Clinical Quiz

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A child with abnormal lower limb hypertrophy, skin lesion and varicose veins

Clinical Presentation

A 9 month-old boy was referred with complaints of discomfort of the right lower extremity since birth. He was a product of full-term, normal pregnancy. There was no similar problem in the family. A few days after his birth, a prominent asymmetric lower extremity was observed associated with skin nodules in the tongue and the abdomen. On examination, his growth parameters were normal, otherwise he was clinically stable. The lower extremities showed macular vascular nevus (port-wine stains) with abnormal limb hypertrophy and syndactyly (**Figure 1**) while his CT angiogram involving lower extremities demonstrated varicose veins (white arrows) (**Figure 2**).

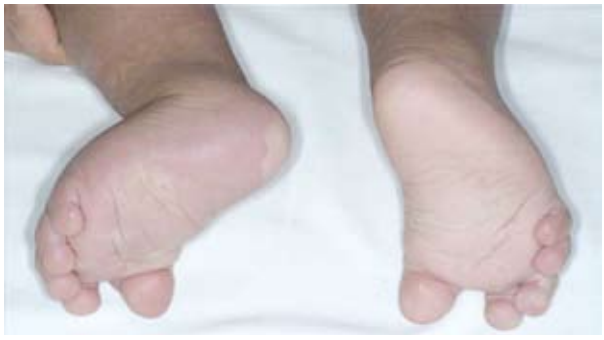

Figure 1 - Patients photograph showing abnormal limb hypertrophy, and syndactyly.

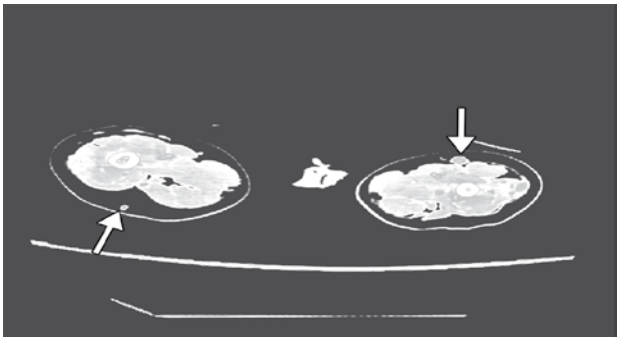

Figure 2 - Patients CT angiogram showing varicose veins.

Questions

1. What is the diagnosis?
2. What is the management?

Clinical Quiz

Answers and Discussion

1. ***Diagnosis.*** Klippel-Trenaunay Syndrome. In 1900, Klippel and Trenaunay,[1] described a syndrome that was characterized by the following 3 features: 1) a cutaneous vascular nevus of the affected extremity (capillary malformation), 2) soft tissue and bony hypertrophy of the extremity, and 3) varicose veins, or venous malformations. Klippel-Trenaunay syndrome (KTS) is a rare congenital generalized mesodermal abnormality characterized by combination of 1) capillary malformations (usually port-wine stains), which frequently are located laterally, need not extend over the entire affected limb, and may be found at sites other than the hypertrophied limb; 2) soft tissue, or bony hypertrophy (or both); and 3) varicose veins, or venous malformations, often with persistent lateral embryologic veins.[2] The cause of KTS remains obscure. It has been suggested that intrauterine injury to the sympathetic ganglia or intermediolateral tract, resulting in dilatation of microscopic arteriovenous anastomoses, may be the underlying cause.[3]
2. ***Management.*** In most patients with KTS, management should be nonoperative. The indication for epiphysiodesis is a leg length discrepancy that exceeds 2.0 cm in the growing child. Amputation of a grossly hypertrophied, poorly functioning digit may be necessary, but a more proximal amputation of a foot, hand, or limb is rarely warranted. Removal of symptomatic varicosities or localized venous malformations can yield good results in selected patients, provided a functioning deep vein system is present.[4,5]

References

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