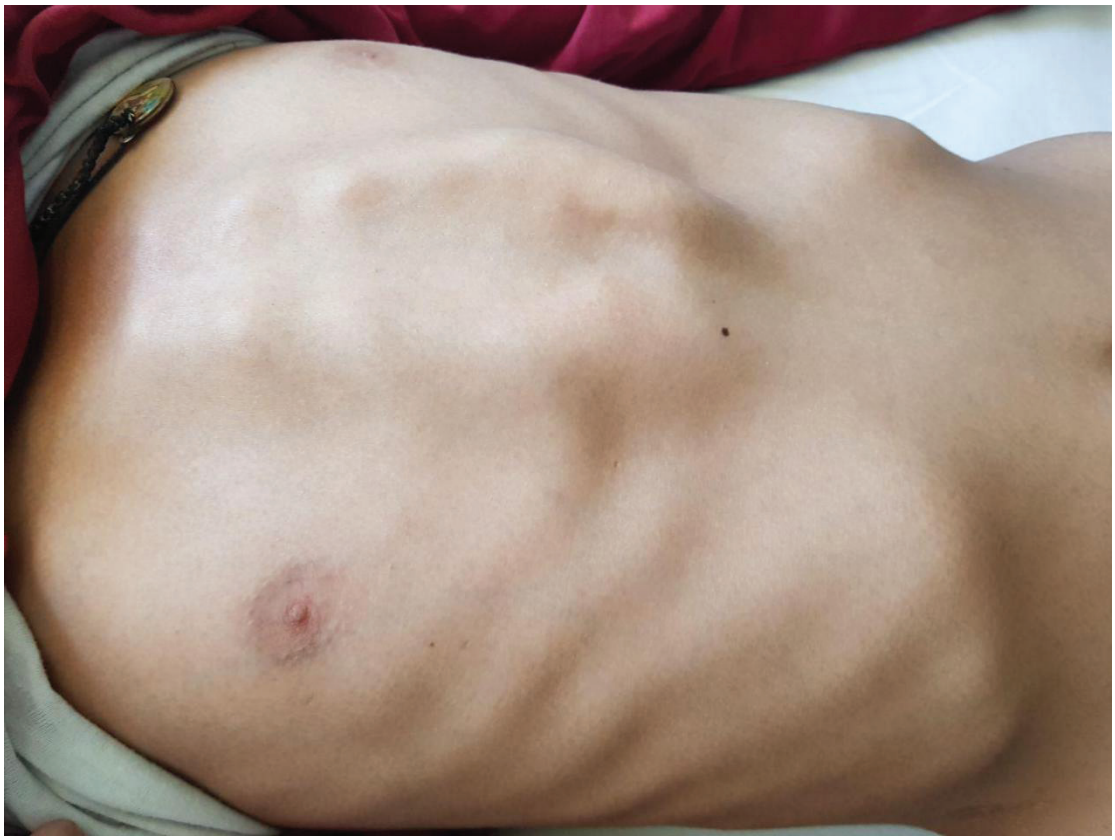

Figure 2: Pectus Carinatum can be seen in this with protrusion of the sternum.

Figure 3: Camptodactyly can be seen in little fingers bilaterally (Left> Right)

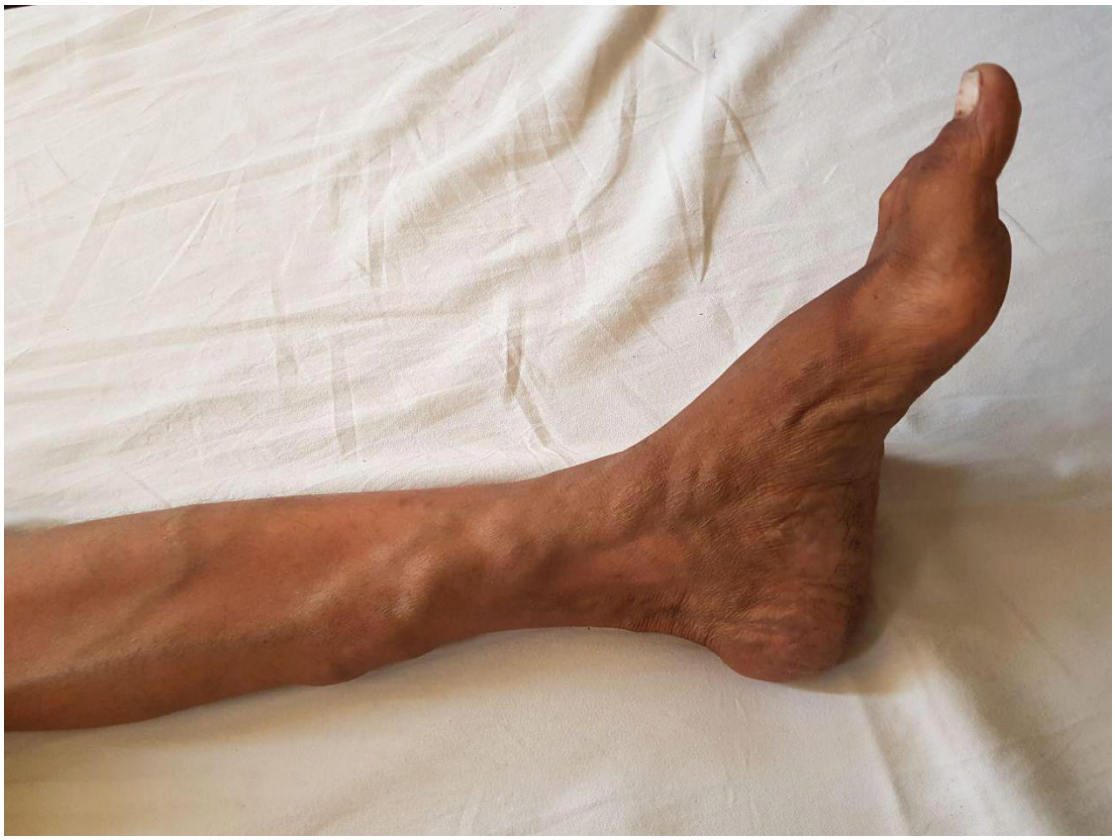

Figure 4: Visible Venous varicosities in lower limbs with grossly tortous veins

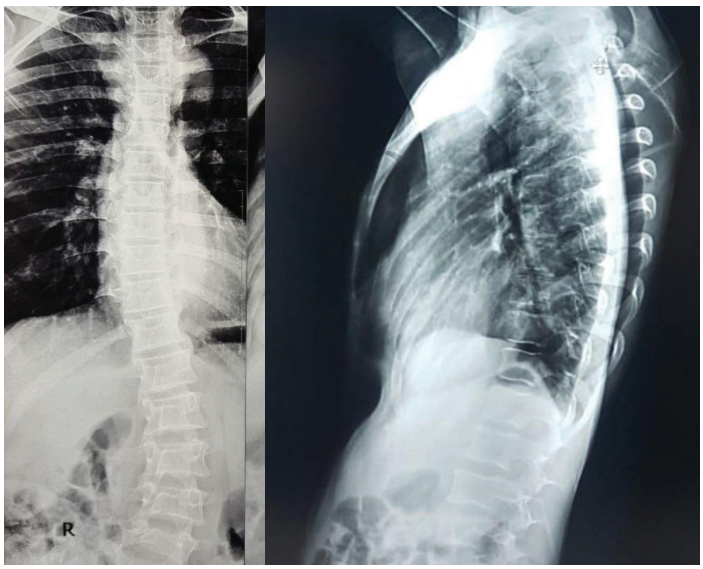

Figure 5&6: Xray chest (Anterioposterior and Lateral view) showing Thoracolumbar scoliosis with cardiomegaly

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