Ehlers Danlos Syndrome: A Case Report

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Introduction

The history of Ehlers Danlos Syndrome (EDS) goes back to centuries where it is much described in early writings as the ‘Elastic Man’. In 1668, Job Janszoon van Meek’ren published the first written description and pictorial depiction of an individual with EDS. The first available photograph of a person with EDS is of Felix Wehrle in 1880. In 1891, A. N. Tschernogubow, a Russian dermatologist provided the first complete description of the syndrome in a 17 year-old and as such, EDS in Russian literature is to date described as “Tschernogubow syndrome”. Tschernogubow’s report is in the Russian language and failed to find a widespread audience.1 The current description of EDS in literature is the result of the works of Edvard Ehlers (1901) and Henri-Alexandre Danlos (1908). The syndrome was coined as EDS in 1936 by Weber.2

Diagnosis and Classification

Ehlers Danlos Syndrome is a clinically and genetically heterogeneous group of disorders that result from defect in the synthesis or structure of collagen. The diagnosis of EDS is done clinically. The common features of EDS are skin hyperextensibility, joint hypermobility, and fragility of tissues. EDS is classified into sub-types based on the cause.3

Classic type

EDS type I and type II come under this category. Inheritance is autosomal dominant. The major diagnostic criteria are skin hyperextensibility, widened atrophic scars, joint hypermobility. The molecular defect is in the structure of collagen V. Some cases have been linked to a mutation in COL5A1 and COL5A2 genes, and other causes are being investigated.

Hypermobility type

EDS type III comes under this category. It is inherited as an autosomal dominant condition. The major diagnostic criteria include hyperextensibility and/or smooth, velvety skin and generalized joint hypermobility. Patients have recurring joint dislocations with chronic joint and limb pain. The inherent cause in some cases can be attributed to a mutation in COL5A3 gene.

Vascular type

EDS type IV results from mutations in the gene for type III procollagen encoded by COL3A1. This results in an abnormal collagen III with defective secretion. Inheritance is autosomal dominant with a significant number of cases being new mutations. Clinical diagnostic criteria include thin skin with visible veins, arterial/intestinal/uterine fragility or rupture, easy bruising, and characteristic facial appearance.3 EDS type IV is the only sub-type in which the patient has an increased risk of dying.5 Most affected patients survive the first and second major complications, but Ehlers-Danlos syndrome type IV results in premature death.

Kyphoscoliosis type

EDS VI is caused by a deficiency of lysyl hydroxylase, which is a collagen-modifying enzyme. Inheritance is autosomal recessive. The major diagnostic criteria are generalized joint laxity, severe muscle hypotonia at birth, scoliosis at birth that is progressive, fragile sclera and rupture of ocular globe. Diagnosis is made at birth by the presence of any three major criteria and laboratory testing.

Arthrochalasia type

This includes EDS type VIIA and type VIIB.6 Inheritance is autosomal dominant for both of these disorders. The major clinical diagnostic criteria associated with this syndrome include bilateral congenital dislocation of the hip and severe generalized hypermobility of the joints with recurrent subluxations. The basic defect is caused by mutations that result in deficient processing of amino-terminal end of collagen type I. The genes that are responsible are COL 1A1 and COL 1A2.

Dermatosparaxis type

EDS type VIIC follows autosomal recessive inheritance. It is the result of deficiency of procollagen I N-terminal peptidases caused by mutant alleles. Major diagnostic criteria are severe skin fragility, and sagging and redundant skin.

Other types

EDS type V has X-linked inheritance.7 EDS type VIII patients exhibit periodontic fragility. EDS type IX was reclassified as “occipital horn syndrome.” EDS type X has currently been shown in a single family. EDS type XI is now termed “familial joint hypermobility syndrome.”

Case Presentation

History

The patient is a 28 year-old Caucasian female who presents with the chief complaint of “pain in all joints.” Her rheumatologist who suspected a diagnosis of Ehlers Danlos Syndrome referred her to the OMT clinic. The patient complained of daily dislocations of various joints, which began at the age of three.
The most common joints affected include her thumbs, right shoulder, right knee and right hip. She had noticed loss of feeling and tingling associated with the dislocations. She reported chronic rib pain, especially with sneezing and coughing.

**Past Medical History** is significant for a pituitary tumor, which is not operable, migraine headaches, EDS, and bipolar disorder. She stated that she had a horseshoe kidney as well as a history of seizures.

**Past Surgical History** is significant for right shoulder surgery in 2005 secondary to chronic dislocations. She had a right knee surgery in 1993 for a patellar reattachment. She has had sinus surgeries at ages 12, 13, and 14. She also reported a jaw surgery to correct an overbite in 2002. She has had an ovarian cyst removed in the past.

**Social History**: The patient was, until recently, an executive in the Sales and Marketing department of a company, but is currently unemployed secondary to medical conditions. She denies any tobacco or illicit drug use, but reports occasional alcohol consumption.

**Family History**: Family history is positive for fibromyalgia, hypertension, and hypercholesterolemia in her mother as well as bipolar disorder. Patient’s father has no significant medical history.

**Current medications** include Amiodarone pm soroquel 400mg daily, Topamax 50mg daily, Bromocriptine 2.5mg daily, Prozac 40mg daily, Clonazepam as needed.

Patient is allergic to sulfa drugs, Compazine, and Thora.zine.

**Review of Systems**: ROS is positive for above chief complaint. The patient does report some weight gain over the past year. She is unable to exercise secondary to her hypermobility syndrome. The rest of the review of systems is negative.

**Physical Examination**: Vital signs were within normal limits. The patient rated her pain anywhere from a 2 currently, to a 10 when she has dislocations.

The physical exam was essentially negative except for some musculoskeletal findings. The patient had an overall increased ligamentous laxity and increased joint mobility throughout multiple joints in her body, particularly in her thumbs, shoulders, knees and hips. Her knees had tenderness bilaterally. The L5/S1 tender points were noted and in the right lower extremity at the knee she had tender points in the popliteal fossa, consistent with Jones Strain/Counterstrain points of the ACL and PCL. She also had peripatellar tender points around the entire right kneecap.

**Assessment**: Ehlers Danlos Syndrome, knee pain, low back pain, and bipolar disorder, migraine headaches, and pituitary tumor by history.

**Treatment/Plan**: OMT was performed utilizing Jones strain/counterstrain to alleviate tenderness at the above-mentioned tender points. The patient tolerated the procedure well. She reported decreased pain after the procedure. The patient was educated and given information on how she could self-treat her tender points. The patient was to consider possible physical or occupational therapy to strengthen muscle attachments in an effort to stabilize her joints. Water aerobics was also suggested as a low impact way to exercise. A postural survey would be done and treated to minimize any postural forces that would encourage joint dislocation. The patient was asked to return to the clinic in one week for further evaluation.

On her next visit, the patient reported a pain level of 2 in right knee, right shoulder, and left thumb. She also stated that she had difficulties in self-treating her tender points as her joints dislocated during the treatment due to the pressure she applied. Treatment options were discussed with the patient with great emphasis on her hypermobility. The patient was advised to complete a postural survey to check for any sacral base inclination to ensure proper loading of joints and minimize any postural stress contributing to the dislocations.

Postural studies showed that the patient had a 1.4 cm leg length discrepancy when she was measured at the apex of the femoral heads, right greater than left. A 1.5 cm iliac crest discrepancy was measured, right greater than left. This resulted in a four-degree sacral tilt, right side higher than left. Using Cobb’s method, an eight-degree levoscoliotic curvature of the upper lumbar spine was measured from superior T12 to inferior L3 end plates. On lateral view, Ferguson’s angle was found to be 120 degrees.

**Literature Review and Discussion**

EDS is a heterogeneous group of heritable disorders of the connective tissue. Its incidence is estimated to be around 1 in 5000 with no racial or ethnic predisposition.

Treatment of patients with EDS proves to be a challenge, as there is no cure for the condition. Most treatments provided are palliative that revolve around the ramifications and complications of the condition. The three major clinical manifestations of EDS are skin hyperextensibility, joint hypermobility and tissues fragility. Joint hypermobility poses a huge problem for EDS patients as it leads to frequent dislocations of various joints. A major consideration in the management of an EDS patient would be to decrease the episodes of dislocations. A number of these patients suffer from spinal deformities like scoliosis. One leading cause of scoliosis is short leg syndrome. Short leg syndrome leads to sacral tilt, which further leads to a compensatory scoliotic curve in the spine. The sacral tilt and the scoliotic curve increase the risk of dislocations due to a non-optimal center of gravity. A heel lift can decrease the sacral tilt, thereby improving posture and providing a more stable sacral base and spinal alignment. This will lead to fewer episodes of dislocations. Therefore, a postural evaluation of an EDS patient should, therefore, be done to rule out any leg length discrepancy.

A compromised postural alignment leads to stress on surrounding muscles leading to chronic, recurrent trigger points. Osteopathic manipulative treatment can help better prepare the muscles, joints, ligaments and supporting soft tissue for the postural change brought about by the heel lift. Chiropractic modalities have been used successfully to treat subluxations of joints in EDS patients. They have had improved function and decreased dependency on pain medicines. It should also be important to consider a light, strength-building exercise program to enhance muscle tone which can substitute for the decreased connective tissue support. Postural corrective exercises can stretch shortened soft tissues and strengthen postural muscles. Hydrotherapy can be a low impact way of exercising and is also enjoyable.
EDS patients suffer from chronic pain secondary to their frequent dislocations and soft tissue damage. Pain begins early in their life and progresses over time. It is generally refractive to pharmacological interventions and significantly interferes with daily functioning. Patients have used analgesics, opioids, massage, chiropractic manipulation, etc. to help cope with the chronic pain. A normal physiological reaction to a painful hypermobile joint is for muscles surrounding the joint to splint the joint and protect it from excess motion. Physical examination of the patient will reveal a restriction of motion. A high-velocity thrust technique may work to free up the restriction, resulting in a decrease in pain and an improvement in motion, but the treatment contributes to joint instability. The more the HVL A technique is used, the looser the joint will become. Due to the fragile tissues in an EDS patient, it is prudent to limit the treatment to relatively gentle techniques such as indirect, myofascial release, Jones’ strain-counterstrain, balanced ligamentous tension. These mild treatments can relieve tender points, resolve muscle spasms, improve structural alignment, and balance tissues in order to provide relief over time with the body adapting to the new homeostatic state.

Chronic pain and disability lead to psychosocial challenges for EDS patients. Many of them suffer from anxiety, depression, anger, and, in general, frustration with the medical system. Patients are commonly referred to a range of specialists prior to accurate diagnosis. Many patients remain undiagnosed for years which leads to frustration. They have concerns about their relationships, social activities, and reproductive health. A significant percentage of patients have a history of use of mental health care. This chronic condition requires patients to cope with the disease over their lifespan and special attention should be given to the psychosocial well being of these individuals. These patients could also benefit from behavioral and cognitive therapy such as hypnosis and relaxation. It has been shown that patients who have worked full-time accepted their disability to a greater extent than patients who were on disability. Patients who accepted their disability also had a greater sense of coherence and better functionality.

Summary

There is paucity in osteopathic literature on the management of Ehlers Danlos Syndrome. Treatment of patients with EDS proves a challenge, as there is no cure for the condition. These patients need several treatment modalities simultaneously to take care, not only of their chronic pain, but the often equally important aspect of psychosocial well being. A multidisciplinary approach is required to help the patients limit their disability, cope with the disease, and rehabilitate them in their chosen profession.

The patient we saw exhibited many of the classic issues of EDS. She was in pain and was frustrated with the lack of cure and loss of employment. In order to manage her symptoms, she should be prescribed heel lift of the appropriate size. With the prescribed lifts, her sacral tilt should improve, which in turn should improve her mild scoliosis. Her significant right knee pain could be secondary to her surgery, but it could also be due to a longer right leg. A heal lift in the left shoe should remove the added stress on the right leg and improve the pain in right knee and hip. OMT should help the tissues and ligaments to adapt to changes in the posture following the use of a heal lift. Continuous assessment of the patient’s pain and specific treatments to alleviate the pain, should go a long way in making the patient comfortable.

EDS is not a rare phenomena. It is estimated to occur in about 1 in 5000 people, regardless of race or gender. Of the known types of EDS, hopefully the biochemical disorders will be amenable to treatment in the future. Until then, osteopathy provides the tools and techniques to help patients cope with the ramifications and complications of Ehlers Danlos Syndrome.

Bibliography


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