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'.[1 3] This case may be included in the mildest spectrum of this clinical condition.

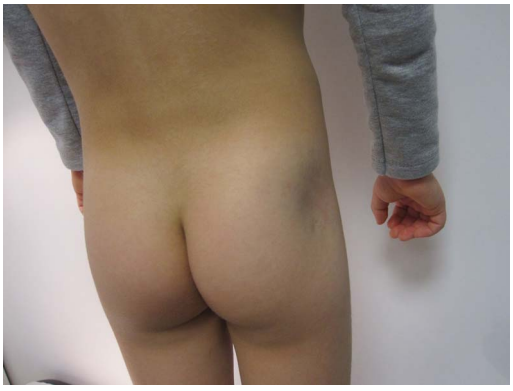

Figure 1 Focal dermal and soft tissue atrophy.

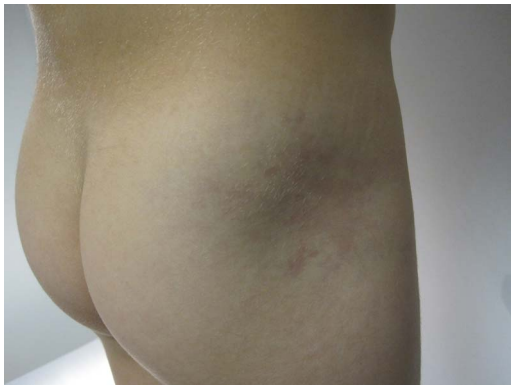

Figure 2 Telangiectatic nevus.

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.

To cite: Queirós G, Diamantino F, Lopes MJ. *BMJ Case Rep* Published online: [*please include* Day Month Year] doi:10.1136/bcr-2013-009188

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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