

# A Novel Exercise Protocol for Individuals with Ehlers Danlos Syndrome: A Case Report

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## Abstract

Ehlers-Danlos Syndromes (EDS) are a group of heritable connective tissue disorders first described by Hippocrates in 400 BC. EDS has undergone multiple changes in classification and diagnostic criteria since its initial observation. Once thought to be extremely rare EDS has seen a significant increase in prevalence corresponding with these changes and estimates have reached one in 2500-5000 depending on the subtype considered. Physical therapy is considered an essential therapeutic modality for a number of the different subtypes of EDS. Unfortunately the outcomes have not always been positive and patients have reported increased pain and dysfunction following physical therapy intervention. This case report presents the positive outcomes of a patient who has completed a novel exercise protocol designed specifically for individuals living with EDS.

**Keywords:** Ehlers danlos syndrome; Physical therapy; Exercise protocol; Collagen; Connective tissue

## Introduction

Ehlers-Danlos Syndromes (EDS) are a heterogeneous group of heritable connective tissue disorders (HCTDs) characterized by the variable triad of (i) generalized joint hypermobility and related osteoarticular complications, (ii) dermal dysplasia extending from minor changes of skin texture to clinically relevant skin fragility and defective scarring, and (iii) vascular and internal organ fragility with proneness to traumatic injuries and spontaneous ruptures, dissections and prolapses [1]. Individuals with EDS demonstrate defects in the body's connective tissues, manifesting as altered strength, elasticity, integrity, and healing properties of the tissues [2]. The diagnosis and classification of EDS has seen significant changes since Hippocrates first described individuals with joint laxity and multiple scars in 400 BC. In 1901 Edvard Ehlers recognized the condition as a distinct entity. In 1908, Henri-Alexandre Danlos suggested that skin extensibility and fragility were the cardinal features of the syndrome [3]. Formalized classification of EDS began in the 1960's with significant changes in nosology being made in 1988 and 1998 respectively [4]. As diagnostic criteria have improved new subtypes of EDS continue to be identified. The most recent work conducted by The International EDS Consortium devised an EDS classification which recognizes 13 subtypes (Table 1). For each of the subtypes they proposed a set of clinical criteria that are suggestive for the diagnosis [5].

Clinical EDS Subtype	Abbreviation
1. Classical EDS	cEDS
2. Classical-Like EDS	clEDS
3. Cardiac-valvular	cvEDS

4. Vascular EDS	vEDS
5. Hypermobility EDS	hEDS
6. Arthrochalasia	aEDS
7. Dermatosparaxis EDS	dEDS
8. Kyphoscoliotic EDS	kEDS
9. Brittle Cornea syndrome	BCS
10. Spondylodysplastic EDS	spEDS
11. Musculocontractural EDS	mcEDS
12. Myopathic EDS	mEDS
13. Periodontal EDS	pEDS

**Table 1:** Clinical classification of Ehlers-Danlos Syndromes.

While EDS was once considered to be a rare condition with a prevalence rate reported between 1/400,000-500,000, the creation of new evaluation criteria has led to a diagnostic rate that is becoming surprisingly common. Prevalence rates have been reported to reach between 1/2500-1/5000 under the new diagnostic criteria and currently there are more than 1.5 million cases of EDS worldwide when taking into account all subtypes, with this number likely to increase substantially. Classical and hypermobility types account for more than 90% of cases. The third most common type is vascular EDS and may affect 1 in 250,000 people [6]. The hypermobility type is considered the most common. It is referred to as type V EDS under the new classification and follows an autosomal dominant inheritance pattern. It involves types I and VI collagen, specifically gene TNX-B on chromosome 6 [7]. The prevalence is determined to be 1 in 5,000 to 20,000 [7]. Characteristic clinical manifestations include soft,

smooth, mildly hyper extensible skin with easy bruising and joint and spine hypermobility [7]. Joint subluxations and dislocations are common. Some consider the hypermobility subtype of EDS to be the least severe because it is less likely to involve internal organs. However, these patients may experience the most debilitating musculoskeletal function [7]. They are more likely to require surgery, experience more pain, and function at a lower level [7].

Ehlers-Danlos syndromes (EDS) are caused by various abnormalities in the synthesis and metabolism of collagen and other connective-tissue proteins in the extracellular matrix (ECM), such as elastin, proteoglycans, and macromolecular proteins [8]. Collagens are a group of proteins found within all organs of the body. Defects in the genes that provide instructions on how to use the proteins and assemble collagen can result in abnormal size, shape and organization of these various proteins depending on the syndrome subtype.

Physical therapy is considered a foundational treatment approach for a number of the different types of EDS. Unfortunately the outcomes associated with physical therapy management are not always positive. Consistently patients have reported that physical therapy has exacerbated symptoms and have focused on one single joint rather than treating all of the patients' deficits [9,10]. Due to the propensity for misdiagnosis and paucity for understanding of the pathophysiology of EDS it is no surprise that a vast number of the individuals suffering from these conditions are poorly managed. Traditional treatment plans can result in further injury if not properly sequenced and their intensity titrated due to continued collagen and joint damage.

The purpose of this case study was to present the possible benefits of a novel exercise protocol for an individual with classical EDS. To the best of our knowledge this is the only EDS specific exercise protocol being used clinically to date.

## Case Presentation

This case discusses the presentation and physical therapy management using a novel exercise protocol of a young female patient presenting with complaints of widespread joint pain, especially involving the lumbar and cervical regions and the lower extremities. The patient was initially diagnosed as having "growing pains" by a pediatrician and then "generalized laxity", based on symptoms, including chronic musculoskeletal pain, widespread muscle weakness and history of hypermobility and chronic injury with physical activity. The patient had been seen for wide spread pain and multiple musculoskeletal injuries at five other physical therapy clinics and one chiropractic clinic within the preceding six years. The patient was evaluated and placed on therapeutic exercise programs based on the current evidence and standard of care for the individual conditions she was presenting with at the time of evaluation, assuming an otherwise healthy individual of her age. Through each course of treatment, the patients' condition worsened and she complained of increased pain and further decreases in function. These attempts at therapeutic intervention resulted in the patient "failing" physical therapy and being discharged to other providers in search of more answers. The continuing progression of pain and loss of functional mobility coupled with the inability to receive a proper diagnosis and prognosis led to depression, social withdrawal and overall decreased quality of life (QOL). Finally, at the end of 2014 the patient received a diagnosis of hEDS and was referred to our clinic.

## Initial evaluation

The patient was a 17-year-old female when she presented to our clinic in February of 2015. She was 5'1" in height and weighed 117 lbs, patient presented to physical therapy complaining of widespread pain and fatigue for the past six years, most prominent in the lumbar and cervical regions as well as the bilateral knees and feet and to a lesser extent the upper extremities. Patient stated pain worsening as the day progresses, associated with increased physical activity in most cases but also with prolonged sitting. Pain score varied from 6/10 to 9/10 on the numeric pain rating scale. The increased pain was associated with progressive weakness more prominent in the bilateral lower limbs. Other findings included; dizziness, fainting, nausea, seizure, and postural orthostatic tachycardia syndrome (POTS). Patient has had a joint hypermobility history since childhood. Previous evaluations diagnosed the patient with ligament laxity and hypermobility and prescribed progressive resistance exercise with activity modification. Other past medical history included depressive disorder manifesting as her function progressively decreased. Recent diagnostic testing included; X-rays (cervical, thoracic, lumbar, pelvis, bilateral knees and feet-all multiple views), MRI (cervical, lumbar, bilateral knees) and two bone scans. A healed L4 transverse process fracture was seen on x-ray but not clinically correlated to the patients' current condition, all other diagnostic testing was normal.

On the day of the initial evaluation the patient presented as follows:

**Subjective:** 6/10 pain bilateral upper extremities

7/10 pain cervical spine

6/10 pain thoracic spine

9/10 pain lumbar spine

8/10 bilateral lower extremities

Further subjective complaints included tension type headaches, and feel unsteady and progressive fatigue lower extremities > upper extremities.

**Objective:** Beighton Scale - 9/9

Range of Motion (ROM) – Within Normal limits or exceeded normal limits for all joints

Manual Muscle testing (MMT) – Globally 4/5

Ligamentous Laxity Testing - Positive

Multiple vertebral and joint subluxations present

Reflexes – Normal

Sensation (peripheral and Central) – Normal

Balance – Within normal limits

Coordination – Within normal limits

Gait – Antalgic gait secondary to pain in bilateral knees and feet

**Outcomes:** *Oswestry Disability Index (ODI)* – The ODI is a validated and reliable self-report questionnaire currently considered by many as the gold standard for measuring degree of disability and estimating quality of life in a person with low back pain [11].

The patient received a score of 47% at the initial evaluation indicating severe disability.

**Neck Disability Index (NDI)** - The Neck Disability Index (NDI) is a valid and reliable self-report questionnaire. Currently it is the most widely used outcome instrument for the assessment of patients with neck related disorders [12].

The patient received a score of 26 at the initial evaluation indicating moderate disability.

**Lower Extremity Functional Score (LEFS)**The LEFS is a valid and reliable self-report questionnaire. The capacity of the LEFS to detect change in lower-extremity function appears to be superior to other outcome measures such as the SF-36 physical function subscale [13].

The patient received a score of 33.75% at initial evaluation indicating significant disability.

**Upper Extremity Functional Index (UEFI)** - The UEFI is a valid and reliable self-report questionnaire which measures disability in people with upper extremity orthopedic conditions [14].

The patient received a score of 59 at the initial evaluation indicating moderate difficulty with upper extremity tasks.

## Intervention

Following a comprehensive evaluation, the patient participated in a specialized graduated exercise protocol developed by Kevin Muldowney PT, owner of Muldowney physical therapy an outpatient clinic dedicated to EDS treatment.

The basis of the exercise protocol is that it must be specifically sequenced in order to gain the maximal benefit using EDS manual therapy techniques as an adjunct to the exercise protocol. The protocol is divided into two phases. Phase one focuses on graduated strengthening of the joints throughout the body. In this phase the body is divided into three exercise progressions; 1) Sacroiliac joint and lumbar spine, 2) Cervical spine, thoracic spine and upper extremities, and 3) Lower extremities. The distinction allows the therapist to focus their manual and modality specific treatment on one area of the body while the patient is strengthening the same area within the protocol. Once phase one has been completed the majority of patients should be able to perform most activities of daily living without increased pain or subluxations. Once phase one has been completed phase two will begin. Phase two is further divided into three exercise progressions; 1) Twisting progression, 2) Dynamic balance progression, and 3) Throwing progression. Once the top level has been reached for each phase the patient will receive a finalized home exercise program. The full protocol can be found in the book, 'Living Life to The Fullest with Ehlers Danlos Syndrome' [15]. The protocol averages between six months to one year for full completion depending on patient compliance, tolerance and condition severity. The current patient participated in treatment three times per week between February 2015 and February 2016.

At the beginning of the protocol, the patient began with Level 1 Mat exercises. She performed all five exercises for 1 minute, 30 seconds. The next day, all five exercises were performed again, but at 1 minute, 40 seconds each. The patient continued this pattern of adding 10 seconds per day for each exercise until she could perform all five exercises for 3 minutes each. This marked the completion of Level 1 mat. At this point, the patient was ready to begin Level 2 Mat and add Level 1 Ball exercises. The patient began each of these at 1 minute, 30 seconds and follow the same progression of adding 10 seconds per day until reaching 3 minutes for each exercise. Once completed, the patient performed Level 3 Mat and Level 2 Ball. Once completed, the patient

performed Level 3 Mat at 3 minutes twice a week and will begin Level 3 Ball at 1 minute. Once she reached 3 minutes of the level 3 Ball exercises she added Level 1 Neck exercises every day at 1 minute, 30 seconds and progress 10 seconds per day until reaching 3 minutes. Once completed, the patient performed Level 2 Neck exercises every day with same progression while continuing Level 3 Mat and Level 3 Ball each at 3 minutes twice a week. The patient then followed the same progression for the Neck exercises from Level 2 to Level 7.

Once these were completed, the patient added Level 1 Lower Extremity exercises. All Lower Extremity exercises are progressed as follows: begin at 10 repetitions, add 2 repetitions per day until reaching 26 repetitions for each exercise. Once all exercises are performed at 26 repetitions at a specific level, the patient can progress to the next level. Once the patient reached Level 7 Lower Extremity exercises, she performed Level 3 Mat and Level 3 Ball twice a week, Level 7 Neck twice a week and Level 7 Lower Extremity twice a week, and then began Phase II of the protocol. Any set back will cause the patient to drop down one level and begin all exercises at that level and progress as described above.

## Results

One year and one day from the initial evaluation the patient was discharged with a home exercise program. At discharge the patient presented as follows:

### Subjective

- 0/10 pain bilateral upper extremities
- 0/10 pain cervical spine
- 0/10 pain thoracic spine
- 0/10 pain lumbar spine
- 0/10 bilateral lower extremities

Further subjective comments included: no longer suffering from headaches, no longer feeling unsteady or fatigued.

### Objective

- Beighton Scale - 9/9
- Range of Motion (ROM) - Exceeded normal limits for all joints
- Manual Muscle testing (MMT) - Globally 5/5
- Ligamentous Laxity Testing - Positive
- No joint subluxations present at discharge
- Reflexes - Normal
- Sensation (peripheral and Central) - Normal
- Balance - Within normal limits
- Coordination - Within normal limits
- Gait - Normal

### Outcomes

**Oswestry Disability Index (ODI)**The patient received a score of 2% at discharge indicating no disability.

**Neck Disability Index (NDI)**The patient received a score of 0 at discharge indicating no disability.

**Lower Extremity Functional Score (LEFS)**The patient received a score of 97.5% at discharge indicating no disability.

**Upper Extremity Functional Index (UEFI)** - The patient received a score of 79 at discharge indicating no difficulty.

## Discussion

This case report shows the efficacy of an EDS specific exercise protocol to reduce pain and dysfunction and improve functional mobility in a patient with hEDS. Exercise is an essential technique in rehabilitation; however, evidence to determine the most advantageous type, frequency, dosage, duration, or delivery method is lacking in the growing population of individuals diagnosed with EDS [16]. To the best of our knowledge this is the first exercise protocol developed specifically for the EDS population.

Review of evidence based exercise programs typically used with EDS patients can show a lack of systematic, low intensity titrated progression and discrepancies in treating whole body deficits, choosing more commonly to focus on one or two body regions during the course of treatment and increasing intensity too quickly. Another common issue with traditional physical treatment plans when addressing patients living with EDS is the inclusion of static and dynamic stretching exercises. While these can have a tremendous positive impact for a number of Neuromusculoskeletal disorders the inclusion of global stretching for the population of patients with EDS may have a significant negative impact. Due to the various abnormalities in collagen metabolism and synthesis, the tendons and ligaments may be getting stretched more than the muscles resulting in increased laxity and instability and therefore more subluxations, pain and dysfunction. Providing a specific, stepwise and global exercise protocol may elevate the majority of these issues and lead to improved functional outcomes and greater overall quality of life.

While not highlighted in this case the use of manual therapy as an adjunct to the exercise protocol remains a focal component to the steady progression of an individual with hEDS. The vast majority of patients with this condition suffer from multiple chronic and disabling joint subluxations and dislocations. The use of manual therapy techniques to restore normal arthrokinematics in hypomobile spinal segments or peripheral joints is appropriate to address mechanical pain and dysfunction [17,18] but must be used in a judicious manner and in many cases techniques must be modified for this population to account for collagen and subsequent joint dysfunction. There is no doubt EDS specific manual therapy techniques are being developed and refined in clinics with a growing number of EDS patients and future research should look to developed standards in regard to best practice.

Practitioners and patients both must realize that the proposed treatment progression is a long and arduous journey lasting between six months to one year on average. Setbacks are a common occurrence and could lead to noncompliance with the exercise protocol if not properly monitored and treated but with proper education, treatment and care individuals with EDS can live life to the fullest.

## Conclusion

The importance of a progressive EDS specific exercise protocol designed to decrease pain, correct biomechanical deficits and improve

overall functional mobility should not be understated. While EDS research is still in its infancy and considerable work remains to be done in the development of evidence based guidelines and treatment plans, the Muldowney exercise protocol provides a firm start point for a population that has been consistently mismanaged by well-meaning and otherwise competent healthcare providers.

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