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Case Series

Prevalence of Female Athletes with Anterior Cruciate Ligament Tear Presenting with Ehlers-Danlos Syndrome: A Case Series

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ABSTRACT

Ehlers–Danlos syndrome (EDS) is a connective tissue disorder characterized by joint hypermobility, which may predispose individuals to musculoskeletal injuries such as Anterior Cruciate Ligament (ACL) tears. This case series examines the prevalence of EDS in female athletes aged 14-25 who have sustained ACL tears. Our findings suggest a higher occurrence of EDS among this population than previously reported in the general population, highlighting the need for early screening and preventive measures for all athletes, with emphasis on females due to their higher risk.

Keywords: Ehlers Danlos Syndrome (EDS); Anterior Cruciate Ligament (ACL); Injury; Diagnosis; Physical therapy

INTRODUCTION

Ehlers Danlos Syndrome (EDS) is a group of disorders which affect connective tissues found supporting the skin, blood vessels, visceral organs and joints. A mutation in collagen, the protein that integrates both flexibility and strength in connective tissues, has been found to cause lax, hypermobile joints prone to dislocation and subluxation [1].

The ACL is a common musculoskeletal injury which occurs in the knee. This injury has been observed at much higher levels in females than in males due to multiple contributing factors such as estrogen levels, a sharper intertrochlear groove and a higher Q angle [2]. In congruence with the pathophysiology of EDS with augmented flexibility and strength of joints, it could be hypothesized that abnormal stability of the knee as found in some EDS patients could contribute to ACL tears at a higher rate than in non-EDS populations, especially in females. The prevalence rate is estimated to be 1/5000 in screening of patients with EDS, however, the hypermobility of the knee joint found in young females with EDS could propagate the tearing of the ACL alongside the other contributing factors [3]. Scerpella et al., found a significant correlation between generalized joint hypermobility and increased incidence of ACL injuries, underscoring the biomechanical vulnerabilities inherent in hypermobile individuals [4]. Sundemo et al., conducted a systematic review revealing that generalized joint hypermobility is associated with an increased risk of ACL injuries and may lead to inferior outcomes following ACL reconstruction [5]. It could prove to be beneficial for paediatricians to screen for EDS in young female patients, especially in ages 14-26, to prevent tearing of ligamentous structures, such as the ACL.

CASE PRESENTATION

Subjects consisted of female athletes ranging in age from 14-25 years (mean ± SD age, 19.13 ± 2.68 years) that were currently being seen for ACL rehabilitation or finished within the last 4 months. All subjects that qualified were included.

METHODS

Subjects were assessed using the Beighton Scoring System which measures joint hypermobility using a 9-point scale [6]. Only information from criterion 1 was considered. If the subject was under 18, they required a 6/10 Beighton score and over 18 required a 5/9 (Figure 1).

RESULTS

A one sample z-test for proportions was used to compare the difference in prevalence of our sample population to the general prevalence using 95%. Confidence intervals including continuity correction. Z-scores were computed for raw scores. VassarStats statistical computation web site was used to compute the difference between the two independent proportions. Fifteen females (mean ± SD age, 19.13 ± 2.68 years) agreed to participate. Of these, six met the Beighton Scoring System criteria for EDS which resulted in a proportion of 0.4. The prevalence of the general population with EDS is one out of 5000 which results

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in a proportion of 0.0002. The difference between the two proportions is .398. The two tailed p value=0.0002 with a z score of 23.289.

Case Summaries

- **Case 1:** A 16-year-old soccer player with a non-contact ACL tear; Beighton score: 7/9.
- **Case 2:** A 20-year-old basketball player injured during a pivot; Beighton score: 6/9.
- **Case 3:** An 18-year-old gymnast injured upon landing; Beighton score: 8/9.
- **Case 4:** A 22-year-old volleyball player injured while jumping; Beighton score: 5/9.
- **Case 5:** A 19-year-old track athlete injured during sprinting; Beighton score: 6/9.
- **Case 6:** A 17-year-old dancer injured during performance; Beighton score: 7/9.

DISCUSSION

The purpose of this study is to assess the correlation of EDS and ACL injury in female adolescents. The results indicate there is a correlation between the diagnosis of EDS and susceptibility of musculoskeletal injury. The long-accepted prevalence rate of EDS has been noted as 1 in 5,000 [3]. However, in the population utilized in this study of 15 females ages 14-26 receiving physical therapy intervention post ACL tear, there was a 40% prevalence of EDS according to the Beighton Score criteria. These results bring into question the potential under diagnosis of EDS in pediatric patients, and what impact that may have on their musculoskeletal health. Early screening for EDS is warranted for children if the prevalence of the diagnosis is indeed higher than previously thought. If diagnosed, medical professionals could implement an injury prevention program for strengthening and stability of the joints in hopes of reducing the number of injuries that occur due to the hypermobility of joints. Ericson Jr and Wolman et al., emphasize the necessity of individualized orthopaedic management strategies for patients with EDS to address joint instability and prevent injuries [7]. It might even be necessary in extreme cases to steer a child away from certain activities in sports to maintain the integrity of the musculoskeletal system. Without knowledge of a diagnosis, a child might be more susceptible to an injury, such as an ACL tear, that could have been prevented with proper strengthening, compensatory strategies and nutrition. Childhood screening and education about EDS for both children and parents could improve the quality of play and longevity of career for pediatric athletes. In addition, once injury has occurred, Lindskog et al., found that patients with generalized joint hypermobility had lower rates of returning to their pre-injury level of activity two years after ACL reconstruction, highlighting the challenges in rehabilitation for this population [8].

LIMITATIONS

Some limitations of this study include a small sample size and a specific population that may limit generalizability. Future research should investigate a causational relationship between EDS and ACL tears. As ACL tears are more prevalent in females than males despite the presence of EDS, it may be that EDS does not play as big of a role in the injury as sex of the patient itself plays. Further research should also investigate the prevalence in a male vs. female population.

Implications for practice

This includes the screening, prevention programs and education:

- **Screening:** Routine screening for joint hypermobility using the Beighton Scoring System in young athletes (i.e., paediatricians).
- **Prevention programs:** Developing individualized training programs that focus on muscle strengthening, proprioception and neuromuscular control.
- **Education:** Informing athletes, parents and coaches about the risks associated with joint hypermobility and strategies to prevent injuries.

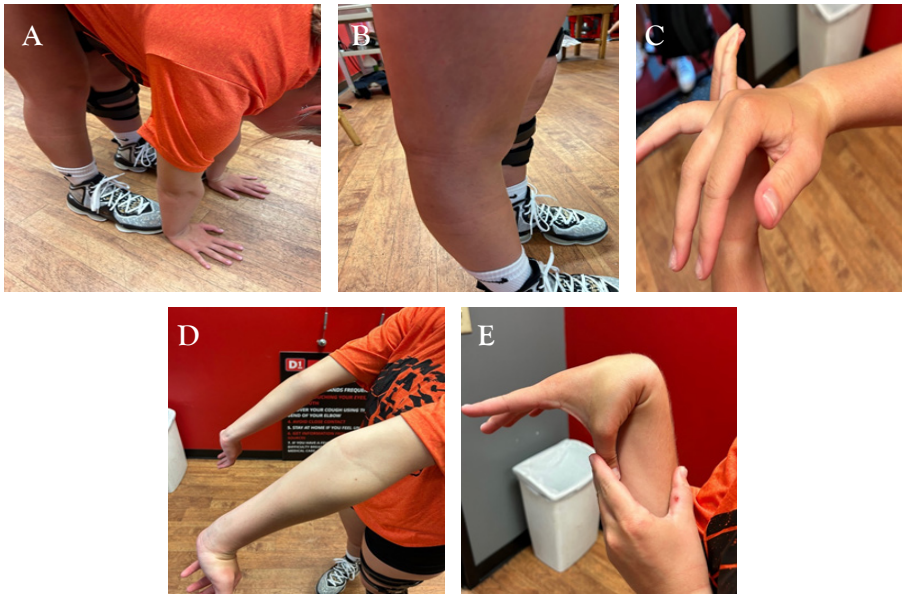

Figure 1: Positive primary criteria findings for diagnosis of Hypermobile Ehler's-Danlos Syndrome. **Note:** A) The first image is worth one-point; B-E) The latter four images have potential for two points each if hypermobility is detected bilaterally totally up to a nine total point Beighton Score.

CONCLUSION

The potential link between EDS and increased susceptibility to ACL injuries in athletes underscores the importance of early recognition and preventive strategies. By identifying individuals with EDS early, healthcare professionals can implement targeted interventions to enhance joint stability and reduce the risk of serious musculoskeletal injuries. This proactive approach could improve the quality of play and extend the athletic careers of pediatric athletes, with particular emphasis on females due to their higher risk.

ACKNOWLEDGMENTS

We acknowledge the participants for their contribution to this case series. Early screening for hypermobility conditions like EDS is crucial. By fostering awareness and encouraging proactive measures, we can contribute to the well-being and athletic success of all athletes, with an emphasis on females who are at higher risk for ACL injuries.

CONFLICT OF INTEREST

There are no financial conflicts of interest to disclose.

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