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Case Report

Classical Ehlers-Danlos Syndrome in a Pediatric Patient: A Case Report and Review of Surgical Implications

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Abstract

Ehlers-Danlos Syndrome (EDS) is a group of hereditary connective tissue disorders characterized by joint hypermobility, skin hyperextensibility, and tissue fragility. The classical type of EDS (cEDS) is caused by mutations in the COL5A1 or COL5A2 genes, which encode type V collagen. This case report highlights the importance of early recognition and specialized surgical management in pediatric patients with cEDS. We present the case of a 5-year-old child with a known diagnosis of cEDS who presented with a lumbar spine wound following minor trauma. The wound was managed in accordance with established guidelines for cEDS, including meticulous suturing techniques and postoperative care. This case underscores the value of a proactive, multidisciplinary approach in optimizing outcomes for patients with cEDS.

Keywords: Ehlers-Danlos Syndrome; Classical type; Pediatric surgery; Connective tissue disorder; COL5A1; COL5A2; Case report

Introduction

Ehlers-Danlos Syndrome (EDS) encompasses a spectrum of inherited connective tissue disorders, with the classical type (cEDS) being one of the most common forms. cEDS is primarily caused by mutations in the COL5A1 or COL5A2 genes, leading to defects in type V collagen synthesis. This results in hallmark features such as skin hyperextensibility, atrophic scarring, and joint hypermobility [1]. Pediatric plastic surgeons may encounter patients with cEDS in various clinical scenarios, including wound management, hernia repairs, and complications related to tissue fragility. Early recognition of cEDS and adherence to specialized surgical protocols are critical to minimizing complications and improving outcomes. This case report illustrates the successful management of a pediatric patient with cEDS, emphasizing the importance of tailored surgical techniques and a multidisciplinary approach.

Case Presentation

A 5-year-old child with a known diagnosis of classical Ehlers-Danlos Syndrome (cEDS), previously confirmed through genetic testing at a European center, presented to the emergency department with a lumbar spine wound following minor trauma **(Figure 1)**. The child's medical history of cEDS prompted immediate referral to the pediatric plastic surgery department for specialized wound management.

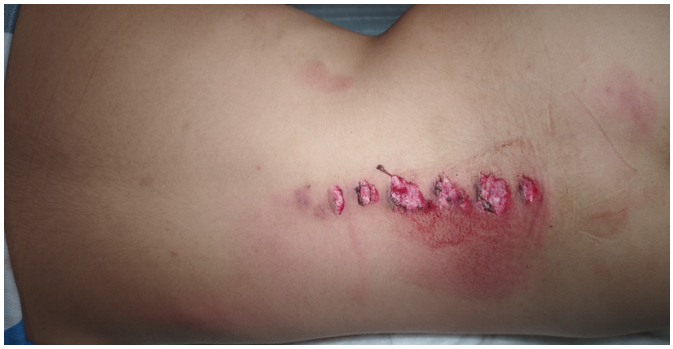

Figure 1: Image showcasing the lumbar wound.

On examination, the child exhibited characteristic features of cEDS, including skin hyperextensibility and joint hypermobility. The lumbar wound was irregular, with thin, atrophic edges indicative of tissue fragility **(Figure 2)**. Given the patient's known diagnosis, no additional diagnostic tests were required, and the focus shifted to optimizing wound closure and healing. The wound was meticulously repaired using a two-layered suturing technique with non-absorbable sutures (Prolene) to minimize tension and reduce the risk of dehiscence. Special attention was paid to ensuring deep, layered closure to provide adequate support to the fragile tissue. Postoperative care included prolonged wound support with adhesive strips and close monitoring for signs of complications **(Figure 3)**.

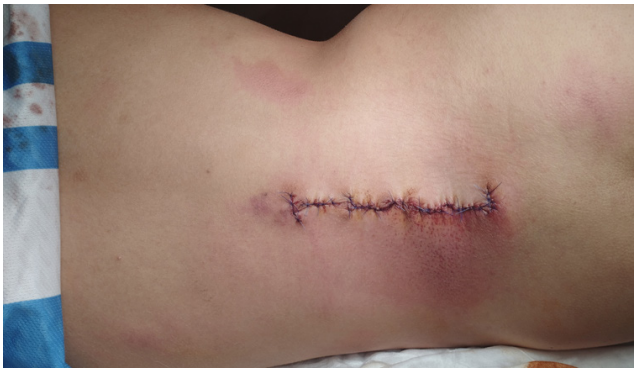

Figure 2: The wound after proper suturing

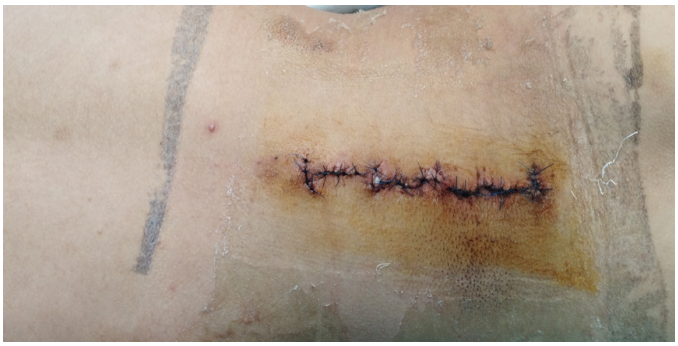

Figure 3: Follow-up of the wound care.

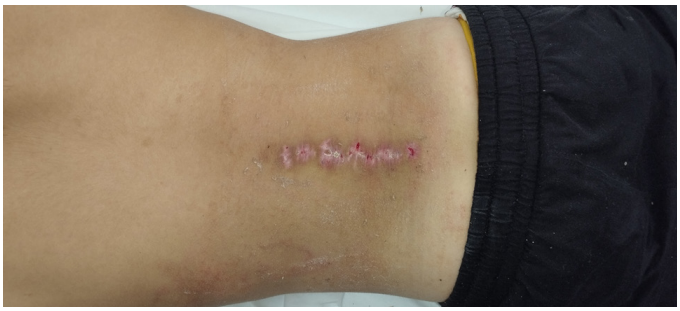

Figure 4: The. wound healed.

The patient's recovery was uneventful, with the wound healing well over the following weeks. Regular follow-up visits confirmed good wound evolution **(Figure 4)**, and the patient was referred to a genetic counseling service for ongoing management and family screening.

Discussion

Clinical Features of Classical EDS

cEDS is characterized by skin hyperextensibility, atrophic scarring, joint hypermobility, and tissue fragility [2-5]. These features necessitate a high index of suspicion and specialized management strategies, particularly in pediatric patients who may present with wounds or other surgical complications.

Genetic Basis and Diagnosis

cEDS is primarily caused by mutations in the COL5A1 or COL5A2 genes, which encode type V collagen [6]. While genetic testing confirms the diagnosis, clinical recognition of cEDS is essential for timely and appropriate management. In this case, the patient's prior diagnosis allowed for immediate implementation of tailored surgical techniques, highlighting the importance of early genetic and clinical evaluation.

Surgical Considerations for Pediatric Surgeons

Pediatric surgeons must be aware of the unique challenges posed by cEDS:

Wound management: Delayed wound healing and abnormal scarring necessitate meticulous surgical techniques, including deep, layered sutures with non-absorbable materials and prolonged wound support [9].

Preventive approach: Early recognition of cEDS allows for proactive management, reducing the risk of complications such as wound dehiscence and recurrent hernias [10].

Multidisciplinary care: Collaboration with geneticists, physical therapists, and other specialists is crucial for comprehensive management and improved patient outcomes [13].

Multidisciplinary Approach

The management of cEDS requires a coordinated effort among various healthcare providers. Early intervention, patient education, and family screening are essential components of care. This case demonstrates the value of a proactive, multidisciplinary approach in achieving favorable outcomes.

Limitations

This case report is limited by its focus on a single patient. However, the successful management of this case underscores the importance of adhering to established guidelines for cEDS and highlights the need for further research to refine surgical techniques and improve outcomes.

Conclusion

This case highlights the importance of early recognition and specialized management of classical Ehlers-Danlos Syndrome in pediatric surgical practice. The child's known diagnosis of cEDS allowed for immediate implementation of tailored surgical techniques, resulting in successful wound healing and an uneventful recovery. A proactive, multidisciplinary approach is essential for optimizing outcomes in patients with cEDS. Further research is needed to enhance surgical protocols and improve long-term outcomes for these patients.

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Ethical Approval: No ethical approval is required for de-identified case reports and case series based on our institutional policies.

Consent: Written informed consent was obtained from the patient's legally authorized representatives.

References

1. Malfait F, Francomano C, Byers P, et al. The 2017 international classification of the Ehlers-Danlos syndromes. Am J Med Genet C Semin Med Genet. 2017;175(1):8-26. doi:10.1002/ajmg.c.31552
2. Beighton P, De Paepe A, Steinmann B, Tsipouras P, Wenstrup RJ. Ehlers-Danlos syndromes: revised nosology, Villefranche, 1997. Am J Med Genet. 1998;77(1):31-37. doi:10.1002/(sici)1096-8628(19980428)77:1<31::aid-ajmg8>3.0.co;2-o
3. Castori M, Tinkle B, Levy H, Grahame R, Malfait F, Hakim A. A framework for the classification of joint hypermobility and related conditions. Am J Med Genet C Semin Med Genet. 2017;175(1):148-157. doi:10.1002/ajmg.c.31539
4. Pepin MG, Schwarze U, Rice KM, Liu M, Leistritz D, Byers PH. Survival is affected by mutation type and molecular mechanism in vascular Ehlers-Danlos syndrome (EDS type IV). Genet Med. 2014;16(12):881-888. doi:10.1038/gim.2014.72

5. Symoens S, Syx D, Malfait F, et al. Comprehensive molecular analysis demonstrates type V collagen mutations in over 90% of patients with classic EDS and allows to refine diagnostic criteria. Hum Mutat. 2012;33(10):1485-1493. doi:10.1002/humu.22137
6. Colombi M, Dordoni C, Chiarelli N, Ritelli M. Differential diagnosis and diagnostic flow chart of joint hypermobility syndrome/ehlers-danlos syndrome hypermobility type compared to other heritable connective tissue disorders. Am J Med Genet C Semin Med Genet. 2015;169C(1):6-22. doi:10.1002/ajmg.c.31429
7. Byers PH, Belmont J, Black J, et al. Diagnosis, natural history, and management in vascular Ehlers-Danlos syndrome. Am J Med Genet C Semin Med Genet. 2017;175(1):40-47. doi:10.1002/ajmg.c.31553
8. De Paepe A, Malfait F. The Ehlers-Danlos syndrome, a disorder with many faces. Clin Genet. 2012;82(1):1-11. doi:10.1111/j.1399-0004.2012.01858.x
9. Brooke BS, Arnaoutakis G, McDonnell NB, Black JH. Contemporary management of vascular complications associated with Ehlers-Danlos syndrome. J Vasc Surg. 2010;51(1):131-139. doi:10.1016/j.jvs.2009.08.019
10. Lum YW, Brooke BS, Black JH. Vascular Ehlers-Danlos syndrome: pathophysiology, diagnosis, and prevention of vascular complications. Cardiol Rev. 2012;20(1):4-7. doi:10.1097/CRD.0b013e3182342316
11. Wiesmann T, Castori M, Malfait F, Wulf H. Recommendations for anesthesia and perioperative management in patients with Ehlers-Danlos syndrome(s). Orphanet J Rare Dis. 2014;9:109. doi:10.1186/s13023-014-0109-5
12. Tinkle B, Castori M, Berglund B, et al. Hypermobile Ehlers-Danlos syndrome (a.k.a. Ehlers-Danlos syndrome type III and Ehlers-Danlos syndrome hypermobility type): clinical description and natural history. Am J Med Genet C Semin Med Genet. 2017;175(1):48-69. doi:10.1002/ajmg.c.31538
13. Hakim A, Grahame R. Joint hypermobility. Best Pract Res Clin Rheumatol. 2003;17(6):989-1004. doi:10.1016/j.berh.2003.08.001
14. Rombaut L, Malfait F, De Wandele I, et al. Medication, surgery, and physiotherapy among patients with the hypermobility type of Ehlers-Danlos syndrome. Arch Phys Med Rehabil. 2011;92(7):1106-1112. doi:10.1016/j.apmr.2011.01.016
15. Murray ML, Pepin M, Peterson S, Byers PH. Pregnancy-related deaths and complications in women with vascular Ehlers-Danlos syndrome. Genet Med. 2014;16(12):874-880. doi:10.1038/gim.2014.53