Ehlers-Danlos Syndrome in a College Tennis Athlete

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Abstract

Background: Athlete is a female tennis athlete in College NCAA Division III. She has a history of Ehlers-Danlos Syndrome (EDS) and correlated diagnoses such as multifocal Dystonia, Wolff-Parkinson-White syndrome, and Ventricular Septal Defect. The history of the patient includes a heart surgery at the age of 12 and two ankle surgeries on the left extremity during sophomore year in high school. She reported to the AT room with ankle pain. Evaluation revealed (+) anterior drawer and (+) Talar tilt. Referred for MRI confirming Grade III ATFL and Deltoid ligament sprain. She was put on a brace 24/7 for 4 weeks. She began rehabilitation for 2 weeks and peroneal subluxation was found during inversion isometrics. Continued rehabilitation for 4 weeks, had surgery for a lateral reconstruction shortening the ligaments anchoring it to bone reinforced with synthetic grafts. Additionally, she had a peroneal reconstruction/ SPR repair tying the peroneal tendons together. **Treatment:** After 9 days, she was removed from the cast, sutures, and was placed in a non-weight bearing boot and crutches. Athlete began regaining ROM, except inversion until 6 weeks postop. Then, she will transition to ASO brace and regular shoes. From week 6 to week 12 postop she will begin low-impact exercises. And after week 12 postop, she will have no restriction for her rehabilitation. **Uniqueness:** Ehlers-Danlos syndrome is an inherited connective tissue disorder that modifies the collagen in the tissue and makes the it more elastic and fragile. This syndrome can only be diagnosed by doing a complete study of the genome sequence of the individual. After combining elements in the athlete's life such as being diagnosed with Multifocal Dystonia, Wolff-Parkinson-White syndrome, Ventricular Septal Defect, and multiple sprains in R ankle after surgery; doctors concluded that she was eligible for Ehlers-Danlos Syndrome. Thus, a complete genome study which was positive. **Conclusion:** The athlete has suffered from the consequences of Ehlers-Danlos Syndrome her entire life. This includes instability of her ankles that caused the latest injury; ATFL and Deltoid ligament tear. The surgery will take time to heal and she will be facing several weeks of rehabilitation to regain ROM and strength on the muscles related to the ankle. In the future, she will have more stability on her ankles, and she will be able to go back to playing tennis. Regarding the Multifocal Dystonia, her current medicine prevents her body from spreading any further or to other muscles; and, the treatment on her Biceps Brachii and Anterior Tibialis will continue to be Botox injections, and on her 4th and 5th digits on the hand will continue to be a brace that prevents the involuntary contraction.

Introduction

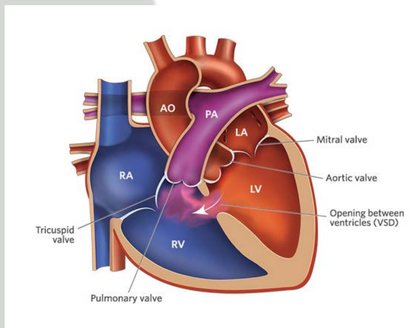
Ehlers-Danlos Syndrome is a group of connective tissue disorders which main cause is a mutation in the genome leading to a malformation in the collagen. When the collagen is malformed, it affects tissues such as skin, joints (ligaments and tendons), bones, blood vessels, and internal organs, creating pain, a lack of structure, instability, hypermobility, and fragility which makes the individual to be prone to injuries or other disorders/syndromes. There are 13 different subtypes of EDS that present in the genome as different mutations. The most common type is Hypermobile EDS or hEDS affecting athletes the most since it predispose them to injuries due to the hypermobility, instability, and fragility of the joint.

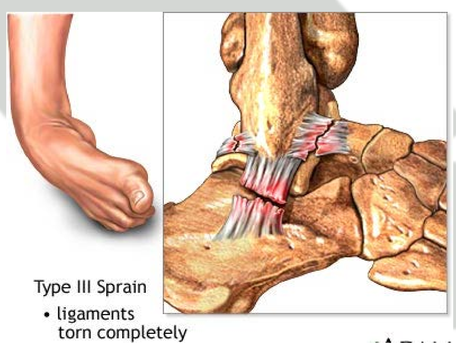
Purpose

The purpose of this case report was to introduce a female Division III tennis athlete who presented with multiple ankle sprains due to Ehlers-Danlos Syndrome. Patient has a history of Multifocal Dystonia, Wolff-Parkinson-White syndrome, and Ventricular Septal Defect. Ehlers-Danlos Syndrome is a connective tissue disorder that presents differently in every individual. Hypermobility is one of the presentations that creates instability within the joints; therefore, with case there were multiple ankle sprains and instability affecting her athletic career. Due to the complexity of EDS, an overview of the syndrome will be presented.

Etiology

Ehlers-Danlos Syndrome is caused by a genetical mutations in at least 19 different genes. Depending on the mutations, the clinical presentations may vary determining which subtype of EDS the individual might have. Mutations in one or more genes encoding fibrillar collagen or enzymes involved in the synthesis of it are responsible for the presentation of EDS. The mutations can be created by the individual, which means it was not inherited by ancestors. However, most commonly, parents are carriers of the mutation, one or both parents have EDS, or one of the parents have EDS and the other one is a carrier. This means that if an individual is diagnosed with EDS, there is a big chance that the parents are carriers or have EDS. Since it is a commonly misdiagnosed syndrome, there is a chance that an individual has the syndrome and never know it. Therefore, their children are very likely to present it EDS.





Clinical Presentation

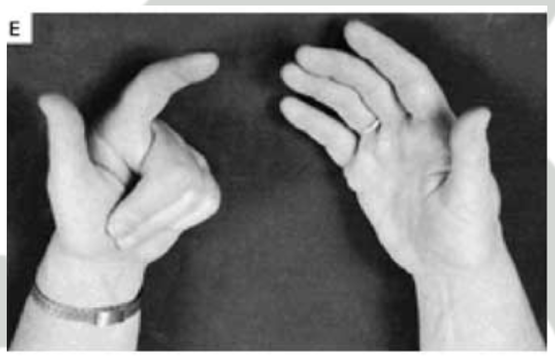
EDS presents differently; however, most of them have correlated signs and disorders. The most common signs include excessive skin stretchiness by 73% of the cases and joint hypermobility by 96% of the cases in which some include small functionality of the impact absorption in the joints. Other manifestations include multiple pain, fatigue, sleep disorders, motor proprioception disorders, dystonia, skin fragility, dysautonomia, hemorrhagic tendency, respiratory dysfunction, cutaneous hypersensitivity, disorder relating auditory, olfactory, vestibular and binocular functionality, digestive disorders, oral disorders, bladder-sphincter disorders, dyspareunia, obstetric, and cognitive disorders. As well as abnormal bruising and bleeding, unexplained vessel rupture or dissection, tissue fragility, atrophic scarring or skin hyperextensibility, symptomatic joint hypermobility, dislocations, recurrent sprains, hollow organ rupture, cardiovascular diseases or syndromes, translucent skin, dystonia, and scoliosis.

The tennis athlete's presentation included multifocal dystonia in her hamstring, biceps brachialis, and 4th and 5th digit of the hand. Cardiovascular syndromes such as Wolff-Parkinson-White syndrome and Ventricular Septal Defect which include 2 different holes in the septum. She also presented hypermobility in her joints as well as instability that lead her to have multiple sprains (Grade I, II and, II) in both her ankle.

Diagnosis

EDS is misdiagnosed or not diagnosed at all because it can present as other disorders or injuries that are not correlated one to another in the moment. When EDS is suspected, the first step is to collect a complete medical history, and physical examination. Cardiovascular and family history should be included. In case of positive EDS in the family, a complete genome study should be done. A blood sample or skin biopsy can be used for the study of the genome.

The tennis athlete was born with 2 different heart problems, later, she continued to have multiple problems if instability in her joints, and multifocal dystonia. When the doctors put all the signs together, they concluded that she was eligible to have a complete study of her genome. This led to a positive for EDS.



Treatment

The treatment for EDS will vary depending on the presentations of each case. Normally, before diagnosing the syndrome, it is more relatable to treat the individual conditions. For example, if an individual is facing ankle instability, it would be treated with physical therapy. If the individual is facing a heart disease, the treatment would involve making the heart condition better and control it to the point it is safe for the individual to continue with daily activity. There are surgical and non-surgical treatments which a specialist will decide upon.

In the case of the tennis athlete, her treatment for both heart conditions were surgical. She had surgery when she was a newborn due to the high risk of the disease. The multiple dystonia in her biceps brachii is controlled with Botox injections. The dystonia in the 4th and 5th digit of the hand is controlled with a brace; this means that she hold the tennis racket with only 3 digits. Lastly, her instability in the ankles have been a recurrent problem. Some of the moments she had ankle sprains; they were treated non surgically; however, she has had 2 surgeries in her right ankle, and 1 surgery in her left ankle. The decision of the surgery was made because a complete tear of the ATFL was present.

Conclusion

Ehlers-Danlos Syndrome is mainly inherited by parents that present the syndrome or are asymptomatic carriers. EDS subtypes differ from one to another by different mutations that at the same time shows different clinical presentation. Depending on the signs and symptoms presented, the treatment can be focused directly to them; however, some of the signs can be life threatening disorders that involve the life of the organs. In case of sports, it is necessary to be precautious because individuals with EDS are more likely to get injuries than people without EDS.

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