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Ehlers Danlos Syndrome

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Continuing Education Activity

Ehlers-Danlos syndrome (EDS) is a genetic disorder affecting collagen formation and function. It affects virtually every organ system, which can result in significant morbidity and mortality. Complications of this disease include arterial rupture, organ rupture, joint dislocation, chronic pain, and fatigue, among many others. Proper diagnosis of EDS is essential to improving the overall health and well-being of affected patients as well as mitigating these complications. This activity outlines the background, presentation, evaluation, and management of Ehlers-Danlos syndrome and its complications. It further highlights the role of an interprofessional team in ensuring the best patient outcomes.

Objectives:

- Review the diagnostic approaches and the importance of proper identification of this condition.
- Describe the various differential diagnoses for Ehlers-Danlos syndrome.
- Summarize the multitude of disease complications of Ehlers Danlos syndrome.
- Outline the importance of an interprofessional and collaborative approach to the care of patients with Ehlers-Danlos syndrome (EDS) to improve overall patient well-being and satisfaction in the face of this chronic disease.

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Introduction

Ehlers Danlos syndrome (EDS) is a group of hereditary connective tissue disorders that manifests clinically with skin hyperelasticity, hypermobility of joints, atrophic scarring, and fragility of blood vessels.[1][2] It is largely diagnosed clinically, although identifying the gene encoding the collagen or proteins interacting with it is necessary to identify the type of EDS. Identifying the type of EDS to guide management and counseling is important.[1] In 2017, a new international classification of EDS was proposed with 13 different variants.[3][4] This syndrome is heterogeneous and has been classified into six types (classical, vascular, hypermobile, arthrochalasis, kyphoscoliotic, and dermatosparaxis), with the causative collagen pathology being different for each type.

Asymptomatic, nonsyndromic joint hypermobility, Ehlers-Danlos syndrome, and hypermobility spectrum disorders (particularly the hypermobile type) are the commonest phenotypes associated with joint hypermobility. Compilations of these syndromes can be chronic pain, dysautonomia, gastrointestinal dysmotility, mast cell activation, and anxiety and phobic states.[5] Many new variants have been identified with the advancements, which tend to be more complex and clinically overlapping.[6] Clinical recognition of EDS variants is important. For instance, it is important to know that the patient has the vascular type, which is associated with arterial rupture and organ perforation, with potentially

life-threatening consequences.[7]

Etiology

Ehlers-Danlos syndrome is an intriguing group of inherited collagen and extracellular matrix protein disorders with a wide array of phenotypic expressions. EDS is most often an autosomal dominant trait, but up to 50% of patients can have a de novo mutation.[8] EDS has long been characterized by subtypes using the 1997 Villefranche classification system, an update from the original Berlin system initially published in 1988.[9] It is noteworthy that this is a collection of syndromes with similar genetic etiologies that have become more clearly described in the literature over the last several decades. In 2017, the International EDS Consortium published a new international classification system to replace the outdated numerical Villefranche classification. Due to the heterogeneity and overlap of the clinical presentation of the EDS subtypes, the goal of the new classification system was not only to describe suggestive clinical criteria for each subtype but also present data for genetic and molecular diagnostic confirmation for all subtypes except the hypermobile type.[2]

There are 13 subtypes described in the 2017 classification system.[2] The top 5 and most clinically significant subtypes (based on incidence and their respective etiologies) will be in focus here.

Classical EDS involves an autosomal dominant (AD) inheritance pattern, and associated mutated genes include COL5A1 and/or COL1A1, which code for type V and Type I collagen, respectively. Major clinical criteria include atrophic scarring, skin hyperextensibility, and generalized joint hypermobility. Minor clinical criteria include epicanthic folds, skin fragility, soft “doughy” skin, easy bruising, hernia, complications of joint hypermobility, molluscoid pseudotumors, subcutaneous spheroids, and a family history of a first-degree relative who is affected by and meets the same clinical criteria.[2]

Classical-like EDS involves an autosomal recessive (AR) inheritance pattern and is associated with a mutation in the TNXB gene, which codes for tenascin XB. Major clinical criteria include skin hyperextensibility without atrophic scarring, generalized joint hypermobility, and easy bruising. Minor clinical criteria include non-cardiogenic lower extremity edema, mild muscle weakness, atrophy of hand and foot muscles, axonal polyneuropathy, foot deformities, and cavitary prolapse (uterine, vaginal, rectal).[2]

Cardiac-valvular EDS involves an AR inheritance pattern and is associated with mutations in the COL1A2 and/or NMD genes, which code for type I collagen. Primary clinical criteria include skin hyperextensibility, atrophic scarring, easy bruisability, restricted or generalized joint hypermobility, and progressive cardiac-valvular problems. Minor clinical criteria include foot deformities, pectus deformity, joint dislocations, and inguinal hernias.[2]

Vascular EDS involves an AD inheritance pattern and is associated with mutations in the COL3A1 and/or COL1A1 genes, which code for type III and type I collagen, respectively. Major clinical criteria include arterial rupture at a young age, uterine rupture (specifically third trimester with no risk factors), the formation of a carotid-cavernous sinus fistula without trauma, and a family history confirmed via genetic testing. Congenital hip dislocation and spontaneous pneumothorax are two of the minor criteria that should be reasonably identifiable to employ further diagnostic measures.[2]

Hypermobile EDS involves an AD inheritance pattern and has no known associated gene mutation(s) as of the publication of the 2017 classification system. This subtype is entirely a clinical diagnosis that must meet three strict criteria. The specific diagnostic detail of these criteria goes beyond the scope of this review.[2]

Epidemiology

The prevalence of Ehlers-Danlos syndrome is estimated to be between 1 in 5000 and 1 in 100,000, based on the EDS subtype, but this could be an underestimation.[6] The accurate prevalence of the different EDS subtypes is still not known. In our 2019 review of the literature, epidemiologic data based on the new 2017 classification system is minimal as most of the data is based on the 1997 nosology, in which genetic etiology was not yet a mainstay for

diagnosis. What can be delineated is that hypermobility type EDS is the most common subtype, with an incidence between 1 in 10000 and 1 in 15000.[10]

EDS classical type is estimated to affect 1 per 10000 to 1 per 20000. Aside from the common subtypes, most others are exceedingly rare, with specific subtypes like EDS kyphoscoliotic type identified in about 60 total patients and EDS dysfibronectinemic type identified in only one single family. However, it is important to note that there is likely underestimation of the overall incidence of EDS as the more mild presentations with only minimal joint or skin involvement may not have clinically significant disease and, therefore, may not seek medical care. If they do, subtle signs and symptoms can be difficult to diagnose, and genetic testing is not indicated or pursued.[8][2][11][12]

Pathophysiology

The pathophysiology of most Ehlers Danlos syndrome subtypes involves heritable mutations in collagen synthesis and/or processing. The inheritance pattern of these mutations is variable, including autosomal dominant and recessive inheritance involving different mutations; however, it is worth noting that there are reports of spontaneous mutations causing identical genotypes and phenotypes. The collagen affected by these mutations is integral to every body system, from the skin to the integrity of the vasculature, and as such, the symptoms of the disease can be variable and widespread, as discussed throughout this manuscript.[8][2][11][9]

The collagen defect is identified in at least six of the variants of EDS. The vascular type, sometimes referred to as type IV, is secondary to a decreased amount of type III collagen. It is caused by genetic mutations in the *COL3A1* gene resulting in excessive connective tissue fragility causing arterial, uterine, and intestinal ruptures and premature death.[13][14] Types V and VI are caused by deficiencies in hydroxylase and lysyl oxidase, important posttranslational modifying enzymes in collagen synthesis. The deficiency of amino-terminal procollagen peptidase characterizes type VII. Type IX is caused by abnormal copper metabolism, and nonfunctioning plasma fibronectin is found in type X.

In Ehlers-Danlos syndrome types I and II, causative mutations may include the *COL5A1, COL5A2,* and *tenascin-X* genes and are suggested to be in the *COL1A2* gene. Nevertheless, in most families with autosomal dominant EDS, the disease seems to be associated with loci that bear the *COL5A1* or *COL5A2* genes. A significant number of the mutations cause low levels of mRNA due to the mutant allele as a result of nonsense-mediated mRNA decay.[15]

Wenstrup et al. observed haploinsufficiency of the *COL5A1* that encodes type V collagen in the classic type of EDS. 8 of 28 probands with classic EDS had complete or almost complete loss of expression of 1 *COL5A1* allele. One-third of patients with classic EDS were observed to have mutations of *COL5A1,* resulting in haploinsufficiency. These findings indicate that the normal synthesis of the heterotypic collagen fibrils containing types I, III, and V collagen needs the expression of both *COL5A1* alleles.[16] In classic Ehlers-Danlos syndrome, type V collagen mutations are pivotal.

Autosomal recessive–type VI EDS, also referred to as the kyphoscoliotic type, manifests as neonatal kyphoscoliosis, widespread joint laxity, severe muscle hypotonia, and skin fragility at birth. Biochemically, the deficiency of lysyl hydroxylase (LH) is responsible for this type. More than 20 mutations have been identified in the *LH1* gene causing LH deficiency and clinical EDS type VI.[17]

History and Physical

Patient presentations vary widely based on the respective underlying subtype; however, certain features of this condition are essential for the practitioner to identify and appropriately investigate to aid in diagnosis and progression of care. The biochemical collagen pathology is present at birth; however, clinical manifestations become apparent later. Skin hyperextensibility and hyperflexible joints are two of the most common presenting signs of EDS.[2]

Cutaneous manifestations are the hallmark of Ehlers-Danlos syndrome. The most common ones are hyperextensibility, smooth and velvet-like texture, fragility, delayed wound healing, and thin atrophic scars after wound

healing.[8] Shoulder dislocation is usually the initial sign of EDS. The skin features provide the diagnostic criterion of EDS, but there is no supportive laboratory test.[18] Children with this type tend to have various complaints, particularly orthostatic intolerance, diarrhea, urinary incontinence, impaired postural control, pain, and fatigue.[19] Often, muscle weakness is a prominent feature, with patients having a tendency to fall. Sometimes, patients report difficulty walking.

Mental development is generally normal.

Dental pathologies are commonly observed in these patients, such as hypodontia of permanent teeth, delayed eruption, and dentin dysplasia.[20] A lack of attached gingiva could be a pathognomonic feature.[21] Thus, dentists have a crucial role in early diagnosis and management.[22]

Common musculoskeletal manifestations include hypermobility leading to repeated subluxation and dislocation, leading to early osteoarthritis and chronic pain. This hypermobility may manifest as early as peripartum with hip dislocations in the newly delivered infant. Recurrent fractures may also be present.[9]

Other tissues are susceptible to friability due to underlying collagen dysfunction, including hollow and solid internal organs. They can be subject to both spontaneous and traumatic rupture or perforation. Additionally, hernias and rectal prolapse are common features.[8][9]

Physical Examination

The skin is usually white in color and soft to the touch, and underlying vessels can become apparent. The skin has a doughy feel and is easily hyperextensible. It is easily stretchable and immediately returns to its original state after release. Molluscoid pseudotumors are small, spongy outgrowths observed over scars and pressure points. They are commonly found in patients with type I EDS.

Smaller, deep, and movable nodules are often palpable in the subcutaneous tissue. They can be observed in the arms and over the tibia. Radiography may reveal calcification. The fragility of dermal skin with frequent bruises and lacerations is common. The joints are hyperextensible, but the degree of involvement varies. The digital joints are most commonly influenced, but alterations can be present in all the joints.

Evaluation

Diagnosis largely has its basis in identifying a collection of symptoms previously described that alert the practitioner to the possibility of Ehlers-Danlos syndrome. Following that suspicion, an initial evaluation of a patient's complaints should be tailored to the specific system identified to determine if they are indeed consistent with EDS. The initial workup after diagnosis of EDS should focus on determining the extent to which the underlying pathology affects the body. A recommended approach from Malfait et al. for classic-type EDS includes a thorough examination of the skin followed by applying the Beighton criteria. In infants and children, motor development should be assessed. A baseline echocardiogram for children under age 10 is also recommended. If easy bruising is a concern, evaluation of clotting factors should be considered to determine if another concomitant bleeding disorder is present.[8]

Skin hyperextensibility is evaluated and differentiated from other etiologies such as cutis laxa by assuring that easily stretched skin immediately returns to its original shape and form following manual manipulation.[8]

If and when EDS is suspected, CT, MRI, and echocardiography can be utilized to evaluate for common cardiovascular concerns such as mitral valve prolapse and aortic dilatation. MRI may show white matter lesions in line with the diagnosis of EDS.[23][24]

Bloodwork is generally not helpful for diagnosing EDS, though some specialized laboratory studies may be positive. It is worth noting that patients who present with easy bruising and bleeding will have normal coagulation studies and bleeding times unless another underlying disorder exists. Although rare reports of hemophilia A have been observed in the vascular type EDS.[25]

Ultimately, referral to a geneticist who can perform diagnostic genetic testing is recommended for specific subtype-level diagnosis. Additionally, tissue biopsy with electron microscopic analysis to evaluate for classic abnormalities in the appearance of collagen can be performed. However, this technique is seldom utilized.[8][2][9] Further evaluation is guided by presenting symptomatology. For example, if the cardiovascular workup is negative in a patient with suspected EDS and complaints of pre-syncope are present, a tilt table test may be ordered to evaluate for an associated syndrome such as postural orthostatic tachycardia syndrome (POTS) or another dysautonomia.[26][27]

Special consideration is necessary for EDS patients in the setting of trauma, given the increased risk for joint dislocation and vessel and organ rupture. Connective tissue disorders such as EDS merit consideration in young patients with physical exam findings out of proportion to reported trauma. Of course, non-accidental trauma must also be a consideration and ruled out.[28]

Treatment / Management

Any provider caring for a patient with Ehlers-Danlos syndrome should be aware of the multitude of complications of the disease and potential preventative measures. Treatment and management of patients with EDS should use a multidisciplinary approach that focuses on preventing disease progression and subsequent complications as there is no cure for the disease. Specialists generally manage specific care within the field of which the patient has concerning pathology. For example, cardiovascular concerns will be monitored by a cardiologist; likewise, musculoskeletal pathology is monitored and treated by an orthopedist. Often, a geneticist or family medicine provider acts as the primary provider referring the patient to these specialists.[8]

The underlying collagen abnormalities predispose the patient's skin to poor wound healing and dehiscence.[9][29] Therefore, wound repair should be meticulous, including the use of deep closures, and sutures should remain in repaired tissue for extended periods.[12][29]

With hypermobile joints comes the increased risk of joint subluxation and dislocation.[8][2] Prevention is the most crucial aspect of medical management as each injury increases the likelihood of recurrence and complications such as osteoarthritis. Prevention revolves around robust patient counseling regarding limiting at-risk activities like contact sports or weightlifting.[8][12]

Cardiovascular screening should be done regularly to mitigate risk factors as best as possible. Hypertension, for example, increases strain on already fragile vasculature and increases the risk of complications over time and should be managed aggressively. Screening for structural abnormalities of the heart, such as aortic root and mitral valve abnormalities, should be conducted using echocardiography as these may require surgical intervention to avoid the complications of rupture or congestive heart failure, respectively.[24]

Pregnant EDS patients should be followed and managed by Obstetricians trained in high-risk pregnancy. See "Complications" for further information.[29]

Differential Diagnosis

Osteogenesis imperfecta (OI) type I can be confused with some Ehlers-Danlos syndrome subtypes due to significant morbidity following minor trauma and joint hypermobility. However, a combination of symptomatology, in addition to genetic testing, can distinguish the two disease processes. Many patients with non-deforming OI (OI type I) present with blue sclerae, sensorineural deafness, wormian bones, and/or dentinogenesis imperfecta. It bears mentioning that patients with either disorder may present with the blue sclera. Also, mutations can be present in the COL1A1 and COL1A2 genes. Other OI subtypes are not part of the differential.[30]

Marfan syndrome shares many similar characteristics with different subtypes of Ehlers-Danlos syndrome (specifically the hypermobility subtype); however, the overall diagnosis of Marfan syndrome can be clinically and genetically separated.[8][2] Genetically, mutations are often identified in the fibrillin-1 gene. Clinically, when joint hypermobility is present in a patient with marfanoid habitus, ectopia lentis, and/or aortic root pathology, both EDS and Marfan

syndrome should be suspected.[2] However, aortic root pathology, specially ectasia, is unlikely to progress in severity into adulthood, unlike the more common progression in MFS.[24] Additionally, elbow involvement is generally spared, and marfanoid habitus is generally more evident in MFS with arm span: height ratio greater than 1.05. Furthermore, genetic testing of MFS can reveal a mutation in the fibrillin 1 protein, which does not occur in EDS.[8][30]

Loeys-Deitz Syndrome can be confused with Ehlers-Danlos syndrome due to its autosomal dominant inheritance pattern and early diagnosis of aortic pathology, specifically aneurysms. However, there is a clinical triad that is characteristic of LDS that helps the clinician guide diagnosis. The triad includes bifid uvula/cleft palate, hypertelorism, and aortic aneurysm. More specifically, the aneurysms discovered in a patient with LDS can be detected throughout the arterial system rather than clustered around the aortic root, as is more commonly seen in certain EDS subtypes.[30][31]

Cutis laxa may be mistaken for Ehlers-Danlos syndrome on the initial exam. However, a distinction is possible from the integumentary exam. While many patients with EDS possess hyperextensible skin, their skin will almost immediately return to its original form following blunt manipulation or palpation. The skin of cutis laxa patients is characteristically known to return to its original form from distention slowly. Heart valve, vascular involvement, and hernia formation are three characteristics with significant overlap between the two disease processes. Genetic analysis of patients with cutis laxa will often identify mutations in the fibulin-5 gene.[8][30][32]

Patients with EDS, especially those with EDS hypermobility type, are often misdiagnosed with conditions such as fibromyalgia, chronic fatigue syndrome, or depression, given the overlap of symptoms and the psychosocial impact they have on the patient.[8][12][33] While these conditions may exist concomitantly, careful attention and a high index of suspicion should be applied, as misdiagnosis puts the patient at risk for the complications associated with EDS and not the alternate diagnoses mentioned.

Prognosis

Prognosis varies considerably based on subtype and whether morbidity or mortality is a more significant consideration. Regarding morbidity, it correlates highly with the patient's environment and subtype. If a patient with Ehlers-Danlos syndrome hypermobile type receives early education and heeds the warnings regarding the avoidance of potentially traumatic activities such as weightlifting, they may experience relatively low disease morbidity throughout their life. On the contrary, significant morbidity can be expected if that patient undergoes physical trauma, causing repeated joint injuries that may require surgical intervention.[12]

In general, the mortality of patients with hypermobile and classic subtypes is not affected by the disease. However, the lifespan of patients with vascular and kyphoscoliotic subtypes is significantly affected. A patient with the vascular subtype will likely experience a major vascular event by age 40 with a median lifespan of 48.[34][35] Similarly, patients with the kyphoscoliotic subtype experience vascular insults and even restrictive lung disease, which are both known to shorten life expectancy.[36]

In addition to the purely medical prognosis of Ehlers-Danlos syndrome, there is now more of an understanding of the psychosocial impact of the disease. Due to the lack of a cure and what can be seen by the patient as failed treatments by their provider, patients can lose faith in the healthcare system; this may discourage EDS patients from accessing needed healthcare as the disease progresses.[33] Although the treatments may have the desired impact, for example, beta-blockers for vascular protection, the patient often views them as failures because they are not feeling better in their day-to-day life, and the undesired outcome, vascular injury, may indeed be the eventual outcome regardless of their adherence to the treatment.

As with any chronic condition, EDS can have significant psychosocial implications. Not only due to the symptoms and sequelae of the disease itself but also from the activity restrictions recommended by clinicians meant to reduce risk to the patient. Also, patients report a lack of support from clinicians, especially when reported symptoms don't correlate

with visible or quantifiable pathology.[8][33]

Complications

Vascular and organ rupture are the two most feared and mortal complications of EDS. They are most common in the vascular and kyphoscoliotic subtypes but have been reported in the more common subtypes as well.[8][2][9][37] Vascular rupture can occur anywhere in the arterial system, most commonly in the thorax and abdomen. Organ rupture most often occurs in the gravid uterus, sigmoid colon, spleen, and liver.[35][37] Sudden onset of pain in the setting of suspected or known EDS should prompt emergent medical evaluation regardless of the presence or absence of trauma.

Mitral valve prolapse may exist with or without regurgitation.[8][2][24] The degree of regurgitation can range from mild to severe, though the more severe forms are less common. Mitral regurgitation may not be clinically significant until physiologic stress occurs (such as in pregnancy) and may present in the typical fashion with congestive heart failure and pulmonary edema.[24][29]

Due to underlying connective tissue abnormalities, patients have an increased risk of cervical incompetence, and labor can progress prematurely and/or precipitously. Vaginal births correlate with a higher risk of significant vaginal and perineal lacerations, which can increase both morbidity and mortality. Additional complications in the pregnant patient may include premature rupture of membranes, breech presentation, uterine and bladder prolapse, uterine rupture, and great vessel rupture.[8][9][29]

The risk of bleeding is much greater, given the previously discussed vascular fragility.[8] Lastly, pre-existing pathology such as a known mitral valve prolapse or aortic dilation should be monitored regularly during pregnancy for an acute worsening secondary to the physiologic changes associated with pregnancy. Birth injuries to both the fetus and mother are more common, including dislocations for the child and higher-grade injuries to the laboring mother. Despite these increased risks, it is unclear whether delivery via planned cesarean section over vaginal delivery to the patient or newborn is advantageous.[29]

It is essential to note that any surgical procedures, including those necessary for the prevention of life-threatening complications of Ehlers-Danlos syndrome, pose a significant risk of complications due to blood vessel and organ fragility, as well as prolonged healing time, among others. However, it is important to note that surgery, in general, poses an increased risk of complications in EDS patients due to prolonged healing time and increased bleeding risk.[9][12]

Orthostatic intolerance should prompt evaluation for vascular injury as it may be a sign of hypovolemia. However, a detailed history of the symptoms is especially important as orthostatic intolerance can be common in EDS patients, specifically hypermobile type, and may have a more benign etiology, such as low blood pressure or postural orthostatic tachycardia syndrome (POTS), a known association with EDS.[26] POTS is a minimally understood dysfunction of the cardiovascular autonomic system, primarily affecting young females. Patients who suffer from POTS will often seek medical care for palpitations, dizziness, and/or syncope. Some events or “POTS attacks” have identifiable triggers, like orthostatic changes or infections, but not all have known triggers. Runs of significant sinus tachycardia are commonly associated with symptoms out of proportion to the patient’s activity.[27]

Sudden onset of dyspnea in an Ehlers-Danlos syndrome patient should prompt emergent evaluation for pneumothorax.[2][9][38]

Repeated joint injuries can lead to early-onset osteoarthritis and potentially a multitude of orthopedic procedures, especially in hypermobile and classical subtypes. While these procedures may be warranted based on the insult, the understanding is that the laxity of the affected connective tissue increases over time, which may predispose to additional insult.[8][39]

Sleep disturbance and sleep-disordered breathing, termed genetic obstructive sleep apnea (OSA), has a known

association with EDS.[2] The causality of the association is postulated to be due to craniofacial soft tissue abnormalities.[40] Chronic fatigue may result, and the patient should obtain a referral to a sleep specialist.

Deterrence and Patient Education

The most critical aspect of patient education regarding Ehlers-Danlos syndrome is how to prevent or slow the progression of the disease optimally and what to expect as life progresses; this includes lifestyle modifications and adherence to physical therapy and other medical treatment. Avoidance of potentially traumatic activities and involvement in low resistance activity may decrease their risk of long-term musculoskeletal morbidity.[8][12][39] It is crucial to educate the patient that adherence to beta-blockers or other anti-hypertensive drugs may improve their prognosis regarding potential future vascular events.[35]

Enhancing Healthcare Team Outcomes

Patients with Ehlers-Danlos syndrome benefit most from an interprofessional healthcare team approach to their care. Care begins with clinical practitioners (MDs, DOs, NPs, or PAs) having a high index of suspicion to ensure an accurate diagnosis based on personal and family histories and a physical exam followed by appropriate genetic testing.[9] While there are no clear-cut evidence-based algorithms to direct management, experts in the field recommend enlisting the help of proper specialists based on the risk of disease complications, as discussed previously. As with any multi-system disease process, a knowledgeable primary care physician should have involvement in coordinating the care of the patient with EDS for both surveillance and treatment needs.

Arming the patient with the knowledge needed to identify worrisome symptoms and seek immediate medical care is invaluable. Universal recommendations for wearing a medical alert bracelet or another device should be given to any EDS patient with vascular pathology as these devices can help facilitate emergent medical care if necessary.

It's also important for the patient to understand that there is no cure for the disease, and as of now, no medical treatment can completely resolve their symptoms. This fact may be problematic for many patients to internalize and accept, which is why psychosocial support, including expectation management, for patients suffering from EDS is paramount in their care.[8][33] The Ehlers-Danlos Society, among others, aims to provide some support and educate its members about improving their quality of life and accessing proper medical care.

Specific guidance is necessary for patients regarding the genetic inheritance of the disease. Genetic testing can help delineate a patient's specific subtype and inheritance pattern. With the help of this information, patients will understand when making decisions regarding risks associated with their children.[8][29] The risks of pregnancy and labor should be discussed in-depth with women of childbearing age.[29] This is where a nurse with specialized training on the disorder can assist with counseling, answering questions, and explaining management options.

Psychosocial support is paramount for EDS patients, who experience many psychosocial challenges related to the variety of symptoms they suffer and the activity restrictions imposed by both their symptoms and as recommendations by clinicians. Open communication between interprofessional team members is vital if one wants to improve outcomes.[8][12][33] [Level 5]

Review Questions

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