

Case Report

Klippel–Trenaunay syndrome in combination with congenital dislocation of the hip

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Abstract

Klippel–Trenaunay syndrome (KTS) is a rare and sporadic disorder characterized by the triad of capillary malformations, venous varicosities, and limb hypertrophy. The clinical manifestations of KTS are heterogeneous. In this report, we present a unique case of KTS in combination with congenital dislocation of the hip (CDH) in a 4-day-old female neonate. The patient had a widespread port-wine stain surrounded by regions of unaffected skin in a mosaic pattern, cutaneous hemangioma on the upper lip, left-sided hemihypertrophy involving the entire body, and also evidence of left CDH (based on the results of a physical examination and radiographic interpretation). We present this case for the rarity of presentation, discuss the relationship between KTS and CDH, and the treatment options available with a brief review of the literature.

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1. Introduction

In 1900, two French physicians, Maurice Klippel and Paul Trenaunay, originally described a congenital syndrome characterized by the triad of capillary malformations, venous varicosities, and limb hypertrophy.¹ Klippel–Trenaunay syndrome (KTS) is a rare condition with obscure etiology and variable expression besides these three main symptoms. The prevalence rate of KTS is approximately 1 per 20,000–40,000 live births.² Children of any ethnic group can be born with KTS, and it equally affects both genders. Although KTS is present at birth, often the only visible sign in babies is a port-wine stain and the diagnosis may not be confirmed until the venous varicosities or limb hypertrophy becomes more noticeable.³ However, the patient reported herein, a 4-day-old

female neonate, had a very rare form of obvious KTS, and furthermore, suffered from congenital dislocation of the hip (CDH).

2. Case report

A 4-day-old female neonate was admitted to the Department of Pediatrics, the First Affiliated Hospital of Anhui Medical University, Hefei, in March 2009 for an evaluation of widespread port-wine stain and hemihypertrophy. She was born at 37 weeks' gestation, to a G2P1, 22-year-old woman by lower cesarean section. The baby weighed 3800 g and had Apgar scores of 9 and 10 at 1 and 5 minutes, respectively. The placenta and amniotic fluid were grossly normal. Medical history of her mother was not significant, and the mother's family had no history of similar disorders. This mother's first pregnancy was terminated by induced abortion at 5 weeks' gestation.

A physical examination of the baby showed that there was a widespread port-wine stain on both sides of her face, which

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extended to both lower extremities, surrounded by regions of unaffected skin in a mosaic pattern. In addition, the left-sided hemihypertrophy was prominent in the face, trunk, and leg. A bright red cutaneous hemangioma was also present on the upper lip (Fig. 1). Although the left leg was larger in circumference than the right leg, there was no significant difference in the length of both lower limbs. More specifically, asymmetric skin folds were noted in the groin, below the buttock, and an unstable left hip was identified by Ortolani and Barlow's test (Fig. 2). Results of hematological, biochemical, urinary laboratory tests, coagulation tests, and the chest radiograph were all normal. An abdominal ultrasonography and cranial computed tomography scan did not show any pathological findings. However, plain radiographs of the hips and lower limbs revealed left CDH, and hypertrophy of bones and soft tissues (Fig. 3).

The patient was diagnosed with KTS in combination with left CDH, according to the results of a physical examination and radiological evaluation. Subsequently, she was referred to the Department of Plastic Surgery, Yuying Children's Hospital of Wenzhou Medical College, for further consultation. After substantial discussion, we concluded that no current intervention should be conducted to treat the patient's KTS. Even after 2 weeks of intensive observations, the left CDH continued to persist. Under the circumstances, a Pavlik harness was used to dynamically position the hips in flexion and abduction positions, while continuing to allow motion.

3. Discussion

The triad of capillary malformations, venous varicosities, and limb hypertrophy has been found to occur in 98%, 72% and 67% of children with KTS, respectively, according to the report of Jacob et al.⁴ The diagnosis of KTS can be commonly



Fig. 1. The face showed asymmetry with the left side exhibiting hypertrophy compared with the right side. A bright red cutaneous hemangioma was present on the upper lip.

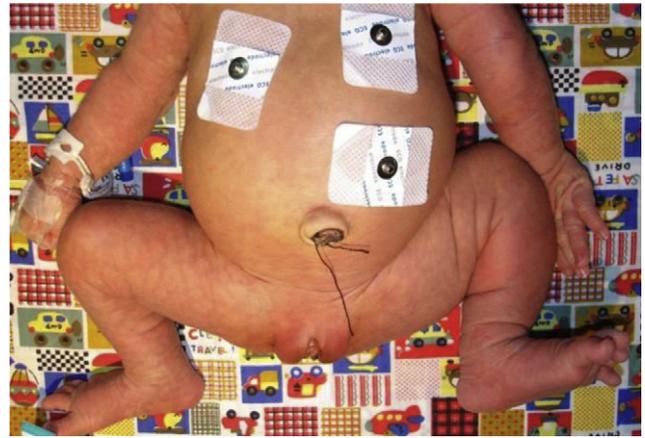


Fig. 2. Although the left leg was larger in circumference than the right leg, there was no significant difference in length between the lower limbs. More specifically, asymmetric skin folds were noted in the groin, below the buttock, and unstable left hip was identified by Ortolani and Barlow's test.

made when any two of the three cardinal features are visible. In fact, KTS is present at birth but only involves the port-wine stain in the neonatal period. Venous varicosities and limb hypertrophy may gradually become obvious as these babies grow older.⁵ This disease must be differentiated from other diseases that involve reticulated vascular lesions. Cutis marmorata telangiectatica congenita (CMTC) is a rare, sporadic condition that presents at birth as a localized or generalized reticulated, blue-violet vascular network in the skin.⁶ The cutaneous lesions of CMTC commonly occur on the legs, arms, and trunk, but rarely involve the face and scalp; characteristically, they become more prominent in a cold environment but generally do not disappear with rewarming.⁷ Whereas in our case, we encountered a 4-day-old female



Fig. 3. Anteroposterior plain radiographs of the hips and lower limbs revealed left congenital dislocation of the hip, and hypertrophy of bones and soft tissues. The Hilgenreiner's line is drawn horizontally through the triradiate cartilages of the pelvis, while the Perkin's line is drawn perpendicular to the Hilgenreiner's line at the lateral edge of each acetabulum. Note that the femoral head on the right (normal) lies in the inferomedial quadrant formed by these lines. The left hip is dislocated and its femoral head lies in the inferolateral quadrant.

neonate who is considered as an interesting and definite case of KTS, which can be attributed to the presentation of wide-spread port-wine stain, cutaneous hemangioma, and left-sided hemihypertrophy at birth.

Although the exact etiology of KTS remains obscure, various theories are being argued by the medical community. Genetic and environmental factors destroying the blood vessel system, which may develop during pregnancy can lead to the onset of KTS.⁸ Thus, capillary malformations, such as port-wine stain and cutaneous hemangioma, are the most significant pathophysiological changes other than the earliest clinical manifestation. Limb hypertrophy is closely correlated with increased angiogenesis, which might explain the hypertrophy of bones and soft tissues in our patient at birth.⁹ However, based on this hypothesis, one question confused us. Why were the distributions of capillary malformations and limb hypertrophy inconsistent? Further investigation of our patient is warranted through molecular and genetic methods. To the best of our knowledge, venous varicosities, the third main symptom, have not been documented in infantile KTS cases until now. This characteristic was consistent with the findings of the current case. However, this still highlights the continuing medical challenge and eventuality that these varicose veins would become more prominent and problematic, accompanying growth and development as these patients get older, due to the increasing venous pressure, valve incompetence, or outflow obstruction.¹⁰

Associated complications derived from KTS are pain, cellulitis, ulceration, thrombophlebitis, gangrene, consumptive coagulopathy, high-output cardiac failure, and tumor, etc.² However, KTS may rarely occur in combination with CDH. Interestingly, in this report, we presented a 4-day-old female neonate with obvious KTS, who also suffered from left CDH. A physical examination of the child showed asymmetric skin folds in the groin and below the buttock, with an unstable left hip identified by Ortolani and Barlow's test, which was identified with the findings of the plain hip radiographs. The diagnostic procedures of CDH were conformed to those procedures in previous studies.¹¹ There are several factors that contribute to CDH, including breech presentation, female gender, positive family history, firstborn status, and oligohydramnios.¹² However, in the case of our patient, none of these susceptible factors except for the female gender existed. About 80% of children with CDH were female, according to Wilkinson's report.¹³ This was postulated to be related to the effects of additional levels of estrogen produced by the female fetus, which increases ligamentous laxity. Moreover, we speculated that the limb hypertrophy might put the homolateral hip out of line in the neonate with KTS.

KTS itself cannot be treated, but the symptoms associated with it can be improved to a great extent. Laser therapy and sclerotherapy are useful in some patients for decreasing the intensity of capillary malformation and for cosmetic reasons. Physiotherapy and compression can help ameliorate hypertrophy and venous varicosities.¹⁴ If a discrepancy in leg length exceeds 2 cm, the growth in the affected leg can be slowed down using an operation called epiphysiodesis, or the shorter leg could be lengthened.¹⁵ The common complications such as

pain, cellulitis, and thrombophlebitis can be managed using analgesics, antibiotics, and anticoagulants, respectively.² Because the severity of symptoms of KTS varies from one patient to another, the suggested treatment options are alternative, and not every patient will require all these treatments. In addition, a multidisciplinary team is also necessary to provide the best medical care for such patients.¹⁶ In the current presentation, taking into account the growth and development of babies, we concluded that no interventions should be conducted to treat KTS in this patient. CDH often corrects spontaneously and may be observed for 2 weeks without treatment.¹⁷ Unfortunately, even after 2 weeks of intensive observation, the left CDH of our KTS patient continued to persist. Under the circumstances, a Pavlik harness dynamically positioned the hips in flexion and abduction positions, while still allowing motion. Lastly, it is also very important to develop an optimal doctor–patient relationship to provide adequate social support during the management of KTS.

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