RESEARCH PAPER | Medical Science | Volume : 5 | Issue : 2 | Feb 2015 | ISSN - 2249-555X

Klippel Trenaunay Syndrome with Angiokeratoma - A case report with brief review of the literature.

KEYWORDS: Angiokeratoma, Klippel Trenaunay Syndrome, Nevus Flammeus.

Dr. Meenal Patil. | Dr. Manoj N. Kulkarni

ABSTRACT *Klippel Trenaunay Syndrome is a sporadic disorder presenting at birth, childhood or in early adult life , consisting of a triad of cutaneous , vascular & soft tissue involvement. It may have a mild to severe form of disease with life- threatening bleeding manifestations. It usually affects one extremity and may have associated vascular lesions such as angiokeratomas. We report a case of multiple angiokeratomas in Klippel Trenaunay syndrome in an 8 -year old girl.*

Introduction :
Klippel Trenaunay (K-T) Syndrome is a rare syndrome characterized by a triad of port-wine stain (Nevus Flammeus), soft issue hypertrophy with or without bony hypertrophy and venous varicosities (1).

Angiokeratomas represent true ectasias of blood vessels of superficial dermis. They can have different clinical presentations of which the Angiokeratoma circumscriptum type may be associated with Klippel- Trenaunay (K-T) Syndrome.

Case report :
An 8-year old girl, presented with multiple black coloured nodular, hyper- keratotic lesions 0.5 cm- 1.0 cm, over the thigh and shin of right leg of 5 years duration . Some of the lesions were coalescing and few showed crusting. The lesions were located on the background of a large diffuse pinkish macular patch on the thigh. (Fig 1). There was history of bleeding from few lesions on minor trauma.

Prominent dilated superficial veins were noted on the lateral aspects of the knee and leg (Fig 2) .

The right leg showed increased girth as compared to the left leg and hypertrophy of the middle toe of right leg was prominent (Fig 3).

Hematological and biochemical assays were within normal limits.

Colour Doppler of leg veins showed hypoplastic saphenofemoral vein and popliteal vein along with superficial varicosities draining into an abnormal lateral vein. (Fig 4).

MRI angiography confirmed the findings of the colour Doppler .

Ultrasonography of abdomen and pelvis did not reveal any vascular malformations in internal organs.

We received a skin biopsy from one of the nodules measuring 0.3cm X0.3 cm in size.

Microscopic Examination- H&E stained sections showed hyperkeratosis and acanthosis of epidermis . The dermis showed ectatic vascular channels, some of which showed organizing thrombus. Some of them were enclosed in a collarette of epidermis.(Fig 5). There were numerous dilated vessels of varying caliber filled with blood (Fig 6). Endothelial lining of the vascular channels was continuous on the upper side with the basal layer of epidermis.

Subcutaneous tissue was uninvolved.

A diagnosis of angiokeratoma in Klippel Trenaunay syndrome was made based on the

histopathological and clinical findings.

Discussion:
Klippel Trenaunay Syndrome was first described in 1900 by Maurice Klippel and Paul Trenaunay, who called it Naevus Vasculosus Osteohypertrophicus. It is a rare disorder presenting at birth or in childhood, exact incidence of which is not known. It is characterized by nevus flammeus and varicosities in superficial veins and soft tissue hypertrophy usually affecting one extremity.

It is sporadic in nature and believed to be because of some mutation in genes that regulate fetal vasculature in embryonic life..A mutation in angiogenic factor VG5Q has been proposed by Tian et al (2).Varicosities may occur in deeper leg veins posing a risk of pulmonary embolism. Varicosities in organs like colon , urinary bladder and brain may lead to rectal or urinary bleeding and seizures (3). Children with KTS may present with. seizures due to venous malformations in the brain (4).

Apart from complications resulting from chronic venous stasis and visceral bleeding, patients may have polydactyly or syndactyly and orthopaedic complications like gait disturbances and scoliosis may occur. Psychological problems due to cosmetic appearance may also occur.

Investigations like MRI and Angiography are helpful in assessing vascular lesions.

Angiokeratoma or Lymphangioma circumscriptum are some of the vascular lesions which may be associated with K-T syndrome. Fabry gave the first description of Angiokeratoma in 1915 (5).

Angiokeratomas may arise as corporis diffusum type in bathing trunk area in association with Fabry's disease.

Angiokeratomas of Mibelli are seen in fingers and toes in childhood.

Volume : 5 | Issue : 2 | Feb 2015 | ISSN - 2249-555X

Angiokeratomas of Fordyce are seen on scrotum or vulva.

Solitary or multiple angiokeratomas , as seen in K-T syndrome arise on the lower extremities and thrombosed lesions may clinically simulate malignant melanoma.

Angiokeratomas arise as one or several hyperkeratotic blue to black, firm nodular lesions ranging from 2-10 mm in size. Histologically, numerous dilated thin-walled, congested vessels are seen in the papillary dermis, underlying an epidermis showing variable acanthosis with elongation of rete ridges and hyperkeratosis. (6).

Prabhavathy et al described a case of angiokeratoma in K-T syndrome (7).

Differential diagnosis :
Park Weber's syndrome –It affects arms where there is a high flow arteriovenous malformation rather than capillary hemangioma as described above in K-T syndrome.

Cloves syndrome - It shows port wine stains and high flow shunts with congenital lipomatous overgrowth, along with epidermal naevi and skeletal abnormalities or seizures.

Proteus syndrome – It is a rare hamartomatous disorder causing asymmetrical hypertrophy of a range of tissues with macrocephaly (the so called " Elephant man ").

Sturge – Weber syndrome- It is characterised by angiomatosis in the distribution of Trigeminal nerve and ipsilateral leptomeningeal vascular anomalies.

Management :
Treatment of K-T syndrome is mainly with conservative measures like graduated compression garments and pain management.

In females , oestrogen containing contraception is contra indicated. Pregnant women with K-T syndrome need careful monitoring due to range of hematological, obstetric and anaesthetic complications. Agale et al have described a mild form of K-T syndrome in a 25 year old married female (8).

Angiokeratomas, if small, can be treated by diathermy or cautery. Larger lesions may require laser ablation or surgical excision.

Surgical intervention is required only for severe cases like hypoplastic veins or some times for cosmetic improvement of head and neck cutaneous lesions (9).

Thrombosis prophylaxis is required prior to any surgery. Detailed pre-operative assessment of venous system with careful assessment for possible complications like consumptive coagulopathy should be worked up.

Limb amputation may be required in severe deformity (10).

Conclusion :
Klippel Trenaunay Syndrome is a rare sporadic disorder which if diagnosed early, can be treated with a multi-disciplinary approach. Patients can have a normal life span by a combination of the different modalities of treatment for the spectrum of lesions characterising this condition.

Fig 1.Hyperkeratotic nodules over a port-wine stain on right thigh.

Fig 2. Dilated superficial veins on lateral aspect of right leg.

Volume : 5 | Issue : 2 | Feb 2015 | ISSN - 2249-555X

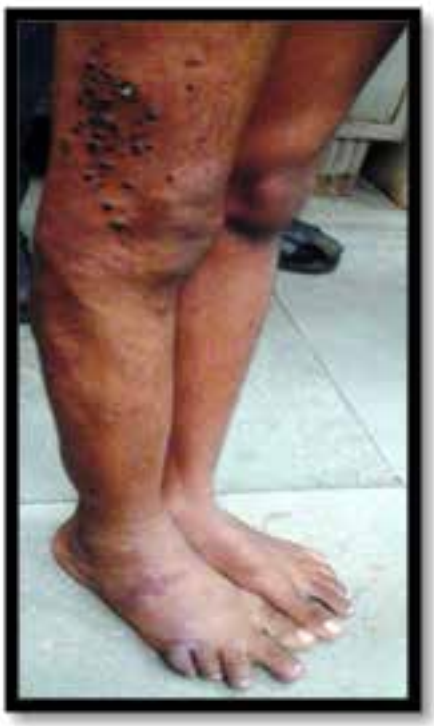

Fig 3. Increased girth of right leg and hypertrophy of middle toe.

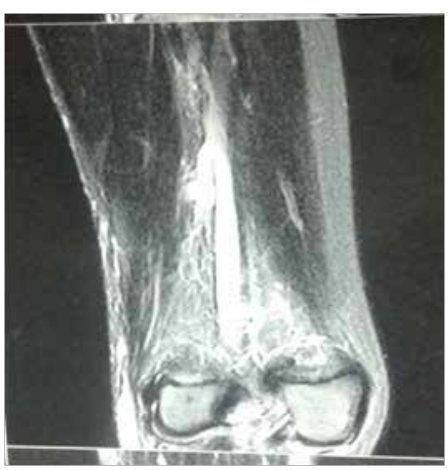

Fig 4. Colour Doppler showing superficial varicosities draining into an abnormal lateral vein.

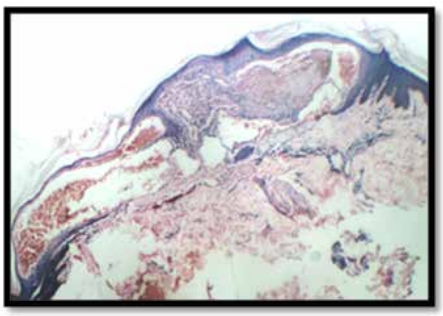

Fig 5.Microphotograph showing dermal ectatic vessels with thrombus, enclosed in epidermal collarette. Subcutaneous tissue is uninvolved.

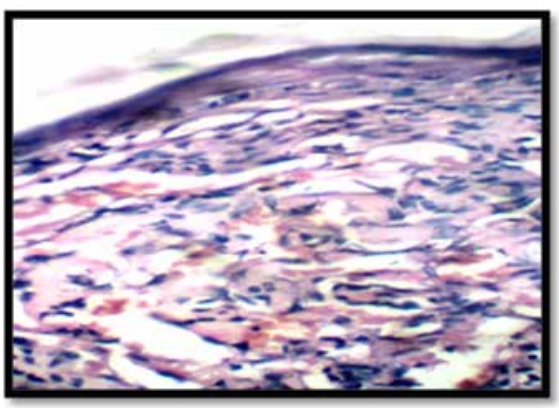

Fig 6.Microphotograph showing multiple dilated vascular channels in superficial dermis.

REFERENCE

1. James JD, Berger TG, Elston DM. Dermal and Subcutaneous tumours. In: James JD, Berger TG, Elston DM, editors. Andrews Diseases of the skin. Clinical Dermatology.11th ed, Philadelphia: Saunders Elsevier. 2011.p.576-8. | | 2. Tian XL, Kadaba R, You SA, Liu M, Timur AA ,and Yang L, et.al. Identification of an angiogenic factor that when mutated causes susceptibility to Klippel-Trenaunay syndrome.Nature.2004; 427:640-5. | | 3. Moss C., Shahdull H. Naevi and other developmental .defects. In: BurnT Breathnach S, Cox N,editors. Rooks Text Book of Dermatology. 8thed.UK: Wiley Blackwell. 2010.p.18.60. | | 4. Chhajed M, Pandit S, Dhawan N, Jain A. Klippel-Trenaunay and Sturge-Weber overlap syndrome with phakomatosis pigmentovascularis. J Pediatr Neurosc .2010;5:138-40. | | 5. Bruce DH. Angiokeratoma circumscriptum and angiokeratoma scroti. Report of a case. | Arch Dermatol 1960 Mar 81:388-93. | | 6. Elder DE. In:Rosalie E, Johnson BL, Murphy GF, editors. Lever's Histopathology of the Skin. 10th ed. Philadelphia: Lippincott Williams and Wilkins. 2009.p.1011 | | 7. Prabhavathy D, Ratnavelu P, Sundraram M, Sugantha. Klippel- trenaunay syndrome. | Indian J Dermatol Venereol Leprol 1994; 60 :161-2. | | 8. Agale SV, Bharambe BM, Pawar R, Valand AG. Klippel trenaunay syndrome: report of a rare , mild form of syndrome. JEMDS 2012,1: 693-6. | | 9. Gloviczki P. Stanson AW. Stickler GB et al. Klippel trenaunay syndrome: the risks and benefits of vascular intervention. Surgery 1991: 110.469-79. | | 10. Jacob A G, Driscoll D J, Shaughnessy W J, Stanson AW, Clay RP , Gloviczki P. | Klippel-Trenaunay syndrome : Spectrum and management. Mayo Clin Proc 1998 ; 73:28-36. |