Case report

# Lateral ankle instability in a young patient with Ehlers-Danlos syndrome

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**Abstract**

Ehlers-Danlos Syndrome (EDS) is a group of hereditary disorders of connective tissue that are varied in the ways they affect the body and in their genetic causes. It is generally characterized by joint hyper mobility, joint instability, dislocation, scoliosis, skin hyper extensibility, and abnormal scarring, and other structural weakness such as hernias and organ prolapse through the pelvic floor. This case reviews a 33- year-old male with a classical history of EDS, severe aortic regurgitation and left ankle instability of greater than 6 months. Instability was confirmed with clinical evaluation, stress inversion and anterior drawer test. Conservative treatment of lateral ankle instability was done, including rest, elevation, initial range of motion, progressive weight bearing guided by symptoms tolerance and physical therapy. Lateral ankle instability is a complex condition that can, at times, prove difficult to evaluate and treat. This activity reviews the etiology, presentation management and evaluation of lateral ankle instability. In addition, the role of interprofessional team in evaluation, diagnosis and management.

**Keywords:** Ehlers-Danlos, lateral ankle instability, Bighton score.

## Introduction

Ehlers-Danlos syndrome (EDS) is an umbrella term for a group comprising heritable soft connective tissue disorders characterized by generalized joint hypermobility, skin texture abnormalities, and visceral and vascular fragility or dysfunctions [1]. Major and minor diagnostic criteria have been defined for each type and complemented whenever possible with laboratory findings; the clinical Villefranche classification identifies six major EDS variants [1]. Advances in molecular testing have made it possible to identify the causative mutation in the majority of patients showing that abnormalities in type V collagen are the cause of a classical type EDS [2]. There is a clinical overlap with the joint hypermobility syndrome (JHS) [3]. Joint hypermobility is relatively abundant, found in up to one-third (30%) of children [4]. It can present in a variety of signs and symptoms such as musculoskeletal and neuropathic pain, joint instability, fatigue, anxiety, and cardiovascular problems [5-7].

Adults diagnosed with JHS/EDS-hypermobile type often experience joint pain or widespread pain of different levels of severity [8]. In each affected individual, clinical decision making should be theoretically applied for now, underpinned by the available evidence, making each of the patients with EDS unique in a therapeutic approach.

**Case presentation**

A thirty 33 years old male lives in a village near Shendi presented to the clinic with recurrent attacks of multi- joints pain since he was 9 years old, the pain mainly affects his ankle joint bilaterally and his right wrist and right carpometarcpal joint, he also reported chronic rib pain especially with sneezing and coughing, he saught medical advice in Khartoum and seen by an orthopedic consultant and received oral and topical analgesics.

In 2018, he was diagnosed with aortic regurgitation and mitral regurgition when he started to develop fatigability, exertional dyspnoea and lower limbs oedema. Echocadiogram was done and revealed: severe aortic regurgitation and mitral regurgition.

On examination, patient looks unwell, slightly pale, fullyconscious, not dyspnoic or jaundiced. Has leptoprosopic face with orbital hypertrolism. The skin over extremities is Loose and easily stretchable. Has flat feet and hypermobility of Knees, ankles, and joints of the hand.

On the Bighton score, he received (6/9) score: passive apposition of the thumb to fore arm and passive dorsal hyperextension of the metacarpophalangeal joint >90° on both sides were done with no strain; he was able to actively hyperextend both knees on both sides. Right ankle instability was confirmed with clinical evaluation; stress inversion and anterior drawer test (Fig 1).



**Figure 1.** An ankle stress radiograph showing lateral ankle instability with nearly a 40-degree talar tilt angle

**Discussion**

Etiologically, lateral ligament instability can result from either of two means; functional or mechanical instability. Mechanical instability arises either via an acute injury or chronic repetitive stress resulting in attenuation and alteration of the mechanical structures of the ligaments. The most common mechanism of injury in lateral ankle sprain occurs with a plantar-flexory force on an inverted ankle as the body moves over the foot. The ATEL is the most commonly injured ligament followed by the CFL, and then the PTFL [9]. Ligaments often heal in an elongated position which can lead to plastic deformation, which further reduces the ability to provide restraint. Functional instability is the feeling of ankle instability or recurrent, symptomatic ankle sprains due to proprioceptive deficits. Lateral ankle

instability can also be caused by hereditary ligamentous laxity that is associated with Ehlers-Danlos syndrome, Marfan's syndrome and Turner's syndrome.

Epidemiologically, 40% of all athletic injuries are ankle sprains, making ankle sprains the most commonly sustained injury during sporting events [10]. Over 50% of basketball injuries and almost 30% of soccer injuries can be attributed directly to ankle injuries [11]. Over 10% of the emergency room visits in the United States are due to ankle sprains with an incidence of 30.000 per day [12]. 75% of ankle injuries involve the lateral ankle with an equal distribution between male and females. Some literature has reported a 25% higher rate of grade 1 sprains in females compared to males, and a higher predisposition to suffering future sprains once having sustained an ankle sprain [13-16]. 90% of all ankle sprains involve the ATFL, while 50-75% of the time the CFL is engaged, and only 10% of the time is the PTFL involved [17-20]. 55-72% of patients who sustain a lateral ankle sprain suffer residual symptoms. In the USA the estimated incidence rate of ankle sprains in the general population presenting to emergency departments is 2.15 per 1.000 person-per year. The incidence of ankle sprains in the military has been reported to be up to 27-fold higher than in the general population [21].

Evaluation of lateral instability consists of the history, and physical described previously but also includes radiographs of the ankle to assess for syndesmosis widening, foot or ankle deformities which may predispose patients to instability. Stress radiographs such as with the anterior drawer test or talar tilt test are also essential to evaluate for structural abnormalities. Patients with mechanical instability will display radiological evidence; patients with functional instability may have a negative anterior drawer or talar tilt tests. MRI can be useful to evaluate the lateral ankle complex, and it can also identify concomitant pathologies such as talar dome osteochondral defects, peroneal tendon pathology and can be of assistance in surgical planning.

Conservative treatment of lateral ankle instability consists of early functional rehabilitation; including rest, ice, elevation, compression, initial range of motion, progressive weight-bearing guided by symptom tolerance, and physical therapy. Even with proper functional rehabilitation 10 to 40% of patients will go on to develop chronic ankle instability following acute ankle sprains. Multiple studies have been published which show both the benefits of conservative therapy as well as surgical treatment for cases that do not adequately respond to conservative treatment. There

have been more than 70 different operative techniques described in the literature for correcting ankle instability, and can be divided into three main categories; anatomic, non-anatomic and anatomic augmented tenodesis reconstruction. Anatomic reconstruction consists of the Brostrom procedure, which includes direct repair of the ATFL in pants over vest fashion; this reduces any redundancy due to ligamentous laxity and can be used for completely torn ligaments. The Brostrom procedure has been modified and can include the use of the extensor retinaculum to increase the efficacy of the repair.

In conclusion, Ehlers-Danlos syndrome is a rare disease of the connective tissue and a diagnostic challenge. Late diagnosis leads to chronic pain and complications, limiting daily activity and mobility. This case report serves to remind the rehabilitation clinicians that it is important to identify all affected patientsin order to prevent complications instead of just providing the therapy against pain.

## Consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images.

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