Mild form of inverse Klippel-Trenaunay syndrome?

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DESCRIPTION

Klippel-Trenaunay syndrome (KTS) is defined by a coexistence of nevus flammeus and overgrowth of one or more limbs. Remarkably, however, deficient growth of an affected limb may likewise be noted. It has been speculated that either a ‘plus’ or ‘minus’ allele at the responsible gene locus in the postzygotic cells might be involved in the occurrence of overgrowth or underdevelopment of the affected limb, respectively.1

Orthopaedic complications are related to limb asymmetry and bone hypertrophy. Arteriovenous malformations may lead to thromboembolic events or high-output cardiac failure.2

A 5-year-old boy was referred to the dermatology clinic for evaluation of a congenital port-wine strain in the right buttock. He was otherwise healthy, apart from past surgery for cleft palate correction.

On examination we observed a telangiectatic nevus with focal dermal and soft tissue atrophy in the right buttock (figures 1 and 2). There was no significant difference in limb length or circumference.

MRI excluded major organ or vascular abnormalities.

In principle, a pre-requisite of KTS is partial or complete overgrowth of a limb. However, there are also patients with shortening or smaller muscle mass of the affected limb. This fact lead to the recently proposed concept of ‘inverse Klippel-Trenaunay syndrome’.13 This case may be included in the mildest spectrum of this clinical condition.

Learning points

▸ The association between port-wine nevus and soft tissue/limb hypotrophy or hypertrophy should recall the Klippel-Trenaunay syndrome (KTS) spectrum.
▸ A child with KTS requires limb growth monitoring in order to prevent orthopaedic complications.
▸ When feasible and clinically relevant, imaging studies may be helpful in excluding major vascular abnormalities.

Competing interests None.

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REFERENCES
