ORIGINAL RESEARCH PAPER

Volume-8 | Issue-3 | March-2019 | PRINT ISSN No 2277 - 8179

INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

A NEONATE WITH LIMB HYPERTROPHY: RARE CASE OF KLIPPEL TRENAUNAY SYNDROME

Neonatology

Dr. Jothi DS Assistant professor department of Neonatology, Madurai Medical college,Madurai Tamil Nadu

ABSTRACT

Klippel-Trenaunay syndrome (KTS) is a rare disorder characterized by the triad of vascular malformations, venous varicosities, and bone and soft-tissue hypertrophy. We present a case of Klippel-Trenaunay syndrome with limb hypertrophy without bone hypertrophy .

KEYWORDS

Klippel-Trenaunay syndrome, Port wine stain.

INTRODUCTION

Klippel Trenaunay Syndrome (KTS) is a rare congenital disorder characterized by asymmetric limb hypertrophy, usually of the lower limbs with capillary malformations. This condition presents at birth and affects both males and females equally. We present a typical KTS in new born baby and simple method of evaluation to differentiate major capillary – venous –lymphatic malformation.

Case Presentation

A one day old female neonate was admitted to the department of newborn ward, Madurai with a swelling of left leg since birth. Baby was born at 40 weeks of gestational age to a primi 24 year old woman by lower segment caesarean section. Medical history of the mother was not significant, antenatal scan done at 8 months of gestation was normal and the mothers family had no history of similar disorders. The baby weighed 3500g and had APGAR score of 8 and 9 at 1 and 5 minutes, respectively.

A physical examination of the baby had good cry, no respiratory distress, heart rate 150/min, capillary refill time <3 secs, peripheral pulses were felt normally and there was a diffuse swelling of the left lower limb (hypertrophy of the limb) extending from mid thigh to the dorsum of the foot, skin over the swelling was shiny with bluish discoloration(portwine stain).not warmth. Swelling was firm in consistency, and non tender. (Fig 1) Left leg circumference(15.6cm) was larger than the right leg (9cm) and there was no significant difference in the length of both lower limbs(49.5cm). (Fig 2) No other skin lesions were noted and other examination findings were normal. Hematological parameters done on day 2 showed hemoglobin of 14.6gms, total count of 10,000 and a platelet count of 76,000/cmm. Biochemical, coagulation test and the chest radiograph were all normal. Echocardiography, abdominal ultrasonography, neurosonogram were normal. Doppler ultrasound done on (D7) showed dilated superficial venous systems , slow flow across the superficial venous systems and normal flow across deep venous system seen without Arterio venous fistula. MRI left leg showed extensive soft tissue mass lesion involving muscular compartment of the left leg as well as the intervening fat planes extending to thigh muscular compartment showing high signal intensity on T2 weighted images with no bone hypertrophy was noted.(FIG 2,3) The baby was diagnosed as a case of Klippel trenaunay syndrome based on clinical examination and radiological examination . Patient was managed conservatively with IV antibiotics, IV fluids, direct breast feeds started on day 2. C reactive protein done on 4th day of was negative ,antibiotics stopped on 5th day and the platelet count became normal (210000/cmm) on 7th day. Baby was discharged on10th day.

Discussion

Etiology and epidemiology

The Klippel-Trenaunay syndrome is a rare mesodermal abnormality characterized by a triad of vascular nevus, varicose veins, and soft tissue and bony hypertrophy of limb. It can be diagnosed on the basis of any two of these three features. It was first described by French physicians Maurice Klippel and Paul Trenaunay they associated vascular malformations with hypertrophy in the affected limb. Subsequently, Parkes Weber described arteriovenous fistulas in these patients. It is rare with uncertain origin with an incidence of approximately 1:100,000 live births. It has no predilection for gender or race, and most of the cases are sporadic and appear at birth. The etiology of the disease can be embryonic mesodermal changes resulting in increased angiogenesis lead to increased vascular flow causing tissue hypertrophy and vascular changes or sporadic polygenic mutations. The association between the angiogenic factor gene AGGF1 and KTS appears to be significant.

Presentation

Skin malformations in KTS are mostly capillary hemangiomas. Skin capillary malformations will be visible from birth in the vast majority of cases (98%) are diffuse or mostly located on the hypertrophic extremity side. The skin lesions are characteristics port wine stain appearance usually red purple or bluish in color. Lower extremities are affected in about 95% of cases. Changes can be limited to the skin only or can affect subcutaneous tissue, muscles, and bones. 56% of patients have visceral vascular malformations including hemangiomas and/or lymphangiomas.

The limb involvement in case of KTS is mainly due to venous and/or lymphatic malformation. This causes the soft tissue as well as the bone to become hypertrophied and the limb overall is longer than the uninvolved side. The skin and muscles contain multiple areas of venous malformation. Venous malformation in the medullary canal is also described. The bones are usually osteopenic. And these patients have high risk of fall and subsequent fracture of long bones. Limb hypertrophy is often greater distally. The digits may be affected with macrodactly; syndactly .An increase in limb girth may be the main feature if soft tissues are predominantly affected. Leg length discrepancy can occur due to meduallary bone involvement with increase in vertical length. In our case the baby had portwain stain, venous varicosity and unilateral limb hypertrophy without any signs of arteriovenous fistula.

Complications

Clinical presentation of KTS syndrome has wide spectrum from asymptomatic state, chronic venous stasis to potentially life-threatening complications. Such as hypercoagulability, deep vein thrombosis and pulmonary thromboembolism (PTE).The chronic venous stasis can result in venous dermatitis. Venous ulcers, Thrombophlebitis and venous thromboembolism. In addition to chronic venous stasis the lower limb involvement can cause limb disparity and subsequent, gait disturbance, chronic pain in the limb and scoliosis.

Investigation and management

Large lower limb can be caused by many disorders like lymphatic malformation, combined vascular malformation, capillary malformation, KTS, hemi hypertrophy, posttraumatic swelling, Parkes Weber syndrome, venous malformation, rheumatologic disorder, infantile hemangioma, kaposiform hemangioendothelioma, or lipofibromatosis. The major differential diagnosis in our patient was Klippel–Trenaunay syndrome or Parkes Weber syndrome. MRI showed features of soft tissue hypertrophy with subcutaneous edema, adipose hypertrophy, and normal sub facial compartment with no arterio venous fistula. Doppler showed dilated superficial venous system with low flow suggestive of no arteriovenous fistula. These non invasive investigation is more suggestive of KTS.Our baby had moderate thrombocytopenia with normal coagulation profile and had

clinically stable course with no bleeding manifestation and follow up platelet count was normal which ruled out associated Kassabech Meritt syndrome .Klippel Trenaunay syndrome with kassabech Meritt syndrome were reported in some case studies which was not present in our case.

Management

- Management requires a multidisciplinary and individualized approach, aiming to ameliorate the patient's symptoms. The most common treatment for capillary malformation pulsed dye laser therapy, and surgical intervention is indicated when the patients become excessively symptomatic. Varicosities and pain due to venous malformation can be managed with compression therapy, (garments/bandages) it is efficacious for patients with limb hypertrophy, venous stasis, and phlebitis, Ulceration and bleeding can be managed with excision if the deep venous system is intact. If the deep venous system is blocked it can be treated with reconstruction of bypass. Ultrasound-guided foam sclerotherapy can be considered for chronic venous insufficiency. The leg length discrepancy has to be closely monitored in this syndrome .If the leg length discrepancy is between 1.5 to 2cm it requires orthotic insole, more the 2 cm needs epihysiodesis which is usually scheduled after 10 years of age.

Up to now, there has been no certainly established prognostic factor of KTS. Life expectancy is largely normal, depending on the severity of the malformation and thus the likelihood of complications. Venous thromboembolism has been reported to occur in 8-22% of Klippel-Trenaunay syndrome patient .Possibly about 10% of patients develop a pulmonary embolism.

Conclusion

Capillary –venous –lymphatic malformation has to be considered in new born bay with limb hypertrophy .Non invasive investigation like MRI and Doppler will be sufficient to differentiate the major capillary –venous –lypmphatic malformation in the neonate. Visceral involvement can ruled out with ultrasonography, neurosonography. Invasive investigation like lymphscintigraphy and angiography can be considered later.

REFERENCES

1. Mneimneh S, Tabaja A, Rajab M. Klippel-Trenaunay Syndromewith Extensive Lymphangiomas.Case Rep Pediatr. 2015;2015:581394.
2. Sung HM, Chung HY, and Lee SJ et al Syndrome. Arch Plast Surg. 2015 Sep;42(5):552-8.
3. Sharma D, Lamba S, Pandita A et al Klippel-trénaunaysyndrome- a very rare and interestingsyndrome.Clin Med Insights Circ Respir Pulm Med. 2015 Mar 5;9:1-4 Schook CC
4. Mulliken JB, Fishman SJ, Alomari AI, Grant FD, Greene AK. Differential diagnosis of lower extremity enlargement in pediatric patients referred with a diagnosis of lymphedema. Plast Reconstr Surg. 2011 Apr;127(4):1571-81
5. Ikpeme AA, Usang UE, Inyang AW, Ani N. Klippel Trenaunay Syndrome: A Case Report in an Adolescent Nigerian Boy. Open Access Maced J Med Sci. 2015 Jun 15;3(2):322-5
6. Yara N, Masamoto H, Iraha Y, Wakayama A, Chinen Y . Diffuse Venous Malformation of the Uterus in a Pregnant Woman withKlippel-TrénaunaySyndromeDiagnosed by DCE-MRI. Case Rep Obstet Gynecol. 2016;2016:4328450 Gupta Y
7. Jha RK, Karn NK, Sah SK, Mishra BN, Bhattarai MK Management of Femoral Shaft Fracture in Klippel-Trenaunay Syndrome with External Fixator. Case Rep Orthop. 2016;2016:8505038 Chu ST
8. Han YH, Koh JA et al A Case of Klippel-Trenaunay Syndrome with Acute Submassive Pulmonary Thromboembolism Treated with Thrombolytic TherapyJ Cardiovasc Ultrasound. 2015 Dec;23(4):266-70
9. Upadhyay H, Sherani K, Vakil A et al . A case of recurrent massive pulmonary embolism inKlippel-Trenaunay-Weber syndrometreated with thrombolytics. Respir Med Case Rep. 2016 Jan 25;17:68-70
10. Bhat L, Bisht S, Khanijo K. Klippel-Trenaunay-Weber Syndromewith Kasabach-Merritt Coagulopathy and Hydronephrosis. Indian Pediatr. 2015 Nov;52(11):987-8.