Case Report

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Surgical management of a thoraco-lumbar extradural cyst in a pediatric patient with Klippel–Trenaunay syndrome: a case report

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Introduction and importance: Klippel–Trenaunay syndrome (KTS) is a congenital disorder characterized by the abnormal development of blood vessels, soft tissues, bones, and the lymphatic system. The syndrome is rare, with few cases reported worldwide, especially those describing an association between KTS and spinal extradural meningeal cysts (SEMC). This report highlights a rare case of a pediatric patient with KTS who underwent successful surgical decompression of a thoraco-lumbar extradural cyst, highlighting the importance of reevaluating surgical interventions in KTS patients.

Case presentation: A 15-year-old girl diagnosed with KTS 4 days postnatally, was referred to our clinic due to chronic back pain and spastic paraparesis. These symptoms were attributed to a compressive extradural thoraco-lumbar cyst. Diagnostic evaluations confirmed the presence of the cyst, leading to the decision to proceed with surgical intervention.

Clinical discussion: The surgical approach involved a laminoplasty at T11–T12–L1, resulting in the total removal of the cystic lesion. The patient exhibited a complete resolution of symptoms postoperatively, with no significant complications reported during the surgery.

Conclusion: With this case, the authors question the fear of surgical intervention in KTS patients, which is often avoided due to concerns of high-risk complications like excessive bleeding or poor wound healing, and hint at a possible association between KTS and extradural meningeal cysts.

Keywords: Klippel–Trenaunay syndrome, mesodermal abnormality, pediatrics, spinal extradural meningeal cyst, thoraco-lumbar

Introduction

Klippel–Trenaunay syndrome (KTS) is a rare condition that occurs at birth and involves abnormal development of blood vessels, soft tissues, bones, and the lymphatic system[1]. This condition is characterized by three key features: a red birthmark (port-wine stain), abnormal overgrowth of soft tissues and bones, and vein malformations with or without lymphatic anomalies[2,3]. Typically, KTS affects only one limb, often a leg, and the severity can vary significantly among individuals[4]. The exact cause of KTS is still unclear, but it's believed to involve gene mutations responsible for vascular development, particularly the (PIK3CA) gene[5].

People with KTS often deal with a wide set of challenges, including pain, bleeding, and varicose veins, and there's no one-size-fits-all treatment. Instead, clinicians focus on managing symptoms and improving the patient's quality of life, which may include surgery for more severe cases like our study subject.

HIGHLIGHTS

- Demonstrates the feasibility and safety of surgical decompression for spinal extradural cysts in pediatric patients with Klippel–Trenaunay syndrome (KTS), challenging existing reservations about surgical risks in such complex conditions.
- Emphasizes recognizing and researching the rare association between KTS and spinal extradural cysts, suggesting a deeper exploration into their shared pathophysiological mechanisms.
- Highlights the need for a thorough diagnostic process and a tailored, multidisciplinary treatment strategy in managing rare and complex disorders like KTS, focusing on improving patient outcomes through individualized care.
- Underlines the success of surgical interventions in resolving symptoms of spinal extradural cysts in KTS patients and stresses the importance of regular follow-up for ensuring sustained recovery and monitoring long-term health.

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Every case of KTS is unique, making its management quite complex. Though uncommon, a variety of spinal pathologies have been documented in patients with KTS. These conditions included spinal arteriovenous malformations (AVMs), spinal lipomas, spinal angiolipomas, spinal angiomas, cavernous angiomas, and vertebral hemangiomas[6–11].

Spinal extradural cysts by itself are exceedingly uncommon in pediatric patients, so it wasn't surprising that to this day and with an in-depth search of various databases, we've only come across two reported cases of spinal extradural cysts in patients with KTS[12,13]. Therefore, our documentation of a thoraco-lumbar extradural cyst in a 15-year-old patient with KTS syndrome which was managed at Tishreen University Hospital is valuable input to medical literature. Through this illustrative study, we aim to emphasize the feasibility and safety of surgical intervention in patients with KTS, contrary to the prevailing non-surgical approach typically favored for treating this condition[14].

This particular case has been documented and reported adhering to the comprehensive standards guidelines by the SCARE 2023 criteria, ensuring thoroughness and consistency in reporting surgical case reports[15].

Case report

Presentation

A 15-year-old female, diagnosed with KTS 4 days after birth, was referred to our neurosurgical clinic. Her medical history was complex, marked by a wide range of pathologies both related to and separate from KTS.

She reported chronic back pain for about 2 years and symptoms indicative of spastic quadriparesis, such as intermittent paresthesias and 'numbness' in her lower extremities. The pain was managed with painkillers and NSAIDs at first, then with opioids (tramadol). It's noteworthy that a prior diagnosis of osteoporosis had misleadingly led her to attribute her chronic back pain to this condition, thereby delaying her pursuit of specialized neurosurgical consultation. Initially, osteoporosis was suspected to be a concurrent pathology with KTS, this suspicion was due to restricted access to diagnostic tools. However, a subsequent DEXA scan provided a proper diagnosis of osteoporosis, clarifying her medical situation.

Despite these symptoms, a neurologic examination primarily identified S1 paresthesias, with no significant evidence supporting spastic quadriparesis. Muscle strength and tone in her lower limbs were normal, as were her reflexes, and she reported no incontinence.

Upon examination, the patient exhibited several phenotypic anomalies, including port-wine stains, limb hypertrophy, pronounced vein abnormalities, syndactyly, and a shortened left lower limb, which contributed to mild scoliosis (Fig. 1).

These physical markers were accompanied by rich history of medical challenges, starting with a notable episode at two months of age where recurrent bleeding led to a diagnosis of colonic polyps through colonoscopy. However, the cessation of bleeding

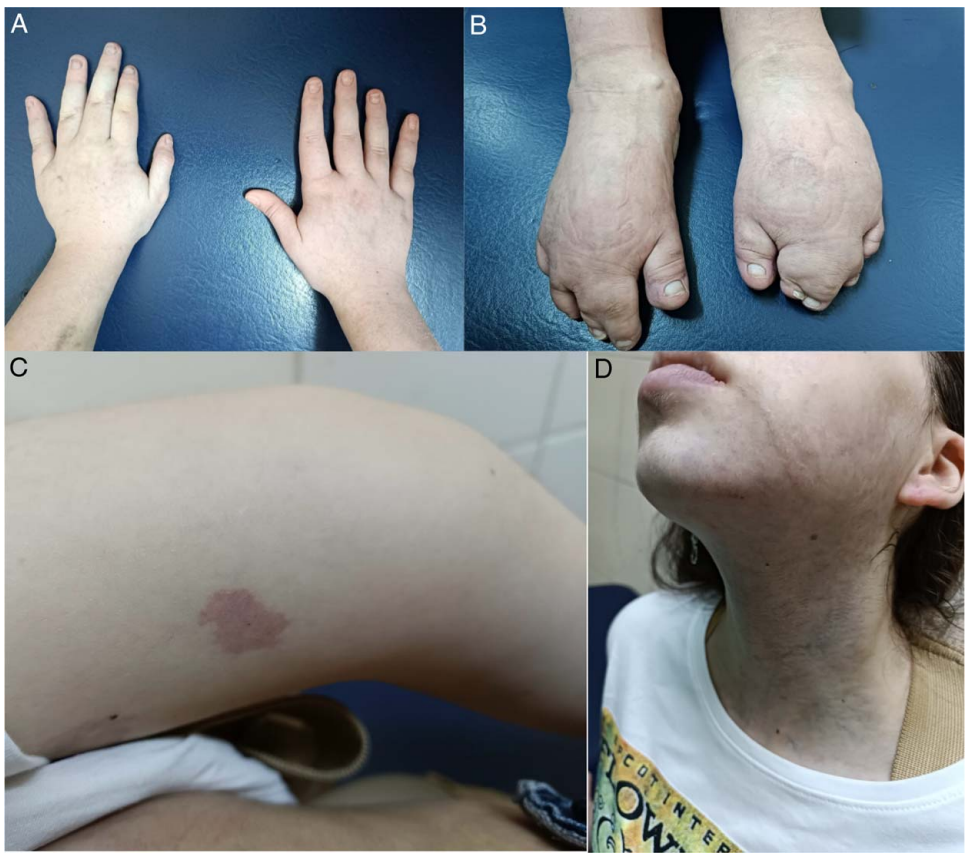

Figure 1. (A, B) Hypertrophy of both upper and lower limbs. (B) Syndactyly in both feet. (C) Port-wine stain. (D) Pronounced venous malformation.

after discontinuing milk intake suggested a potential allergy rather than a direct consequence of the polyps.

At 13 years of age, the patient was diagnosed with severe saphenous insufficiency and was treated pharmacologically with vasoconstrictors. Shortly after that, at 14 years old, abdominal pain, nausea, and vomiting led to the discovery and surgical removal of a bleeding ovarian cyst, adding another layer to her complex medical history.

The patient's family history prominently features a range of cardiovascular conditions. The patient herself had an atrial septal defect (ASD) that spontaneously closed. Her brother was affected by patent ductus arteriosus (PDA), ASD, coarctation of the aorta, and both mitral and aortic stenosis. Additionally, her sister was diagnosed with a rare and distinctive case of Lown–Ganong–Levine syndrome (LGL)

An MRI of the lumbar spine revealed a cystic lesion in the thoraco-lumbar region, pressing on the spinal cord. Its characteristics, consistent with cerebrospinal fluid (CSF), were evident in both T1 and T2 weighted images, spanning from T11 to L1 vertebrae. This lesion spanned from T11 to L1 vertebrae (Fig. 2).

Subsequent imaging of the remainder of the spine did not reveal any additional areas of spinal canal compromise. The cystic content, probably CSF as indicated by MRI imaging characteristics, did not undergo laboratory confirmation to verify its nature.

According to the pathology department, the histopathological analysis revealed that the cystic lesion's wall consisted of fibrous connective tissue, lined internally by a single-cell layer of arachnoid cells, identifying the lesion as an extradural meningeal cyst. No nerve content was found within the cyst; however, it exerted a mass effect, resulting in the neurological symptoms observed in the patient.

Spinal extradural meningeal cysts (SEMC) are uncommon in the general population, and their occurrence in patients with KTS is exceptionally rare. This rarity does not diminish the potential significance of the association but rather underscores the importance of documenting such cases. The presence of SEMC in a KTS patient, as reported in our case, contributes valuable insights into the spectrum of neurological manifestations that can occur in KTS. It suggests that, despite their rare co-occurrence, there may be underlying pathophysiological mechanisms linking KTS to the development of SEMC. Further research into these rare associations can enhance our understanding of both conditions and potentially lead to more targeted approaches to diagnosis and treatment.

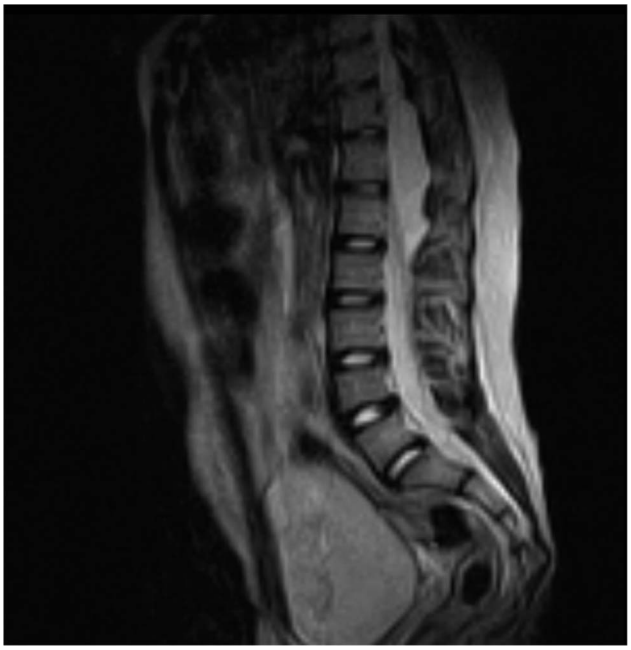

Figure 2. Sagittal MRI of the lumbar spine, revealing a compressive cystic lesion extending from T11 to L1.

Surgical and postoperative details

The literature is currently still primarily in favor of non-operative intervention in KTS patients[16]. However, in this case, the patient's worsening back pain and reduced quality of life necessitated intervening surgically. Moreover, the previous uncomplicated surgery, (removal of an ovarian cyst), encouraged the decision for spinal surgery.

The primary goal of the surgical intervention was to perform thoraco-lumbar decompression, specifically targeting the alleviation of radicular symptoms, primarily presented as S1 paresthesias.

Although S1 paresthesias were the sole radicular symptom explicitly reported, the decision for surgical decompression was based on a comprehensive evaluation of the patient's symptomatology, including chronic back pain and functional impairment, which suggested a broader impact of the spinal extradural cyst on her neurological function.

Laminoplasty on T11–T12–L1 vertebrae -which is not typically performed on the lumbar spine- was selected and operated by the head surgeon, and the rationale for that was to provide wide meningeal exposure and the ability to avoid fixation tools, benefiting from the patient's younger age for bone fusion and therefore preserving spinal stability. Echography was utilized during surgery to define the cystic lesion borders accurately. Once the laminae were drilled and lifted, the cyst appeared clearly (Fig. 3), and got precisely and carefully excised from the meninges. After cyst removal, the spinous processes were secured with Proline 0.2.

The surgery necessitated the transfusion of two blood bags (900 ml) to manage the expected blood loss in such patients. Although blood transfusion in this context might not typically pose a significant risk, the specific circumstances of this case—namely, the minimally invasive nature of the procedure and the patient's young age and small physical stature—make the 900 ml transfusion somewhat concerning. To mitigate these risks, extensive pre-surgical planning was critical, encompassing the preparation of adequate blood supplies and employing intraoperative blood conservation strategies, including echography for precise surgical execution. The operation lasted 3.5 h, from anesthesia administration to wound closure. A Redovac drain was placed, and the wound closure adhered to standard protocols. Postoperatively, a full back brace was recommended for two months to ensure proper fixation and bone fusion.

On the initial day postoperatively, the patient ambulated independently and was subsequently discharged on the fourth postoperative day. An MRI of the lumbar spine conducted 1-month post-surgery, confirmed the total and complete removal of the cyst, verifying the success of the surgical intervention (Fig. 4).

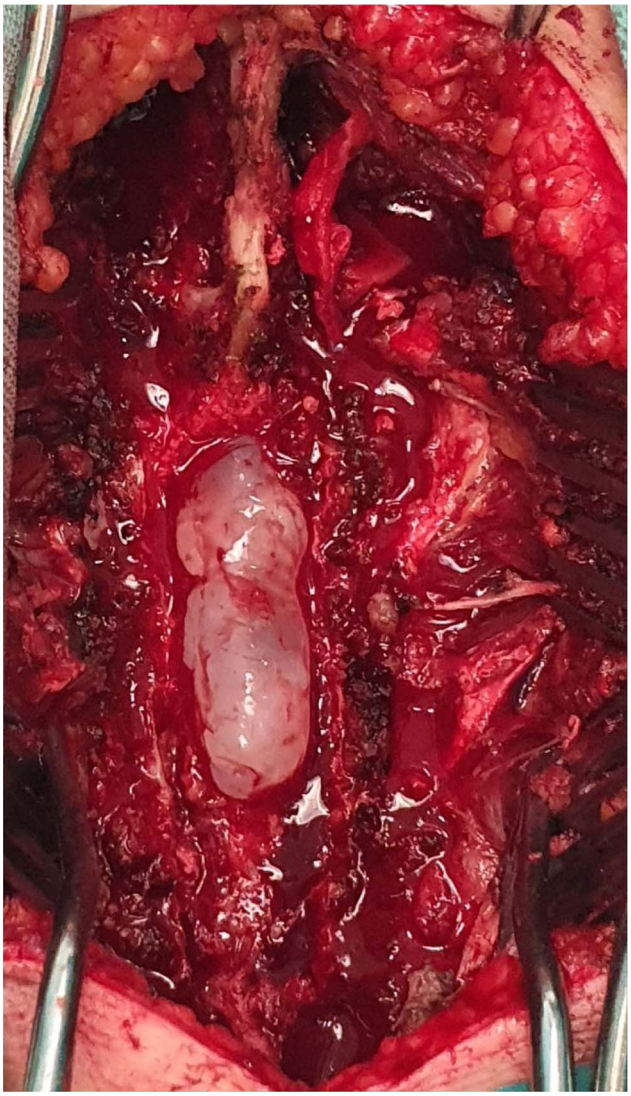

Figure 3. Intraoperative image of the cystic lesion clearly defined.

Scheduled follow-up appointments at 1, 3, and 6 months post-surgery were conducted as part of the postoperative care plan. The patient consistently reported a complete resolution of the preoperative symptoms during these appointments. Furthermore, neurological evaluations performed at each visit revealed a return to normal neurological function. The successful outcome not only underscores the efficacy of surgical intervention in this context but also offers hope for similar cases, challenging the conventional hesitancy towards operative management in KTS patients.

Discussion

SEMC are one of the rare pathologies to cause radicular pain, accounting for 1–3% of all primary spinal space-occupying lesions[17,18]. Our case falls into the type I category of SEMC, as defined by Nabor *et al.*[19], who classified SEMCs into three major categories: extradural meningeal cysts without nerve root fibers, extradural meningeal cysts with nerve root fibers, and intradural meningeal cysts.

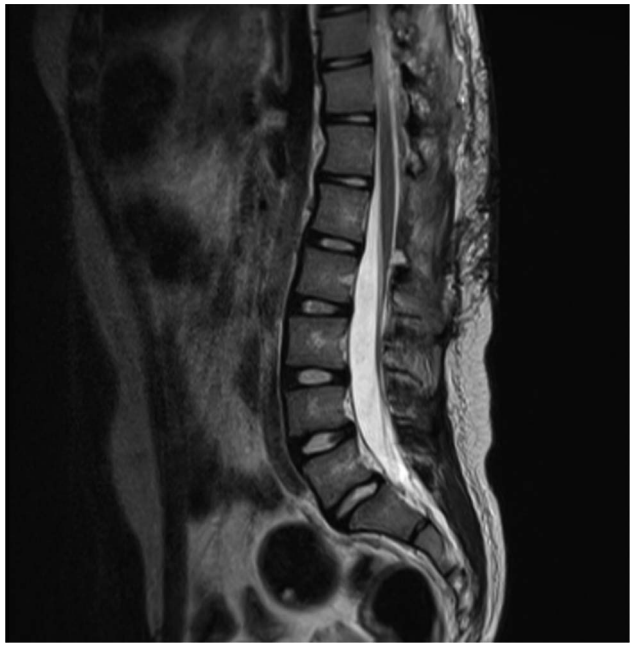

Figure 4. Postoperative sagittal MRI of the lumbar spine demonstrates complete extradural cyst excision.

The etiologies of SEMCs are diverse, including congenital, inflammatory, and trauma-related factors[20]. Particularly, Type I SEMCs are often linked to congenital dural defects or arachnoid herniations. The rarity of SEMCs, especially among patients with KTS, presents a unique intersection of uncommon pathologies.

Given KTS's association with mesodermal development issues, which can lead to vascular and skeletal issues, SEMC may represent another side of these mesodermal irregularities. This is further supported by KTS's link to various central nervous system malformations. Moreover, the vascular malformations characteristic of KTS, including venous and lymphatic abnormalities, could contribute to abnormal fluid dynamics and pressures in spinal epidural spaces, potentially leading to the formation of SEMCs[2]. A 2021 study involving 116 patients with confirmed KTS who underwent neuroimaging found a 16.4% prevalence of neurovascular malformations, with paraspinal and epidural anomalies being the most common[21].

One of the newer hypotheses involves the concept of fibroadipose vascular anomaly (FAVA), which is thought to be based on a similar pathophysiological basis as KTS[22]. FAVA consists of fibrofatty infiltration of muscles along with abnormal vessels. This infiltration could structurally weaken spinal structures or create inflammatory responses that promote cyst formation[23].

The presence of somatic mutations in the (PIK3CA) gene in patients with FAVA, is supported as part of a broader spectrum of related disorders including KTS. Studies have shown that these PIK3CA mutations are prevalent in various syndromes characterized by overgrowth and vascular malformations, suggesting a genetic underpinning that could contribute to FAVA's structural and vascular abnormalities and potentially influence the development of spinal cysts in KTS[22].

Most tissues affected by these conditions show a low percentage of mutant cells, indicating a need for sensitive detection methods to uncover the underlying genetic causes and understand

their impact on disease pathology. Unfortunately, both studies do not confirm a definitive association between KTS and spinal cysts but support the narrative of a potential connection between the two etiologies.

The diagnosis and treatment of SEMC in individuals with KTS are complicated by the syndrome's variable expression. MR) is essential for identifying SEMC, aiding in understanding cyst characteristics, and informing surgical decisions. Our case demonstrates the effective surgical treatment of SEMC in a KTS patient, challenging the usual preference for conservative management. The surgery decision was based on the patient's symptoms and MRI findings, consistent with evidence suggesting favorable outcomes for surgically treated SEMC, depending on factors like age and neurological involvement. We emphasize the necessity of a comprehensive examination of the etiology, progression, and management of SEMC within the context of KTS. We call for additional research to delve into the potential pathological links—if any exist—between mesodermal anomalies and spinal conditions. Our aim is to advance the precision of diagnoses and the effectiveness of treatment approaches for individuals affected by these disorders.

Conclusion

In this report, we have presented a case of successful surgical intervention for an extradural cyst in a pediatric patient diagnosed with KTS. This case highlights the potential safety and efficacy of surgical approaches in treating complex spinal conditions associated with KTS. Notably, the successful resolution of symptoms following the surgical removal of the cyst underscores the possibility that such cysts, though rare, may be an underrecognized manifestation in patients with KTS. The association between extradural spinal cysts and KTS, as suggested by this case, invites further research to explore the underlying mechanisms that may link these conditions. Establishing a clearer connection could lead to more targeted diagnostic protocols and therapeutic strategies, enhancing outcomes for KTS patients with similar presentations.

Ethical approval

There's no active ethical committee, a proper signed consent from the patient and her caregiver were obtained.

Consent

Written informed consent was obtained from the patient's parents/legal guardian for publication and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

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Author contribution

B.Z.: writing - original draft, writing—review and edit, investigation, resources, visualization. H.I.: review original draft, data collection, resources. M.J.: review original draft, data collection, resources. G.J.: resources. M.A.: supervision, project administration. G.A.: supervision.

Conflicts of interest disclosure

The authors report no conflict of interest concerning the materials or methods used in this study or the findings specified in this paper.

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Not applicable.

Guarantor

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Data availability statement

Not applicable.

Provenance and peer review

Not applicable.

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