Klippel-Trenaunay syndrome: diagnosis in a neonate

Cátia Pereira, Rita Espírito Santo, Joana Saldanha

Department of Paediatrics, Neonatology Unit, Hospital Santa Maria, Centro Hospitalar Lisboa Norte, Lisboa, Portugal

Correspondence to
Dr Rita Espírito Santo, a.r.
espiritosanto@gmail.com

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DESCRIPTION

A male newborn was evaluated due to a port-wine stain. Mother, 40 years old, father and brother were healthy. Gestation was uneventful. Amniocentesis revealed a normal male karyotype. A caesarean delivery was performed at 38 weeks. First physical examination showed a port-wine stain affecting the abdomen, back and left limb (figure 1) and hypertrophy of the affected limb. A biopsy was performed and histological findings revealed capillary malformations. Therefore, a Klippel-Trenaunay syndrome was diagnosed. Abdominal and lower limb doppler ultrasound and brain MRI excluded other vascular abnormalities. He was followed by a multidisciplinary team. At 9 months, there was a slight difference in the length of legs and the circumference of thighs (figure 2).

Klippel-Trenaunay syndrome is a rare disease, characterised by capillary and venous malformations and soft tissue or bone hypertrophy with overgrowth of the affected extremity.[1] Its aetiology remains unknown.[2] Klippel-Trenaunay syndrome affects both genders equally.[1] The cutaneous capillary malformation presenting as a port-wine stain and limb hypertrophy are usually noted at birth.[2] Some cases presenting with atrophy and reduced growth of the affected limb have been described. Venous malformations may be present at birth or appear during infancy.[1] The lower extremity is the most commonly affected. However, it can involve upper limbs and extends to the trunk.[2] Doppler ultrasound allows to identify abnormalities of the venous system and MRI helps to characterise vascular malformations.[1 2] Complications may include limb-length discrepancy leading to impaired gait and pain, thromboembolism, bleeding, venous insufficiency and soft-tissue infection.[1 3] Treatment is mainly supportive and includes management of complications.[1]

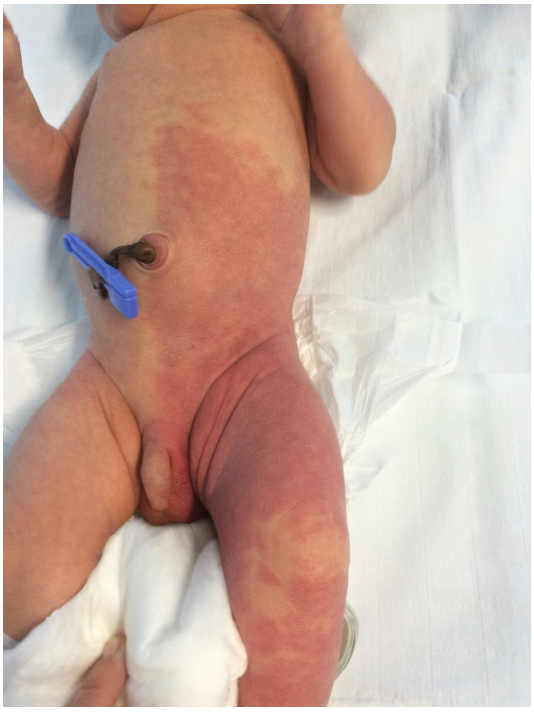

Figure 1 A port-stain affecting the abdomen and left limb at first physical examination of the neonate.

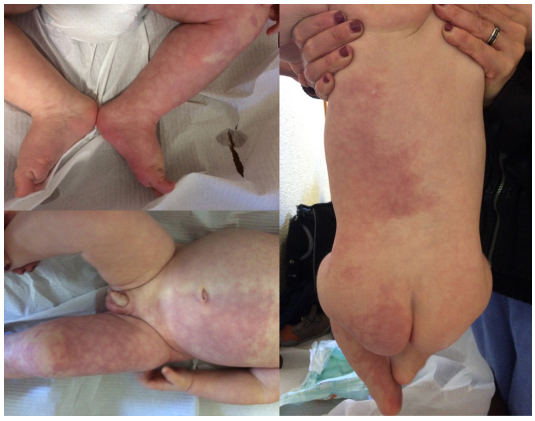

Figure 2 Appearance of the abdomen, trunk and limbs at 9 months of age.

Learning points

- Klippel-Trenaunay syndrome is a rare disease, presenting with a port-wine stain and limbs asymmetry.
- Physicians, namely neonatologists and paediatricians, should be aware of Klippel-Trenaunay syndrome in order to be able to make an accurate diagnosis and treat complications.

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Competing interests None declared.

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