Section: Paediatrics

Case Report

KLIPPEL-TRENAUNAY SYNDROME WITH SEVERE ANEMIA SECONDARY TO VASCULAR MALFORMATIONS: A RARE PEDIATRIC PRESENTATION

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ABSTRACT

Background: Klippel-Trenaunay Syndrome (KTS) is an uncommon congenital vascular disorder characterized by capillary malformations, venous anomalies, and limb overgrowth. Anemia resulting from chronic bleeding secondary to vascular malformations is an unusual and underreported complication, particularly in pediatric cases.

Clinical Description: We present the case of a 4-year-old male with KTS who developed recurrent severe anemia due to bleeding from vascular lesions. The child exhibited macrocephaly, dolichocephaly, pectus carinatum, and bilateral lower limb hypertrophy. Hemoglobin levels dropped as low as 2.3 gm/dL, necessitating multiple transfusions.

Management: Magnetic Resonance Imaging (MRI) revealed extensive lymphovenous malformations in the pelvis and lower limbs. The patient received blood transfusions and symptomatic care, followed by referral to a multidisciplinary team for surgical evaluation. He was discharged with stable hemoglobin and advised regular follow-up.

Conclusion: This case highlights the importance of recognizing chronic bleeding and severe anemia as potential complications in KTS, warranting early imaging and coordinated multidisciplinary management.

Keywords: Klippel-Trenaunay Syndrome, severe anemia, pediatric vascular malformations, multidisciplinary management.

INTRODUCTION

Klippel-Trenaunay Syndrome (KTS) is an uncommon congenital vascular disorder characterized by a classical triad of capillary malformations (typically presenting as port-wine stains), venous malformations, and soft tissue and/or bony overgrowth of an extremity1. The estimated incidence is approximately 1 in 100,000 live births, though the clinical presentation can vary significantly, ranging from mild, asymptomatic cases to severe, potentially life-threatening complications.[1,2]

While the hallmark features of KTS limb hypertrophy and vascular anomalies—are well-established in the literature,[2,3] the occurrence of severe anemia due to chronic bleeding from cutaneous vascular malformations is exceedingly rare, particularly in the pediatric population. Such anemia, often under-recognized, can lead to significant morbidity if not promptly identified and managed. Early recognition of this rare complication, combined with comprehensive imaging and multidisciplinary care, is essential to improving outcomes in affected children. In this case report, we describe a 4-year-old male with KTS who presented with recurrent, transfusion-dependent severe anemia caused by bleeding

vascular lesions. This case underscores the importance of early diagnosis, comprehensive imaging, and multidisciplinary care in managing complex presentations of KTS, while adding to the limited pediatric literature on this rare but serious complication.

Clinical Description

A 4-year-old male presented with a 10-month history of intermittent shortness of breath and occasional bleeding from vascular lesions over the right knee, accompanied by a two-day history of fever. The child had a history of recurrent severe anemia, necessitating multiple hospital admissions and blood transfusions at 9 months and again at 4 years of age, when hemoglobin levels were recorded at 5 gm/dL and 2.3 gm/dL, respectively.

He was born via cesarean section at 36 weeks gestation, with a birth weight of 2.5 kg. Developmental assessment revealed global delay, with motor milestones achieved at 18 months and speech development by 2 years of age. The family history was notable for a sibling with glaucoma and hydrops cornea, managed surgically with tarsorrhaphy.

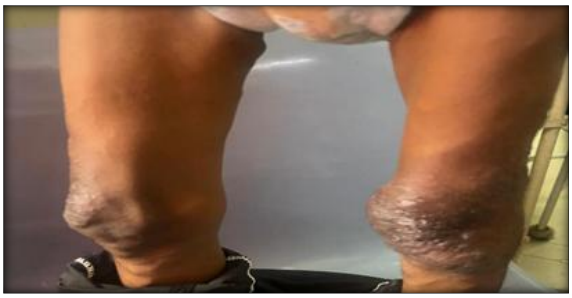

Figure 1: Bilateral Knee Vascular Malformations with Skin Changes in Klippel-Trenaunay Syndrome

Physical examination revealed distinct craniofacial and skeletal features, including macrocephaly (cephalic index: 0.69), dolichocephaly, sutured eyelids from previous glaucoma surgery, megalocornea, pectus carinatum, and bilateral lower limb hypertrophy. Cutaneous examination showed vascular malformations over the right knee, some of which were actively bleeding. Vital signs on admission were stable, with a heart rate of 124 beats per minute, respiratory rate of 28 breaths per minute, and oxygen saturation of 94% on room air.

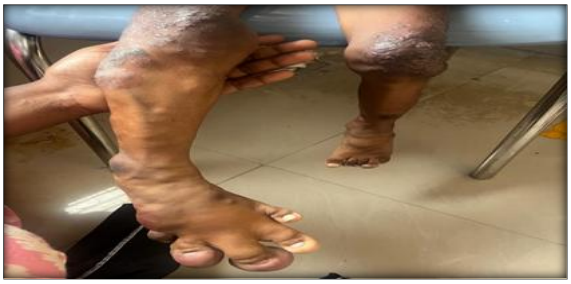

Figure 2: Hypertrophy of Lower Limbs with Cutaneous Vascular Malformations in a Child with Klippel-Trenaunay Syndrome

Laboratory investigations revealed severe anemia with hemoglobin of 4.7 gm/dL. Peripheral smear demonstrated pronounced anisopoikilocytosis with predominantly microcytic hypochromic red cells, pencil cells, target cells, and macroovalocytes, indicative of a dimorphic anemia. No hemoparasites were detected, and the white blood cell count and platelet count (261 × 10^3/μL) were within normal limits.

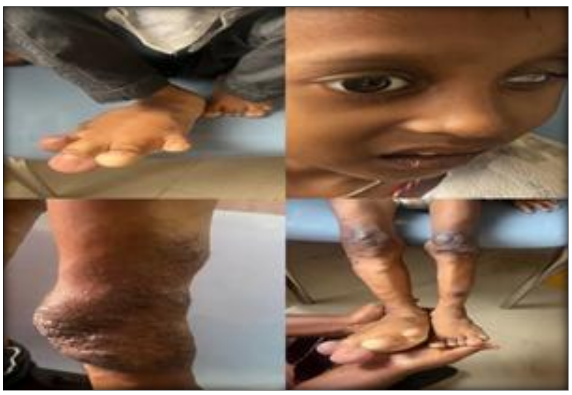

Figure 3: Clinical Manifestations of Klippel-Trenaunay Syndrome in a Child: Limb Hypertrophy, Cutaneous Vascular Malformations, and Craniofacial Anomalies

Management and Outcome

The initial management focused on stabilizing the child's condition by correcting the severe anemia through packed red blood cell transfusions, which successfully increased the hemoglobin level to a safer range. Given the presence of the classical clinical triad capillary malformations (bleeding cutaneous lesions), venous malformations, and soft tissue overgrowth a comprehensive evaluation was undertaken to confirm the diagnosis and assess the extent of vascular involvement.

Magnetic Resonance Imaging (MRI) of the abdomen, pelvis, and lower limbs revealed extensive lymphovenous malformations involving the gluteal and pelvic musculature, extending into the ischio-rectal fossa and both lower limbs, including the thighs, calves, and ankles. Importantly, no arterial abnormalities were identified. The recurrent severe anemia was attributed to chronic low-grade blood loss from these fragile vascular malformations.

Supportive care was provided to manage the associated fever, and a multidisciplinary team including pediatricians, hematologists, vascular surgeons, and radiologists—was engaged to plan long-term management. Surgical or interventional options were considered for controlling recurrent bleeding and addressing progressive limb overgrowth.

The child was discharged after four days of hospitalization with hemoglobin improved to 9.8 gm/dL. Outpatient follow-up was arranged to monitor for recurrence of anemia, signs of infection, or further progression of vascular or skeletal abnormalities. Longitudinal follow-up is also planned to assess treatment adherence, functional

outcomes, and quality of life for both the patient and caregivers, although these aspects were not evaluated at the time of discharge.

DISCUSSION

The diagnosis of Klippel-Trenaunay Syndrome (KTS) in this case was established based on the presence of the classical clinical triad: capillary malformations, venous anomalies, and soft tissue and/or bony overgrowth. The identification of extensive lymphovenous malformations on imaging further supports this diagnosis, reflecting the well-recognized heterogeneity in the vascular presentations of KTS.[3]

While limb hypertrophy and vascular malformations are hallmark features of KTS, the development of severe, transfusion-dependent anemia as a primary complication remains exceedingly rare, particularly in the pediatric population.[4] The most plausible pathophysiological explanation for anemia in this case is chronic, low-grade hemorrhage from fragile cutaneous vascular lesions a mechanism that has been sparsely reported in children. This highlights an often-overlooked but potentially life-threatening aspect of the syndrome. The presence of a dimorphic peripheral blood picture, alongside a history of multiple transfusions, also suggests the possibility of iron deficiency anemia and/or hemolysis, emphasizing the need for thorough hematological assessment in such presentations.

Magnetic Resonance Imaging (MRI) played a critical role in this case, not only confirming the extent of the vascular malformations but also excluding arterial involvement—information essential for both diagnosis and the planning of surgical or interventional management strategies3. Early and comprehensive imaging remains a cornerstone in the evaluation of KTS, particularly in cases where complications such as bleeding, thrombosis, or progressive limb deformities are suspected.[3,4]

Previous reports of KTS in children have predominantly described complications such as thromboembolism, limb overgrowth, or gastrointestinal involvement.[5,6] In contrast, this case draws attention to severe anemia as an underrecognized yet significant clinical manifestation. While adult cases have occasionally documented bleeding and anemia, pediatric instances remain exceptionally rare, with limited cases reported in the literature6.

The presence of craniofacial anomalies, macrocephaly, and a sibling with congenital glaucoma in this patient raises the possibility of a broader syndromic or genetic association that could not be fully explored due to the unavailability of genetic testing. Such overlaps have been described in some case reports, suggesting that KTS may, in certain patients, represent part of a wider spectrum of developmental anomalies.[6]

Additionally, KTS is associated with an increased risk of thrombotic complications, including deep vein thrombosis and pulmonary embolism, which can further complicate management.[7] Although these were not observed in this child, awareness and surveillance for such events remain crucial in long-term follow-up.

The limitations of this case include the lack of genetic evaluation, which could have provided insights into potential syndromic overlaps, and the short duration of follow-up, which limits conclusions about long-term outcomes, disease progression, and quality of life. Nevertheless, the case underscores the critical importance of multidisciplinary management involving pediatrics, hematology, vascular surgery, and radiology to optimize outcomes in KTS.

CONCLUSION

This case highlights the importance of considering Klippel-Trenaunay Syndrome (KTS) in children presenting with vascular malformations and unexplained severe anemia. Although limb overgrowth and venous anomalies are classical features of KTS, chronic bleeding from cutaneous vascular lesions leading to life-threatening anemia is a rare but significant complication, particularly in pediatric patients. Early recognition, prompt imaging, and a multidisciplinary approach are essential for accurate diagnosis and comprehensive management. Clinicians should maintain a high index of suspicion for atypical presentations of KTS to prevent delayed diagnosis and improve patient outcomes.

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