

Case Report

A rare association of angiokeratoma circumscriptum with Klippel Trenaunay syndrome

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ABSTRACT

Klippel-Trenaunay syndrome (KTS) is a rare congenital disorder of blood and lymphatic vessels that is characterised by a combined vascular malformation of the capillaries, veins and lymphatics, congenital abnormalities, and associated limb hypertrophy. It may also involve gastrointestinal tract which can lead to life threatening bleeding. This syndrome presents since birth to early infancy or childhood with equal distribution in both genders. An adolescent female presented with right lower limb hypertrophy with port wine stain and overlying hemorrhagic vesicles since birth. Histopathology of these hemorrhagic vesicles angiokeratoma. On the basis of history and classical clinical triad patient was diagnosed as KTS with angiokeratoma, an association rarely reported in literature.

Keywords: Klippel Trenaunay syndrome, port wine stain, limb hypertrophy

INTRODUCTION

Klippel Trenaunay syndrome (KTS) is a non-heritable rare disorder characterized by the triad of macular vascular naevus (port-wine naevus), bony and soft tissue hypertrophy and venous varicosities. Incidence of KTS is about 2-5 per 100,000. It affects males more often than females.¹ The vascular malformation is usually limited to a single extremity, though multiple extremities can be involved. Legs are more often affected than arms. Inverse Klippel Trenaunay syndrome term is used to describe upper limb hypertrophy.

We encountered a female child with Klippel Trenaunay syndrome associated with angiokeratoma, cleft palate and urethral bleeding. Occurrence of angiokeratoma with KTS is rare with only a single case reported till date.²

CASE REPORT

A 13 year female child born out of non-consanguineous marriage came with a huge swollen right leg with overlying painful reddish lesions since birth gradually progressing to present size. She also complained of per urethral bleeding with burning micturition since one year. Since last 3-4 years she developed small reddish, painful lesions which bled on friction, initially appearing over right ankle and later involving knee, thigh and buttock. She also complained of perurethral bleeding. She denied history of breathlessness, bone pain, jaundice, palpitation, convulsions. Her systemic examination revealed no significant abnormality.

Examination revealed scars over left side of lip and palate suggestive of repaired cleft lip-palate. Left leg was within normal limit. Multiple hemorrhagic keratotic papulo-

vesicles were present in linear pattern along right leg (Figure 1). Varicosities were noted over involved limb with deformed toes due to soft tissue and bone involvement.



Figure 1: Multiple hemorrhagic keratotic papulovesicles over ankle.



Figure 2: Limb discrepancy.

Right leg was longer than left side by three centimeters (cm). Leg circumference at level of calf region was greater by six cm and mid foot circumference greater by nine cm than left leg (Figure 2). There was a large ill-

defined blotchy reddish non-blanchable macule without bruit suggestive of portwine stain over back extending to right thigh and leg (Figure 3). She had no macroglossia, abdominal wall defects, ear lobe creases or cafe-au-lait macules.



Figure 3: Portwine stain.

We kept differential diagnosis of Klippel –Trenaunay syndrome, Parkes Weber Syndrome, CLOVE (Congenital lipomatous overgrowth, vascular malformations, and epidermal nevi) syndrome. For hemorrhagic vesicles over right leg, we considered lymphangioma circumscriptum, verrucous hemangioma and angiokeratoma as differentials.

On investigation, complete blood cell count, renal function tests, liver function tests, urine and stool routine and microscopy were within normal limits. Chest x-ray, ultrasound of abdomen and pelvis and electrocardiogram were normal.

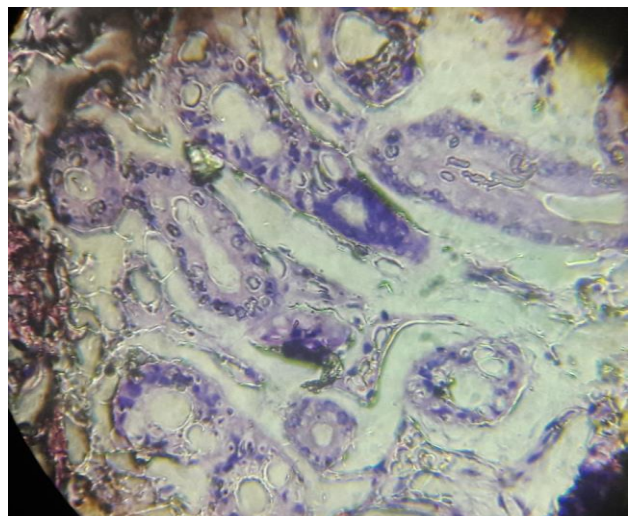


Figure 4: Histopathological examination showing numerous, dilated, thin-walled, congested capillaries mainly in the papillary dermis underlying an epidermis.

Local x-ray of right leg was suggestive of soft tissue hypertrophy predominantly over second and third toes with increase of length of metatarsal and phalanges bone of second and third toes.

Ultrasound of right leg demonstrated increased soft tissue thickness with multiple tortuous, vascular channels with normal compressibility, flow and lumen suggestive of vascular malformation.

Histopathology showed numerous, dilated, thin-walled, congested capillaries mainly in the papillary dermis underlying an epidermis that shows variable degrees of acanthosis with elongation of the rete ridges and hyperkeratosis suggestive of angiokeratoma with nevus flammeus (Figure 4).

Final diagnosis of Klippel Trenaunay syndrome with angiokeratoma was reached based on the classical triad of unilateral limb hypertrophy, port wine stain and varicosities. The patient was managed conservatively with local wound care and compressive bandages.

DISCUSSION

Klippel-Trenaunay Syndrome is characterized by triad of findings considered to represent KTS includes [1] a localized vascular nevus on a body part, [2] hypertrophy of affected tissues, and [3] varicosities in that same body part. Vascular abnormalities occurring in KTS include capillary malformations (such as port wine stains), lymphatic malformations, venous malformations, and arterio-venous malformations (AVM). Typical venous anomalies in KTS include venous hypoplasia and persistence of fetal veins. The overall decreased venous drainage and subsequent congestion that results is thought to cause the varicosities that can be seen on the skin surface, as seen in our patient.³ The affected limb is either larger or smaller than the unaffected limb. Most cases of KTS are caused by somatic mutations in the *PIK3CA* gene. Additionally, a wide range of other skeletal and skin abnormalities sometimes co-exists.

According to Husmann et al, incidence of genitourinary manifestations of Klippel-Trenaunay syndrome is 30% Of 218 patients with Klippel-Trenaunay syndrome, 30% had genitourinary involvement, including 7% with cutaneous genital abnormalities, 7% with visceral genitourinary involvement and 16% with each type. Intermittent bleeding from cutaneous genital abnormalities, as observed in our case, developed in 65% of these patients.⁴

There are several additional conditions that are either historically associated with KTS or confused with KTS. In Parkes Weber syndrome, the same triad is present as in KTS but the limb hypertrophy is caused by multiple arteriovenous fistulae and hence a continuous bruit may be present. Application of a tourniquet often results in bradycardia (Branham sign).

CLOVE syndrome can be distinguished from KTS by the presence of spinal or paraspinal AVM, scoliosis and sometimes seizures.

Our patient satisfied diagnostic criteria proposed by Charle`ne et al, thereby confirming the diagnosis of KTS.⁵

Angiokeratoma is a cutaneous vascular disorder characterized by dilated vessels in the upper part of the dermis. KTS is rarely associated with angiokeratoma. Angiokeratomas are mainly of five types: (1) Solitary or multiple angiokeratomas, (2) angiokeratoma of Mibelli, (3) angiokeratoma of Fordyce, (4) angiokeratoma corporis diffusum, and (5) Angiokeratoma Circumscriptum (AC). AC is the rarest form among all.⁶

In previously reported case of angiokeratoma with KTS, which was seen in 2 ½ year old male child, there was no associated genitourinary complaints and cleft lip and palate defect. However the hemorrhagic vesicles were extensive as compared to our patient.²

Management is largely conservative and the extent of diagnostic evaluation is determined by the planned treatment. Compression is the hallmark of conservative management. A multidisciplinary approach to management of KTS is warranted.⁷ Angiokeratoma lesions can be treated with electrocoagulation, excision, cryotherapy, or laser treatment.

CONCLUSION

Klippel Trenaunay syndrome is a rare congenital disease comprising pathognomonic clinical triad of capillary, lymphatic and venous malformations, congenital abnormalities, and associated limb hypertrophy. Angiokeratoma circumscriptum is a nevoid form which presents as dark red papules and plaques with a verrucous surface. This is itself a rare entity and its association with KTS is even rarer.

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Ethical approval: Not required

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