International Journal of Contemporary Pediatrics
Kumar KS et al. Int J Contemp Pediatr. 2017 Jan;4(1):294-296
http://www.ijpediatrics.com
pISSN 2349-3283 | eISSN 2349-3291
DOI: http://dx.doi.org/10.18203/2349-3291.ijcp20164624

Case Report

Klippel Trenaunay Syndrome: a rare case report in a neonate

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Received: 08 November 2016
Accepted: 03 December 2016

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ABSTRACT

Klippel Trenaunay Syndrome (KTS) is a cutaneous vascular malformation in combination with bone and soft tissue overgrowth, with or without lymphatic malformation. Symptoms appear by birth and approximately 90% of the cases are diagnosed by the age of 12 years. Prompt diagnosis at the earliest and conservative management with regular follow up is crucial in prognosis of KTS.

Keywords: CLOVES syndrome, India, KTS, Newborn

INTRODUCTION

Klippel trenaunay syndrome (KTS) is rare but yet has a distinct clinical identity. KTS is characterized by a classical triad of capillary, venous malformations and limb overgrowth with or without lymphatic malformation.[1,2] Incidence and prevalence are not known, there is no gender or ethnic predilection.[3] The symptoms and their intensity may vary from discrete hemangiomas to large venous ecstasias, causing deformity of the limbs.[4] Patients with KTS have increased risk of complications with deep vein thrombosis and pulmonary thromboembolism.[5] We report a rare case of KTS in a newborn diagnosed at birth.

CASE REPORT

A term girl neonate, appropriate for gestational age was delivered to a 23 year old Primi mother, non-consanguineously married couple by normal vaginal delivery and said to have cried immediately after birth with good apgar scores and birth weight of 2750 grams.

On examination vitals were stable. Local examination revealed a large port wine stain over the left antero lateral abdominal wall measuring 8*12 cms with palpable multiple soft lump's over the anterior abdominal wall.

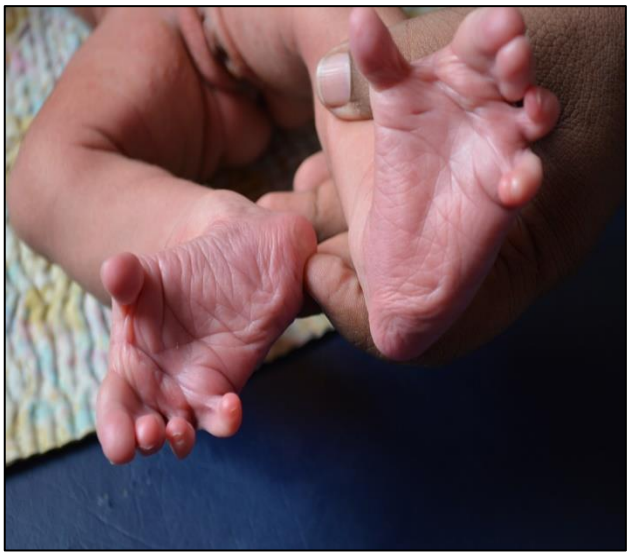

Figure 1: Bilateral foot deformity with soft tissue hypertrophy.

Presence of bilateral foot deformities with soft tissue hypertrophy of all digits with syndactyly of 2nd and 3rd toe of foots were noted with wide digit angles. Mild hypertrophy of right lower limb with length discrepancy of 1 cm was noticed on measurements.

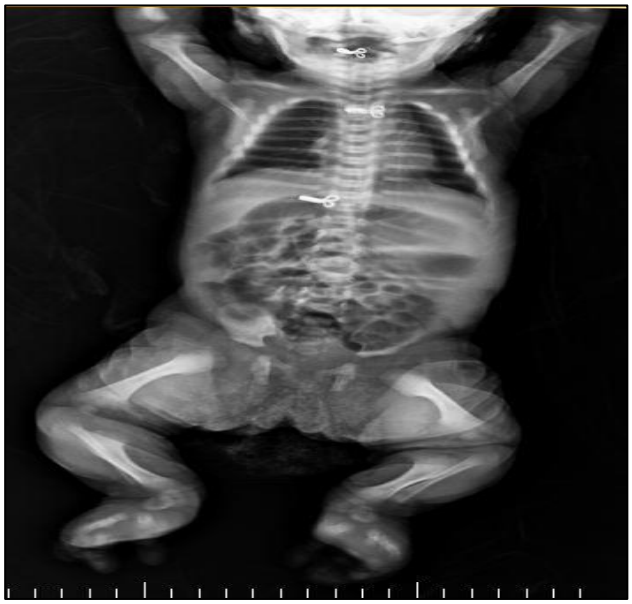

Figure 2: Infantogram consistent with clinical findings.

Neonate was investigated for full blood picture with coagulation profile which was within normal limits. Neonatal infantogram were consistent with clinical findings. Neurosonogram was within normal limits.

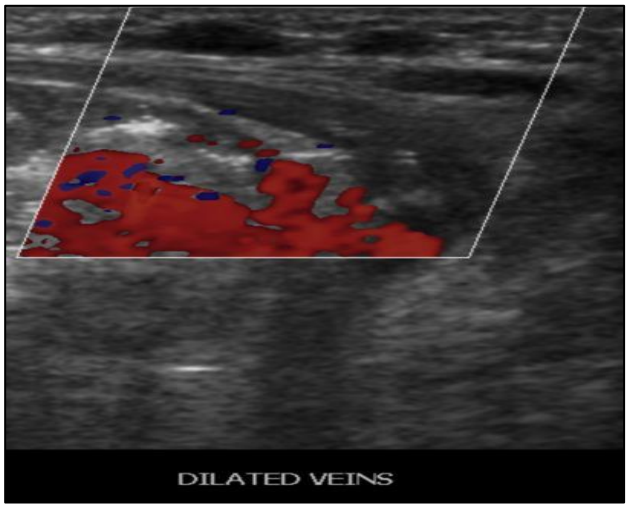

Figure 3: Dialted posterior, anterior and peroneal veins.

Color doppler of bilateral lower limbs for arteries and veins were suggestive of dilated posterior tibial, anterior tibial and peroneal veins were noted. No pseudo aneurysm or arterio venous malformation. ultra sonogram of abdomen and pelvis were within normal limits with large heterogenous lesion of the skin and subcutaneous region of the left antero lateral abdomen wall. Multiple slow flow dilated veins were noted.

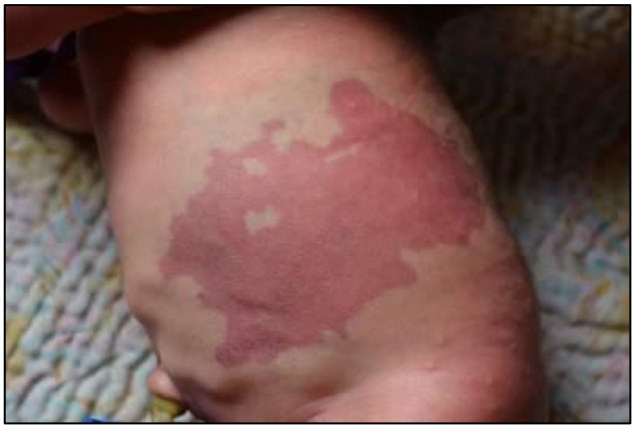

Figure 4: Port wine stain in the abdominal wall.

This neonate was diagnosed with the above findings of port wine stain of abdominal wall, varicose veins in the abdominal vessels, lower limbs, and bony and soft tissue hypertrophy involving the extremities at birth. Neonate is on regular follow up with no complications at present.

DISCUSSION

The etiology of KTS is unknown, although various theories have been proposed.6 Most of the cases are sporadic, but few with familial history have been reported.[7] Several patients with KTS carry post zygotic somatic mutations in the phosphotidyl inositol-4, 5-Bisphosphate S Kinase, catalytic subunit (PIK3CA) gene.[8]

KTS is a combination of capillary malformation, venous varicosities or malformation and underlying soft tissue and/or bony deformities of the involved extremities with or without lymphatic malformations representing key clinical features.[2] In a retrospective study of 40 patients with KTS, 21 of 22 with geographic stains had definite or probable lymphatic disease. 16 0f 17 with blotchy stains had no evidence or only possible lymphatic malformation. KTS also have associated limb anomalies including macrodactyly, syndactyly, polydactyly, clinodactyly, camphodactyly, ectrodactyly and congenital hip dislocation.[10,11]

Diagnosis of KTS is primarily by clinical features but radiology and laboratory tests may be useful securing the diagnosis. Imaging studies, conventional venography, plain radiograph, laboratory tests and biopsy are part of investigations in diagnosis of KTS.

Differential diagnosis of our index case were CLOVE syndrome (congenital lipomatous growth, vascular malformations and epidermal nevus), parkes weber

syndrome and DCMO (diffuse capillary malformation with overgrowth).

CLOVE syndrome has characteristic epidermal nevi, which may appear later in our index case. The substantial overlap between CLOVE and KTS is explained by similar somatic PIK3CA mutation in both conditions.[13] Parkes weber syndrome is a combination of capillary malformation, arterio venous fistulae and limb overgrowth. differentiates from KTS with presence of arterio venous fistulae.[14] DCMO, approximately 1/3rd of cases have prominent varicosities, needs to be monitored for long term to differentiate them from KTS.[15]

KTS treatment involves multi-disciplinary approach. consultation with pediatric, dermatology, interventional radiology, plastic surgeon, orthopedic surgeon, vascular surgeon, hematology, urology and gastroenterology is required. Conservative management includes compression garments, lymphatic massage, physical therapy and physical activity.[16] Patients with KTS need to be on regular follow up with quarterly laboratory investigations.

CONCLUSION

KTS diagnosis in our Index neonate was diagnosed at birth and investigated further for confirmation of diagnosis. Prompt inter-disciplinary approach has been initiated with conservative management.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Kumar KS, Kumar NM. Klippel Trenaunay Syndrome: a rare case report in a neonate. Int J Contemp Pediatr 2017;4:294-6.