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CASE REPORT

A case report and brief literature review of Klippel-Trénaunay syndrome

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Abstract: Klippel-Trénaunay syndrome 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-Trénaunay syndrome with limb hypertrophy, port-wine stains, angiokeratoma, and venous varicosities in the limbs.

Keywords: Klippel-Trénaunay syndrome, sporadic, venous varicosities, port-wine stain, angiokeratoma

Introduction

Klippel-Trénaunay syndrome (KTS) is a rare, complex developmental disorder characterized by the triad of cutaneous hemangioma, varicosities, and asymmetrical hypertrophy including the limbs and trunk. It is associated with life-threatening complications such as high-output cardiac failure, consumptive coagulopathy, and internal hemorrhages.[1]

Case report

A newborn male child of a nonconsanguineous marriage presented with right-sided limb hypertrophy, port-wine stains all over the body, angiokeratoma on the right upper limb, and venous varicosities on the right lower limb (Figure 1). Multiple discrete and grouped red and white port-wine stain-lesions were present on the trunk and limbs. Generalized gigantism was seen predominantly on the lower right side of the limb (Figure 2).

His birth weight was 2.8 kg, length was 52 cm, and head circumference was 34.5 cm. The baby was of full-term gestation and delivery was normal. There was no history of similar children in the family or close relatives without any significant antenatal history in the mother during delivery. Limb discrepancy was present between two limbs (Figure 2). Results of a physical examination at the time of admission were normal for gestational age and limb hypertrophy.

On physical examination, no cardiac anomalies were found, there was equal bilateral air entry, and the abdomen was distended with a palpable liver and spleen. External genitalia were normal for a male with right-sided limb hypertrophy, soft-tissue hyperplasia, and multiple vascular anomalies in the form of port-wine stains (Figure 2). The remainder of the general and physical examination was normal.

A complete blood count showed a hemoglobin of 18.6 g/dL, total lymphocyte count of 9800, N-62%, L-31%, and platelet, 2.3 lacs/μL; blood sugar and calcium

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Figure 1 A full-term androgenetic alopecia male baby showing cutaneous hemangioma, angiokeratoma, and hemihypertrophy of the right lower limb.

levels were normal and C-reactive protein was negative. An ultrasonography of the abdomen showed no vascular malformation and magnetic resonance imaging of the brain was normal, while Doppler imaging showed decreased blood flow and stasis in the affected parts. Contrast venography and arteriography were not performed because of parental refusal. The baby needed no active intervention during the hospital stay and was discharged on day 4. Parents were advised to regularly monitor their baby for limb length, girth discrepancies, cellulitis, thrombophlebitis, congestive heart failure, stasis dermatitis, cutaneous ulcers, and bleeding.

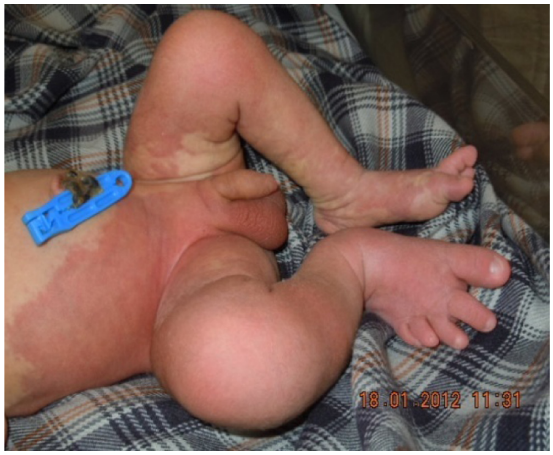

Figure 2 Showing the close view of right lower limb hypertrophy, varicose veins, and port-wine stains.

Discussion

KTS is a capillary-lymphatic venous malformation characterized by the triad of vascular malformation, venous varicosity, and hyperplasia of the soft tissue and bones. It was first described by Maurice Klippel and Paul Trénaunay, and it is also known as angioosteohypertrophy syndrome, congenital dysplastic angiectasis, osteohypertrophy nevus flammeus, and elephantiasis congenital angiomatosa.[1] The etiology of KTS is unknown, but a paradominant inheritance pattern is suggested, and incidence is two to five cases per 100,000 live births, with male dominance.[2] Port-wine stains are the most common type of vascular malformation. These macular telangiectatic vascular nevi are generally present at birth, but at times may appear in early childhood. In a study at the Mayo Clinic, port-wine stains were seen in 98% of patients, varicosities or venous malformations in 72%, and limb hypertrophy in 67%. Atypical veins, including lateral veins and persistent sciatic veins, were present in 72%.[1] In a study of 144 patients, 95% had a cutaneous vascular malformation, 93% had soft tissue and bone hypertrophy, 76% had varicosities, and 71% had involvement limited to one extremity.[3,4] Small angiokeratomas and lesions resembling granuloma telangiectaticum may occur. These patients are more prone to bleeding after trauma.[2] The KT vein is a large, lateral, and superficial vein that begins in the foot or the lower limb and travels proximally until it enters the thigh or the gluteal region. It is associated with pain, thrombophlebitis, and hemorrhaging. Hypertrophy of the affected part is due to soft tissue and fat overgrowth (girth discrepancy), although bony overgrowth (length discrepancy) may be present.[5] There may sometimes be atrophy of a limb rather than hypertrophy, which is known as inverse KTS and is caused by postzygotic recombination.[6] Associated developmental defects include polydactyly,[7] oligodactyly, macrocephaly, blue nevi, pulmonary vein varicosities, cerebral aneurysm, gastrointestinal hemorrhage,[8] pulmonary embolism,[9] incompetent valves in veins, and visceral venous malformations.[2,3] Complications are most often related to the underlying vascular pathological condition. Vascular malformations involving the gastrointestinal and genitourinary tract are a significant source of morbidity and even mortality. Involvement of the gastrointestinal tract occurs in 20% of the patients, can present at any age, and may go unrecognized in patients without overt symptoms. Bleeding is the most common symptom reported in KTS patients with gastrointestinal involvement. The most frequently reported sites are the distal colon and rectum.[10,11] Genitourinary involvement in patients with KTS seems to occur in more severe cases and presents late in life.

Rectal and bladder hemorrhage are serious complications of pelvic vascular malformations and have been reported in 1% of cases. Vascular malformations are often located on the anterior bladder wall and dome, while the trigone and bladder neck are rarely involved. Genital lesions usually do not cause problems for patients with KTS.[12] Pulmonary embolism, cerebral aneurysm, and pulmonary vein varicosities are incidental findings and give rise to life-threatening complications.[9]

Regular clinical and radiographic monitoring of the affected limbs, compression stockings for chronic venous insufficiency, intermittent pneumatic compression devices for reducing limb size, flash lamp-pumped pulsed dye laser for port-wine stains, and surgical correction of varicose veins are needed as required.[13]

Conclusion

We report this case because of its rarity and to reiterate the importance of regular follow up of these patients to reduce complications.

Acknowledgments

MG and ZU were involved in taking patient details, patient management, review of literature, and manuscript writing. RN and CK supervised patient management and drafted the manuscript. MG acted as a guarantor.

Disclosure

The authors report no conflicts of interest in this work.

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