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Klippel-Trenaunay Syndrome: A Case Report

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ABSTRACT

Klippel-Trenaunaysyndrome(KTS) is a rare congenital condition usually presenting with port wine stains, excessive growth of bones and soft tissue and varicose veins which most commonly occurs in the legs, but it also may affect the arms, face, head, or internal organs. We report a case of term male neonate with clinical findings of Port-wine stain, multiple cystic swellings with ultrasonographic findings suggestive of vascular malformations and limb abnormalities in the form ofsoft tissue hypertrophy of right upper limb, polydactyly of right hand and syndactyly of left hand consistent withKlippel-Trenaunay syndrome.

Keywords: Klippel-trenaunay syndrome; port-wine stain, vascular malformations; soft tissue swelling;polydactyly.

INTRODUCTION

KlippelTrenaunay Syndrome is a rare condition, characterized by the triad of capillary malformations, usually port-wine stains, soft tissue and bone hypertrophy(occasionally hypotrophy) and varicose veins or venous malformations. Not all cases have the full triad of features. There is wide variation in the clinical manifestations of the condition. Mostly, it is idiopathic in origin; however, sometimes it may occur as an autosomal dominant trait. KlippelTrenaunay Syndrome is estimated to affect at least 1 in 100000 people worldwide irrespective of sex and races. [1]The characteristic capillary haemangiomaisvisible from birth in the vast majority of cases (98% in one series). [2]The vascular malformation is usually limited to a single extremity, though multiple extremities can be involved. Limb abnormalities may present initially as gait disturbances. The digits may be affected with macrodactyly, syndactyly, polydactyly or oligodactyly.

Here, we report a case of KlippelTrenaunay Syndrome diagnosed in newborn infant as we couldn't find any case report of KlippelTrenaunay Syndrome diagnosed in newborn reported in Nepal so far.

CASE REPORT

A 2 days male baby born of 22 years old Gravida 02, Para 01 hailing from Thokarpa VDC, Sindhupalchok, presented to our emergency department, Kanti Children's Hospital, with complaints of multiple cystic swellings over trunk and upper limbs since birth.The baby was born at term by spontaneous vaginal delivery at home.On examination, the neonate had 4000 grams weight on admission with normal Vital signs. There was a soft, ill-defined, cystic swelling about 4 x 5 cm2 in the left chest wall extending into axilla and left arm. There was another cystic swelling about 3x2 cm2 in right axilla(Fig 1) extending into right arm. Multiple Port-wine stains(Fig 2) were found predominantly in left axilla, left upper back and left upper limb associated with hypertrophy of right upper limb. Additional clinical features noted were syndacytyly(fused third and fourth digits) in left hand and postaxial polydactyly in right hand(Fig 3).There was no cardiac murmur; no distended vessels, Arteriovenous malformations in retina

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were found on ophthalmologic evaluation.

On investigation, thrombocyte count was normal(2,28,000/cu mm)and routine septic screen were within normal limit. Ultrasonography of the cystic swellings showed ill-defined irregular multi-loculated cystic lesion in bilateral anterior chest wall and left axilla suggestive ofVascular malformation. Ultrasonography of abdomen and pelvis were normal. Echocardiography showed 2 tiny fenestrated Secundum ASD 3 mm, 2 mm Left to Right Shunt. Cardiothoracic team concluded that the baby didn't need any vascular intervention. The neonate was clinically diagnosed as a case of KlippelTrenaunay Syndrome.

DISCUSSION

Klippel-Trenaunay Syndrome was first described by two French doctors, Klippel and Trenaunay in 1900. It is a triad of capillary malformations, usually port-wine stains, soft tissue and bone hypertrophy (occasionally hypotrophy) and varicose veins or venous malformations (Klippel & Trenaunay, 1900). [3]Port-wine stains may be present at birth. 1 These vascular malformations consist of mature dilateddermal capillaries. These lesions are macular, sharply circumscribed, pink to purple in color and tremendously variable in size. Port-wine stains can occur as a component of Klippel-Trenaunay-Weber Syndrome associated with soft tissue enlargement (i.e. hypertrophy of an extremity or a part of it).[4] In this case, upper limbs are hypertrophied (Fig 2) and port-wine stains (Fig 3) are present predominantly in left axilla, left upper back and left upper limb which is consistent with other studies. There may be varicose veins, in this case no such findings were found on physical examination. Limb lengthening may present initially as gait disturbances. The digits may be affected with macrodacytly, syndactyly, polydactyly or oligodactyly; in this case, syndactyly is present is left hand and postaxial polydactyly is present in right hand(Fig 1).Based on the clinical findings of Port-wine stain, soft tissue hypertrophy, syndactyly, polydactyly since birth and Ultrasonography of the cystic swelling suggestive of vascular malformations, adiagnosis of Klippel- Trenaunay Syndrome was made. Sometimes there may be associated arterio-venous malformations;9 in this case, no such lesion was found. There may be some rare complications, e.g. thrombophlebities, dislocation of joints, gangrene of

the affected extremities, heart failure, hematuria secondary to angiomatous involvement of urinary tract, rectal bleeding from lesions of gastrointestinal tract,pulmonary lesions and malformation of lymphatic vessels.10No such complication was found in this case.A series of 252 patients with KTS was studied at Mayo Clinic, Rochester between January 1956 and January 1995. It showed presence of capillary malformations (port-wine stains) in 246 patients (98%), varicosities or venous malformations in 182 (72%), and limb hypertrophy in 170 (67%). All three features of KTS were present in 159 patients (63%), and 93 (37%) had two of the three features. Atypical veins, including lateral veins and persistent sciatic vein, occurred in 182 patients.[2]

There has been one case report of KlippelTrenaunay Syndrome in Bangladesh, in aone day term neonate, with clinical findings ofport wine stain, varicose veins and excessive growth of soft tissue of lower limbs consistent with Klippel-Trenaunay syndrome. We found only one case reported in Nepal about KlippelTrenaunay Syndrome, in a 15 years old Female, with clinical findings of port-wine stain, capillary malformation and soft tissue hypertrophy of lower limb since birth consistent with Klippel-Trenaunay syndrome, which was initially misdiagnosed and treated as lymphatic filariasisby local health practitioner.

Although the cause of KTS is still unknown, it is hypothesized that it is caused by a mesodermal abnormality during fetal development leading to vascular and soft tissue malformations in the affected limb (Baskerville et al, 1985). McGrory&Amadio(1993) believed that an underlying mixed mesodermaland ectodermal dysplasia was responsible for development of KTWS. Klippel-Trenaunay Syndrome

might develop due to a single gene defect. Rarely it can be inherited as an autosomaldominant trait. Whelan et al reported a case of a girl with translocation: KTW syndrome associated with a

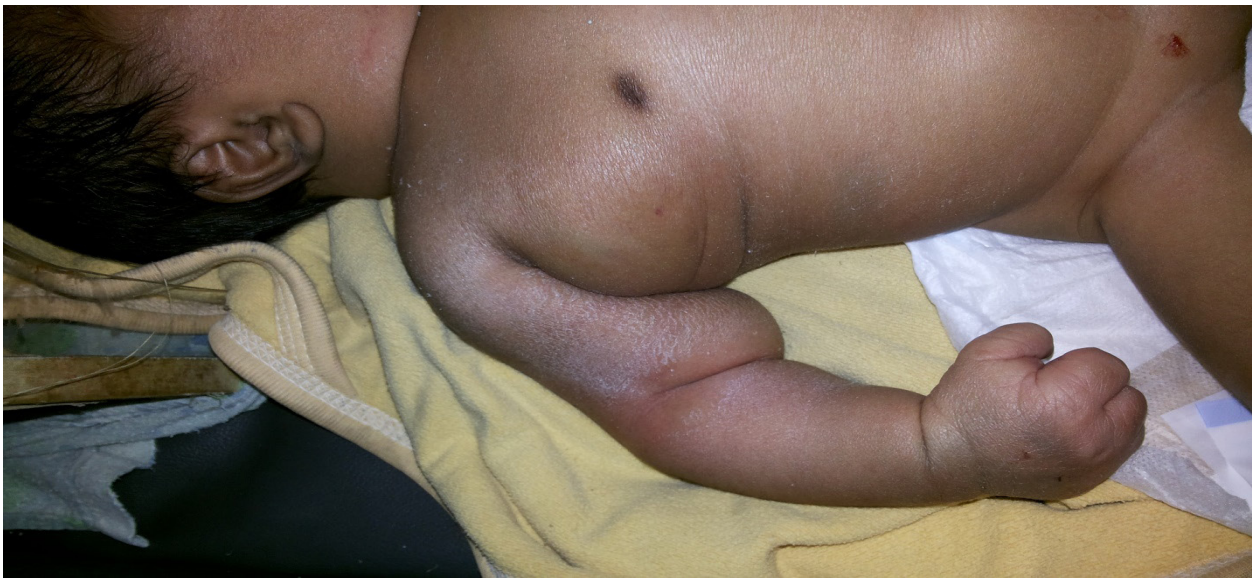

Fig 1.soft tissue hypertrophy with cystic swelling(USG suggestive of Vascular malformation).

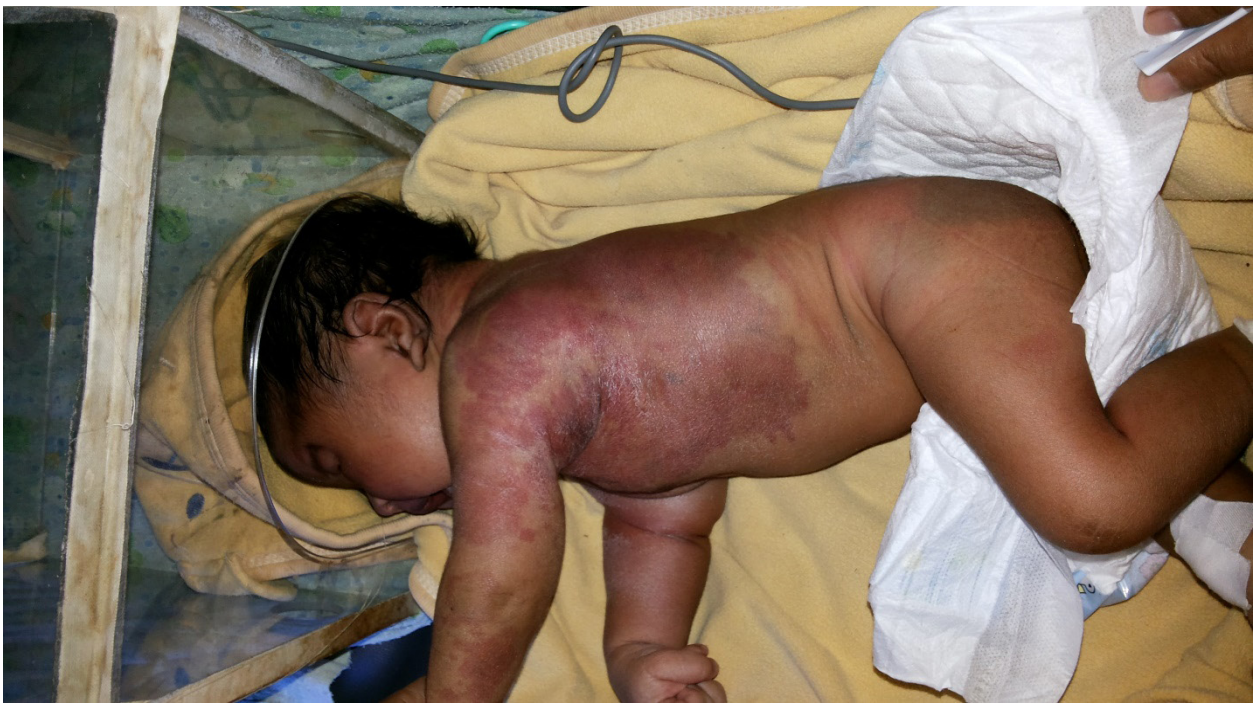

Fig 2.Portwine stain.

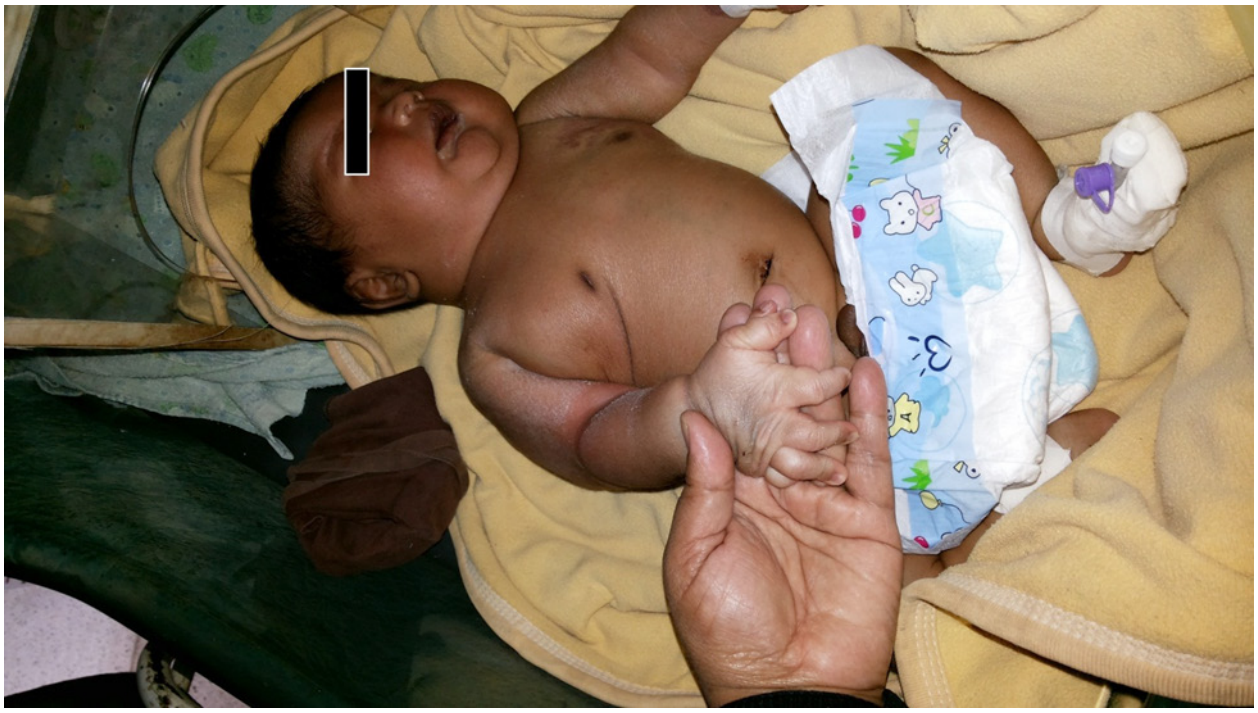

Fig 3.Polydactyly.

reciprocal t(5;11)(q13.3;p15.1). The de novo translocation t(8;14)(q22.3;q13) has also been reported byWang et al. The association between the angiogenic factor gene AGGF1 and KTS appears to be significant.

In many patients, a thorough medical history and physical examination are sufficient to make the diagnosis. However, a number of imaging studies are useful when there are complications.There is no curative therapy. Management requires a multidisciplinary and individualized approach, aiming to ameliorate the patient's symptoms and correct the consequences of limb-length discrepancy.

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