Case Report

Klippel Trenauny Syndrome

Jawad Jalil[1], Mobeen Shafique[2], Tariq Ghafoor[3], Umer Amin[4]
Paediatric Department[1,3], Radiology Department[2,4], Combined Military Hospital, Bahawalpur.

Abstract

Klippel Trenauny Syndrome is a rare congenital syndrome characterized by port wine stain (capillary malformation), limb or hemihypertrophy and other vascular malformations. We present a case of this rare disorder in a young boy at an early stage of this disease.

Introduction

Klippel-Trenauny Syndrome (KTS) is a rare congenital malformation which is known by various names including Klippel-Trenauny Weber syndrome, angio-osteohypertrophy, nevous varicosus osteohypertrophicus syndrome, hemangiectasia hypertrophicans and nevous verucosus hypertrophicans.[1]

The clinical manifestations may include cutaneous capillary malformations i.e. port wine stain (PWS), soft tissue and bony hypertrophy, venous malformations and lymphatic abnormalities. The lower limb is involved in 90% of cases. Complications may include bleeding, cellulites, venous thrombosis or pulmonary embolism. Other associated abnormalities include hand and feet anomalies, lymphoedema or involvement of abdominal and pelvic organs.[2] The gene responsible for this syndrome has recently been identified; however sporadic cases may also be seen.[3]

Case Report

A four year old boy presented to paediatric out patient department at Combined Military Hospital, Bahawalpur with the complaints of limping gait and disproportion in the size of the lower limbs along with pain off and on in the right lower limb. The parents revealed that the child started having difficulty in weight bearing on right foot and was limping at approximately 18 months of age which gradually aggravated. There was also a history of tripping over and occasional fall, when the child attempted to run or walk quickly. The right lower limb of the child appeared large and developed a large dark spot over the skin involving the sole of the right foot, ankle and right lower calf.

On physical examination the child was active and cooperative with normal anthropometric and developmental status. There were multiple hyperpigmented macules and patches of different sizes all over the body. The largest port wine stain extended over the right lower calf, ankle and sole of the foot (Figure 1). The right lower limb was appreciably larger and bulkier than the left lower limb (Figure 2). The portion of the right leg under the port wine stain was soft, non tender with diffuse margins. Soft bruit was audible over this area. There was over riding of the second on the third toe of the left foot (Figure 2). The rest of the body including face, trunk and upper limbs was proportionate in size. There were no signs of infection, cellulites or trauma.

Radiologically the limbs were normal in size with no disproportion in the size of the long bones; however there was increased soft tissue mass extending from lower half of right calf to mid foot. Doppler ultra-sonography of the right lower limb showed arterio-venous malformation consistent with cavernous haemangioma under the area of port wine stain. Abdominal sonographic study revealed no abnormality.

As the child was not having any significant clinical problem, he was advised symptomatic analgesics. Parents were assured and advised to have regular follow up.

Discussion

KTS was originally reported in 1900 by Klippel and Trenauny as an entity consisting of a capillary-venous malformation, an early onset of varicosities and underlying soft tissue and bony hypertrophy.[4] In 1907, Frederick Parkes Weber noted the association of arterio-venous fistulas with these findings.[5] Initially this syndrome was named "KTW syndrome". Later "Weber" was dropped to avoid confusion with the Parkes Weber syndrome which was characterized by arterio-venous malformations leading to extremity similar to KTS.[6]

The exact etiology of KTS remained obscure for a long time but recent advances in molecular genetics has made it possible to localize genes responsible for many AV malformations. The gene for KTS has been identified as AGGF1.[7]

The syndrome is currently defined as a combination of (a) capillary malformations (port wine stains) which may not extend all over the affected limb, and may be found at other places on the body (b) soft tissues or bony hypertrophy, or both (c) varicose veins. However, diagnosis can be made only with the presence of any of the two features. In our case the main port wine stain was on the right leg, however there were multiple hyperpigmented patches all over the body but mostly on the trunk. He also had a large cavernous haemangioma on the sole of the right foot extending on the posterior aspect of the same leg. It was confirmed by Doppler ultrasound study. His right leg was obviously larger and bulkier than the left leg but there was no radiological disparity in the sizes of the long bones of two legs. There

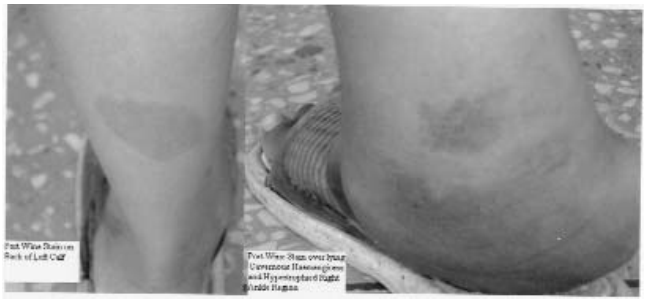

Figure 1. A-Port Wine Stain on the back of left calf. B-Port Wine Stain overlying cavernous haemangioma and hypertrophied right ankle region.

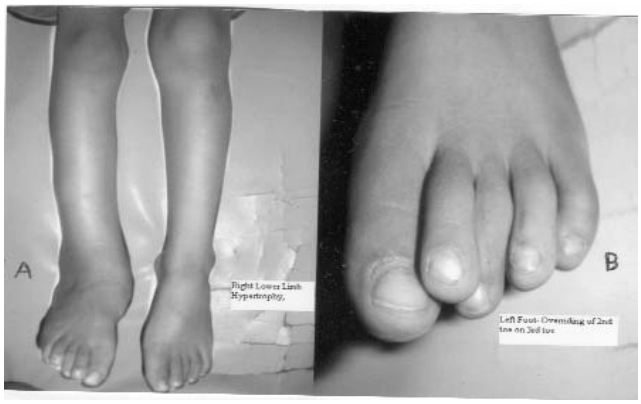

Figure 2. A-Right lower limb hypertrophy. B-Left foot, Over-riding of 2nd toe over 3rd toe.

was no clinical evidence of partial or complete hemihypertrophy. About 10% cases of KTS have urinary tract anomalies[8], however radiological evaluation of urinary tract in our case showed no abnormality. KTS may be associated with other developmental anomalies such as polydactyly, syndactyly, and macrocephaly.[9] The reported patient only had over riding of the left second toe over the third toe. Bleeding, cellulites and pain are common problems in KTS[1]; however the reported case had no such problem except pain which was aggravated on walking. Apparently this patient was diagnosed at a younger age and the disease process was at an early stage of evolution so most of the complications of the disease did not manifest in the child.

The usual agents used for the treatment of haemangiomas, like prednisone and interferon alpha are of no benefit in KTS.[10] Laser therapy may reduce or eliminate the PWS. KTP (532 nm) laser is an effective and safe tool in the management of capillary malformations of PWS type.[11] Surgical procedure may be necessary to de-bulk the excessive tissue, to excise excessive veins or haemangiomatous tissue and to correct uneven growth.[12]

References

1. Zubairi NA, Arfan ul Bari, Mahmood T. Klippel-Trenauny syndrome. JCPSP 2004; 14:423-4.
2. Al-Salman MM. Klippel-Trenauny syndrome: clinical features, complications and management. Surg Today 1997; 27: 735.
3. Wang QK. Update on the molecular genetics of vascular anomalies. Lymphat Res Biol 2005; 3:226-33.
4. Klippel M, Trenaunay P. Du naevus variqueux osteahypertrophique. Arch Gen Med 1900; 185: 641-72.
5. Weber FP. Angioma formation in connection with hypertrophy of limbs and hemihypertrophy. Br J Dermatol 1907; 19: 231-5.
6. Lindenauer SM. The Klippel Trenauny syndrome: varicosity, hypertrophy and hemangioma with no arteriovenous fistula. Ann Surg 1965; 162: 303-14.
7. Wang QK. Update on the molecular genetics of vascular anomalies. Lymphat Res Biol 2005; 3:226-33.
8. Rubenwolf P, Roosen A, Gerharz EW, Kirchhoff-Moradpour A, Darge K, Riedmiller H. Life-threatening gross hematuria due to genitourinary manifestation of Klippel-Trenaunay syndrome. Int Urol Nephrol 2006; 38:137-40.
9. Sunar H, Halici U, Duran E. Klippel-Trenauny syndrome associated with polydactyly. Clin Anat 2006; 19:78-81.
10. Liu J, Gou SJ, Han ZK. Diagnosis and treatment of Klippel Trenauny syndrome. Zhongguo Xiu Fu Chong Jian Wai Ke Za Zhi 2002; 16: 379-81.
11. Latkowski IT, Wysocki MS, Siewiera IP. Own clinical experience in treatment of port-wine stain with KTP 532 nm laser Wiad Lek 2005;58:391-6
12. Lam SM, Williams EF 3rd. Practical considerations in the treatment of capillary vascular malformations, or port wine stains. Facial Plast Surg 2004; 20: 71-6.