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CASE REPORT

Klippel-Trenaunay Syndrome Presenting in a Child with Vascular Malformation

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ABSTRACT

Klippel-Trenaunay syndrome is an uncommon disease mainly affecting the paediatric population. This syndrome is rare among Chinese people. This is a report of a patient with Klippel-Trenaunay syndrome with emphasis on magnetic resonance imaging, arteriogram, and venogram. The overall features and treatment of Klippel-Trenaunay syndrome are reviewed.

Key Words: *Arteriovenous malformations, Child, Klippel-Trenaunay-Weber syndrome*

CASE REPORT

A 3-year-old Chinese girl presented with pigmented lesions over the right thigh since birth. Progressive enlargement and episodes of unprovoked bleeding from the lesions brought her to medical attention at the age of 2 years. Physical examination at that time revealed multiple pigmented lesions extending from the right hip over the thigh to the knee region, and a provisional diagnosis of cavernous haemangioma was made. Lower limb venogram (Figure 1), however, revealed the absence of normal deep veins. Only a single dilated valveless superficial varicose vein was present on the lateral aspect of the thigh and leg, which drained the entire lower limb. A diagnosis of Klippel-Trenaunay syndrome was made.

Repeated attempts of sclerosing therapy using hypertonic saline injection were not successful. Throughout her childhood years, the patient suffered from episodes of bleeding from the thigh lesion and per rectum bleeding that necessitated repeated blood transfusions. Progressive hypertrophic changes in the right lower limb resulted in an increase of length and girth compared with the healthy left limb. At the age of 12 years, magnetic resonance imaging (MRI) was performed for interval assessment (Figure 2). The scan depicted extensive serpiginous and roundish foci with hypointense signal intensity on T1-weighted images and hyperintense signal intensity on T2-weighted images in the subcutaneous fat over the right lateral and posterior aspect of

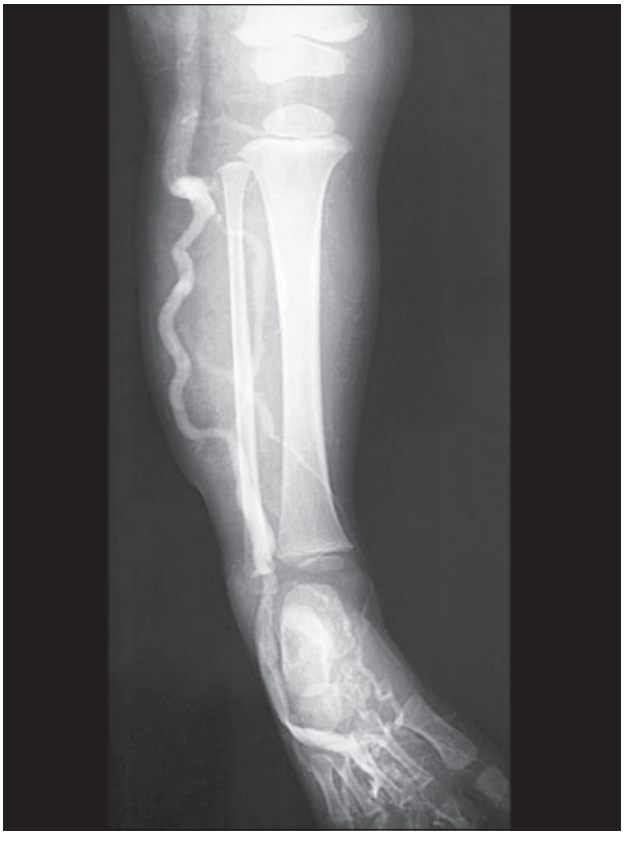

Figure 1. Right lower limb venogram showed only a prominent superficial vein in the lateral aspect of lower limb. The normal deep venous system was not shown.

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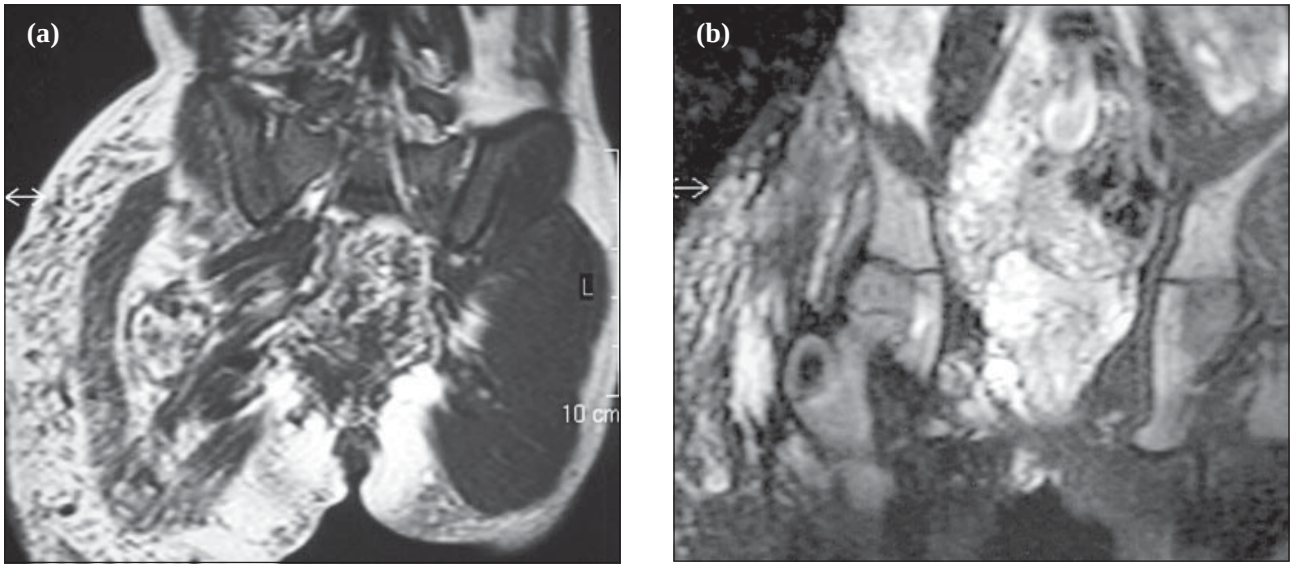

Figure 2. (a) Coronal T1-weighted spin-echo magnetic resonance image: there were diffuse hypointense lesions in the subcutaneous fat of the pelvis, perineum and right gluteal maximus muscle. (b) Coronal T2-weighted turbo spin-echo magnetic resonance image showed hyperintense lesions extending to the right thigh. The soft tissue on the affected side was thicker than on the healthy side with possible leg-length discrepancy.

the pelvis, with involvement of the gluteal maximus muscle and right ischiorectal fossa, and extension to the inguinal region. The lesions in the right ischiorectal fossa abutted the right anal and rectal wall, but without involvement. There was also extension of lesions into the fat space of the right perineum and labia majora and minora. The vagina was intact. Similar serpiginous lesions were diffusely distributed in the right hypertrophic lower limb. The muscles and soft tissue were involved.

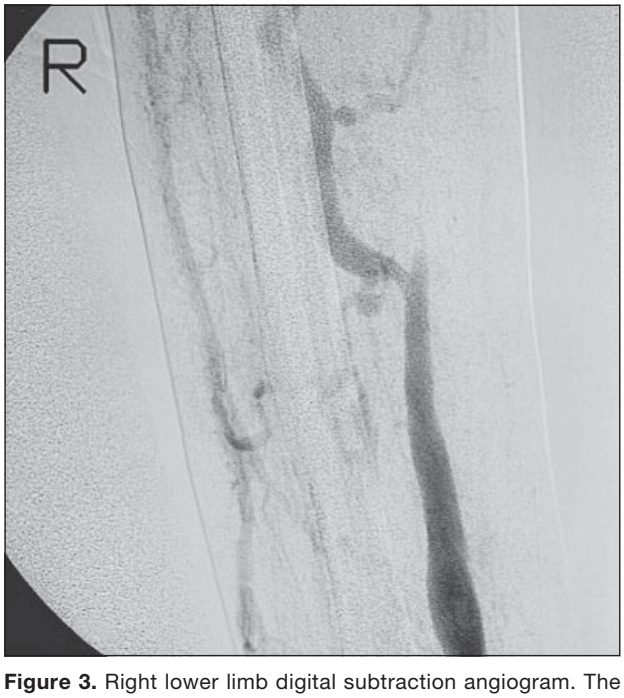

Figure 3. Right lower limb digital subtraction angiogram. The venous phase showed a single superficial vein in the lateral aspect of the lower limb.

The overall features were those of a diffuse soft tissue capillary haemangioma.

Due to increasing frequency of bleeding from the thigh and buttock region, a second lower limb arteriogram, which demonstrated essentially similar findings to the previous venogram, was performed in 1998 when the patient was 13 years old. There was no abnormal arterial vascular malformation but the large superficial vein was well shown in the venous phase. Embolisation was not feasible as the dilated superficial vein was probably the only draining vein of the whole right lower limb. The patient was conservatively treated with repeated admission to hospital for blood transfusion.

DISCUSSION

Klippel-Trenaunay syndrome is a sporadic rare mesodermal abnormality that usually affects a single lower limb. The lower extremity is 10- to 15-fold more likely to be affected than the upper extremity. Bilateral involvement is relatively rare and is found in less than 5% of patients. The syndrome usually involves children and there is no sex bias.

The syndrome is characterised by a triad of soft tissue hypertrophy of the extremety, diffuse venous malformations, and capillary haemangioma. Capillary haemangioma is unilateral and cutaneous, often distributed dermatomally on the affected limb and is clinically seen as a flat port-wine nevus. Involvement of the whole side of the body or of the contralateral limb may be seen.[1,2]

Limb overgrowth is apparent during the adolescent growth spurt. The enlargement is due to bone elongation or circumferential soft tissue hypertrophy.[1,2] Varicose veins are found on the lateral aspect of the affected limb in the majority of patients,[1-3] and these may be prominent. For more than two-thirds of patients, a characteristic incompetent lateral venous channel arises near the ankles and extends a variable distance up the extremity to the infrainguinal or pelvic deep venous system.[2] Extremity pain, spontaneous cutaneous haemorrhage, chronic venous insufficiency, or thrombophlebitis are commonly encountered.[1,2]

In this patient, lower limb venography shows the absence of a deep venous system and the presence of an ipsilateral prominent vein, extending from the ankle to the groin, which is responsible for the entire venous drainage of the lower limb. This finding is important for planning the management because resection or embolisation would be contraindicated. Worsening of the clinical situation has been reported when resection of varicose veins was performed for dilated superficial collateral veins associated with deep vein hypoplasia.[1] Extensive capillary haemangioma is present in the pelvic region in the patient described here. Diffuse involvement of the perineum, soft tissue, and gluteal muscles is uncommon, although extension of venous malformation into the pelvis resulting in recurrent rectal bleeding or haematuria have been documented in a few patients.[4]

X-ray could demonstrate bone elongation contributing to leg length discrepancy, soft tissue thickening, or calcified phleboliths. Venographic findings are more characteristic of this syndrome, however. Extensive dilation of superficial veins and segmental absence or hypoplasia of the deep venous system may be found. Angiography is necessary for the exclusion of arterial-venous malformation, since the disease may be simulated by Parkes-Weber syndrome in which limb overgrowth and varicosities due to congenital persistence of multiple arteriovenous malformation is characteristically observed.[5]

MRI provides lesion morphology in another perspective. The hypointensity on T1-weighted images and hyperintensity on T2-weighted images allows a clear delineation of the venous and lymphatic malformation.[6] Deep extension of these lesions into muscular compartments and the pelvis, and their relationship to adjacent organs and structures, and the bone or soft-tissue hypertrophy can be displayed on sectional images in different planes.[1] More recently, delineation of significant features of extremity venous malformation on MR venography using 2D time-of-flight technique has been reported to be comparable to conventional venography. Specifically, a global picture of the superficial varicosities, enlarged perforating veins, and absent or hypoplastic deep veins characteristic of the syndrome can be effectively demonstrated.[7]

Treatment for the majority of patients with Klippel-Trenaunay syndrome is conservative, including application of graded compressive stockings or pneumatic compression devices to the enlarged extremity. Percutaneous sclerosis of localised venous malformations or superficial venous varicosities may be indicated for some patients.[7] Surgical treatment includes epiphysiodesis to control leg length discrepancy, excision of soft tissue hypertrophy, stripping of superficial varicose veins or, less commonly, reconstructive surgery at sites of deep venous obstruction.[2,3]

CONCLUSION

The constellation of extremity soft tissue hypertrophy, diffuse venous malformations, and the capillary haemangioma in this patient are characteristic of Klippel-Trenaunay syndrome. This syndrome should be borne in mind as a differential diagnosis of hemihypertrophy with haemangioma.[8] MRI allows an exquisite anatomical delineation of the lesion. Angiography and venography to assess the underlying vascular anatomy are also important parts of the complete examination.

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