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Case Report

A case study of a 10-year-old child with Ehlers Danlos syndrome alongside hematoma and cellulitis at the left lower limb

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ABSTRACT

A 10-year-old child was admitted with a soft, non-tender, fluctuant swelling over the left lower limb following minimal trauma at school, complicated by overlying cellulitis. The patient had a history of joint hypermobility for one year and easy bruising for six years after trivial injuries. Examination revealed numerous atrophic scars, skin hyperextensibility of 1.5 cm, and a Brighton hypermobility score of 9/9, confirming generalized joint hypermobility. Classical features included elbow and knee hyperextension >10°, fifth metacarpophalangeal joint extension >90°, thumb-to-forearm opposition, and positive Gorlin's sign. Growth parameters were normal, with an arm span-to-height ratio of 0.95 and upper-to-lower segment ratio of 0.85. A diagnosis of Ehlers-Danlos syndrome (EDS) with secondary hematoma and cellulitis was made. Laboratory tests, including blood counts and coagulation profiles, excluded bleeding disorders, while chest X-ray and echocardiography were performed to assess systemic involvement. Management was multidisciplinary: physiotherapy for joint stabilization, hydrotherapy for muscle strengthening, vitamin C supplementation to enhance collagen synthesis, and psychological and genetic counseling. Due to risks of aortic root dilatation and mitral valve disease, regular cardiac monitoring was initiated. The patient improved with conservative therapy, cellulitis resolved, and functional status improved. Long-term follow-up included annual echocardiography, orthopedic review, and genetic counseling. This case highlights the importance of recognizing connective tissue disorders like EDS in children presenting with unexplained bruising, poor wound healing, and skin changes, as early diagnosis and comprehensive management are essential to improve outcomes and prevent complications.

Keywords: Ehlers-Danlos syndrome, Connective tissue disorder, Joint hypermobility, Hematoma, Cellulitis, Brighton score, Pediatric genetics, Collagen disorders, Tissue fragility, Wound healing

INTRODUCTION

Ehlers-Danlos syndrome (EDS) is a complex group of heritable disorders of connective tissue with joint hypermobility, skin hyperextensibility, and fragility of tissues, with an estimated prevalence of 1:5,000-1:20,000 worldwide.[1,2] The 2017 international classification identifies 13 subtypes, of which the most common is hypermobile EDS (hEDS).[3] Pathologically, EDS is due to mutations in collagen-related genes (collagen types I, III, V, VI), disturbing extracellular matrix integrity and leading to mechanical tissue weakness.[4] Clinical presentations include joint laxity, bruising easily, impaired wound healing, and thin "cigarette paper" scars.[5] Pediatric presentations may feature developmental delay, frequent injuries, and persistent pain, making early diagnosis challenging.[6] Systemic complications range from cardiovascular (aortic dilatation, mitral valve prolapse) to gastrointestinal (dysmotility, perforation) and musculoskeletal (dislocations, osteoarthritis) systems, requiring careful observation.[7] Diagnosis is clinical (e.g., Brighton score) and genetic analysis, where indicated.[8]

Treatment is multidisciplinary, focused on symptom relief, injury prevention, and surveillance for complications, as there is no cure. Increased clinician awareness is necessary to avoid delays in diagnosis and maximize outcomes.

CASE REPORT

This is a case of Farhan Islam, a 10-year-old Bangladeshi boy who was admitted to a tertiary care hospital with a history of progressive swelling in the left lower limb for 10 days following a trivial trauma during a football game. The swelling was initially a painless small lump but gradually grew to 5×3 cm with overlying reddish-purple skin discoloration and developed into its cellulitis of 7×5 cm, which was warm, tender, and erythematous. The mother provided additional history with features of an underlying connective tissue disorder of long duration, such as dramatic joint hypermobility during the past year in which he could bend fingers backward, hyperextend elbows and knees beyond the normal range, and perform odd contortions without pain initially, although he recently started to develop joint pains after physical activity. More importantly, from age 6, he had had easy bruising out of proportion to trivial trauma, with extensive, dark bruising occurring after minimal knocks and taking an unusually long time to settle, which in the past had prompted concerns of potential abuse that had been investigated and excluded. His previous medical history was of delayed wound healing after a cycling accident two years earlier that had resulted in a typical broad, thin, atrophic "cigarette paper" scar on his forearm. Birth history was normal vaginal delivery at 38 weeks with normal birth weight, but developmental milestones showed subtle delays like independent walking only by 24 months of age. Motor development continued to exhibit poor coordination and easy fatigability with physical activities despite normal cognitive development and academic progress. There was no contributory symptomatology in the parents or siblings on a non-consanguineous parental background, although the parental non-consanguinity did not rule out de novo mutations. On physical examination, Farhan was well-nourished and cooperative with stable vital signs: pulse 75 bpm, respiratory rate 20/min, blood pressure 80/60 mmHg, and temperature 37.2°C. Anthropometry was within normal percentiles (weight 35 kg at 75th percentile, height 139 cm at 80th percentile, BMI-18.1 kg/m^2) with a normal upper/lower segment and arm span/height ratio. Cutaneous and musculoskeletal findings were most notable. The left leg had a 5×3 cm fluctuant hematoma with surrounding 7×5 cm cellulitis with erythema, heat, and tenderness. There were multiple atrophic "cigarette paper" scars on limbs, chin and scalp, and 1.5 cm skin hyperextensibility on forearm stretching and earlobe hypermobility. Musculoskeletal examination on the Brighton hypermobility score revealed a maximum 9/9 score with bilateral elbow and knee hyperextension of greater than 10°, passive thumb opposition to the forearm, fifth finger dorsiflexion of greater than 90°, and was ability to place palms flat on the ground with the knees straight. He had a positive Gorlin's sign (tongue-to-nose contact) and temporomandibular joint hypermobility. The rest of the systemic findings, including cardiovascular, respiratory, abdominal, and neurological, were unremarkable without organomegaly, murmurs, or focal deficits. The clinical presentation-with widespread joint hypermobility, skin hyperextensibility, easy bruising, delayed healing of wounds, and characteristic atrophic scarring-was very suggestive of hypermobile EDS (hEDS), the most common EDS subtype.[9] The absence of family history considered the possibility of a de novo mutation. Investigations launched were full blood count and coagulation profile to exclude bleeding disorders, ultrasound to assess hematoma and exclude abscess, and echocardiography to exclude cardiovascular features like aortic root dilation or mitral valve prolapse. While genetic testing for mutations in COL3A1, COL5A1, or COL5A2 would be confirmatory, resource limitations affected availability. Acute management included cellulitis treatment with intravenous antibiotics (e.g., clindamycin) alongside hematoma observation, analgesia, and compression.[10] Long-term management plans emphasized physical therapy for joint stabilization, activity modification with restriction of high-impact sports, protective skin care, vitamin C supplementation to enhance collagen synthesis, and annual echocardiograms for cardiovascular surveillance. Psychosocial management included family and school education about the condition to prevent misinterpretation of symptoms and genetic counseling for autosomal dominant inheritance patterns.[11] This case highlights the classical presentation of hEDS and emphasizes how increased awareness of subtle clinical findings like joint hypermobility, abnormal scarring, and easy bruising in a child is important. The delayed milestones and exercise intolerance also demonstrate the systemic nature of the condition. While acute complications require urgent intervention, management in the long term is focused on the prevention of ongoing injuries and monitoring for systemic involvement with multidisciplinary care. The case emphasizes the need to suspect EDS clinically to facilitate early diagnosis, appropriate counseling, and personalized interventions that can significantly improve quality of life in those affected. It also illustrates diagnostic challenges in resource-poor settings where genetic confirmation may not be possible, and how one has to rely on clinical criteria. The psychosocial aspects, including previous suspicions of abuse due to unexplained bruising, emphasize patient and family education as a critical component of management. Furthermore, the case demonstrates how a thorough history and physical can reveal a unifying diagnosis for seemingly unrelated symptoms that had been present for years before presentation. The management plan demonstrates the need for individualized care plans for both the acute and chronic manifestations while considering the patient's stage of development and lifestyle demands. This big-picture thinking is also important to simplify outcomes for children with connective tissue disorders like EDS.

Figure 1: Passive apposition of the thumb to the forearm. Sign of joint hypermobility.

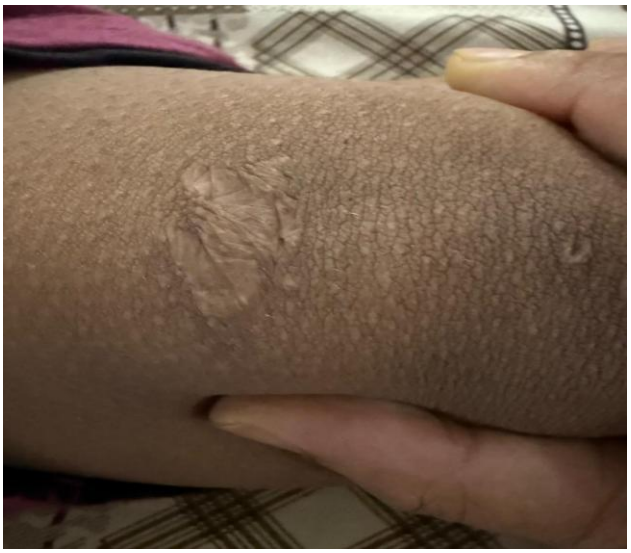

Figure 2: Change in the texture of the skin. Skin is looking like it may be velvety or doughy.

Figure 3: Passive dorsiflexion of the little finger. Sign of joint hypermobility.

Figure 4: Placing of palms on the floor with straight legs.

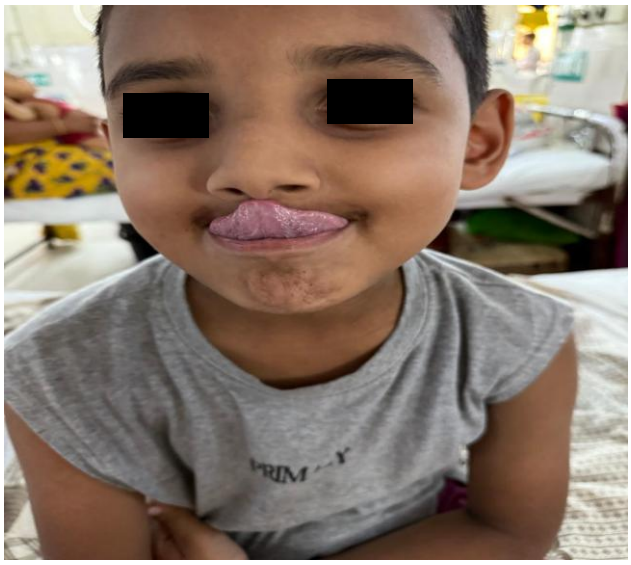

Figure 5: The ability to touch the tip of the nose (Gorlin sign).

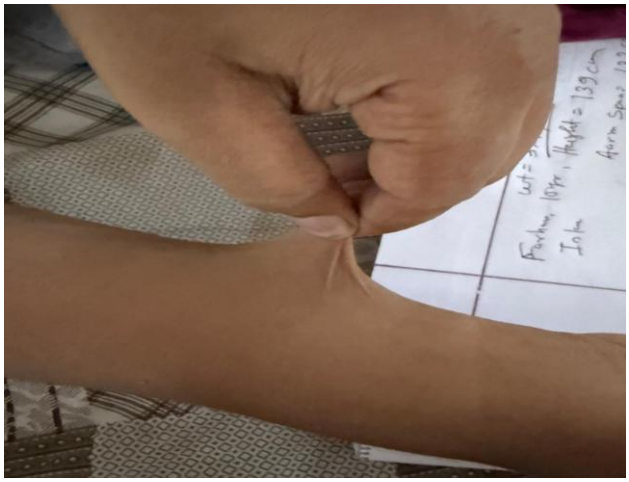

Figure 6: Hyperextensibility of skin. Gently stretching of skin at the forearm and it stretches abnormally far and snaps back quickly.

DISCUSSION

This case study demonstrates a common presentation of hEDS with the classic triad of joint hypermobility, skin hyperextensibility, and tissue fragility. Generalized joint hypermobility is established by the patient's maximum Brighton hypermobility score of 9/9, now within the range of hEDS. Severe hematoma following minor trauma is an excellent demonstration of the vascular frailty of EDS resulting from the deranged structure of collagen, thus weakening the walls of the blood vessels and of supporting connective tissues.[12] Such a pathophysiologic background also explains secondary cellulitis since compromised tissue integrity and compromised wound healing create conditions favorable to bacterial infection by imperfect skin barriers. The bilateral atrophic "cigarette paper" scars on the patient's skin are pathognomonic for EDS and are caused by impaired collagen production, resulting in mechanically inferior scar tissue. The skin manifestations and the patient's history of delay in motor milestones (particularly walking at age 24 months) and ongoing coordination issues are characteristic of the systemic nature of connective tissue impairment in EDS, affecting both musculoskeletal and proprioceptive functions. Genetically, the absence of family history does not exclude EDS, as the majority are a result of new mutations or variable expressivity, while the non-consanguineous parents lower the chance of autosomal recessive forms, genetic counselling remains relevant for awareness of patterns of inheritance (more commonly autosomal dominant in hEDS) and recurrence risk.[13] The diagnostic process utilized the Brighton criteria correctly, though the 2017 EDS classification system has more specific definitions of subtypes that can guide future genetic testing if available.[14] Differential diagnosis with Marfan syndrome was effectively ruled out by upright anthropometric measurements and the presence of classic skin features of EDS.[15] Care for this patient went as per established best practices, emphasizing multi-disciplinary care. Physiotherapy forms the backbone of treatment, and joint stabilization by periarticular strengthening of muscles and proprioception to utilize natural ligamentous laxity. Hydrotherapy benefits particularly in allowing exercise in a low-impact environment. Vitamin C supplementation was appropriately added as adjuvant therapy to potentially enhance collagen synthesis, but with a modest expected benefit.[16] Multidisciplinary care planning also heavily involves psychosocial issues, as EDS children usually experience chronic pain, limitation of activities, and socialization problems that require individual psychological management. Genetic counseling serves a number of purposes-educating families about inheritance, assisting with family planning, and identifying other potentially affected relatives who can present subtly. The annual routine echocardiogram is a necessary preventive therapy because cardiovascular complications develop insidiously in the EDS patient. While this patient is currently without cardiac malformations, observation for dilation of the aortic root, mitral valve prolapses, and other late complications will remain unavoidable. Treatment of acute cellulitis requires standard antibiotic therapy with vigilance heightened secondary to EDS patients' impaired healing of tissues.[17] Long-term care implications also involve watchfulness for orthopedic complications (joint degeneration, premature arthritis), gastrointestinal manifestations (gastroparesis, constipation), and potential ophthalmologic complications (lens subluxation, retinal detachment) and reflect the multisystemic nature of EDS. Prognostically, prompt recognition and management, as in this case, optimize the outcomes for hEDS patients. While life expectancy is usually normal, quality of life may be appreciably compromised by chronic pain and restriction of function, requiring long-term intervention. The educational factor is also important, educating the patients, families, and schools regarding appropriate activity adaptations, injury prevention, and signs of complications. The present case well demonstrates the diagnostic challenge in resource-limited settings with no genetic testing, and one is forced to rely on clinical criteria. It also underscores how meticulous history and examination can uncover a unifying diagnosis for ostensibly heterogeneous complaints, possibly years in the making. The psychosocial component, emphasized by prior suspicion of abuse due to unexplained bruising, highlights the need for rigorous patient and family education on manifestations of the disease.[18] The management approach shows the importance of individualized care plans addressing both acute presentations and chronic expressions based on developmental stage and lifestyle requirements. Here in a pediatric patient, this involves balancing protection with age-appropriate activities to permit normal psychosocial development. The case illustrates that EDS management must evolve as patients age; childhood is all about providing developmental support and prevention of trauma, adolescence is about managing increasing joint laxity and pain, and adulthood is about preventing degenerative complications. This longitudinal perspective is required to optimize outcomes across the lifespan. Several key take-home messages emerge in this case. First, it reminds us to consider connective tissue disorders in the evaluation of children with easy bruising, joint hypermobility, or unusual scarring. Second, it demonstrates that a careful clinical evaluation using standardized tools like the Brighton score can formulate diagnoses independent of genetic testing. Third, it underscores the coordination of multidisciplinary care to manage the many systemic features of EDS. Finally, it reinforces the role of early diagnosis in achieving preventive interventions that can significantly improve long-term prognosis. This multifaceted model of EDS care-involving physical therapies, medical monitoring, psychosocial management, and patient education-offers a paradigm for the care of complex connective tissue disorders in children. The case is particularly effective at illustrating how clinical observation is able to detect underlying syndromes behind a variety of seemingly unrelated symptoms, and in the process render more targeted and efficient interventions. By recognizing the triad of EDS presentation and implementing appropriate multidisciplinary treatment,

clinicians are able to significantly improve the quality of life for involved children and families. The discussion highlights that the best care for EDS involves treating both the immediate clinical issues, such as the presenting hematoma and cellulitis, and, at the same time, instituting long-term monitoring and care systems to effectively deal with the chronic condition throughout the patient's life.

CONCLUSION

This patient demonstrates the challenge in diagnosing and managing EDS in children, demonstrating how subtle signs of hyperextensible joints, easy bruising, and unusual scarring can be symptomatic of this connective tissue disorder. The acute onset of a massive hematoma and cellulitis after minor trauma revealed the clinical impact of damaged connective tissues, which required immediate treatment as well as ongoing multidisciplinary management. The patient presents an example of the importance of early diagnosis by clinical means such as the Brighton Score, followed by thorough assessment to enable timely interventions. Proper treatment involves the integration of physiotherapy to achieve joint stability, management strategies for wounds, cardiovascular assessments, and psychosocial support to counteract the systemic impacts of the condition. Family education and genetic counseling are important factors in informing families about inheritance and everyday management requirements. Even while the patient's acute complications were successfully treated, EDS requires ongoing follow-up to monitor for such complications as joint deterioration or cardiac issues. The case points out the importance of clinician recognition of early signs of EDS to institute aggressive treatment that significantly improves patients' quality of life. Subsequent research needs to be directed towards the development of more specific therapies and standardized guidelines to best treat people stricken by this multi-system, complex condition.

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