EDSers United

Stay Strong

Stay Connected

2012 EDNF Learning Conference Notes

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The following notes are Nadia Bodkin's interpretation of the information presented at the 2012 EDNF Learning Conference.

This year's EDNF Learning Conference was a dramatic improvement from the 2011 EDNF Learning Conference. About 550 EDSers attended the conference from all over the United States, Canada, Colombia, Italy, Norway, and Australia. Last year, the EDNF had the largest turn out with 580 attendees. This year, the attendance number was roughly the same. The venue was way more accommodating in terms of size. There was a Hospitality Suite/Relaxation Room, which proved to be a great addition to the conference because the seminar schedule was intense.

Shane Robinson is the new Executive Director of the EDNF. He came on board in September of 2011, a month after last year's Learning Conference. Simply judging from the dramatic improvement of this year's EDNF Learning Conference to last year's speaks volumes to Mr. Robinson's affect on the overall improvement of the entire organization. Mr. Robinson is obviously a valuable addition to the staff and is no doubt directing the entire EDNF organization towards the right direction.

Elliot Clark's, Chairman of the Board of the EDNF, fundraising presentation was way more inspirational this year. He referred to us as "EDSers," not "sufferers." He delivered statements of a positive future, and showed sincere concern for our current situations. Mr. Clark did step up and admit that the EDNF has made some mistakes in the past, in which some have called them out on. However, they currently look to the future and have made some changes to the staff and have announced that the foundation is "fundamentally sound," unlike in the past. So the EDNF is no longer focused on what they need to do to "survive," but are now focused on what they need to do next. So what are they planning on doing next? The EDNF wants to develop an EDS Clinical Care and Research Center. This center will be a one-stop shop of various specialists and home to a central database for EDS research consisting of blood and tissue samples taken from EDSers throughout the world. To quote Elliot Clark, "If they want to treat us like an orphan, then we need to take over the orphanage" (2012 EDNF Learning Conference). This quote references the fact that Ehlers Danlos Syndrome is still considered an orphan disorder.

EDS Today & Tomorrow

Presented By: Brad Tinkle, MD, PhD, Chair Genetics/Pediatrics

EDS is a group of inherited (genetic) disorders of connective tissue. Because EDS is comprised of a group of disorders, you may see several different EDSers with ailments that you've never heard of.

There are 6 Classifications of Ehlers Danlos Syndrome:

1. Classic Type (Type I & II)
2. Hypermobile Type (Type III)
3. Vascular Type (Type IV)
4. Kyphoscoliosis Type (Type VI)
5. Arthrochalasia Type (Type VII A & VII B)
6. Dermatosporaxis Type (Type VII C)

Other Variants of EDS include:

- X-linked EDS (EDS Type V)
- Periodontitis type (EDS Type VIII)
- Familial Hypermobility Syndrome (EDS Type XI)
    - Benign Joint Hypermobility Syndrome
    - Hypermobility Syndrome
- Progeroid EDS
- Marfanoid habitus with joint laxity
- Unspecified Forms

It is important to note that some of above listed variants of EDS are from the older terminology and some don't exist in the current classification scheme (Dr. Tinkle did not specify which variants are from the older terminology or don't exist in the current classification scheme). Dr. Tinkle did, however, explain that Joint Hypermobility Syndrome is now being considered to be the same as EDS Type 3, thanks to the work of Dr. Grahame.

<u>The **Beighton** Criteria for Generalize Joint Hypermobility</u>
(one of the most well recognized and used criteria for EDS)

1. Passive dorsiflexion of 5th digit to or beyond 90°
2. Passive flexion of thumbs to the forearm
3. Hyperextension of the elbows beyond 10°
    a. >10° in females
    b. >0° in males
4. Hyperextension of the knees beyond 10°
    a. Some knee laxity is normal

   b. Something difficult to understand posture-forward flexion of the hips usually helps
5. Forward flexion of the trunk with knees fully extended, palms resting on floor
   a. May be negative in those with flat feet and tight hamstrings

Dr. Tinkle sometimes suggests to his patients that upon entering an emergency room, to show that they are hypermobile by flexing the thumbs to the forearm, etc. Dr. Tinkle claims depicting yourself as a hypermobile patient upon arrival to an emergency room is just as important as telling them your name. You want them to know that your joints work a little different than they may think.

Beighton's Scores vary based on age, race/ethnicities, sex, and injuries. According to Dr. Tinkle, the overall scoring system has a lot of caveats. And physicians using the Beighton Criteria for Generalized Joint Hypermobility need to realize that it is simply a guideline and that they essentially need to think past these guidelines. If patients have had injuries at given joints, they may not be as hypermobile at that joint that suffered an injury. It is important that physicians using the Beighton Critieria incorporate previous injuries into the evaluation. Essentially, physicians have to think beyond these guidelines.

Ehlers-Danlos Syndrome Etiology

EDS Type	Genetic Defect	Inheritance
Classical	Type V collagen (60%)	Dominant
Hypermobile	Largely unknown	Dominant
Vascular	Type III collagen	Dominant
Kyphoscoliosis	Lysyl hydroxylase (PLOD1)	Recessive
Arthrochalasia	Type 1 collagen	Dominant
Dermatosporaxis	ADAMTS2	Recessive

Some other conditions/disorders that look like EDS are Brittle cornea syndrome (PRDM5; ZNF469), Spondylocheiro dysplastic, Musculocontractural/adducted thumb clubfoot/Kosho (D4ST1 deficient EDS), and Tenascin-X deficiency.

The 2012 International Symposium in Belgium will look at possibly reclassifying (and even renaming) the different types of EDS and perhaps possibly even add some new types of conditions (like those listed above that appear to "look like" EDS) to the classification list.

Clinical Features of the most common EDS Types
(Classical & Hypermobile)

CLASSICAL EDS

- Major Diagnostic Criteria include:
    - Skin hyperextensibility
    - Widened atrophic scars
        - Atrophic scarring is often pre-tibial. It develops early in childhood from the "bumps and bruises," that we all develop from just being children. A lot of the time, child services gets involved since the fragility and friability of the skin may appear, to the untrained eye, to be child abuse.
        - Cigarette Paper Skin is the descriptive term for markedly attenuated skin with a shiny, velvety surface, which is typical of classical EDS. "Mild" Classic EDS may not have atrophic scars initially until injury or surgery. Therefore, it may be difficult to distinguish from the hypermobile type.
    - Generalized joint hypermobility
    - Inheritance is autosomal dominant

- Redefining Classical EDS
    - Using all of the major criteria, more than 90% of classical EDS patients had mutations in the type V collagen genes (COL5A1/COL5A2). Which is a great probability, when it comes to genetic testing. It has been proposed that patients with classical EDS only be defined with these criteria, making those with milder features that overlap with type 3 (hypermobility) likely to be grouped together. Thus, EDS Type 2 may actually be re-classified as EDS Type 3 and no longer under the classification of Classical EDS.

HYPERMOBLE EDS

- Major Diagnostic Criteria
    - Mild skin hyperextensibility with velvety texture
        - This criterion is highly subjective. Doctors constantly disagree with each other on what they interpret as soft and velvety skin. A big problem arises when doctors reject the diagnosis of EDS based solely on the fact that they perceive the patient's skin as being "normal," regardless of the patient exhibiting all the other signs and symptoms of EDS.
    - Normal or near-normal scarring
    - Generalized joint hypermobility
    - Inheritance is autosomal dominant

  - Genetic causation is unknown
  - Incidence is greater than (or equal to) 1 in 5,000

- Minor Diagnostic Criteria (what an EDSer doesn't necessarily need to have, but is found to occur amongst 10% of EDSers)
  - Recurring joint dislocations
  - Chronic joint/limb pain
  - Positive family history

- The 1998 Revised **Brighton** Criteria (for Hypermobility Syndrome)
  - Major Criteria
    - Beighton score of 4 (either currently or historically)
    - Arthralgia for 3 months in 4 joints
  - Minor Criteria
    - Beighton score of 1,2, or 3 (0-3 if age 50 years or older)
    - Arthralgia (3 months) in 1-3 joints or back pain (3 months)
    - Spondylosis, spondylolisthesis
    - Dislocation/subluxation in 1 joint, or in 1 joint on 1 occasion
    - Soft tissue rheumatism 3 lesions (e.g., epicondylitis, tensosynovitis, bursitis)
    - Marfanoid habitus (tall, slim, span:height ratio 1.03, upper:lower segment 0.89, arachnodactyly)
    - Abnormal skin:striae, hyperextensibility, thin skin, papyraceous scarring
    - Eye signs: drooping eyelids or myopia or antimongoloid slant
    - Varicose veins or hernia or uterine/rectal prolapse

  Note: The **Brighton** Criteria is different from the **Beighton** Criteria.
  The diagnosis of benign joint hypermobility syndrome (BJHS) requires the presence of 2 major criteria, or 1 major 2 minor criteria, or 4 minor criteria, or 2 minor criteria and an unequivocally affected first-degree relative. BJHS is excluded by the presence of Marfan's syndrome or Ehlers-Danlos Syndrome (EDS), other than hypermobility-type EDS.

  The term "Benign" doesn't mean the absence of pain. The use of the term "Benign" is also going to be re-considered at the 2012 International Symposium in Belgium.

Hypermobility, Fibromyalgia, & Chronic Pain

Presented By: Rodney Grahame CBE, MD, FRCP, FACP

Joint Hypermobility Syndrome (1967) was thought to be a rheumatologic issue diagnosed and treated by a Rheumatologist. Ehlers-Danlos Syndrome (1968) was thought to be a completely separate condition diagnosed by a geneticist. Research over the years has shown that Joint Hypermobility Syndrome and EDS are the same.

In 1967, Hypermobility Syndrome (HMS) was originally described. The main description included musculoskeletal pain due to joint instability. As the years went by, new evidence came forward about various symptoms associated with HMS. The following is a table that describes the progression of associating symptoms to HMS.

Year	Symptom
1967	Musculoskeletal pain/ join instability
1970	Overlap with HDCT/Skin/Habitus
1980	Urine/Rectal prolapse
1990	Chronic pain syndrome Anxiety/Phobias
2000	Dysautonomias GI Dysmobility
2010	Progressive Disability

According to Dr. Grahame's collected data for the last decade, the prevalence of rheumatologists that understood HMS (also known as EDS) was 45%. Another interesting point that emerged was that the prevalence of EDS in males is lower than in females; and the prevalence of EDS in non-Caucasian females is higher than Caucasian females.

Dr. Grahame does not believe EDS is an orphan (rare) disorder…."This is not an orphan disease; this is an everyday condition. I personally see about 18 new cases per week."

Clinical Features of Joint Hypermobility Syndrome (JHS)

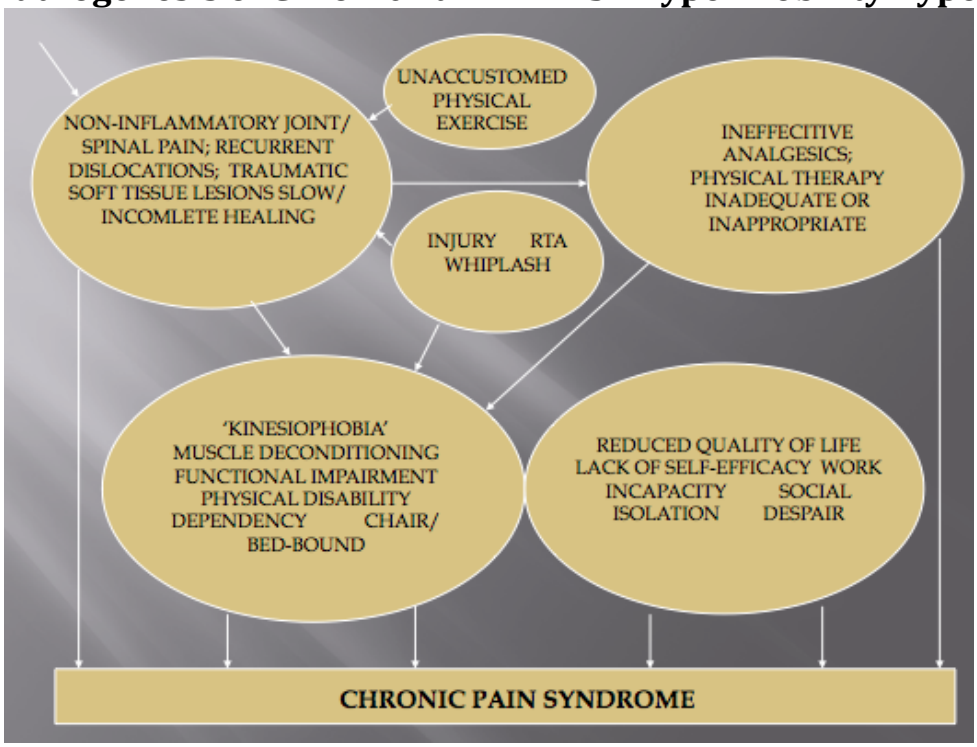
Joint	Soft Tissue	Spine	Extra-articular
Hip Dysplasia Late walking Growing Pains Arthralgia/Myalgia Dislocation/Subluxation Joint Synovitis Chondromalacia Patellae Osteoarthritis	Ligament/Muscle/Meniscustear Epicondylytis Tendonitis/Capsulitis Tenosynovitis Entrapment Neuropathy Baker's Cyst Fibromyalgia	Loose-Back Syndrome Disc Prolaps Pars Defects Spondylolysis – Olisthesis Spinal anomalies Spinal stenosis S.I. Joint Instability Dysautonomia	Stretchy skin Thin scars Hernia Vericose Veins Fractures Uterine/Rectal Prolapse GI Dysmotility Chronic Pain Syndrome Depression Anxiety

Fibromyalgia was coined in the 1990's and was very much a favored diagnosis amongst Rheumatologists in North America. Fibromyalgia is another way of saying chronic pain.

Acute vs. Chronic Pain

Acute	Chronic Pain
Sudden injury	Not injury related
Overuse injury	Not degree of H/M-related
Severe Pain	Gradual build-up
Self-limited	Distribution non-anatomical
Responds to: - Heat/Cold - Rest - Analgesics	Associated with: - Fatigue - Depression - Diminished QOL (Quality of Life)
	More diffuse
	Un-responsive to analgesics Takes the Edge of the pain

Pathogenesis of Chronic Pain in EDS –Hypermobility Type



A theory is that this chronic pain is increased…it is amplified. In fibromyalgia, the pain signal is distorted and as a result amplified. So the pain in hypermobile EDS is disproportionate and misleading because it makes the patient feel like something really wrong is happening to your body. This pain leads to Kinesiophobia, which is the fear of movement. If movement causes pain, people will intuitively avoid movement to avoid pain. This is understandable, but not good for hypermobiles because you loose muscle function making it more difficult to move and balance, causing even more joint instability, which in itself can lead to more injury. Since a lot of doctors can't figure it out, they assume that chronic pain is psychogenic.

Fibromyalgia is not a specific entity, which it is largely regarded as, but more of a manifestation of a condition, a symptom.

The frequency of joint hypermobility syndrome in chronic pain patients is between 40-50%.

"Chronic pain is a manifestation of EDS" –Sachetti et al (1997) Journal of Chronic Pain 14(2): 88-93

> In this study about half of the participants suffered from frequent headaches and a third suffered from intermittent abdominal pain. 40 out of 45 adults reported having symptoms began in childhood or adolescence. 70% of

EDSers suffered from sleep disturbance; 70% of EDSers also reported impaired physical activity. About 45% of EDSers reported impaired sexual function.

In conclusion, EDSers experience frequent and severe lifetime pain, which is unfortunately unrecognized in medical literature. A pain management protocol is desperately needed. And above all, EDS should ALWAYS be considered in the differential diagnosis of chronic musculoskeletal pain.

The Natural History of EDS/JHS

First, the EDSer exhibits musculoskeletal tissue laxity. This involves non-inflammatory joint/spinal pain; dislocations/subluxations; ligament muscle, tendon, enthesis injury/overuse; flat feet; and pelvic floor, herniae, varicose veins.

Second, the EDSer may exibit non-articular symptoms. This involves pain amplification; Kinesiphobia; deconditioning widespread chronic pain (fibromyalgia); fatigue; orthostatic intolerance; and postural tachycardia (POTs).

Third, the EDSer may be introduced to a psychosocial sequellae. This involves anxiety/depression; obesity; work incapacity; isolation; and dispair.

EDS is the new Rheumatological Disability! A lot of EDSers are severely physically disabled, regardless of their muscular skeletal system being largely intact. Chronic pain is a very serious ailment that often leads to Kinesiophobia. And a majority of EDSers suffer from pain that is unresponsive to analgesics including opiates. A lot of EDSers are mostly young and highly motivated individuals that are cut down in their prime. Unfortunately, EDSers are often told that it is all in their minds causing them to feel dispirited, abandoned, angry, and desperate. Most EDSers require intensive physical rehabilitation and pain management (CBT). EDS-dedicated programs are now available at UCH and KNOH.

The COPE Program (Changing Outlooks on the Pain Experience) helps people manage their pain and reduce its effects on their lives. It is important that every EDSer understand chronic pain and EDS. The COPE Program can help EDSers reduce pain related distress, especially fear; reduce avoidance and engage in activity; manage increases in pain; and gain greater independence in health care. The programs aim is to change the relationship to the pain. It is not a cure for pain. The entire length of the program is a total of 42 hours (involves 6 weeks and 8 days). Groups of 9-10 EDSers with chronic pain meet with a team comprised of 2

clinical psychologists, 1 physiotherapist, 1 nurse and 2 rheumatologists. Outcomes are assessed using standard questionnaires with a one month follow-up.

Common Misdiagnoses

Adults	Children
Fibromyalgia	Congenital hypotonia
Osteoarthritis	Laziness
Seronegative arthropathy	School phobia
Psychogenic rhematism	Dysfunctional family
Depression	Fabricated for induced illness (FII) aka Munchausen's Syndrome by Proxy
Chronic Fatigue Syndrome	

Living with Pain and Ehlers-Danlos Syndrome

Presented By: Clair A. Francomano, M.D.

Chronic pain in Ehlers-Danlos Syndrome is common, severe, and associated with functional impairment. Both pain and fatigue are important possible determinants of disability in patients with EDS hypermobility type.

When comparing Hypermobile EDSers to patients with rheumatoid arthritis, the EDSers showed significantly higher levels of pain severity and life interference due to pain and a lower level of perceived life control. However, Hypermobile EDSers showed significantly lower levels of pain severity, life interference, and affective distress when compared to a group of Fibromyalgia patients.

Sources of Pain in EDS include: headaches, neck & back pain, muscle spasms, myofascial pain, and neuropathic pain. So many EDSers seek alleviation for headaches. It is important to discover what the underlying factor of the headache is, because the way we go about treating the headache will depend on what is causing the headache. We know that muscle spasms and myofacial pain can cause really terrible pain in the head. Increased Cerebral Spinal Fluid (CSF) pressure (hydrocephalus) as well as decreased CSF pressure (CSF leaks) can result in headaches. Temporo-mandibular joint dysfunction (TMJ) is very common in EDSers and should be investigated as a potential cause of headaches. Many EDSers with TMJ don't know that they have it because it isn't something that is readily recognized. In addition to headaches, many EDSers need to address their neck pain as well. Muscle spasms as well as Myofascial pain may be the cause of many EDSers' neck pain. Disc disease should also be taken into consideration because many teen EDSers are actually already showing signs of deterioration. Syrinx (syringomyelia) as well as neuropathic pain should also be investigated as potential causes of neck pain for

EDSers. Joint pain is very common amongst EDSers and is in majority due to joint dislocations and subluxations. Myofasical pain (referred pain to the joints), meniscal tears, bursitis, tendonitis, and synovitis are all potential causes of joint pain that should be investigated. Arthritis is a secondary condition that many EDSers end up developing due to joint instability. Muscle pain is another very common issue for EDSers that can be caused my muscle spasms, myofascial trigger points, and muscle tears all over the body. Some EDSers may want to work on figuring out what their myofascial trigger points are and work on alleviating these points at home with the guidance of a physical therapist. Low back pain can result from muscle spasms, degenerative disc disease, neuropathic pain, syrinx, and/or tethered cord syndrome. Abdominal pain is a huge issue for many EDSers. Gastrointestinal dysmotility is a very common cause of abdominal pain. Micro-tears in the abdominal musculature is actually another cause of abdominal pain that gets overlooked a lot. There is actually a way that an individual can palpate the abdominal area and locate tears. Finally, mast cell disorders also aggravate and add to abdominal pain.

There are 3 general approaches to pain management: physical strategies, cognitive/behavioral strategies, and medications. In terms of physical strategies, the number one step to manage pain is physical therapy for muscle relaxation. This can be done via deep heat treatment, ultrasound, Transcutaneous Electrical Nerve Stimulation (TENS), and massage. Getting muscles relaxed is very important before doing any exercises for toning/strengthening of muscles. If the muscles are tight before exercising them, you risk tearing. All EDSers going to physical therapy should tell their physical therapist that they want to do some sort of activity that will relax their muscles first, before engaging in toning and strengthening exercise. If your physical therapist does not want to relax your muscles before exercising them, then get a new physical therapist because relaxation is KEY in helping to relieve pain. Myofascial trigger point release is another technique used to relieve pain. Dr. Francomano recommends "The Trigger Point Therapy Workbook – Your Self-Treatment Guide for Pain Relief" by Clair Davies. Pilates, Alexander, and Feldenkreis techniques are becoming very popular amongst EDSers and can be used to strengthen the core. Dr. Francomano recommends that EDSers use Epsom Salt baths/footbaths. Epsom Salts are magnesium salts and they absorb very well through the skin. One thing about taking magnesium orally, it can build up and cause diarrhea and abdominal discomfort. In terms of physical therapy, it is very important to stay active; but to avoid high impact activities. Avoid heavy weight lifting. Instead, use lower weights with more repetitions. Aquatic exercise is an excellent option and Dr. Francomano recommends it. Warm water can help relax muscles and provide a low impact environment for strengthening exercises. Dr. Francomano always recommends to all her patients to stay active and to shoot for 30 minutes of physical activity daily. Just be practical about your chosen physical activity and listen to your body. For example, you may be better on an elliptical

machine rather than a treadmill because of the lower impact on you joints. Cognitive-Behavioral therapy can be extremely helpful in coping with chronic pain. There are limitations to what we can do with medications and learning relaxation techniques and mindfulness-based stress reduction is really important when it comes to learning to manage chronic pain conditions. Medications aren't always the answer and we really can't rely solely on them. With that sayed, non-steroidal anti-inflammatories, muscle relaxants, anti-depressants, and analgesics like tramadol and some narcotics, can be helpful in managing chronic pain. Gabapentin has been shown to be very effective for managing nerve pain as well. Dr. Francomano really believes that narcotic use for chronic pain should be really be used short-term only to help individuals get over their hump and start moving and engaging in more activities. Dr. Francomano emphasized how she does not think long-term use of narcotics is the answer in terms of managing chronic pain. She thinks that EDSers should take advantage of the other techniques in terms of managing their pain. Dr. Francomano introduced a topical cream that she generally prescribes to her patients. It is called SEC "Formula H + Magnesium" and needs to be filled by a compounding pharmacy. The ingredients of this cream include: Diclofenac (anti-inflammatory), Baclofen (muscle relaxant), Cyclobenzaprine (muscle relaxant), Gabapentin (for neuropathic pain), Lidocaine (local anesthetic), Magnesium (for muscle relaxation), & Lipoderm base (for optimal absorption). Directions for this transdermal analgesic cream are as follows: Use 1-2 grams, 3-4 times daily. Apply heat to the area to be treated for 3-5 minutes. Dispense 1 gm (pre-measured dispenser). Rub in ½ gm for 30-60 seconds. Then rub in the second 3/2 gm for another 30-60 seconds. Wait 10-15 minutes to see if the pain is relieved. If not, then apply a second gram as need. A lot of her patients have told her that this has been a great product that relieves their pain effectively.

There is a reason that the KGB used sleep deprivation as a form of torture. Our bodies need restorative sleep!! Depression and chronic pain are closely intertwined. Addressing depression will help with chronic pain and vice versa. It is important to develop a strong mind in order to address the many issues that go on with our bodies.

Hypermobility: Getting Healthy, Improving Function & Strength

Presented By: Opal Riddle, PR, DPT

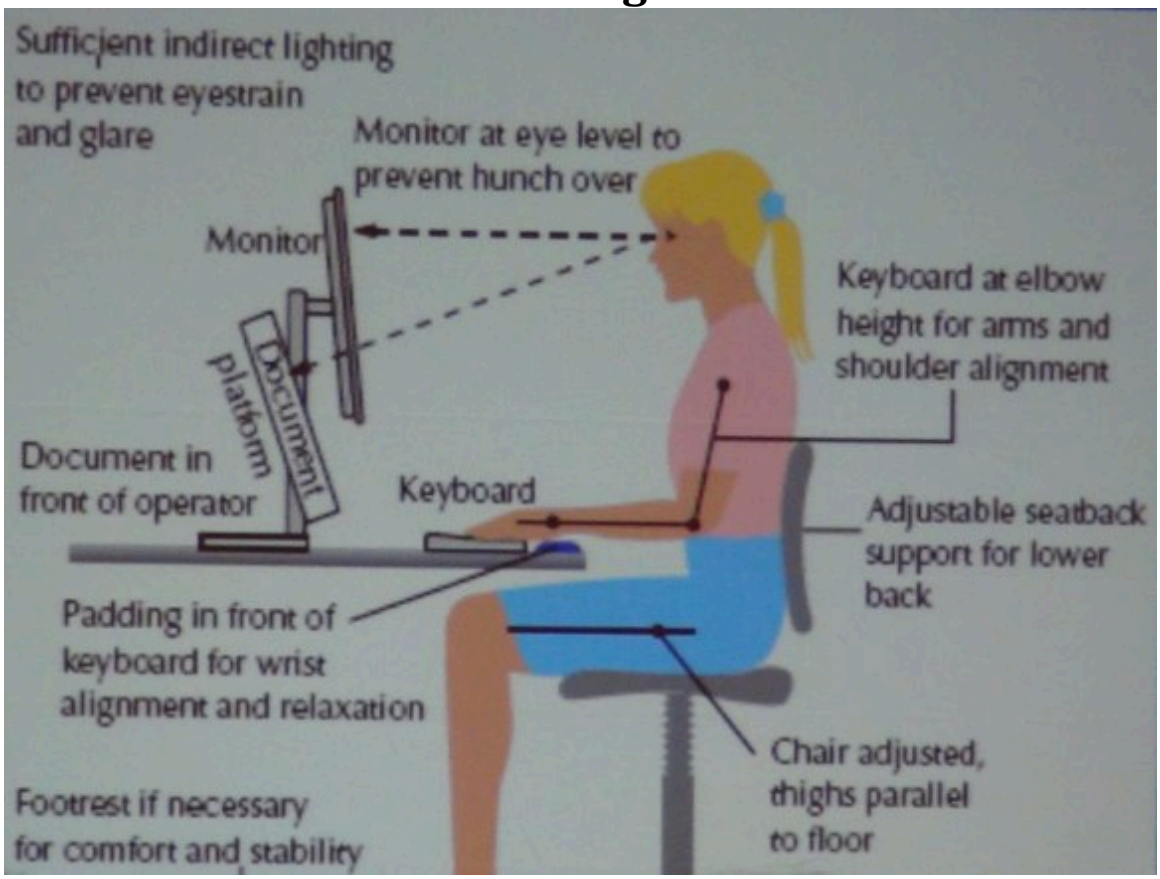
There are 2 types of **factors that contribute to joint stability**.

1. ***Static Contributors***: include bones, capsules (fluid filled sacks surrounding the joints) and ligaments
2. ***Dynamic Contributors***: include muscles, tendons and the nervous system which controls movement of the muscles

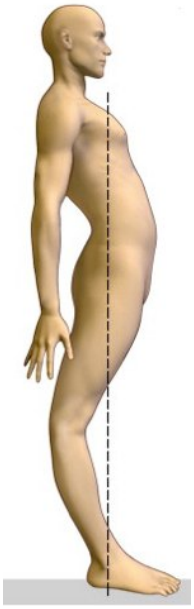
The main symptoms that are associated with JHS/EDS are pain, fatigue, weakness, early degenerative joint disease, headaches, poor sleep, dizziness/vertigo/POTS, nerve compression disorders, temporomandibular joint dysfunction, congenital conditions, recurrent sprains, subluxations, dislocations, and decreased bone density. According to Opal, bone density decreases as a result of being not very active as a child.

Education is the most important aspect of managing JHS/EDS in regards to having the ability to make appropriate decisions about the careers, sports, and activities that we choose to engage in. For example, symptoms associated with spinal/wrist hypermobility can improve with education in ergonomics and body mechanics as can joint protection can decrease pain/traumatic injury. The selection of jobs, sports and recreational activities is important to lessen exacerbation of the condition.

Sitting



Genu Recurvatum is when both knees are hyperflexed in the standing position.



Proper Lifting Techniques

- Squat to lift and lower. Do **not** bend at the waist.
- Keep your lower back bowed in while bending over.
- Keep the weight as close to you as possible.
- Bow your back in and raise up with your head first.
- If you must turn, turn with your feet –not your body.
- **Never jerk or twist.**
- Put the weight down by keeping your lower back bowed in.
- Keep your feet apart, staggered if possible.

Joint Stress During Activity

Activities	Hips	Knees	Ankles	Shoulders	Hands
Walking	•	•	•		
Swimming				•	•
Running	••	•••	•••		
Rowing	•	••	•	••	••
Climbing Stairs	•••	•••	•		
Cycling	•	••	•		
Tennis/Racquet	•••	•••	•••	•••	•••

Strengthening and proprioceptive exercises surrounding the affected joints is very important when it comes to functional stability re-training for controlling mechanical dysfunction.

Why should you exercise?

- Strengthens muscles, ligaments and tendons, which increases support
- Increases aerobic capacity/endurance
- Increases bone density
- Reduces dementia
- Reduces pain
- Increases balance and proprioception
- Reduces blood lipids
- Improves GI motility
- Increases metabolic rate
- Decreases depression and improves mood
- Improves self confidence and quality of life

We loose 5% of muscle strength in our 20's. By our 30's, the amount of muscle strength lost goes up to about 15%. That is why exercise is so important as we age. We must continue to exercise in order to increase the strength of our muscles, reduce pain, etc.

The BORG Scale of Perceived Exertion

One way to gauge how hard you are exercising is to use the Borg Scale of Perceived Exertion. The Borg Scale takes into account your fitness level: It matches how hard you *feel* you are working with numbers from 6 to 20; thus, it is a "relative" scale. The scale starts with "no feeling of exertion," which rates a 6, and ends with "very, very hard," which rates a 20. Moderate activities register 11 to 14 on the Borg scale ("fairly light" to "somewhat hard"), while vigorous activities usually rate a 15 or higher ("hard" to "very, very hard"). Dr. Gunnar Borg, who created the scale, set it to run from 6 to 20 as a simple way to estimate heart rate—multiplying the Borg score by 10 gives an approximate heart rate for a particular level of activity.

How you might describe your exertion	Borg rating of your exertion	Examples (for most adults <65 yrs old)
None	6	Reading a book, watching television
Very, very light	7 to 8	Tying shoes
Very light	9 to 10	Chores like folding clothes that seem to take little effort

Fairly light	11 to 12	Walking through the grocery store or other activities that require some effort but not enough to speed up your breathing
Somewhat hard	13 to 14	Brisk walking or other activities that require moderate effort and speed your heart rate and breathing but don't make you out of breath
Hard	15 to 16	Bicycling, swimming, or other activities that take vigorous effort and gets the heart pounding and makes breathing very fast
Very hard	17 to 18	The highest level of activity you can sustain
Very, very hard	19 to 20	A finishing kick in a race or other burst of activity that you can't maintain for long

Many different physical activities can improve your health and independence. The goal is to gradually work your way up to include endurance, strength, balance, and stretching exercises.

Here are some points to keep in mind as you begin increasing your activity:

If you stop exercising for several weeks and then return, start out at about half the effort you were putting into it when you stopped, then gradually build back up. Some of the effects of endurance and muscle-building exercises deteriorate within 2 weeks if these activities are cut back substantially, and benefits may disappear altogether if they aren't done for 2 to 8 months.

When an exercise calls for you to bend forward, bend from the hips, not the waist. If you keep your entire back and shoulders straight as you bend forward, that will help ensure that you are bending the right way, from the hips. If you find your back or shoulders humping in any spot as you bend forward, that's a sign that you are bending incorrectly, from the waist. Bending from the waist may cause spine fractures in some EDSers.

We can't tell you exactly how many pounds to lift or how steep a hill you should climb to reach a moderate or vigorous level of exercise, because what is easy for one EDSer might be strenuous for another. It's different for different EDSers.

We can, however, provide some advice based on scientific research: **Listen to your body.** Strength vs. Endurance Training all depends on what extent the patient has EDS.

For **endurance activities**, you should **gradually** work your way up to **level 13** - the feeling that you are working at a somewhat hard level. Some people might feel that way when they are walking on flat ground; others might feel that way when they are jogging up a hill. Both are right. Only *you* know how hard your exercise feels to you.

Strength exercises are higher on the Borg scale. **Gradually** work your way up to **level 15 to 17** - hard to very hard - to build muscle effectively. You can tell how hard an effort you are making by comparing it to your maximum effort. How hard does your current effort feel compared to when you are lifting the heaviest weight you can lift? Once you start exerting more than a moderate amount of effort in your muscle-building exercises, your strength is likely to increase quickly.

As your body adapts and you become more fit, you can gradually keep making your activities more challenging. You might find, for example, that walking on a flat surface used to feel like you were working at level 13 on the Borg scale, but now you have to walk up a mild hill to feel like you are working at level 13. Later, you might find that you need to walk up an even steeper slope to feel that you are working at

level 13.

The Borg scale is simple to use. But if your level of effort doesn't match the numbers you see on the Borg scale - for example, if you think you are doing the exercises correctly, but you aren't progressing or you are exhausted by your effort - check with a fitness professional. These experts are likely to understand the science that went into developing the Borg scale, and they can teach you how to match your level of effort with the right number on the scale.

Warm Up and Cool Down

- Begin by warming up for 5-10 minutes
- End by cooling down for another 5-10 minutes

Warming up pumps nutrient-rich, oxygenated blood to your muscles and raises your heart rate/breathing. **Cooling down** slows breathing and heart rate bringing blood back into normal circulatory patterns. This prevents sudden drop in blood pressure causing dizziness.

Principles of Stability Rehabilitation

In order to gain control of your *Neutral Joint Position* retrain tonic, low threshold activation (increase muscle stiffness) by applying a consistent low force hold with activation throughout the range and into functional activities. This helps with recruitment of mechanoreceptor stimulation in a joint ligament. How do you go about doing this? The answer lies in a simple device called the ***Stabilizer Pressure Bio-Feedback*** (a lumbar stabilizer).

The STABILIZER Pressure Biofeedback is used to provide feedback to ensure quality and precision in exercise performance and testing. In the physical therapy clinic, the

STABILIZER monitors the position of the low back and provides feedback to the physical therapist and patient when the abdominal and back muscles are not actively or effectively protecting the spine. The STABILIZER is simple enough to use during your home exercise program to provide sensitive feedback on the positional changes of your lower back, while performing back exercises. It may help prevent exercise-induced sprain/strain of your lower back (the lumbopelvic region).

A university study showed no recurrence of lower back pain for 80% of the subjects performing these exercises. Use the STABILIZER for specific exercises targeting the abdominal corset and multifidi: these are the key muscles that control low back pain.

For those seeking Stability Rehabilitation, it is important to understand that motor control and recruitment are the priority, not strength and flexibility. You must palpate for correct activation, and should not feel any pain (or additional pain for those of us dealing with chronic pain), or fatigue (or additional fatigue for those of us dealing with chronic fatigue syndrome). Use the device with a low force sustained hold with normal breathing (10 sec) and repeat (10 times). Breathe fluidly without rigidity. It is important that you breath when you exert or you can cause a lot of anal/rectal issues.

Pain is an inhibitor; it will not allow you to use the muscle effectively and thoroughly. The two Principles of Stability include:

1. Retrain dynamic control of the direction of stability dysfunction
2. Low-load integration of local and global stabilizer recruitment with only moving through the range in which you have control

For those living with stability dysfunction, perform slow, low-effort repetitions and movement only through the range that the dysfunction is controlled actively. Perform 15-20 slow repetitions until it starts to feel familiar and natural. This helps unload pathology and decrease mechanical provocation of pathology and also assists in symptom management

Active lengthening or inhibition of muscles

- Employ this when there is a lack of extensibility due to overuse or adaptive shortening, compensatory overstrain to maintain function.
- Clinical guide: sustain the correction for 20-30 seconds and repeat 3-5 times

The **Key to Exercising** is to control **neutral joint position** (no hyperflexing). It is important that you identify abnormal resting positions (for example, hyperextending or locking out at the knees or elbows) and practice normal physiological range positions. By learning to control neutral joint position, you will

re-train dynamic control and re-train specific muscles to maintain joint position while moving adjacent joints (like moving your hips or marching wile keeping your spine neutral).

To integrate specific training into functional activities and making recruitment automatic, think about the movement, and/or feel or visualize a specific muscle activating.

Equipment

Balance	Bosu, airex, dynadisc, balance board, exercise ball, balance stones
Resistance	Weight machine, elastic bands, exercise ball, cuff weights, body weight
Aerobic	Pedometer, elliptical, recumbent bicycle, treadmill

Some classes that may function as stability rehabilitation for some EDSers include:
- Swimming
- Tai Chi
- Pilates (reformer)
- Yoga
- Silver Sneakers

Some EDSers may want to look into S3 Braces, Kinesiotaping or McConnel taping, Foot orthotics, and Compression wear.

For those with Pes Planus (Flat Feet), medial knee strain and compression to the lateral portion of knee, as well as muscle tendon strain and compression to lateral ankle, may be prevented with the use of foot orthotics. Foot orthotics are very important when it comes to supporting and preserving the arch. Supporting the arch is important when it comes to providing knee and pelvic support.

Stop if you Experience
- Extreme fatigue
- Lightheadedness
- Tingling and numbness (vague)
- Clunking
- Headaches

Bottom Line

"Don't give up, the beginning is always the hardest."

30 minutes of moderate aerobic exercise at least 5 days/wk

2-3 sessions per/wk of strength training that works the major muscle groups of legs, trunk and arms and shoulders

5-10 minutes of warm-up and cool down periods of each exercise session

Take-Home Message

Evidence supports the use of physical therapy and exercise of various types in reducing pain and disability in patients with EDS and JHS. Improvements were found with both supervised and unsupervised exercise programs. So physical therapy is definitely something that most EDSers should look into and pursue as a treatment option.

Dr. Claire Francomano, Director of Adult Genetics at the Harvey Institute for Human Genetics and a member of the Professional Advisory Board of EDNF

Understanding the Relationship Between Self-Efficacy, Locus of Control, and Self-Management Behaviors in Adult Patients with EDS

Presented By: Sabrina Neelley, PhD, MPH

Self-Management is generally considered the activities that people undertake in order to create order and control as they incorporate a chronic health condition into their daily lives and seek the best possible quality of life.

Some Self-Management Activities for EDSers include:

1. Putting together a healthcare team
2. Problem solving
3. Communicating with health care providers
4. Setting goals
5. Dieting, exercising, resting
6. Practicing preventive care and stress reduction
7. Maintaining mental health

Self-Efficacy involves the degree to which an individual has confidence in their ability to do what needs to be done in order to achieve desired outcomes.

Some Self-Management Activities for EDSers include:

1. Decreasing and managing pain
2. Keeping pain from interfering with your life
3. Regulating activity
4. Keeping fatigue from interfering with your life

5. Helping yourself feel better
6. Dealing with frustration

Locus of Control has to do with who or what individuals attribute the status of their condition. This has to do with, if you have something going on in your life…who do you attribute the positive & negative affects to?

There are 4 different types of Locus of Control

1. Internal (what kind of internal/personal control an individual applies to managing their condition)
2. Chance (how much does an individual believe in chance and faith playing into managing their condition)
3. Doctors (how much health care professionals contribute to management of an individual's condition)
4. Others (can be family, friends, and other forms of social support and the roles they play in helping an individual manage their condition)

So, a survey was performed involving a total of 151 adult EDS patients. These adult EDS patients were acquired via attendees at the 2010 EDNF conference and an online survey.

To establish Self-Efficacy data, 8 questions were asked. A 10 point scale was applied to these questions where 1 indicated not certainty at all and 10 indicated very certainty. A high score means high self-efficacy. High self-efficacy means the EDSer has high confidence in themselves to do certain things to manage his/her condition. About ½ of the questions had mean scores less than 5. None of the questions were scored higher than 6, which shows a huge problem in the overall confidence in EDSers to be able to manage their condition for themselves and ultimately deal with EDS. Some examples of the topics the self-efficacy questions centered around involve the following points:

- Keeping EDS pain from interfering with sleep
- Regulating activity to avoid aggravating EDS
- Keeping EDS pain from interfering with activities
- Keeping fatigue from interfering with activities

The last two highlighted points showed to be particularly problematic for EDSers.

To establish Locus of Control data, 18 questions were asked. A 6 point scale was applied to these questions where 1 indicated strong disagreement and 6 indicated strong agreement. A high score means a high locus of control. Internal locus of control showed to be the highest, meaning that a majority of EDSers have to rely on their own resources, abilities, self-motivations, and

self-determination in order to get what they really need. All the scores were at the approximate scale midpoint.

To establish Self-Management data, 19 questions were asked. A 5 point scale was applied to these questions where 1 indicated Not Very Likely and 5 indicated Very Likely. Generally, self-management scores were very high. This indicates that EDSers are typically taking care of themselves as best as they can.

The Relationship between Self-Efficacy & Self-Management

Self-Efficacy (having confidence in yourself)	**Self-Management Activities**
EDSer having confidence in themselves to regulate their activity to avoid aggravating EDS by implementing Self-Management activities →	• Developing/implementing an exercise plan • Taking care of themselves mentally & physically • Accepting responsibility to manage problems • Speaking up for themselves about treatment/care
EDSer having confidence in keeping fatigue from interfering with life by implementing Self-Management activities →	• Developing/implementing exercise plan • Taking care of themselves mentally & physically
EDSers having confidence in themselves to be able to keep pain from interfering with their daily lives and to be able to decrease pain by implementing Self-Management activities	• Developing/implementing exercise plan • Taking care of themselves mentally & physically • Accepting responsibility to manage problems • Finding ways to reduce stress
EDSers having confidence in themselves to be able to help themselves feel better and deal with frustrations by implementing Self-Management activities →	• Developing/implementing exercise plan • Taking care of themselves mentally & physically • Developing/maintaining support systems • Searching/gathering information about EDS • Accepting responsibility to manage problems

Generally, those with high self-efficacy had high confidence in themselves to be able to regulate their activities to avoid aggravation of their EDS. EDSers with generally

low self-efficacy (self-confidence in being able to manage their condition), reported experiencing more episodes of pain and fatigue interfering with their sleep and their lives. In conclusion, this survey showed that self-efficacy was enhanced with successful goal setting and achievement. Some key self-management behaviors include establishing an exercise plan, attaining physical & mental care, and accepting responsibility for problem-solving.

Ehlers-Danlos and Pregnancy

Presented By: Ron Jaekle, MD

There are 6 major phenotypes of EDS with significant overlap.

- EDS Type I & II (Type II is typically milder)
  - This subtype is classified as Classic EDS. Its inheritance is autosomal dominant and the mutation is found in Type V Collagen (Gene: COL5A1 or A2; Locus: 9q34.2-34.3 or 2q31). MVP and organ prolapse is common amongst Classic EDSers.
  - Classic EDSers are more susceptible to experiencing PPROM and have a potentially increased risk of amniocentesis complications. Preterm Premature Rupture of Membranes (PPROM) in pregnancy is the more serious cousin to PROM (premature rupture of membranes. PPROM/PROM occurs when the membraned sac holding your baby and the amniotic fluid breaks open before you're actually in labor.
  - 40-50% of Classic EDSers have detectable COL5A1 or COL5A2. 10% have detectable pro1(V) or pro2(V). 30% have pro!(V) haploinsufficiency. In order to diagnose classic EDS, a complex work-up is required and most Classic EDSers are commonly declined.
  - **Classic EDSers generally do well during pregnancy. There are multiple reports of episiotomy complications, however. Classic EDSers run a higher risk of experiencing intrapartum/postpartum hemorrhage; and operative vaginal deliver may pose an additional risk for perineal trauma.**
- EDS Type III
  - This subtype is classified as Hypermobile EDS. It involves a clinical diagnosis. The genetics of EDS type III remains unknown. Hypermobile EDSers typically have striking joint findings with less skin changes. The common symptoms are recurrent joint dislocations and limb pain.

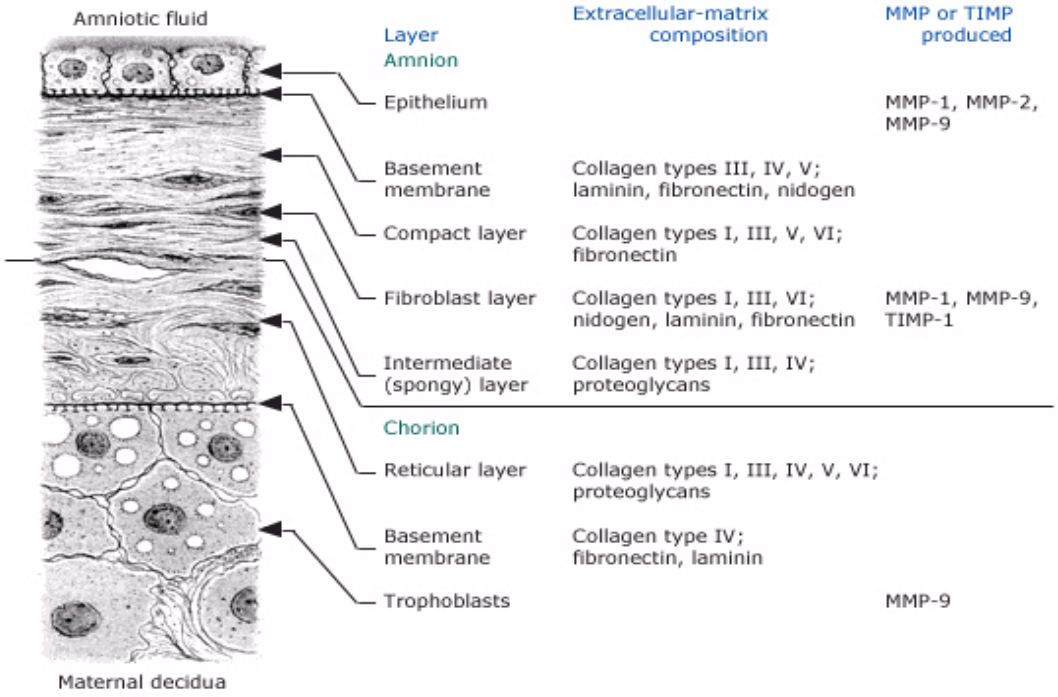
- **Hypermobile EDS doesn't pose any contraindication to pregnancy. Significant pelvic instability and pain is frequently reported. Fortunately, case series suggest overall good obstetric outcomes.**

- EDS Type IV
  - This subtype is classified as Vascular EDS. Its inheritance is autosomal dominant and the mutation is found in Type III collagen (Gene: COL3A1; Locus: 2q31).
  - Vascular markings are typically seen throughout the skin. Vascular EDSers typically risk rupture (vascular, bowel and/or tendon). Vascular EDSers also have a reduced lifespan.
  - **167/183 Vascular EDSers deliver at term. 12/81 Vascular EDSers experience maternal mortalities. Maternal Mortality for Vascular EDSers is 20% for each pregnancy and 38.5% for each pregnant woman.**
- EDS Type VI
  - This subtype is classified as Kyphoscoliosis. Its inheritance is autosomal recessive, with a lysyl hydroxylase deficiency.
  - Vascular EDSers run a high risk of retinal detachment, scoliosis, and exhibit a marfans-like risk of vascular rupture.
  - **Amniotic fluid testing is available (Lysyl hydroxylase activity; Deozypyridinoline/pyroxydinline ratio elevated). Pregnant Kyphoscoliosis EDSers have an increased risk for vascular complications.**
- EDS Type VII
  - This subtype is classified as Athrochalasia. Its inheritance is both autosomal dominant and recessive and the mutation is found in Type I Collagen (Gene: COL1A1, COL1A2 or ADAMST2; Locus: 17q31-22.5, 7q22.1, 5q23-24)
  - Athrochalasia EDSers exhibit hyperflaccid joints with normal skin. They are typically short in stature; and run a risk of congenital skull fractures and hip dislocation.

Some clinical features of EDS include: skin hyperextensibility, joint hypermobility, tissue fragility, poor would healing, and bruising. Few EDSers can have their condition confirmed with laboratory testing. Overlaps of subtypes make specific diagnosis particularly challenging. Genetic consultation and maternal testing is best done prior to pregnancy since the workup may take too much time to offer prenatal diagnosis. Type VI EDS can be diagnosed by Lysyl Hydroxylase in the amniotic fluid.

Physiology of Pregnancy

Layers of amnion chorion



UpToDate

The membranous structure that surrounds the developing fetus and forms the amniotic cavity is derived fro fetal tissue and is composed of 2 layers: the amnion (inner layer) and the chorion (outer layer). The amnion and chorion are separated by the exocelomic cavity until approximately 3 months gestation, when they become fused. Intact, healthy fetal membranes are required for an optimal pregnancy outcome.

An initial evaluation of EDS is important to be completed before pregnancy. Once EDS is confirmed, a maternal echocardiogram to test for MVP and aortic dilation. The aortic root diameter should measure > 4cm. In addition, a carotid and abdominal aorta Doppler analysis should be conducted.

Caring for a pregnant EDS is a multidisciplinary approach (i.e. MFM, Genetics, Vascular Surgery, Cardiology, Rheumatology, General Surgery, Neurosurgery, Ophthalmology, and more). Cervical insufficiency is commonly reported in pregnant EDSers, although no cervical insufficiency was noted in a Dutch cohort study. Case

reports show that prophylactic cerclage associated with a high rate of PPROM and preterm birth is common amongst pregnant EDSers. Unfortunately, there are no recommendations for cerclage in pregnant EDSers. Patients may consider surveillance and vaginal progesterone.

Case series suggest a rate of 25-75% for PPROM/Preterm birth in pregnant EDSers. The PPROM rate for those with an affected fetus is 21/43. The PPROM rate for those with an affected mom is 25/128. Delivery before 37 weeks occurs in 17/43 of those with an affected fetus, and 28/128 of those with an affected mother.

Fetal growth restriction has been described in several case reports. In a Dutch cohort study fetal growth restriction was only seen in affected mothers.

Beta-blockers may be used to control heart rate and pulse pressure in pregnant EDSers. It is important that the mother avoid valsalva or strenuous activity. Vitamin C supplements should be taken in order to maximize strength and quality of collagen crosslinking. Early admission to the hospital may be necessary in some cases.

Vaginal Delivery increases the risk for extensive perineal trauma and for poor episiotomy healing. If an episiotomy is performed, there are arguments for and against the type of episiotomy that can be given being either a midline or a mediolateral. This relates to the angle at which the cut is made. The trend in America is for midlines, in UK and Europe it is a mediolateral. As with everything in Australia, we have a mixture being used. The type of episiotomy given to pregnant EDSers should be discussed with the caregiver and of course the type of EDS the patient is affected with should be taken into consideration. A mediolateral episiotomy involves cutting into more muscle and tissue and does not follow the natural way a woman would tear. This can mean they are harder to repair, have increased bleeding, the cut may not heal as well, it may produce more scarring, and possibly more pain in the weeks following birth. The advantage of a mediolateral episiotomy is that it is less likely to extend to a 3rd or 4th degree tear, which makes it a better option for many EDSers. The midline episiotomy involves cutting through less muscle tissue and following the natural line of the perineum that a tear would take if it occurred. This can mean they are easier to repair, involve less blood loss, heal better, and have less scarring and possibly less pain in the early weeks after the birth. The disadvantages can be that performing a midline episiotomy increases the chances of the cut extending through the anus and causing a 3rd or 4th degree tear. This makes a midline episiotomy very dangerous for EDSers.

Cesarean Delivery increases incisional herniation, poor skin healing, and meticulous hemostasis and retention sutures. The question resides in whether a caregiver

should use suture or glue to close up a C-section on an EDSer. Dermabond, a medical grade version of superglue, was approved by the FDA in 1998 and is currently one of the options for closing c-section incisions. Women are currenty being recruited for trials to compare results between women who's c-section incisions was stapled with those closed using Dermabond. Another trial will compare c-section closures using staples and sutures (stitches). So, there isn't much information available yet to recommend suture or glue for EDSers, but the Dermabond does show great promise and should definitely be considered. It dries very fast (10 seconds) and forms a strong waterproof bond. As the morbidity associated with cesarean delivery, usually results from wound complications, especially infection, and with c-sections accounting for one in every 3 births in the US, knowing which method of incision closure is most effective is paramount.

EDSers also run the risk of both protracted and precipitous labors. The risk of breech occurring is 8% for EDSers (compare to a 3% rate at term normally). Additionally, face and brow presentation is seen in 5/46 affected fetuses.

When using neuraxial anesthesia, there is a risk of epidural hematoma and increased heart rate and pulse pressure in most EDSers. When using general anesthesia, most EDSers run the risk of gingival bleeding, oropharyngeal tissue fragility, and pneumothorax.

Hemorrhage is a huge concern when it comes to pregnant EDSers. It is important to know the rates and take them into consideration when it comes to potential treatment options. Obstetric hemorrhage requiring treatment occurs 20% of the time in EDSers. The risk of hemorrhage is 33% if both mother and neonate is affected. Uterine atony and lacerations contribute to the high rate of hemorrhage in EDSers. 13% of EDSers give birth to infants with Floppy baby syndrome. Unfortunately, affected infants frequently are not diagnosed at delivery.

In conclusion, pregnant EDSers' vessels are at risk due to hormonally induced relaxation. Increased cardiac output, just from being pregnant, also increases the risk of vessel rupture. Pain and joint instability is a common maternal complaint. **Kyphoscoliotic and Vascular Types are associated with significant risk of maternal mortality**. Spontaneous arterial rupture has been described in pregnancy with minimal trauma. Tertiary care is a necessity for pregnant EDSers. PPROM and prematurity is common for pregnant EDSers. When the fetus is also affected with EDS, the risk for PPROM increases. Prenatal diagnosis of EDS is complex but is improving. All pregnant EDSers should consider vitamin C and Beta-blocker therapy during pregnancy. Maternal mortality for specific types of EDS still remains a concern through the postpartum period.

Non-Vascular EDS & Pregnancy: What are the Risks?

Presented By: Anna Mitchell, MD PhD

Some reported complications for pregnant non-vascular EDSers that have been published include:

- Abnormal fetal presentation at delivery
- Incompetent cervix
- Joint dislocation during delivery
- Standing erect becoming increasingly difficult
- Uneventful complications

A study was conducted where the following question was asked,

"What is the obstetrical experience of women with non-vascular EDS?"

A total of 437 surveys were analyzed from woman, 18 years of age and older, with an official diagnosis of non-vascular EDS, of whom which had at least one pregnancy. These woman were recruited from the 2011 EDNF Learning Conference and online. Note: The survey had a total of 517 responses, 34 of which were excluded (because the woman either were never pregnant, didn't know what type of EDS they had or had vascular, or the questionnaire was submitted incomplete). Amongst the 437 woman analyzed, there was a total of 1061 pregnancies.

22 questions were broken up into the following 6 sections:

1. Demographics
2. General pregnancy questions
3. Prenatal care
4. Pregnancy
5. Maternal health during pregnancy
6. Labor and delivery

All sections other than demographics could be answered one time per pregnancy, for up to 4 pregnancies. Open comment boxes were provided throughout the survey, which were categorized and tabulated during data analysis. A one-tailed binomial test was also conducted in order to compare available observed complication rates to published rates. Since there were 61 total first trimester miscarriages reported, only 376 of the 437 woman were included in the obstetrical complication analysis, because the analysis of complications only included second trimester miscarriages and stillbirths.

Please keep in mind the study limitations. All of these questionnaires are self-reported. No control group was collected, and there is ascertainment bias.

The following is a graph of the **Demographic Information of Participants**

		Frequency (n)	Percent (%)
Age (n=435)*	18-19	1	0.2
	20-29	58	13.1
	30-39	186	42.9
	40-49	123	28.2
	50-59	54	12.6
	Over 60	13	3.0
EDS subtype (n=437)	Classic	102	23.3
	Hypermobility	331	75.7
	Kyphoscoliosis	3	0.7
	Arthrochalasia	1	0.2
	Dermatosparaxis	0	0
Had Genetic Testing for EDS (n=113)	DNA analysis	8	7.3
	Protein analysis	38	34.9
	Don't know	68	62.4
	Answer missing	4	3.7

- Two participants did not report their year of birth

Pregnancy Data for Survey Results

		Frequency (n)
Total # of Pregnancies	Population as a whole (n=437)	1061
	Classic EDS (n=102)	258
	Hypermobile EDS (n=331)	796
	Kyphoscoliosis EDS (n=3)	6
	Arthrochalasia EDS (n=1)	1
Average # of Pregnancies per Woman	Population as a whole (n=437)	2.4
	Classic EDS (n=102)	2.5
	Hypermobile EDS (n=331)	2.4
	Kyphoscoliosis EDS (n=6)	2
	Arthrochalasia EDS (n=1)	1

They only analyzed the first pregnancies because multiple pregnancies in one individual presents as a confounding factor. The following is a table that shows the **First Pregnancy Outcomes.**

EDS Subtype	Outcome	Frequency (n)	Percent (%)	General Pop Rate (%)	P-value
Non-vascular (n=437)	Miscarriage (<20 wks)	82	18.8	20[a]	0.281
	Stillbirth (20-24 wks)	2	0.458	0.622[b]	0.496
	Premature delivery (24-36 wks)	63	14.4	12.18[c]	0.09
Classic (n=102)	Miscarriage (<20 wks)	18	17.6	20[a]	0.326
	Stillbirth (20-24 wks)	0	0	0.622[b]	0.529
	Premature delivery (24-36 wks)	13	12.7	12.18[c]	0.475
Hypermobile (n=331)	Miscarriage (<20 wks)	64	19.3	20[a]	0.413
	Stillbirth (20-24 wks)	2	0.604	0.622[b]	0.661
	Premature delivery (24-36 wks)	47	14.2	12.18[c]	0.15

Timing of Miscarriages

Type of EDS	Time of Miscarriage	Frequency (n)	Percent (%)	Gen Pop Rate (%)	P-value
Non-vascular$ (n=432)	First trimester (<13 wks)	61	14.1	16[a]	0.317
	Second trimester (13-19 wks)	16	3.7	4[a]	0.877
Classic% (n=101)	First trimester (<13 wks)	9	8.9	16[a]	0.058
	Second trimester (13-19 wks)	8	7.9	4[a]	0.099
Hypermobile& (n=327)	First trimester (<13 wks)	52	15.9	16[a]	1
	Second trimester (13-19 wks)	8	2.4	4[a]	0.183

Key for the above table:
a Cunningham et al., 2010, Ch. 9
$ Five people did not report when miscarriage occurred
% One person did not report when miscarriage occurred
& Four people did not report when miscarriage occurred

Pregnancy Complications for Non-Vascular EDSers

Complication	Frequency (n=376)	Percent (%)	General Population Rate (%)	P-value
Abnormal fetal delivery position*	55/346	15.9	5.4[a]	<0.001
Incomplete epidural efficacy*	100/191	52.4	12[b]	<0.001
Joint dislocation*	125/330	37.9	<1[d]	<0.001
Post-partum severe bleeding from womb/uterine hemorrhage*	11/332	3.3	1[d]	<0.001
Premature rupture of membranes*	66/343	19.2	3[e]	<0.001

The rate of pregnancy complications for Non-Vascular EDSers is significantly higher than the general population.

Pregnancy Complications for Classic EDS

Complication	Frequency (n=93)	Percent (%)	General Population Rate (%)	P-value
Abnormal fetal delivery position*	13/81	16	5.4[a]	<0.001
Incomplete epidural efficacy*	25/39	64	12[b]	<0.001
Joint dislocation*	27/83	32.4	<1[c]	<0.001
Post-partum severe bleeding from womb/uterine hemorrhage	2/78	2.6	1[d]	0.183
Premature rupture of membranes*	13/86	15	3[e]	<0.001

Pregnancy Complications for Hypermobile EDS

Complication	Frequency (n=279)	Percent (%)	General Population Rate (%)	P-value
Abnormal fetal delivery position*	41/261	15.7	5.4[a]	<0.001
Incomplete epidural efficacy*	74/150	49.3	12[b]	<0.001
Joint dislocation*	96/244	39.3	<1[d]	<0.001
Post-partum severe bleeding from womb/uterine hemorrhage*	9/251	3.6	1[d]	0.001
Premature rupture of membranes*	51/253	20.1	3[e]	<0.001

Pregnancy Complications

		Non-vascular EDS Total N=376			Classic EDS Total N=93			Hypermobile EDS Total N=279		
Complication	Gen Pop Rate (%)	Frequency (n)	Percent (%)	P-value	Frequency (n)	Percent (%)	P-value	Frequency (n)	Percent (%)	P-value
Abnormal fetal delivery position	5.4[a]	55/346	15.9	<0.001 *	13/81	16.0	<0.001 *	41/261	15.7	<0.001 *
Incomplete epidural efficacy	12[b]	100/191	52.4	<0.001 *	25/39	64	<0.001 *	74/150	49.3	<0.001 *
Joint dislocation	<1[c]	125/330	37.9	<0.001 *	27/83	32.4	<0.001 *	96/244	39.3	<0.001 *
Post-partum severe bleeding from womb/ uterine hemorrhage	1[d]	11/332	3.3	<0.001 *	2/78	2.6	0.183	9/251	3.6	0.001*
Premature rupture of membranes	3[e]	66/343	19.2	<0.001 *	13/86	15.0	<0.001 *	51/253	20.1	<0.001 *

Accounting for Fetus' EDS Status

	Fetus has EDS?	Frequency (n)	Percent (%)	General Population Rate (%)	P-value
Abnormal delivery position	Yes* (n=161)	29	18.0	5.4[a]	<0.001
	No* (n=92)	11	12.0	5.4[a]	0.011
Premature delivery (<37 weeks)	Yes* (n=161)	19	18.0	12.18[b]	0.02
	No (n=111)	18	16.2	12.18[b]	0.126
Premature rupture of membranes	Yes* (n=149)	35	23.5	3[c]	<0.001
	No* (n=110)	15	13.6	3[c]	<0.001

Non-Vascular EDS vs. Vascular EDS

Type of EDS	Complication	Frequency (n)	Percent (%)	Vascular EDS Pop Rate (%)	P-value
Non-vascular	Arterial rupture at delivery or post-partum*	11/335	3.3	8.6 [a]	<0.001
	Premature delivery* (<37 wks)	63/437	14.4	19 [b]	0.007
	Premature rupture of membranes	66/343	19.2	19 [b]	0.476
Classic	Arterial rupture during delivery or post-partum*	1/79	1.3	8.6 [a]	<0.001
	Premature delivery (<37 wks)	13/102	12.7	19 [b]	0.064
	Premature rupture of membranes	13/86	15.0	19 [b]	0.221
Hypermobile	Arterial rupture during delivery or post-partum*	9/253	3.6	8.6 [a]	0.001
	Premature delivery* (<37 wks)	47/284	14.2	19 [b]	0.013
	Premature rupture of membranes	51/253	20.2	19 [b]	0.343

The rate of complications in non-vascular EDS is significantly lower than the vascular EDS population.

One-tailed binomial analysis DECREASED rates in the Non-Vascular EDS population

Ruptures resulted in death in the vascular population

Other OB Complications of Non-Vascular EDS include:

- Increases in bone and/or joint pain
- Difficulty standing for more than 5-10 minutes
- Ankle Instability
- Skin tingling, prickling, numbness
- Teeth fragility
- Heavy vaginal bleeding
- Amniotic sac complications, not specified
- Excessive bleeding/Hemorrhage (other than uterus)
- Blood vessel rupture at any time during pregnancy
- Cervical cerclage attached
- Bowel perforation
- Exacerbations of normal EDS Symptoms

Additional Complications Provided by Survey Participants

Complication (if n>5)	Frequency (n)	Examples
Maternal hypertension and pre-eclampsia	40	
Placental problems	28	Previa Abruption
Pelvic complications	26	Symphysis Instability
Cardiac issues and fainting	23	POTS Change in heartrate
Swelling and edema	16	
Oligohydramnios	18	
Gastrointestinal manifestations	14	GERD Dysmotility
Hyperemesis gravidum	13	
Emergency c-section	12	
Stalled labor	9	
Gestational diabetes	7	

The results of this survey suggest that the pregnancy outcomes for women with non-vascular EDS don't differ from those of the general population in terms of miscarriage, still birth, and premature delivery. However, women with non-vascular EDS may be at a higher risk than the general population to experience the following obstetrical complications:

- Fetal malpresentation, regardless of fetus' EDS status
- PROM, regardless of fetus' EDS status
- Premature delivery, if the fetus is also affected
- Incomplete epidural efficacy
- Joint dislocation
- Uterine hemorrhaging/heavy bleeding.

Women with non-vascular EDS may have a LOWER risk than the vascular EDS population for premature delivery (if mom has hypermobile EDS) and a during-delivery or post-partum arterial rupture.

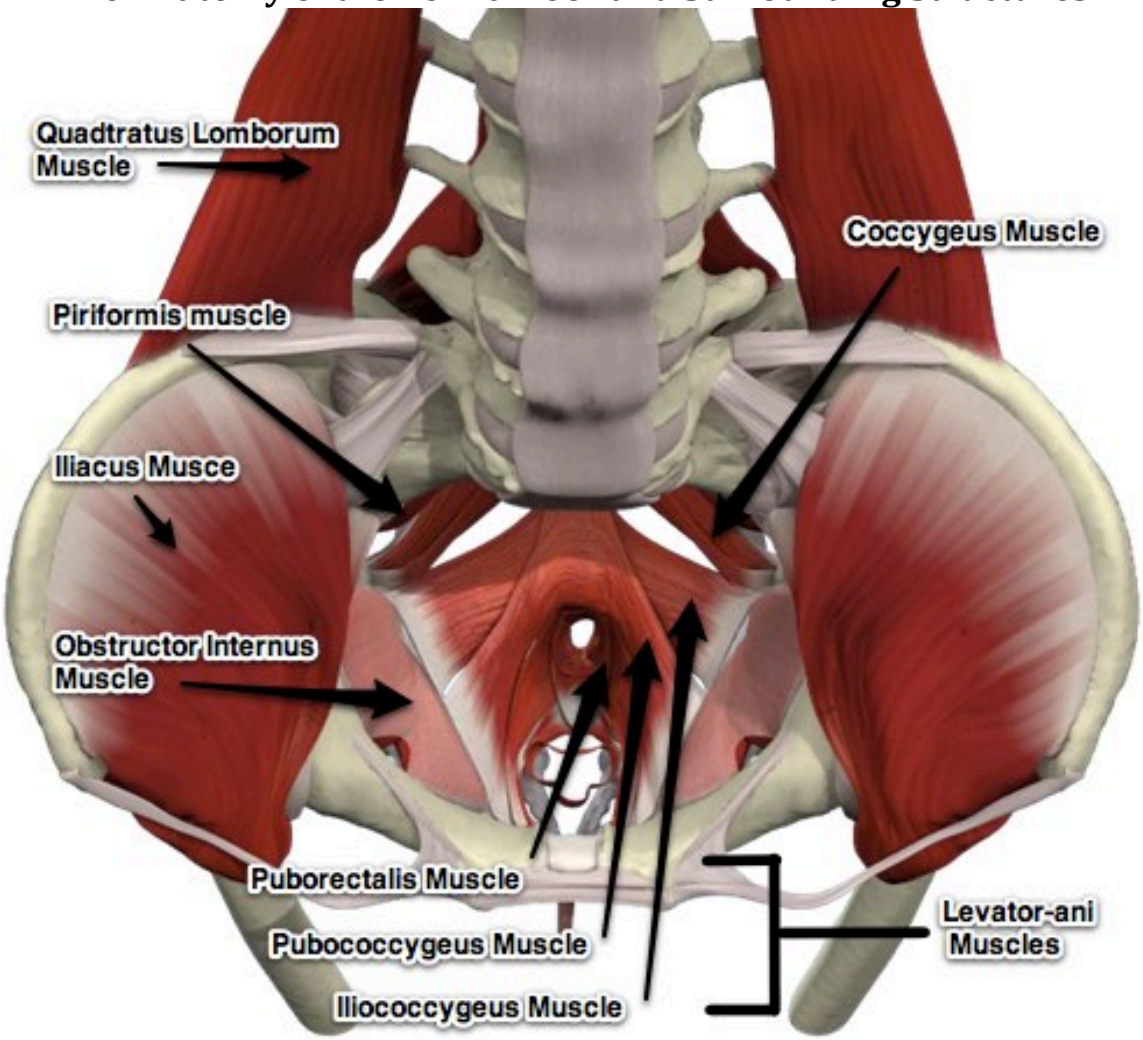
Woman: Understanding, Preventing, and Managing Pelvic Floor Dysfunction

Presented By: Kathleen Zonarich, PT

Pelvic floor dysfunction refers to a wide range of problems that result when pelvic floor muscles are weak, tight, and/or an impaired sacroiliac joint, low back, coccyx and/or hip joint exists. Tissues surrounding the pelvic organs may have increased or decreased sensitivity, and/or irritation resulting in pelvic pain.

Many times the underlying cause of pelvic pain is difficult to determine. Pelvic floor dysfunction can cause incoordination in the contraction and relaxation of the pelvic floor muscles that assist in controlling bladder and bowel function. Pelvic floor dysfunction is **not** a normal course of aging. An estimated 1/3 of all US women are affected by some type of pelvic floor disorder. 1 in 11 women will have pelvic floor surgery. 13 million Americans are effected by incontinence. Stress incontinence is most common for women. Adolescent girls suffer from stress incontinence with sports. Pelvic floor dysfunction occurs in women that have not given birth.

The Anatomy of the Pelvic Floor and Surrounding Structures



The pelvic floor acts as a sling to:

- Support the bladder, uterus, & rectum
- Surround the urethra, & vagina
- Assist with urination and defecation

Types of Pelvic Floor Dysfunction include:

- **Supportive Dysfunction**: a result of the loss of nerve, muscle, ligament, or fascial integrity of the pelvic floor muscles causing weakness and laxity; could be caused by injury incurred during childbearing or gynecologic surgery, chronic constipation, chronic coughing, obesity, or hormonal changes; can cause bowel and bladder incontinence, pelvic pain or pressure, and back pain.

- **Hypertonus Dysfunction**: include symptoms of pain in the abdominal area, back, or vulvar region; patients may report burning, itching, dyspareunia, urinary urgency and leakage, or constipation; can cause bowel and bladder incontinence, pelvic pain or pressure, and back pain.
- **Sexual Dysfunction**: pelvic floor muscles not only assist with bowel and bladder control, but also with sexual stimulation. If the pelvic floor muscles are not stretched or actively strengthened, they lose a part or all of their function, resulting in difficulty with orgasms or no orgasms at all. The puboccygeus muscle and one of the levator ani-muscles are the main assistants with sexual function.

"In a cohort study by Jha et al. (2007), it was found that the prevalence of both urinary and fecal incontinence appears to be significantly higher in women with BJHS [Benign Joint Hypermobility Syndrome] when compared with women without this condition. **Overall prevalence of urinary incontinence in this group was 68.9%.** The estimated prevalence of incontinence in a similar population without this condition was 30%. **Prevalence of fecal incontinence among members of HMSA is 14.9%** (n=22). The prevalence of fecal incontinence in the general adult population by contrast is 2.2%." (Arunkalaivanan).

Symptoms of Pelvic Floor Dysfunction include:

- Urinary frequency
- Urinary urgency
- Pelvic pain
- Low back pain
- Irritable Bowel Syndrome
- Sensation of incomplete urination
- Pain behind vagina
- Decreased urinary flow
- Constipation
- Painful intercourse
- Chronic stinging
- Burning

Some may have just a few or many of these symptoms. Breathing dysfunction is common with pelvic floor dysfunction due to increased intra-abdominal pressure and straining.

Diagnosis of Pelvic Floor Dysfunction requires a physical exam by a doctor or physical therapist. It is important that other conditions, such as Urinary Tract Infection, endometriosis, neurological disease, and cancer are ruled out. There are no specific diagnostic tests for Pelvic Floor Dysfunction.

Pelvic Organ Prolapse occurs when the pelvic muscles and tissues become weak and can no longer hold the organs in place correctly. 90% of women with prolapse do not seek medical treatment.

Types of Pelvic Organ Prolapse include:

- **Cystocele**: bladder bulges into vagina through the tissues separating the bladder and vagina.
- **Rectocele**: anterior wall of rectum bulges into posterior wall of vagina. Symptoms include increased pressure and difficult with bowel movements.
- **Urethrocele**: tissue between vagina and urethra weakens, resulting in the urethra pushing into vagina.
- **Uterine**: the uterus can press down on the vagina causing the uterus to invert and/or come out through the vaginal opening. Symptoms of Urethrocele include backaches, perineal pain, a sense of "heaviness" in the vaginal area, a lump in the vaginal opening, pelvic discomfort, abdominal cramping, relief by lying down, and increase in symptoms by prolonged standing, walking, coughing, or straining.
- **Vaginal**: the top of the vagina loses support and can drop through the vaginal opening.

Some symptoms of pelvic organ prolapse includes:

- A feeling of heaviness or fullness, or as if something is falling out of the vagina
- A pulling, aching or a "bulge" in the lower abdomen or pelvic region
- A kinking in the urethra, making it harder for a woman to empty her bladder completely, or causing frequent urinary tract infections

Bladder Incontinence is defined by involuntary loss of urine as a result of more pressure in the bladder than in the sphincter. Normal function of urination allows for contraction of the bladder with relaxation of the pelvic floor enabling the sphincters to open. Dysfunction occurs when pelvic spasms instead of relaxing and does not allow the bladder to empty efficiently. Types of Bladder Incontinence include:

- **Urge Incontinence**: overactive bladder; frequent urge to urinate; urinates more than 8 times in 24 hrs; unable to hold it before getting to toilet; awakens more than once during the night to urinate; leaks moderate to large amount of urine
- **Stress Incontinence**: urethra is not remaining closed, thus sphincter does not stay closed; small volumes of urine lost with coughing, laughing, sneezing, jumping, or any increased intra-abdominal pressure; is the result of weak pelvic floor muscles

    - Those at risk for stress incontinence includes women (due to genetics, childbirth, and anatomic and neurological factors), the aging population, and those with certain lifestyles (i.e. poor nutrition, obesity, smoking, decreased activity, bad toilet habits, inadequate intake of fluids, certain medications)
- **Mixed Incontinence**: urge and stress incontinence
- **Overflow Incontinence**: bladder is underactive; does not empty adequately; is the result of obstruction or underactive neurogenic bladder
- **Reflex Incontinence**: no sensation of control of sphincters (e.g. spinal cord injury)
- **Functional Incontinence**: person is physically or cognitively unable to make it to the bathroom in a timely manner
- **Enuresis**: bladder contracts without person feeling it (e.g. bed wetting)

Treatment options for pelvic floor dysfunction includes exercise, physical therapy, medications, surgery, and pessaries.

Prevention of pelvic floor dysfunction involves Kegel exercises in addition to Core exercises of the abdominals, hip muscles (hip adductor issues are common in PFD), back muscles, and pelvic floor muscles. Pelvic floor exercises enhance the tone and contractile force generated by essential pelvic floor muscles and sphincter.

Approximately 19-31% of women who believe they perform Kegels correctly, actually perform them correctly.

How to Perform A Kegel Exercise:

1. Find the right muscles. While urinating try to stop the flow of your urine These are the muscles you want to use when performing a Kegel exercise. Do not continue to repeat this action while urinating, as it can cause a urinary tract infection. Do not do Kegel exercises with a full bladder as it can weaken your bladder. If your pelvic floor muscles are very weak, you may need to find them in supine position.
2. Chose your position (sitting for most). Tense your pelvic floor muscles firmly, as though you are lifting up the "sling." Repeat these contractions 4-5 times in a row, holding each for 5 seconds. Be sure to completely relax every time before starting anew repetition. Rest for 5-10 seconds between each repetition. Tips: Remember that quality is better than quantity. Build up over time to 10 second holds for each set, resting 10 seconds between each set. Do not use your abdominal, back, gluteal, or hip muscles. Don not hold your breath. Concentrate on only using your pelvic floor muscles.
3. Build yourself up to doing 10 sets of Kegel exercises, 3 times a day. Additional variations of Kegels include the Flicker Technique – contract and

relax quickly 10-20 times in a row; and the Elevator Technique – contract your pelvic floor as though it is an elevator. Visualize contracting at each floor as you go up in the elevator (holding 3 seconds at each floor. You may only be able to hold one second until you build up to 3 seconds). When you can't squeeze any tighter and hold, it is time to come down on the elevator (repeating the same pattern, but as you descend, you relax the pelvic floor a little more at each floor. When you arrive at the bottom level, your pelvic floor should be completely relaxed. Repeat 5-10 times.

4. As you get stronger, you will be able to do a Kegel while performing another activity at the same time. While doing abdominal exercises, be sure to do a Kegel first and hold while you are contracting your abs. If you are lifting or carrying, do a Kegel first and hold while you lift or carry. If you need to sneeze or cough, do a Kegel first and hold while you sneeze or cough.

Biofeedback shows you how your muscles are contracting and relaxing. Visual input is very beneficial in learning to effectively control these muscles. Relaxation is as important as strengthening. When urinating or having a bowel movement, relax the sphincter muscles, DO NOT use force to push.

E-Stim stimulates your pelvic floor muscles and assists in coordination of muscles to contract and relax and decrease pain and muscle spasms.

Ultrasound allows for visualization of the muscles contracting and relaxing. It also increases circulation and decreases spasm and inflammation.

Breathing and relaxations techniques are great physical therapy interventions as well as manual techniques. Some manual techniques include: soft tissue mobilization, myofascial release, massage, stretching, scar management.

Some contraindications to physical therapy interventions included a lack of consent, immediate post partum 6 weeks, and post-op not before 6 weeks. Some precautions include, but are not limited to, severe vaginitis or atrophy, infection, sexual abuse, pelvic pain, pediatric patients, and pregnancy. Note that Kegel is contraindicated in patients with pudendal neuralgia.

In some cases, your physician may prescribe a low-dose muscle relaxant for pelvic floor dysfunction, analgesics, anti-inflammatories, stool softeners, and/or hormones.

Of course, conservative measures should be tried before surgery. But when they fail, there are many options based on the corrections needed, medical history, and surgeon. You and your surgeon can decide together what is best for you. Surgery

options include: Pelvic Floor Reconstruction (for correction or improvement of prolapse and/or incontinence); Laparoscopic; Traditional; Vaginal; Abdominal; Mesh; Sutures; Grafts.

"In a study by Jarvis et al., **preoperative and postoperative physical therapy was found to improve outcomes** (quality of life questionnaire, urinary symptoms, and maximum pelvic floor muscle squeeze on mamometry) compared to a surgical group without per-operative physical therapy. This is an important indicator that physical therapy can play both a preventative role for surgical intervention but also plays an important role when surgery is indicated as the primary treatment" (Physiopedia).

A device to assist with prolapse is called a Pessary. Pessaries are small plastic devices positioned within the vagina. Pessaries give support and hold prolapsed organs in their proper place. Pessaries are an option for those that are waiting to have surgical correction or unable to have surgical correction.

Pelvic Pain Syndromes occur when pelvic floor muscles do not relax, resulting in pain for more than 6 months. Pelvic Pain Syndromes are associated with Irregular Bowel Function, Bladder Irritation, and Stress Incontinence; and often goes along with poor posture, lack of flexibility or strength, core muscle weakness, pelvic floor muscle weakness, sacroiliac joint pain, and past trauma below the waist or with the spine.

Sacroiliac Joint Dysfunction involves the muscles, bones, fascia, vessels, and connective tissue are mal-aligned. Sacroiliac Joint Dysfunction often results from a fall or strain. Physical therapy involves manual therapy, posture reeducation, strengthening and stretching, and modalities.

Myofacial Pain involves blockage and tissue mal-alignment causing generalized pain. Physical therapy involves finding location of the issue and manual release of fascia that is causing pain.

Constipation can be caused by non-relaxing pelvic floor and dietary issues. Physical therapy involves biofeedback for relaxation, manual therapy, and E-stim.

Vaginal pain can result from childbirth, episiotomy or tearing, and/or past trauma. Physical therapy involves biofeedback, manual therapy/massage, scar mobilization, and E-stim.

If you are having any symptoms, tell your doctor and ask him/her to assist you with:

- Identifying exact location of pain
- Activating the correct muscles for pelvic floor strengthening
- Assessing if you have pelvic floor weakness

Find a physical therapist that has specialized training in Pelvic Floor Dysfunction. "85% of women who have bladder or bowel incontinence and/or low libido do find significant improvement or even a cure with treatment by a Women's Health Physical Therapist" (APTA/Women's Health). Visit APTA.org website to locate a specialized physical therapist (select "Find a PT" at the top of the page; then check "Women's Health" to find a local PT). You can also ask our OB/GYN to recommend a PT.

Sleep, Pain, and Fatigue in Ehlers-Danlos Syndrome

Presented By: Susan Cordes, MS, CGC

The purpose of this online survey was to characterize poor sleep, pain, fatigue, and RLS (restless leg syndrome) and determine if there are correlations to age and/or gender in patients self-identified with EDS. They used SurveyMonkey posted on the EDNF website in order to get participants for the survey.

It is a given that sleep disturbance is correlated with fatigue. But pain and sleep disturbances are also closely correlated. Experience and studies have shown that ***those suffering from different pain disorders have significant sleep disturbances, which are related to pain.*** Conversely, sleep deprivation can cause fatigue and increased sensitivity to pain. Therefore, improving sleep quality and quantity may help contribute to breaking vicious cycles and thus enhance patient's overall health and quality of life.

Fatigue is a common symptom in various conditions with chronic pain. People with chronic pain often experience fatigue as a result of physical strain or emotional stress from dealing with chronic pain. Fatigue can have an impact on pain sensitivity as well.

RLS is characterized in order to identify sleep variables that may explain poor sleep complaints. ***RLS can also occur secondary to chronic pain due to musculoskeletal disorders.*** In addition, some evidence suggests that sensory pain-related perception is altered in RLS patients. RLS is associated with sleep disturbance and excessive daytime sleepiness.

All of these can contribute to quality of life and all of these are common complaints in EDSers. However, research on pain, fatigue, sleep quality, and RLS in EDS is unfortunately limited.

For this survey, they used only those surveys that were completed. They analyzed a total of 888 surveys consisting of hypermobile & classic EDSers, ranging from age 14 to 83.

The gender demographics included 91.6% Females and 8.4% Males. A majority of the participants reported having only hypermobile EDS, a little less than 25% of participants reported having only classic EDS, and a handful reported having both hypermobile and classic EDS.

The survey was made up of 7 components: subjective sleep quality index, sleep latency, sleep duration, sleep efficiency (hours slept divided by hours in bed), sleep disturbance, use of sleep medication, and daytime dysfunction.

SLEEP

- EDSers showed a significant decrease in overall sleep quality as compared to the controls.
- EDSers also showed a mild degree of insomnia as compared to controls.
- EDSers spent a less amount of time sleeping than controls due to reduced sleep efficiency, 8.5 hrs in bed with 6 hrs of actual sleep, and frequent nighttime awakenings (more than 3 times per week).
- EDSers usually have difficulty sleeping due to feeling hot and having pain.
- EDSers often resort to taking sleeping aids.
- EDSers experience difficulty staying awake and maintaining high energy (enthusiasm) levels.
- Reduced sleep quality is correlated with worsening fatigue
- ***Low sleep quality did not correlate with pain severity or pain intensity. As a result, pain may only be a contributing factor to sleep issues in EDS.***

PAIN

- Nearly all patients reported pain (98%)
- Respondents reported 12 sites on average with persistent/recurring pain. The most common were the jaw, neck, back (especially lower), shoulder, wrist, hand, digits, hip, knee, ankle, and feet.
- Pain intensity ranged from a 3-7 (on a scale of 0-10). The average score for pain severity was 5. The average for the worst pain was a 6.8
- Respondents reported an average of 27% pain relief from medications
- EDSers reported experiencing significantly stronger pain severity than the compared controls

- EDSers reported significantly more pain interference (pain affecting general activity, mood, work, relationships, sleep, and enjoyment of life.
- Pain did not correlate with sleep quality or overall fatigue.
- 92% of EDSers reported that they were unusually tired and experienced fatigue in the past week.
- The average, least and worst fatigue levels during the past 24 hours ranged from 5-8 on a 10 point scale where 0 was no fatigue and 10 was "as bad as you can imagine"
- 92.3% of EDSers reported being "usually tired." Most EDSers reported that their fatigue significantly interfered with general activities, mood, walking ability, work, relationships, and enjoyment of life.
- When compared to the global scores of fatigue, EDSers ranked twice as high to those diagnosed with sleep disorders and 3/2 times higher than those with cancer (6.11 v. 3.04 (sleep disorders) v. 4.04 (cancer)).
- Fatigue is correlated with decreased sleep quality.

RESTLESS LEG SYNDROME (RLS) & Periodic Limb Movement (PLM)

- 21% of EDSers have self-reported diagnosis of RLS
- EDSers reported RLS symptoms occurred on average 4-5 times per week
- 7% of EDSers were diagnosed with PLM
- Bed partners of EDSers reported leg twitching or jerking a few times a week while EDSer is asleep
- RLS is a disorder in which there is an urge or need to move the legs to stop unpleasant sensations. RLS often occurs at night while awake. RLS is heritable and can disturb sleep.
- PLM is repetitive cramping or jerking of the legs during sleep
- 20.9% of EDSers were diagnosed with RLS
- 6.8% of EDSers were diagnosed with PLM

In conclusion, pain in EDSers is common, severe, involves multiple sites, and interferes with daily living. Sleep disturbance amongst EDSers is common, but frequent awakenings are more common than insomnia. Sleep disturbances are related to generalized fatigue, but may not be the only factor. Pain is somewhat related to the sleep disturbances but so are other factors. RLS is common amongst EDSers, but only has modest effects on sleep disturbances.

Please note that this study has limiations like ascertainment bias, mostly female respondents, and generalized "controls" taken from other publications.

Some management suggestions include practicing proper sleep hygiene, participating in cognitive-behavioral therapy, and taking medications for sleep improvement and pain control.

Service Dogs

Presented By: Canines for Disabled Kids

A service animal is a dog that is individually trained to do work or perform a task for a person with a disability. The trained work or task must be directly related to the person's disability. Comfort and emotional support does not qualify.

Trained work or tasks of a service animal include:

- Turn lights on and off
- Open doors
- Identify sounds
- Alert to oncoming seizures
- Retrieve objects
- Much more

Service dogs should increase the level of independence of their human partners. These service animals cannot be protective nor aggressive.

Locations that serve the general public must allow service dogs to accompany people with disabilities in all areas the public is allowed to access. It is important to note that the service dog **MUST BE** under the handler's control. Allergies and the fear of dogs are **NOT VALID REASONS** for denying access to a person using a service dog. Facilities (i.e. schools, restaurants, etc.) are **NOT** responsible for the service animal.

Adults and children with physical and/or psychological disabilities **MAY** qualify for a service dog. All candidates must be evaluated individually.

It is important to understand that you must no touch or talk to a service dog without permission from the handler. Some owners resort to signs such as "Please do not pet me. I am working," to extend this knowledge to those of the general population.

Some of the following types of service dogs may help EDSers include:

- **Traditional Service Dogs**
    - These dogs help those who use tools including bit not limited to: wheelchairs, canes, crutches, and walkers, by picking up almost any dropped item, turning light switches on or off, and carrying items.
    - "Laptop Dogs" are a smaller version of the traditional service dog with the agility to jump up on counters, retrieve items, and then to jump with the item into the owner's lap.

- **Walker or Balance Dogs**
    - These dogs are generally large breeds which should have a healthy body weight of half, or more, of the handler's body weight. These dogs will wear harnesses specific to their work –some will be trained to help an individual balance in a standing position or to get up or down from a standing position, while others are trained to help prevent falls while the individual is walking. Some may be trained for any combination of the above. It is important to understand what each training organization considers a finished dog with this style of work, as it may or may not be the right tool for the owner's balance needs.

Aquatic Exercise: Benefits and Principles for the EDS Population

Presented By: Kathleen Zonarich, PT

Benefits of aquatic exercise include the following:

- Reduce stress on joints
- Increase muscle strength and tone
- Decrease pain
- Increase cardiovascular function
- Improve balance and coordination
- Decrease edema
- Improve posture and trunk control
- Increase in limited range of motion
- Improve circulation due to hydrostatic pressure
- Warm water promotes relaxation
- Improve proprioception
- Improve kidney function
- Increase respiratory function – due to hydrostatic pressure

Buoyancy is the upward pressure exerted by a fluid in which a body is immersed. Buoyancy decreases the stress placed on joints resulting in less pain. It also assists with movement by eliminating gravity. Buoyancy provides resistance. **Fluid resistance** is the force that opposes motion of an object through a fluid. It is necessary to push through water in order to move yourself, thus acting as resistance. Fluid resistance supports the body in water and assist you with holding your position. Resistance also allows for increased resistance without the use of weights, limiting the distraction of joints. Fluid resistance improves balance, increases sensory awareness, and improves reaction time in a gentle environment. **Hydrostatic pressure** is the fluid pressure exerted equally on all surface areas of an immersed body at rest at a given depth, thus the deeper the body part is in the water, the greater the force. Hydrostatic pressure reduces joint and soft tissue

swelling. It improves joint position awareness and assists with venous return. Mild resistance of rib expansion –breathing in neck deep water is exercise in itself. Warm water allows for **relaxation** of muscles and blood vessels, improving blood flow.

Relative density is the relation of the mass of an object to the mass of the equal volume of a liquid at standard temperature and pressure. Objects that are more dense than water, will sink. Objects that are less dense than water, will float. Muscle is more dense than adipose tissue (fat). Therefore, a person that is mad up of more fat than muscle will end up having an easier time floating, than a person made up of more muscle.

Turbulence is random motion of water as it responds to a disturbance. Benefits of swirling of water against the body (water massage) include increased circulation and decreased pain. Turbulence overpowers pain message to the brain (like a TENS unit does). Change in speed or direction can alter the turbulence force. Increase turbulence with gloves, paddles, jets, and more people in water around you.

It is normal for muscles to feel sore or tired for 24-48 hours after exercising. It is not normal to have the following symptoms after exercising:

- Muscle cramps/spasms
- Muscle twitching
- Increase in muscle/joint pain
- Decrease in range of motion
- Decrease in functional activity
- Extreme Fatigue

If you exhibit signs of overuse, you did too much. Decrease one or more of the variables. If symptoms continue, discuss with your PT or doctor. Water exercise can be deceiving, it may seem easy and that you can do more than you should. The first time you exercise in the water, stick with an easy workout, until you see how the water has affected you. Warm water can increase fatigue…use caution. Be sure to hydrate as well; you still sweat in water. Wear pool shoes for better traction. And remember your body may be tired after water exercise, so be sure to have a safe way to get out of the pool. It's also a good idea to never go in the water alone.

The follow are some types of aquatic exercise that some EDSers may want to try:

- **Ai Chi**: strengthens and tones body while enhancing relaxation
- **Aquatic PNF**: proprioceptive neuromuscular facilitation, which uses patters of movement in different positions with specific exercises.
- **Bad Ragaz Ring Method**: a series of movements while being supported by a ring or float at neck and hips.

- **Back Hab**: Walking program using varying strides and stretches; useful for individuals with back pain, hamstring injuries or decreased abdominal strength
- **Fluid Movements**: individuals follow a series of movements based on early developmental stages of infancy
- **Halliwick Method**: rotational patterns are performed to improve balance and postural control.
- **Swim Stroke**: using a variety of stroke forms
- **Watsu**: moving the body in a way that enables one part of the body to be stretched at a time

2 Aquatic Exercise Equipment Types include:

- **Assistive**: floats (noodles, vests, belts); dumbbells; webbed gloves; flippers; kickboards
- **Resistive**: webbed gloves; foam dumbbells; paddles; flippers; jets

So, **a warm up** involves 5-10 minutes of gentle movements. It is important to warm up before an aquatic exercise in order to adapt to being in the water. A great way to warm up is to walk in the water for about 5-10 minutes. Stretching and Range of Motion (ROM) can also be used as a part of the warm up. ROM should only be done within the normal range. Stretching should focus on any areas that are tight and should only be done within the normal ROM. Warm water will relax the muscles, enabling muscles to stretch easier. This can lead to overstretching/hyperextension in the EDS population which is contraindicated. So, if you're an EDSer, use caution.

Some variables that impact **strengthening/toning** include: buoyancy, resistance, surface area, turbulence, lever arm length, speed depth, frequency, and repetitions. The deeper the water, the less force will be placed on the submerged joints. If you are at C7 (neck level), you are at 10 % of your body weight. If you are at the xiphoid process, you are at 33% of your body weight. If you are at the level of you belly button, you are at 50% of your body weight. You should utilize buoyancy to provide whatever support your body may need (utilize the floating position on the back; move arms and legs in a way that the water supports you). You should also utilize buoyancy to assist by positioning yourself in a way that the water will assist wit the movement (in neck deep water, palm on the thigh; allow water to assist hand coming to surface; longer lever arm will increase ease of movement). Finally, you should buoyancy to resist your movement (when neck deep, palm on surface of water and pull down to thigh. This same exercise can be made harder with the addition of a floatation device such as a Styrofoam dumbbell. Longer lever arms increases difficulty). You can also increase the resistance by increasing the speed, and vice versa (decrease resistance by decreasing speed). Movement of water around you will challenge your balance and strength. Water propelled by jets will

increase the turbulence of the water and increase the challenge. Moving against turbulence is amongst the greatest of strengthening/toning challenges. Only progress through these levels when you are able to successfully perform at each level. The more people in the water, the more the turbulence. Increasing the surface area of the water being displaced by movement will also increase strength. Make sure your hands are open with your fingers spread apart in are to move through the water easier (closing your hands will make it hard to move through the water). Moving through water with the side of your palm cutting through the water will be easier than if you move through the water with your palm flat. This applies to the concept of surface tension. Some aerobic exercises that can be done in the water include: walking, swimming, deep water bicycling, deep water jogging, and treadmill walking/running. 10-20 minutes of straight aerobic exercise is the goal. You may need to start at 2-3 minutes and gradually work up to a longer time frame.

The **cool down** involves 5 minutes of relaxing movement (either slow walking, or gentle swimming/floating).

Water Temperature Recommendations:

- 85-88 degrees F for active/aerobic exercise
- 88-92 degrees F for passive or gentle active exercise
- Over 95 degrees F at risk for excessive fatigue and dehydration

For those with POTS, movement will minimize issues. Issues can result from change of position (supine to stand) and static postures (sitting/standing for longer than 20 minutes). For Vascular EDSers, low impact aquatic exercises and minimal aerobic exercise is recommended.

The following is a list of **Precautions** for Aquatic Exercise:

- The fear of water
- Impaired mobility getting in and out of the pool
- Significant balance or vestibular disorder
- Orthostatic hypotension
- Recently healed surgical incision
- Absent or impaired peripheral sensation
- Diabetes
- Respiratory dysfunction
- Colostomy
- Difficulty with bowel or bladder control
- Seizure disorder controlled well by medications
- Tracheotomy tube
- Compromised vision without corrective lenses
- Compromised cardiac or respiratory system (poor endurance or asthma)

The following is a list of **Contraindications** for Aquatic Exercise

- Cardiac Failure
- Fever
- Infection
- Urinary infections
- Open wounds
- Infectious diseases
- Contagious skin rash
- Excessive fear of water
- Severely weakened or deconditioned state
- Uncontrolled seizures
- Bowel or bladder incontinence
- Colostomy bag or catheter used by patient
- Cognitive or functional impairment that would crate a hazard to the patient in the pool
- Poor endurance
- Abnormal tone
- Severe or decreased range of motion that limits function

PILATES: What can it offer the EDS Patient?

Presented By: Crystal Seamon, PT, DPT, OCS, ATC

Pilates, along with Yoga, Tai Chi, and other low impact exercises are often recommended to individuals suffering from pain or hypermobility to "strengthen your core" When Crystal is asked whether or not someone should do Pilates – perhaps they have been dealing with low back pain or have heard that it is a great exercise to strengthen your core. Often her response is – "It depends – it depends on the type of Pilates you want to do, the level of difficulty, the skill of your instructor, and what your overall goals are."

Specifically for EDSers – Pilates can be great but at the same time in the wrong hands it can be terrible. Every EDSer in general does not begin their journey with Pilates at the same point and is challenged by different areas of strengths and weakness. What seems simple as you know may leave you in severe pain for several days.

Chances are you have probably heard someone talking about Pilates at some point in your life – maybe a friend, a coworker, your doctor or even your physical therapist has recommended that you try it – either because they are doing it and think it is the greatest thing since sliced bread – or because you've tried just about

everything without fail but haven't found the magic trick yet. When done carefully Pilates can be a wonderful exercise for EDSers – but bear in mind this doesn't happen without some bumps along the way just like with anything else as your learn your body's limits and capabilities.

Many people think that Pilates is a relatively new exercise fad developed in the last decade or so when in fact Pilates was first invented by Joseph Pilates in 1912. Plagued by his own physical and health limitations Joseph Pilates studied several different types of exercise which went into to creating this method of exercise which he then later went on to use in order to rehabilitate injured WW1 soldiers. Joe believed that injuries were caused by imbalances in the body and habitual patterns of movement. He observed that when a person had a weakness or maligned area, that person overcompensated or overdeveloped another area to achieve a certain functional movement. Thus, he found it was critical to correct the malalignment and to re-educate the body to prevent recurrence. Certainly ahead of his time, this is a belief that, 50 years later, is being hailed as the phenomenon of the muscle imbalance theories of physical therapy today. Eventually he moved to New York with his wife Clara and set up the first pilates studio as we know it now – with a primary emphasis on working with dancers. In a small way if you think about it many dancers are very flexible and mobile – sometimes almost too much for their own good. Pilates was a way that they could improve their strength without compromising their form or art of dancing. Originally the focus of his program was on 34 original mat exercises – which still continues to be included in mat classes today. From a rehabilitative sense not all of these exercises come close to looking or being considered safe as you will see in a few slides for even the healthiest person without EDS. He eventually went on to develop several pieces of equipment that allowed people to add resistance their exercise routine.

Matwork Pilates is done on a mat. It is the most widely know and accessible form of Pilates because it is relatively inexpensive approx $10-15/hr or obviously free if you practice on your own, requires minimal equipment, and can be performed anywhere – all you need is yourself and a mat. Matwork promotes core stability, flexibility, endurance, postural and body awareness, and correction of muscle imbalances. One thing to keep in mind though is that matwork may actually be more challenging on your body since you do not have resistance from the equipment helping guide your body and give you feedback to perform correct movement patterns.

Equipment based Pilates assists with achieving proper alignment and control while providing appropriate resistance for strengthening and endurance. It allows for weight bearing exercise and gives proprioceptive feedback to enhance stability and muscle control. The most widely used Pilates machine is the Reformer. Other machines include the Cadillac table, Wunda Chair, and Barrel. Crystal's preference

for Pilates is to practice on a reformer – as someone who struggles with neck pain and headaches it allows her to modify her routine to avoid stressing her neck and back, but at the end of the day still get a great workout. One of the largest downsides for practicing Pilates on equipment is usually the price – private sessions can cost anywhere from $55-95/hr depending on the studio and instructor. And while group classes are less expensive at approx $25-40/hr – it requires a greater understanding of your body's limits, knowing how to modify classes, and may provide you with as much hands on feedback or corrections.

Pilates has become widely popular because it can be a gentle non-aerobic workout which lengthens and strengths our muscles, improves our posture – and most importantly does all of this without stressing our joints or the heart. Similar to tai chi and yoga it also focuses on the mind body connection that emphasizes a balanced body through core strengthening and improved awareness. The exercises can be adjusted or modified by adding more or less resistance through our own body weight or adding springs, weights, or bands.

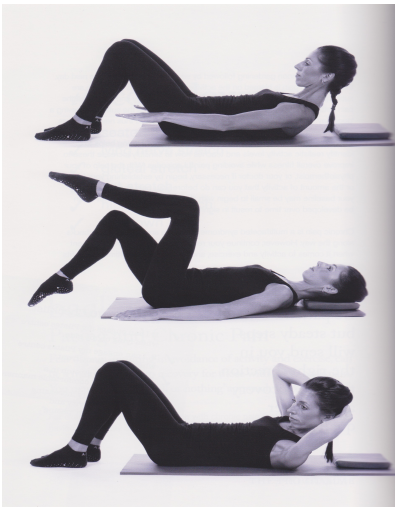
That being said….one of the important things to remember about traditional Pilates or Pilates practiced in a group setting is that it is that it was originally designed for healthy and fit individuals. There are several exercises that are extremely advanced and require not only good control of our muscles but also good awareness and proprioception of where our body is in space – especially for an individual with EDS initially this may be very difficult– and may be dangerous for you to perform.

There are several Pilates mat exercises with similar variations performed on the reformer which contradict Crystal's training and beliefs in regards to what is healthy for our bodies….some of these include: rocker with open legs, neck pull, roll over, jack knife, corkscrew, control balance, boomerang, rocking, bicycle. There is a common theme across these exercises…there is significant pressure and weight being borne across the neck as well as many of these positions place our nervous system on significant tension which can be very harmful for our nerves.

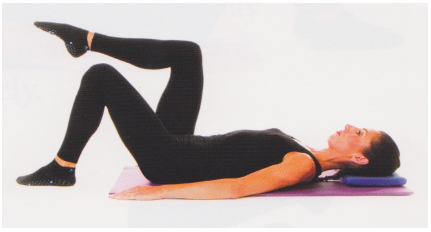
So, as an EDSer, you want to look for modified or therapeutic Pilates exercises. Which simply means you modify the movements of the original exercises that are safer, achievable, and more inline with the way our bodies are supposed to function. This may be achieved by designing a program with your physical therapist or working privately with a skilled and knowledgeable Pilates instructor who can adapt your exercise routine to your specific strength and weaknesses.

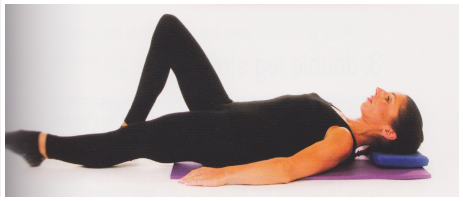
When you are beginning a new exercise remember that modification is key to success. Modifications should always be made to maintain appropriate alignment and muscle engagement by decreasing motion or resistance and performing less

repetitions until endurance and control improves. Avoid end range stretching. It may feel good in the moment, but over the long term you need to find a sense of connection, internal support, and structure in your body.

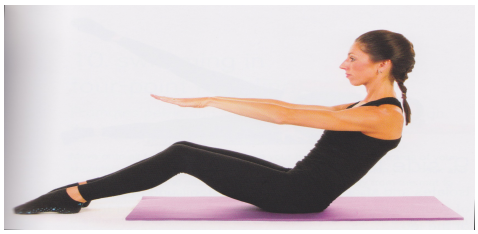


If you experience neck pain…Avoid "C" curve or lifting your head with abdominal work.





Your arms and legs are heavy. Cross your arms over your chest. Keep legs in tabletop or on the floor and avoid lowering them too close to the ground.



Bend your knees or place a small lift under your hips for hamstring tightness, nerve tension, or back pain.

Place a foam wedge, towel roll, or folded edge of the mat under your wrists

Bear weight through your elbow.

At the age of 86 Joseph Pilates said…."I must be right. Never an aspirin. Never injured a day in my life. The whole country, the whole world should be doing my exercises. They'd be happier."

Many types of people, at many levels of fitness, can participate in Pilates and see improvements in ROM, flexibility, circulation, posture, abdominal strength, and decreased pain.

With regular committed Pilates workouts you can expect to:

- Increases the efficiency and effectiveness of exercising
- Develop a strong core
- Learn stability and controlled flexibility
- Create a well balanced body and prevent injuries
- Develop body awareness to transfer to everyday functional movement
- Reduce stress, relieve tension, and boost energy
- Restore postural alignment

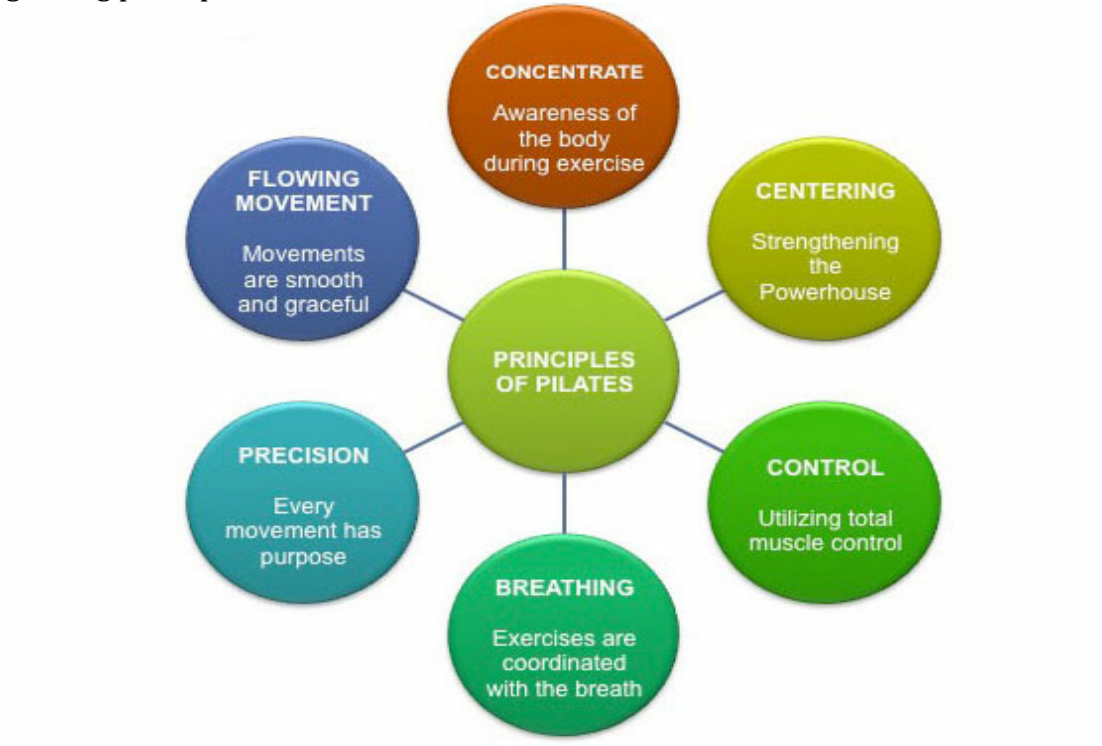
It is gentle and low impact, but also challenging. It is an extensive exercise repertoire that can be modified to fit the needs of everyone.

Our core is made up of two systems – the small local muscles and the large global muscles.

The local muscles are comprised of our diaphragm – which highlights the importance of proper breathing techniques with exercise or exertional activities The transverse Abdominus, multifidus, and pelvic floor. Together these four muscle groups work to create a cylinder or corset like mechanism of stability around our spine. Their primary role is to provide stability and support the movements of our body throughout the day. These muscles are the "center" or "powerhouse" of your core. These muscles are commonly found to be weak or dysfunctional in most individuals who experience spinal pain or hypermobility.

In contrast, our rectus abdominus, glut max, erector spinae, lats, and obliques are our global or power producing muscles. Most often we see that individuals with back pain tend to over activate these muscles – which can become very compressive on our spine – eventually causing more pain and compensations. Pilates teaches us to increase activation of our local system and utilize our global system only when we need it – to generate force or power to strenuous activities – not at rest or with low level activities.

Before you start it is important to understand the 6 Principles of Pilates to maximize your results. Because as Ralph Waldo Emerson once said…."As to methods there may be millions and then some, but principles are few. The man who grasps principles can successfully select his own methods. The man who tries methods, ignoring principles is sure to have trouble. "



Understanding the 6 Pilates Principles are essential to a safe and efficient workout. The emphasis is always on quality over quantity. These principles teach us the ability to concentrate and be aware of how our body is moving with precision so that we utilize our muscles as efficiently as possible. **Learn how to center or activate our abdominals or core while we move our arms or leg**s. **Possess control over how we move our body**. And above all **Master correct breathing patterns.** And if we can put these all together you will create a graceful and smooth flowing movement.

If you decide to do private lessons then the next step is to find a qualified instructor. That individual should understand your hypermobility and monitor and adapt exercises as needed.

They should be well-qualified in Pilates. Ideally you want to work with someone who has completed a comprehensive Pilates training from a reputable source. Some questions you may want to ask when looking for an instructor are:

- Does the instructor have other movement-related teaching experience?
- How long have they been teaching Pilates?
- What is their philosophy and specialty?
- Are they able to handle special needs, injuries, and rehab?

If you decide to participate in Mat class…make sure your mat is dense enough to protect the vertebrae of your spine. A thin yoga mat will not do! When it comes to clothing, generally form fitting clothing is recommended to allow yourself and the instructor to ensure good form and alignment. Ultimately, wear clothes that you are comfortable in and allow you to easily move.

"Patience and persistence are vital qualities in the ultimate successful accomplishment of any worthwhile endeavor" – Joseph H. Pilates. Pilates is not a quick fix! If done correctly you should start to feel some benefit after 4-6 weeks. It is worth persevering…there is much anecdotal evidence that regular Pilates practice can dramatically reduce symptoms suffered by many individuals with hypermobility. Always approach Pilates with care. Be aware of your own vulnerabilities. Take time to choose an instructor. And remember, Pilates should never be painful!

You can find more information out at:

- www.Pilates.about.com
- www.pilates.com
- www.polestarpilates.com
- Back to Life with APPI Pilates: A six-week program to refine, tone and strengthen your body by Elisa and Glenn Withers

- Teaching Pilates for Postural Faults, Illness & Injury: A Practical Guide by Jane Paterson

TAI CHI for Ehlers-Danlos Syndrome

Presented By: Ralph Dehner

Tai Chi therapy increases muscle strength, which supports and protects the joints. It increases stamina, joint stability and improves posture, memory and relaxation. Tai Chi helps balance, thereby reducing falls.

Cervical Instability in the EDS Population

Presented By: A Atiq Durrani, MD

Some cervical spine issues found in EDS include: C1-C2 instability; Cranio-cervical instability; Lower Cervical Kyphosis; Cervical disc degeneration (most common at C4-C5 & C5-C6); and Chiari Malformation.

Common symptoms of cervical instability include occipital headaches, neck pain, passing out at the extremes of lateral rotation, choking sensation, base of skull tenderness, and jaw pain.

If you answer "YES" to the following questions, you may be suffering from cervical instability.

- Do you have pain/pressure at the base of your head?
- Does your pain radiate behind your eyes?
- Does your pain radiate to your neck or shoulders?
- Is it worsened by coughing/sneezing/bowel movements?
- Do you have general neck pain/stiffness?
- Women: Is your headache worsened by menses?
- Do you have pain or pressure behind your eyes?
- Are you sensitive to light?
- Do you have blurred vision?
- Do you have double vision?
- Are you missing a portion of your visual field when looking straight ahead (Field Cuts)?
- Do you have pressure in your ears?
- Do you have dizziness with position changes?
- Do you have feelings of unsteadiness when standing and/or walking?
- Do you have high-pitched ringing in your ears?
- Do you have tremors?

- Do you have decreased hearing?
- Do you have very sensitive hearing?
- Do you have vertigo (feelings that you or the room are spinning)?
- Do you have difficulty swallowing?
- Do you have throat tightness?
- Do you have difficulty speaking?
- Is your voice changing, becoming hoarse?
- Do you have sleep apnea?
- Do you snore?
- Have you ever "passed out"?
- Do you have palpitations?
- Do you ever have shortness of breath?
- Do you have frequent nausea?
- Do you suffer from chronic fatigue?
- Do you suffer from short-term memory loss?
- Do you suffer from long-term memory loss?
- Do you suffer from depression?

If you suspect that you have cervical instability some neurologic tests that your doctor should order for you are:

- MRI of Brain
- Cine MRI (CSF flow study)
- MRI of Cervical Spine
- MRI of Thoracic Spine
- MRI of Lumbar Spine
- CT of Head
- CT of Cervical Spine

Treatment options involve physical therapy, cranio-sacral alignment, and cervical collar braces. Some interventional pain procedures include occipital nerve blocks and cervical epidural/foraminal injections. When conservative procedures fail to control symptoms, then Cervical Spinal Fusion becomes the preferred Surgical Treatment.

Symptomatic C1-C2 instability in EDSers can be surgically treated with a C1-C3 fusion. A study involving 25 patients, all of which underwent stabilization for C1-C2 instability. There was a 1 year follow up for all patients. The mean pre-op pain was reported to be an 8 out of 10. The mean post-op pain at one year was reported to be a 2 out of 10. One patient reported still having residual pain. Another patient reported a screw fracture. Headaches resolved in 92% of patients. When the patients were asked if they would do the procedure again, 95% responded that they would.

Cervical spine instability is a common reason for the headaches and cranio-cervical pain many EDSers experience. It is under-appreciated by the spine community and not very well understood. In many circumstances, patients complaining of such complaints go through extensive work up with no treatment offered in the end. Stabilization of an O-C1-C2 complex resolves cranio-cervical symptoms in EDSers.

Shoulder Instability in Patients with EDS

Presented By: Keith Kenter, MD

A single dislocation does not mean that the patient is going to suffer from recurrent instability. Some definitions to be aware of include:

- **Laxity**: range of motion of the center of the humeral head with respect to the glenoid fossa due to an external force
- **Instability**: symptomatic inability to maintain the humeral head in the glenoid fossa.
- **Subluxation**: partial dislocation; incomplete separation of joint
- **Dislocation**: frank separation of joint
- **Glenohumeral Instability**: complex interaction between physiologic laxity to provide range of motion and joint stability.

The EDS shoulder has increased laxity and is at higher risk for instability (MDI). This introduces 3 different types of constraints: Passive, Static, and Dynamic. Passive Constraints involve negative pressure and/or joint fluid cohesion around and within the humeral head and glenoid fossa structures. Another cause of passive constraints is a fibrocartilagenous lip that increases the glenoid depth and humeral contact area (75% superoinferior; 50% anteroposterior). Static Constraints involve the capsular envelope and glenohumeral ligaments. The glenohumeral ligaments are can exhibit up into 3 types of restraints: Superior; Middle; and Inferior.

- Superior – restraint for inferior translation in adducted shoulder
- Middle – restraint for anterior translation in 45 degree abducted shoulder
- Inferior – restraint for anterior and inferior translation in abducted shoulder

Finally, Dynamic Constraints involve the rotator cuff group, the bicep tendons, and scapular rotators.

Treatment involves immediate reduction of the dislocated shoulder. Two physical therapy programs to consider are Rotator Cuff Strengthening and Scapular Stabilizer Strengthening. Surgical intervention is something that can also be ultimately considered. Above all, patient education and defining the collagen disorder are paramount. Once the patient is diagnosed, modification of activities

should be implemented and work should be done on the patient's mechanics. It is important to work on core strength, spine posture, RC strength, and scapular muscle strength. When considering the surgical option, it is important to understand that surgical treatment results in about 30% recurrence in patients without anatomic lesions.

There has been a rapid evolution in arthroscopic techniques. Early techniques secured the labrum to bone. Now, techniques address capsular laxity via capsular shift, capsular split, capsular plication, and thermal 'shrinkage.' Some advantages to the arthroscopic techniques is the ability to visualize all pathology, that it leads to less stiffness, and is easier to revise. Some disadvantages to arthroscopic techniques is that it is less reliable and technically demanding. There is a higher failure rate and the occurrence of portal scars. Some contraindications to arthroscopic techniques include: capsular deficiency, glenoid bone loss, humera head defect, collision athlete, and the surgeon's skill level not being up to par.

In conclusion, there is a complex interaction between stability and mobility. Engaging a patient in a neuromuscular training and strengthening program for the shoulder girdle is paramount especially in MDL. Surgical emphasis is to restore anatomy and capsular tension. The arthroscopic challenge today is reproducibility of a quantifying amount of capsular redundancy during repair.

Occupational Therapy Management of Hypermobility In Ehlers Danlos Syndrome

Presented By: Michelle O'Sullivan (OTR/L) & Michele Pavlis (OT)

Occupation: the roles you play in your life and the performance of tasks and activities than enable you to fulfill those roles.

Therapy: enabling you to fulfill your roles and find meaning in your activities

Occupational Therapy is a team approach. Occupational therapists work with physical therapists, speech therapists, psychologists, doctors/physicians, and social workers/counselors. The aim of Occupational Therapy is to promote and maintain the patient's independence in as many aspects of their life as possible. In order to see an occupational therapist, you will need to obtain a referral from you doctor. You will most likely need to personally request this from your doctor as not all medical professionals understand occupational therapy roles and services which can be helpful for EDSers. You need to specify to your doctor that you require a full assessment of your activities of daily living.

An occupational therapist may make splints, order assistive devices/equipment through your insurance, do a home assessment (if necessary), make recommendations for modifications or other equipment that you may benefit from, help with problem solving and cognitive strategies, provide recommendations for exercises and activity modification, access programs and services in your community, and build and maintain support systems.

EDSers usually present with physical difficulties after experiencing recurring joint dislocations/subluxations, in addition to pain, fatigue, gastrointestinal problems, and mobility issues.
"We need to learn to move efficiently so that we can save our limited energy for the important things," by keeping frequently used items in a special place which is always in the same place, write checklists, keep everyday items such as cutlery, plates, bowls and glass at waist level, make use of online shopping for groceries, or do multiple small shopping trips per week or take a friend with you to help and save a trip, and use a small bag to put frequently used items in. Use your brain to save your body.

Just because our thoughts impact our pain…it doesn't mean the pain is 'all in our heads.' It doesn't mean that we are somehow willing the pain and making it happen. It doesn't mean that if we have complete control of our thoughts and always think positively, that pain won't occur. However, keeping a thought diary, working out our instinctual response to pain, and thought management may provide some relief. Other strategies for pain management include relaxation, meditation, diversional therapy and goal setting & rewards. Relaxation and meditation takes practice and may be a particularly helpful strategy for falling asleep. Diversional therapy involves watching TV/DVDs, listening to or playing music, reading, meeting up with friends or family and doing something/chatting, painting, craft, going to the movies, baking/cooking, painting your nails (girls), playing games (digital or board games), chatting over the phone or skype with a friend, watching sport, enjoying nature, DIY jobs (home projects).

Other therapies & modalities include heat or ice packs, aquatic therapy, physical therapy, cranio-sacral therapy, reiki and healing therapies, massage therapy, acupuncture, pilates, tai chi, dry needling, trigger point injections. '

Making Life Easier:

- In the Kitchen
    - Check countertop height
    - Easily accessible utensils and frequently used items
    - Minimize the weight of appliances and other items like pots & pans

        - Be careful of how heavy food/drink items are that you purchase at the shops
    - In the Bathroom
        - Can you reach things?
        - Do you need some assistive devices to help you bathe independently?
    - In the Bedroom
        - Easy access to items used most often
        - A good mattress & pillow
        - Decrease the amount of electrical equipment & technology in your room
        - Make your room as dark as possible to promote sleep
    - In the Living Room
        - Check the height of your sofa or couch
        - Are there any trip hazards?
    - In the Car
        - Check steering wheel and pedal height
        - Seat position
        - Handicap placard
    - In the Work Place/Work Station
        - Check desk height
        - Check workstation set up
        - Take Breaks
        - Check lighting and airflow

Some assistive devices that some EDSers may find useful are as follows:

- For Meal Preparation
    - JarKey
    - CanKey
    - Zyliss 5 in 1 opener
    - Dycem Jar opener
    - Dycem Grip Mat
    - Cordless Kettle Tipper
- For Eating
    - Cylindrical foam (for utensils)
    - Rocker Knife
    - Built up cutlery
    - Trolley Cart
    - Sharp Knives
- For Cleaning
    - Long handled Cleaning Brush
    - Long Handled dustpan & broom

    - Long handled toilet brush
    - Robot Vaccum
- For Bathing
    - Long handled toe washer/dryer
    - Long handled back sponge
    - Toothbrush grip
    - Shower stool
    - Toothpaste dispenser
- For Laundry
    - Machine Raiser
    - Easy Clothes pains
    - Drying line
    - Laundry Trolley
- For Work/School
    - Slope Board
    - Footrest
    - Laptop dock
    - Gel wrist support
    - Copy holder
    - Ergonomic Mouse
- Other Assistive Devices
    - Key Turner
    - Swivel Seat
    - Car Handy Bar
    - Tap Turners
    - Blister pack opener
    - Meds organizer
    - Meds reminder

You are not your ability or your disability. Your EDS does not define you. You are a person with EDS, not an EDS patient. There will be setbacks along the road; life might change direction completely…but who you are does not change. When setbacks occur, we need to redefine our expectations and goals, and reassign our energy, skills, and talents to new things. An occupational therapist may be just the resources you need to achieve all of this and lead a life that is meaningful to you.

It's Not In Your Head –Or Is It?

Presented By: Howard P. Levy, MD, Ph.D.

Obviously dislocations/subluxation, acute and chronic muscle spasms, neuropathic pain, degenerative arthritis, and other similar ailments are not in your head. Pain however is a subjective experience and is strongly affected by one's mood, attitude, goals, expecations, and fears. Pain can be exasturbated by isolation, disability, avoidance and other social behaviors. If one can learn to avoid psychological pain escalation and learn psychological pain control, he/she will experience less pain, will have to take less medications, and ultimately will experience fewer side effects.

Factors that alter the pain experience include: emotional state of the patient, thoughts, beliefs, intentions, injuries to social relationships, memories of past injuries, and emotional state of close loved ones. In other words, psychological distress exacerbates pain.
EDSers commonly experience anxiety, depression, low self-confidence, negative thinking, hopeless/helpless thinking, desperation, and low self-efficacy. The thought and belief that "pain will harm me" amplifies the pain experience. In other words, intense self-awareness and hypervigilance (i.e. "waiting for the next shoe to drop") keeps the patient on edge and results in a more intense pain experience.

Many EDSers experience injuries to their social relationships as a result of disbelief by their friends/relatives and a reduced ability to socialize. This can cause resentment, distrust, and hostility between the EDSer and their family and health care team.

Memories of past injuries can cause an increased fear of pain and/or joint instability. By anticipating a negative experience, the EDSer may participate in avoidance behavior, ultimately exacerbating dysfunction and disability.

Endorphins are induced by emotion and exercise and essentially modulate pain. Endorphins are our "natural opioids."

Post Traumatic Stress Disorder (PTSD) is a complicating factor that leads to resistance towards accepting psych etiology as a response to prior misdiagnoses and accusations. Resistance is fed by the stigma of a psych etiology being equivalent to "craziness" and perceived as weakness.

Clinicians must believe pain and other symptoms are real and validated. The patient must believe that there are psych components in the pain experience and management strategies and trust the clinicians. It is important to focus on chronic rather than acute pain management in order to establish reasonable expectations.

This may be done via distraction, hypnosis, and meditation. Counseling for depression, anxiety, PTSD and for accepting, coping and living with pain, dysfunction and disability requires patient acceptance and willingness. It is also to consider the thoughts and feelings of the patient's support system via separate counseling, group counseling, and working on the patient's response to them.

Pain is influenced by cognition, affect, and behavior. The goal is to manage pain and reduce negative consequences. This requires active patient participation.

Some unhelpful thoughts include:

- "Pain means damage; if doing something hurts I should avoid it"
- "…it's hopeless, I should just accept that I'll end up in a wheelchair"
- "I've got wear and tear, better not use my joints or they'll wear out even quicker"
- "I need to rest more, if you feel tired it means you've been doing too much"
- "My pain is a sign of whether I am better, I won't be better until my pain has gone"

Cognitive Behavioral therapy includes: education (and insight); self-efficacy (locus of control); recover function (overcome fears); distraction; relaxation (breathing exercises, muscle relaxation, guided imagery); and biofeedback; reward positive behaviors. Work towards positive thinking by controlling fear and engaging in self-efficacy and assumption of the normal.

Antidepressant medications reduce anxiety and depression and lessen the subjective pain experience. Antidepressant medications have also been shown to directly treat pain, especially neuropathic. Some even improve restorative sleep, leading to less pain.

Note: Unchecked psychological distress can amplify pain. A disciplined mind can reduce pain (mind over matter). "90% of the game is half mental" – Yogi Berra

Vascular Ehlers-Danlos Syndrome: Placing recent studies into context

Presented By: Mitzi L. Murray, MD, MA

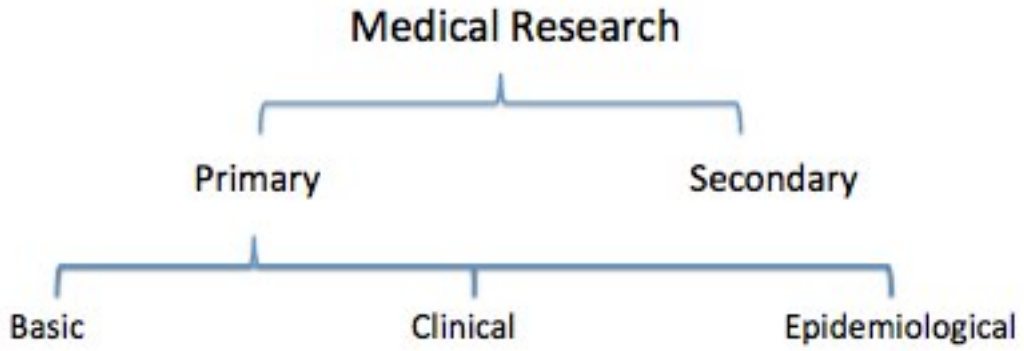
With rare disorders, we often times keep our eyes peeled on relevant research; always with the hope and expectation that the information we need to make health related decisions will become available. However, rarely do we step back to survey the overall research landscape. Understanding the context surrounding new information is important for understanding the significance of that information. We can't always understand everything simply from understanding the context; however, context is critical.

In general, we know that those affected with this vascular EDS are at risk for serious complications – but we also know that every single person with vascular EDS is unique and follows their own path. At this point in time, there are essentially 3 ways to diagnose vascular EDS – clinically based on medical history and physical exam, biochemically by looking at the type III collagen protein and molecularly by studying the gene COL3A1. At this point in time, establishing the diagnosis is certainly important – often offering an explanation and raising awareness of possible future risks, but we still have many questions about how to best care for families living with vascular EDS.

The type of questions that remain unanswered are highly variable and this is just a short list of some examples.

- Why is there variability?
- What factors contribute to risk for complications?
- Should we perform surveillance studies? How?
- Can medications/treatments reduce risk?
- Can we change the course of the condition?
- How are complications best managed?

We oftentimes turn to research to help answer the questions that arise in the clinic. If we each defined what we think of as medical research, we would quickly see that there are many different perceptions.



If we start looking at medical research altogether, we can divide it into primary and secondary research. Secondary research is essentially research studies using other research studies – this can be important for common diseases, but is less often a tool we can use in understanding rare disorders.

Primary research is actually collecting new data in hopes of learning something new that can be used more generally. We can think of it as having three different levels. If we want to look at things smaller than a whole human – such as DNA, cells, non-human organisms - we are talking about basic research. If we want to study one or more humans, we are interested in clinical research. And if we want to step back and look at whole populations, we would be performing epidemiological research.

But the sub-classifying doesn't stop there.

There are several types of basic research. There are studies of animals and animal model of disease – which is when researchers try to re-create a human disease in an animal species for research studies. Some people have strong opinions as to whether this is morally correct, but from a medical perspective, animal models are a very important resource for understanding human diseases.

There is a mouse line that has been created with loss of COL3A1 and recent research has focused on used of a medication called doxycycline in those mice to see if it reduces the risk of vascular complications.

Use of cultured cells can also be very helpful in understanding genetic disorders, as Dr. McDonnell discussed at last year's conference (See 2011 EDNF Conference Notes).

Many times, basic genetic studies focus on trying to identify the cause of rare disorders, such as vascular EDS – but now we can use that knowledge to try to understand whether abnormalities in type III collagen play a role more common types of arterial aneurysms and dissections in individuals without vascular EDS.

There are also different types of clinical research studies. In general, we can think of clinical studies as coming in two different flavors: experimental and observational

"Experimental" studies are what come to mind for many people when they think of medical research – we're doing something to try to influence natural course of the condition. This may be trying a medication, or it could mean performing a procedure or surgery trying to see if it is clearly beneficial.

Observational studies, on the other hand, focus on trying to understand the condition as it is when unperturbed by experimental interference. For example, many of you may be familiar with the paper in the New England Journal of Medicine from 2000 on the natural history of vascular EDS – this provided us with information on the average age of first complications, type of complications and life expectancy of vascular EDSers. Not only is this information important for families and healthcare providers, it is critical to provide baseline information for comparison to determine whether an experimental treatment makes a difference. After all, how can we know if a medication or treatment reduces the risk of a complication if we don't know the likelihood of a complication happening in the first place?

Another type of observational study is called trying to see if there is a 'genotype-phenotype correlation' – this is when we ask whether vascular EDSers but different specific genetic mutations have different clinical pictures. Again, very important information for families but also important for knowing whether participants in experimental research studies actually have different innate risks from other participants based purely on the individual's specific genetic mutation.

Observational studies can also include interviews, questionnaires and surveys studies – these can be important for helping understand aspects of a condition like quality of life and healthcare needs.

In rare disorders, epidemiological studies can be more difficult due to the small number of affected people, but this information is still vitally important to diagnosing and managing rare conditions like vascular EDS.

It is important to note that that a single study rarely is sufficient to answer a question with enough confidence to change clinical care. We want to be sure that the findings are replicable and applicable to patients in a real world setting. A second thing to consider is that using research findings to influence healthcare decisions too quickly can result in harm – one example is the public's response to childhood vaccinations after a study reported an association between childhood

vaccinations and autism. That finding has not only been disproved, it was revealed to be fraudulent. However, we continue to face the ramifications of unvaccinated children suffering preventable illnesses and deaths.

A study from 2000, titled "Clinical and Genetic Features of Ehlers-Danlos Syndrome Type IV, the Vascular Type," published in the New England Journal of Medicine, has been pivotal in better understanding the natural history of vascular EDS. That is, it offers some insight into things like life expectancy and age of first complications. In essence, this is the baseline information that most other studies on vascular EDS will use as a reference.

Shortly following the natural history study was the first report of haploinsufficiency in vascular EDS. In the paper titled "Haploinsufficiency for One COL3A1 Allele of Type III Procollagen Results in a Phenotype Similar to the Vascular Form of Ehlers-Danlos Syndrome, Ehlers-Danlos Syndrome Type IV," now published 11 years ago, the author's commented that individual's with null mutations were seen less frequently than expected. They suggested that this may be because those individuals had a different clinical course from most people with vascular EDS. The reasoning being that those will null mutations had complications less often and, therefore, often never came to clinical attention to be tested.

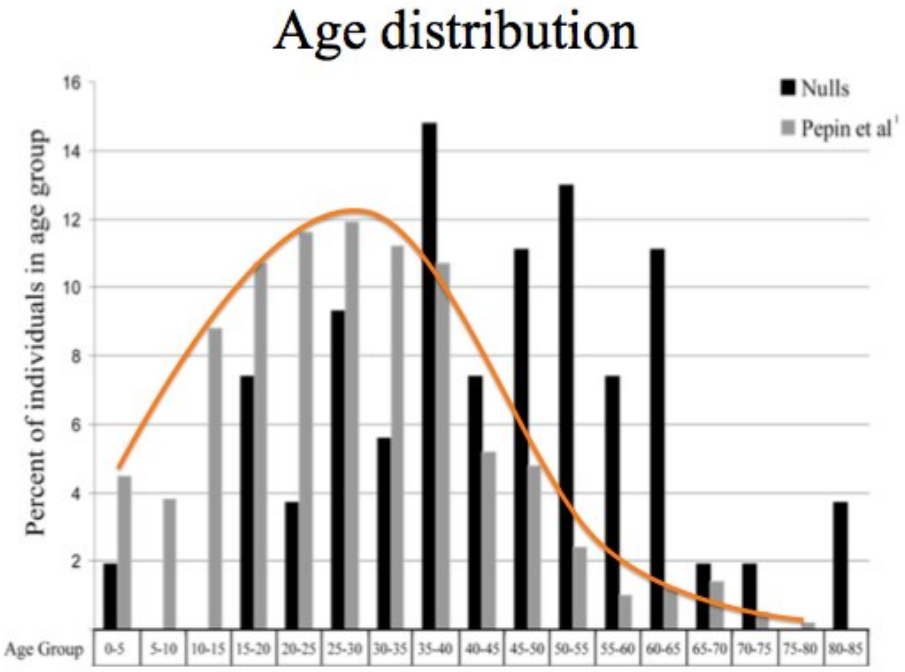
In people without vascular EDS, the typical situation is that there are two copies of the COL3A1 gene, one inherited from the mother and one from the father. Each copy makes protein chains in roughly equal amounts. Three of those chains then randomly come together to form the triple helix of type III collagen. The protein chains do not discriminate in who they associate with – those produced from the maternal copy will associate with those from the paternal copy and vice versa.

Now, if there's a mutation in one copy of the gene that leads to a change in the protein, then half of the chains will be abnormal. The abnormal chains are incorporated into the triple helix of type III collagen and if even one of the three chains is abnormal, it is sufficient for the entire triple helix to be abnormal.

In the case of haploinsufficiency, the mutation is such that it does not cause an abnormal chain to be formed. Instead, it essentially causes one gene copy to not produce any chains at all. All the chains come from the other copy of the gene and are normal. The result is that all the type III collagen made is normal, but it's only half the normal amount because the unaltered copy of the gene doesn't get turned up to compensate for the other. Another way to think of it is quality vs. quantity.

About a year ago, Dru F. Leistritz, MS and others looked at the natural history of families with vascular EDS due to haploinsufficiency in closer detail. Referring back

to our medical research framework, this was a clinical observational study. 19 families with haploinsufficiency were identified with 54 total people. When they looked at complications, most of those who were the first diagnosed in their family had complications – this is expected because there had to be some reason for them to have been tested in the first place. However, when people were tested because a relative was diagnosed, then major complications were seen in less than a third.



In looking at the ages of those tested, it was observed that many more people with haploinsuffiency were alive at older ages compared to the other natural history study mentioned previously. Here we have the ages from birth to 85 years and the bars represent the percent of individuals who were tested. The higher black bards seen outside the orange curve represent more people with null mutations being tested at older ages, likely because they never had a reason to be tested when younger – they are living longer and having less complications to bring them to clinical attention.

If we step away from major complications and look at minor features, we see that the majority of people with haploinsufficiency have no minor features suggestive of a diagnosis of vascular EDS.

In summary, the study found that individuals with haploinsufficiency due to null mutations have a different clinical course with longer life expectancy, later age of first complications, less bowel and pregnancy complications and less minor features.

Of course, there are some limitations we have to keep in mind – the most important being that these are the families with haploinsufficiency who came to testing because someone had a complication – based on what we know of the gene sequence, it's likely that the many families are never identified because there's no complications that lead to testing. For this reason, the risk estimates may be higher in this study than actuality.

What implications does this study have? For families, it may be somewhat reassuring. Or, as one father explained to his daughter – "if you have to have it, this is the one you want". From the research perspective, it will be critically important to take mutation type into account in the study design.

So, it seems that while having half the amount of type III collagen does represent some risk, it may be sufficient to accomplish the goal. On that note, basic research may lead us to further explore use of animal models in vascular EDS research and create a better understanding of the role of type III collagen in more common forms of aneurysms and dissections. There is an ongoing NIH study on natural history, and pregnancy outcomes in vascular EDS. Perhaps in the future we will be able to conduct clinical research testing Doxycycline in humans, celiprolol in people only with COL3A1 mutations, and imaging surveillance and intervention studies.

In summary, we're making progress but we will never know enough fast enough. It'll be important for the individuals and families living with vascular EDS, to drive research to answer their questions. Healthcare providers and researchers tend to be very limited and biased in the types of questions they ask – they ask questions as a doctor, not as someone living with vascular EDS. It really is up to the EDS community to point them in the right directions and that can only be done through being engaged and working together as a team.
The "Effect of celiprolol on prevention of cardiovascular events in vascular events in vascular Ehlers-Danlos Syndrome: a prospective randomized, open, blinded-endpoints trial" was mentioned last year by Dr. Black. In essence, this is the first clinical interventional study of reasonable scale to test a medication in humans with vascular EDS. The goal was to assess the preventive effect of celiprolol on major complications. Celiprolol is a type of blood pressure medication called a beta-blocker and the theory behind using it is that it would decrease blood pressure, which decreases the mechanical stress on the vessels and, therefore, decrease the risk of arterial dissection or rupture. It's important to note that this study began before genetic testing was standardly available – for this reason, many people weren't tested until after they enrolled. Those eligible for the study had either 1 major and 2 minor criteria, as listed here, or 4 minor with no major criteria. 53 people were enrolled, treated half with celiprolol and the other half received no study medication, then they observed the rate of complications. At face value, their

findings are exciting and lead to the study being stopped early – they observed a 69% risk reduction in the group that received celiprolol compared with the group that did not. Interesting, there was no significant difference in the blood pressure of the treated group.

The issue with the two groups being different warrants caution in how excited we get about the results. In addition, if it's true that celiprolol is beneficial, then it warrants questioning how it works since it wasn't observed to lower blood pressure, which was the driving theory behind choosing this medication. In the end, the author's concluded that the results suggest a benefit but that the results must be carefully assessed.

Is there reason to be optimistic – yes! Is further research needed to try to replicate this finding – absolutely! Should we start prescribing everyone with vascular EDS a beta-blocker solely for the purpose of preventing complications – not so much.

Some of you may be wondering…what do we have to lose – why not just do it? It is important to note that because no medication is without potential side effects or harms – beta-blockers, included, it is hard to justify exposing someone to a potential harm unless there is good evidence that there is a potential benefit to balance it out. This study was not sufficient to establish a true benefit. A second reason may be a little more difficult to understand, but it's just as legitimate. If everyone just starts having their doctor prescribe a beta blocker with the hope that it might work, then we forever lose the opportunity of knowing whether it actually does or not. If we want to know the truth, then we have to engage individuals with vascular EDS who decide to take medications for the sole purpose of risk reduction in research studies to that we can learn what works – otherwise, we will find ourselves in exactly the same position when talking to future generations that we are now…no progress will have been made.

Temporomandibular Joint & Cervicocranial Dysfunction in the EDS Patient

Presented By: Dr. John Mitakides

EDS is the name used for a group of connective, often hereditary, tissue disorders. This condition affects the body's collagen, which literally holds the body together, resulting in loose, flexible joints. Among affected joints are those in the neck and jaw, often triggering TMD, requiring specialized care.

Temporomandibular Joint Disorder (TMJ or TMD) is "shorthand" for a complex syndrome of dysfunction of the jaw to the skull, including the cartilage and related muscles including the related pain and symptoms.

Details of the symptoms of TMJ include: Locked jaw (open or closed); jaw that deviates to affected side; problems finding stable bite position; TM joint noise when opening or closing (cracking or popping); and overall limited jaw movement.

Complex and overlapping symptoms include: *Frequent headaches, occurring when upon waking and may possibly redevelop in late afternoon; abnormal and/or painful jaw movements; ear pain; pain in or around the eye area; check pain; and mandibular pain.*

Cervicocranial Disorder or CCD is "shorthand" for a complex disorder emanating from the upper vertebra of the neck, including the related pain and symptoms.

Classic Cervicocranial symptoms include: limited head movement (especially rotation); trouble swallowing; forward head posture; upper back pain; sore, tender or weak neck; frequent "snapping" or "popping" of the neck with regular head movement; and cervical referral pain into the facial area.

The overlap between the Trigeminal nerve, and the greater Occipital and Cervical nerves results in a convergence mechanism. The Trigeminal Nucleus Caudalis extends to the C2 spinal segment and to the lateral cervical nucleus in the dorsolateral cervical area. Symptoms in the trigeminal or cervical territories produce symptoms in either area.

Potential Sources of TMJ & CCD headaches include:

- Muscular spasms & stricture
    - Temples
    - Back of head (Occipital)
- Circulatory (constriction OR dilation)
    - Back of head (Occipital)

    - Below the ear (Mastoid)
- Neurological aberrations
    - Migraine-like headache
    - Referral (source ≠ painful spot)
- Skeletal (Vertebral) Displacement
    - Occipital (or Cervical) Referral

Ear pain mimic an earache. Tinnitus is the term used to describe ringing in the ears Ear pain, if left untreated, can lead to hearing loss and even itching within the ear.

A diagnosis of EDS often precedes a diagnosis of TMJ. A preliminary exam of skeletal joint mobility is often performed to confirm the diagnosis. There are two types of imaging techniques for TMJ.

1. 2-D imaging involves a panograph, transcranial imaging, tomograms, and arthrograms
2. 3-D imaging involves CT scans, MRI (T-1, T-2, gradient), Flair or Fast T-2 (shows edema), and STIR (suppress fat content, which is good for an MS diagnosis)

Some inflammatory treatment options that the patient may want to explore include:

1. Vitamin D-3, 2000 to 10,000 IU per day
2. Doxycycline (50 mg, BID for 3 months)
3. Omega 3 – 2.6 mg/day
4. NSAIDS
5. Glucosamine (1500 mg/day)
6. TMJ splint
7. Muscle relaxants

Start with an in-dept evaluation and diagnosis. In EDSers, management is often preferable to surgical solutions. The best outcomes often involve a combination of treatment modalities. Work closely with a Craniofacial Pain/TMJ practitioner with EDS-specific experience, and you will find your answers!

The TMJ TREATMENT CENTER is located at 2141 N. Fairfield Road, Beavercreek, OH 45431. Phone: (937) 427-3131. Site: www.mitakides.com

Ehlers Danlos Syndrome and Disability Benefits from the Social Security Administration

Presented By: Shoshana R. Pehowic, Esq.

SSA defines "disability" for adults as, "The inability to engage in any substantial gainful activity (SGA) by reason of any medically determinable physical or mental impairment(s) which can be expected to result in death or which has lasted or can be expected to last for a continuous period of not less than 12 months." In other words, you are unable to perform any kind of competitive work on a sustained basis for at least 12 full months (or you are expected to die within 12 months).

SSA defines "disability" for children as, "A child under age 18 […] medically determinable physical or mental impairment(s) that causes marked and severe functional limitations, and that can be expected to cause death or that has lasted [or will last] for a continuous period of not less than 12 months." In other words, a child must have at least one condition which has been diagnosed and which is severe enough to interfere with the child's ability to function at home, in school, and/or in public settings to a "marked" or "extreme" degree, and that limitation must exist for at least 12 months.

There are 2 types of social security benefits for adults, SSD (Social Security Disability) and SSI (Social Security Income). SSD is a way to access your retirement benefits early. You must have earned enough "work credits" in the recent past to qualify. You must prove that you became completely unable to work before a certain date ("date last insured"). SSI is a "needs-based" or welfare program. You need not have worked in the recent past to qualify. Your household income and assets must be below a certain level to qualify.

Social security benefits for children include SSI benefits only. This is a "needs-based" program, so no matter how disabled to child is, the household income and assets must also be below a certain level. Documentation of disability may come not only from medical professionals, but also from schools or social service agencies (i.e. Easter Seals, Boards of MRDD, Head Start).

Eligibility for SSD benefits "expires" approximately 5 years after you last worked on a regular and sustained basis. SSD beneficiaries receive Medicare health insurance, whereas people with SSI benefits can get Medicaid by applying for it at their welfare office. SSI benefits are paid starting in the first full month after you apply or are found disabled (whichever comes firs), while SSD benefits do not begin until 5 months after your disability was found to have started.

With SSI, you are immediately eligible for Medicaid health insurance, which pays for nearly all medical and prescription costs, but which is not accepted by the majority of medical providers. With SSD, you will be eligible for Medicare health insurance 29 months after SSA says your disability started (2 years after you become eligible for SSD benefits). Medicare is accepted by nearly all medical providers, but you are responsible for a portion of the medical bills and you have to pay monthly premiums, much like with private health insurance.

Once you have been found disabled by the Social Security Administration, you will more easily qualify for other services, such as student loan forgiveness, housing resources, and Veterans' Administration disability programs.

On the downside, it can be hard for some people to accept disability benefits, which they consider "charity," and they may feel ashamed for having to accept such help. Certain people treat people with disabilities as "second class citizens" with fewer rights and less intelligence than "normal" people.

The vast majority of claimants do not win disability benefits until the "request for hearing" stage (after the initial and reconsideration stages). All across the country, there is a backlog to wait for a hearing, and while it is better than it used to be, the average wait is still about a year from the time you request a hearing until the time you see the judge. It generally takes at least another month or so to obtain a decision, and then 2-3 months after that for SSA to start paying money.

With SSD benefits, it is possible to work part-time even after you have been found disabled by SSA, but you must stay below a certain amount of gross monthly income. If you surpass that income level more than nine times, you will lose your monthly checks. With SSI benefits, you must report all income of any type to SSA every month, and each month's check can vary, depending on changes in your household income.

There are two main ways to prove medical disability:

1. By proving that your impairments meet or equal the criteria of a "Listing of Impairments,"
2. By proving that you are unable to sustain full-time competitive work at any level of exertion due to the effects of your impairments (your residual functional capacity, or RFC).

SSA considers certain specific impairments to be disabling after those impairments reach a certain level of severity. The Listings of Impairments describe those conditions, and sets forth specific criteria that should be met in order to be found disabled. The Listings are organized by body system (e.g., Section 1.00 relates to

orthopedic impairments, Section 3.00 relates to pulmonary impairments, and Section 11.00 relates to neurological impairments). THERE IS NO LISTING FOR EHLERS-DANLOS SYNDROME!

While Section 14.00 mentions "connective tissue" diseases, it is referring to auto-immune disorders (e.g., lupus or rheumatoid arthritis), rather than to congenital connective tissue disorders such as EDS. Instead of focusing solely on Section 14.00, you should focus on the Listings that correspond to whichever of your body systems are most limited. For some people, that may be the orthopedic Listings in Section 1.00. For others, it might be the cardiac Listings in Section 4.00, or the neurological Listings in Section 11.00. Even less obvious ones may apply, such as the digestive system (Section 5.00), as, for example, in a person who has gastroparesis.

When considering how each impairment limits a person, one looks at what the person can still do, despite those impairments. This is referred to as the persons Residual Functional Capacity (RFC). If a person can do any type of "substantial gainful activity" (SGA), i.e., earn more than $1,010 per month in 2012 before any taxes are taken out, then the person is not "disabled." It does not matter whether the job is something that the person has never done before, or that it does not pay enough; as long as the person is medically capable of performing the job, the person is not disabled. Perhaps the most difficult part of proving disability through RFC is being able to rule out even a sit-down job. SSA says that there are a number of entry-level sit-down jobs, which can be performed even while alternating sitting and standing (such as for people who cannot sit long without pain and must stand briefly to stretch). One way to do this is to show why you cannot sustain an 8-hour day or a 5-day work week. An opinion from your doctor can be strong evidence, but only when that opinion is well-supported by objective medical findings and test results. The SSA is not bound by the opinion of a treating doctor, and it can disregard that opinion when it believes that the opinion is not well-supported by evidence or if it is inconsistent with the treatment notes of that doctor or of others. The SSA can also disregard the doctor's statements if it believes there are inconsistencies between the doctor's opinion and the person's statements of activities of daily living or other functional abilities.

Orthopedic Impairments include generalized arthritis, recurrent dislocations and subluxations leading to "inability to ambulate effectively" and/or "inability to sustain effective fine and gross manipulation" with the upper extremities, and chronic pain.

Pulmonary Impairments include asthma or reactive-airway disease, and collapse of the small airways (even only intermittent).

Digestive System Impairments include GERD (gastro-esophageal reflux disease), Constipation and/or Diarrhea, often lumped under the title of "Irritable Bowel Syndrome," but often the result of too much stretching of the bowel or stomach due to EDS, Gastroparesis and other motility disorders, and TMJ and other jaw-related problems, which can affect not only eating, but also speech.

Cardiac & Neurologic Impairments include palpitations and other arrhythmias, POTS, Orthostatic Hypotension, Neurocardiogenic Syncope, etc., generalized fatigue, Intolerance to Temperature Changes or Increased Activity Levels, and Prolapse of the heart valves.

Blood Vessel Impairments & Tissue Fragility include recurrent abdominal hernias, often requiring multiple, or failed, surgical procedures, and Pelvic Floor Prolapse, causing chronic pelvic pain, incontinence, and other issues which make it difficult to sit, stand, or walk for extended periods of time

Neurological & Psychological Impairments include Depression and/or Anxiety resulting from chronic illness, ADD/ADHD and other impairments of executive function (which may result from functional Chiari malformation or other hidden disorders), "Clumsiness" and Tremors, Neuropathies from Pinched Nerves, and Cranio-Cervical Instability and/or Atlanto-Axial Instability, leading to varying types of vague neurological symptoms which may come and go.

Some practical tips for proving disability:

- Make a list of <u>each</u> of the problems you are currently dealing with and have the doctor's staff copy that to your file for each office visit. The doctor need not address each of the problems at every visit (and most likely will refuse to do so), but it is important to document when a condition first shows up and that it continues to persist over time.
- Ask your doctor to do a brief physical examination at each doctor's appointment and document all positive findings, even if they are findings that you have had for many years. Also be sure to ask your doctor to avoid using default language in the medical records regarding physical examinations: it is common these days for electronic medical records to include default "normal" physical examinations, and those are included in a medical record unless the doctor takes action to remove them. These "false negative" examination results have been the downfall of numerous disability claims. Explain the importance of ensuring that your records are accurate every time.
- Thoroughly describe how your impairments interfere with your activities of daily living (ADLs):

  - For each impairment, consider how you are functionally limited. For example, shoulder pain obviously limits your ability to raise your arm in certain directions, but it may also limit your ability to brush and wash your hair, put on certain types of clothing, cook, or carry groceries.
  - SSA's Function Report and other questionnaires are worded very simply, and do not encourage you to provide details. However, answering these questionnaires in a detailed but focused manner is extremely important in explaining how EDS affects you and limits your ability to function.
  - Don't forget to describe those activities you are able to do, but only with difficulty. Most people simply answer "yes" or "no" without thinking about how they have to change the ways in which they do things, or how their energy level limits them.
- Explain to your doctor any problems you are having affording your medications, and ask the doctor to document it in your medical chart.
- If you need help affording your medications, apply for all available social services, such as Medicaid, and look for outpatient clinics associated with hospitals, which may be able to treat you without insurance for a minimal (or no) cost. Try www.NeedyMeds.org to obtain brand name medications directly from the manufacturers. Search for free clinics in your area.
- Check <u>each</u> of the "$4 Lists" at the chain pharmacies (e.g., WalMart, Target, Kroger) for the generic medications you take: although there is some overlap, each store has different medications on their lists. Also consider obtaining a discount card from a pharmacy (e.g., CVS or Walgreens).

Objective test results include reports of x-rays, CT scans, MRIs, and ultrasounds, along with laboratory results and reports of procedural tests such as EGDs, colonoscopies, EEGs, sleep studies, pulmonary function tests, etc. Some of these results are more "objective" than others. While laboratory results are usually read consistently, radiology studies are very subjective. For example, a damaged spinal disc can be described as "bulging," "protruding," or "herniated," among other words. The radiologist will often use words such as "mild," "moderate," and "severe" when describing how much damage the disc is causing, and will rarely give precise measurements. Unfortunately, the ALJs and other adjudicatory personnel at the SSA are not trained in reading medical imaging, and are forced to rely on the radiologist's report. The way the report is worded can definitely affect the outcome of a case. In a similar fashion, the way that examination findings are described can affect your disability claim. For example, an ideal description of problems with range of motion of a joint would include actual measurements of the joint motion, such as those done by most physical therapists during initial appointments (e.g., "left knee flexion was reduced to 150 degrees out of a normal 180 degrees"). Most

often, though, the clinician estimates the range of motion, or merely refers to it as "reduced" without specifying the severity. Other objective findings, such as spasm, muscle weakness, or tenderness to palpation, may also be described too vaguely. The more specific the report is, the more useful it will be for proving your disability claim.

According to SSA's Regulations, a specialist's opinion, if supported by the evidence and if consistent with the record, is supposed to outweigh the opinion of a general medical doctor, or of a specialist whose specialty is not relevant to the issue. Therefore, if you are able to obtain a favorable opinion about disability from a specialist who is treating you for the impairment in question, that opinion will have even more clout before the SSA. Specialists are also better able to explain the rare conditions and complications, which can occur in a patient with EDS, such as unexpected outcomes of surgery or a surprisingly poor response to a treatment. A well-worded explanation of these issues by a treating specialist can make a big difference.

Do your best to obtain medical treatment for all of your disabling impairments. The SSA assumes that if you have a truly disabling impairment, you will try all possible treatments in order to get relief from that impairment. Conversely, SSA assumes that if you are only seeing a family doctor for a particular condition, the condition is likely not severe enough to be disabling. Therefore, if you are unable to obtain more intensive treatment, ask your doctor to document the reason (e.g., side effects to medication, cannot afford to see a specialist, or surgery would be too risky). Keep a diary or log of any conditions, which cause episodic problems (e.g., seizures, severe headaches, or gout attacks). Bring the log to each appointment with your physician, so that the log can be incorporated into your treatment plan. When submitting medical records to SSA, submit a copy of the log, and then periodically submit updates of the log. That way, there will be a "contemporaneous record" of the frequency of the attacks. Know when to ask for help in presenting your claim to SSA.

Focus on which of your body systems are affected by EDS, and consider how your ability to perform activities of daily living and/or work duties is impaired. You will want to clearly explain these changes: to your doctors, in the questionnaires that are sent to you by SSA, and in your testimony at the hearing. It is also important to obtain opinions from your treating physicians, and to ask that your doctors support their opinions with citations to clinical findings and objective test results. Do your best to be compliant with medication orders and doctor appointments, and avoid "bad habits" such as smoking, drinking alcohol, or using illicit drugs. You will have a much greater chance of winning your disability claim if you seek the assistance of a local attorney or non-attorney representative who is experienced in representing claimants before the Social Security Administration.

How to Communicate With Your Doctor & Build Your Medical Team

Presented By: Linda Neumann-Potash, RN, MN, CBN

Communication is the first step in managing your health situation. The very first important point is to be organized. You want to go to your appointment prepared. Unless it is an emergency, we usually have a couple weeks to prepare for the visit. Use that time to accumulate an accurate list of all your medications and to develop a list of questions you want to ask your health care provider. In most cases, you can simply hand over your list to your doctor so that they can address them one by one. It is highly recommended that you bring someone with you to the visit to take notes. It is very difficult being the patient and having to write down the notes of the visit yourself. It is also important that you make sure you understand the answers to your list of questions. It is the job of the doctor to make sure you understand the answers to your questions before you leave the visit. You should also explore options together with your doctor (i.e. what has worked in the past and what hasn't worked). When a treatment plan is established, it is very important that you, as a patient, follow through with the treatment plan. Before the end of the visit, make sure you find out the best way to communicate with your health care provider whether it be via email, phone, etc.

BEFORE YOUR APPOINTMENT: Find out how long your appoint is when you book your appointment so that you know which version of your story is appropriate for the visit (the 2 minute version vs. the 20 minute one). When preparing your "story," write it down first. In addition to your story, you should also prepare an organized health history, and medication & treatment list. Make copies of your health history, and medications & treatment list, in order to give to your health care provider for their records. When building your list of questions, make sure you include the purpose of the visit. Your questions should relate to the purpose and should be prioritized from most to least important. Lastly, make a list of your expectations for the appointment.

DURING YOUR VISIT: Stay actively involved in the discussion. This is why it is suggested that you bring someone with you to your visit to take notes because it is very hard to stay actively involved in a discussion when you are trying to write everything down. You want the physician to know that you have taken the time out to establish goals for this visit. Make sure you ask for clarification if you don't fully understand something. Make sure you listen to the questions that you are being asked as well by your health care provider as well. A lot of patients don't listen to the entire question being asked to them and they leave out key details. It is important to explain all the details of your symptoms as clearly as possible. Before you leave your visit, make sure you know what the plan of care is. Get the name and

phone number of who to call if you have any questions and the best method of contacting them. If something isn't clear, it's up to you to ask your health care provider to explain. Don't be afraid to keep asking until it is clear in your mind.

AFTER THE VISIT: Follow through with the agreed upon plan of care. Even if you don't agree with the plan try and follow through with it, that way if it doesn't work, you can say I've already tried that, what other options do I have. And of course if you are absolutely against trying out a plan of care, then let your physician know that. But make sure to keep an open mind. Sometimes you can couple something that didn't work with something that might work and ultimately develop an effective plan of care that really works. Keep a detailed list of what is working, what is not working based on the treatment plan. As always, it is important to keep yourself informed about your condition and do further research on your own. Lastly, keep your contact information up to date with your physician. So if you move or your number changes make sure your physician amongst the first to know in order to keep that line of communication open.

When it comes to building you healthcare team, identify experts in their field of practice. You want a health care provider who listens and respects you. You want your health care provider to explain things in a way that you understand. The best health care providers out there are the ones who will tell you honestly when they "Don't know" the answer but are willing to kelp you look for the answer. You want a health care provider who can think outside of the box when needed and step back in the box when it's not. You want a health care provider willing to work with other health care providers and most importantly is a part of your insurance plan.

Your support team includes your healthcare team, your friends and family, the EDS community, and those we meet via social media (i.e. twitter, facebook, etc.).

Organizing your health information is a huge process that a lot of EDSers fall short of. Once you have your medical information organized, obtaining treatment becomes easier and more readily available because the physicians see that you are dedicated to going the extra mile. Physicians respect patients that make their lives easier. You want to create a storage system for your records. Some organize binders, some scan their papers into the computer and carries medical USB's around, others dedicate an entire box to their records. However you decide to organize your health information, make sure you include the following:

- Basic info-allergies, contact info for you, for your emergency contact, etc.
- Personal medical history
    - Medical history must include dates
    - List all surgeries including dates
    - Allergies – what they are and the reaction

  - Blood type/transfusion history
  - Medications – what works…what doesn't
  - Current treatments
  - Tests and procedures
  - Mental health treatment
  - Alternative therapies
- Family medical history
  - Can be just as important as your personal history
  - Ask questions of family members
  - Serious illnesses
  - Chronic health conditions
  - Unusual reactions to medications/anesthesia/surgeries
  - Mental health disorder
  - Cause of death if deceased and age
- Your entire health care professional list – include name/address/phone/ and any records from visits
- Immunizations records
- Medication list – complete list of over-the-counter medications, prescription medications, and supplements.
- Insurance information
- Test results
- Legal documents – power of attorney, living will, etc.

You may also want to create your very own timeline. This will give your health care providers a quick look at your past history. Timelines are great because they are visuals, quick and easy to read. Make sure you update your timeline and keep it current! Update changes as they occur to maintain accurate records. Note: your memory does NOT improve with age!

Pain Management: More than Just a Pill

Presented By: Anne Lynch-Jordan, PhD.

The Nature of Pain: The Gate Control Theory and Beyond

Pain is "an unpleasant sensory and emotional experience associated with actual or potential tissue damage, or described in terms of such damage." Pain is emotional and does not have to be due to an actual injury. The experience of pain focuses on not only the sensation but also the impact of pain. Pain perception is protective. Multiple systems are involved in how we perceive pain (i.e. the peripheral nervous system – which is comprised of sensory nerves & receptors; and the central nervous system – which is comprised of the spinal cord & the brain). Sensory nerves receive input from physical stimuli, while receptor input is transmitted to the spinal cord. Further modifications to the input due occur, finally resulting in signals being relayed to the brain structures for encoding. The Gate Control Theory of Pain explains that pain is reduced or amplified based on descending pathways from the brain due to characteristics like pain history, attention to symptoms, and emotional state. Contributions from genetics, neuroscience, & imaging have refined this theory.

Chronic Pain is the result of central sensitization, the abnormal state of responsiveness to an increased gain of the nociceptive (pain) system." Some causes of central sensitization leading to chronic pain include hyperarousal of the nervous system, spontaneous occurrence of pain signals, and low levels of stimulation causing high levels of pain.

Stress (both physical and/or emotional) disturbs the body's homeostasis. Disruption of the body's homeostasis causes internal immune and hormonal reactions in an attempt to restore balance. When the body's homeostasis is thrown off, the body releases substance to fight infection and repair tissue damage. The hypothalamic-pituitary-adrenal (HPA) system is activated; and cortisol is released. Prolonged cortisol release may not trigger chronic pain, but it may create an internal environment that promotes the trigger of chronic pain.

A multi-modal approach to addressing chronic pain includes medical, psychological, and physical interventions, which has been shown to be most effective. Chronic/recurrent pain affects the patient on a physical, emotional, and social level. Proper treatment addresses pain and functioning simultaneously. The primary goal of Cognitive Behavioral Therapy (CBT) is to improve coping skills. Psychologists have expertise in changing maladaptive behavior and thoughts (cognitions). With enhanced coping skills, functioning should improve, mood should stabilize/improve, and pain & suffering should ease.

Behavior Therapy is based upon the principles of operant conditioning and social learning theory. The goal of Behavior Therapy is to alter behavior. Operant conditioning is based on rewards and punishment; if you are shocked every time you press a lever, you stop pressing the lever; if you are rewarded with candy every time that you pee in the potty, you pee in the potty. Social learning theory suggests that learning occurs in a social context, so as a child, if you live with a parent who has diabetes who exercises, checks blood sugar, and eats healthy, you are more likely to do so based on what you learn from those around you.

Cognitive Therapy is based upon principles of information processing and cognitive processes. The Goal of therapy is to alter thoughts and beliefs. Depression stems from negative beliefs about your self (I am worthless), the world (the world is unfair), the future (the future is hopeless).

Cognitive-behavioral therapy (CBT) was initially developed for the treatment of depression and anxiety disorders. The potential for CBT was quickly recognized for application in pain management by Dennis Turk and Frank Keefe.

Numerous intervention protocols have been developed for adults with low back pain, fibromyalgia, osteoarthritis, rheumatoid arthritis utilizing CBT for pain management. Additionally, increased research attention has been devoted to psychological treatment for youth with chronic pain. Note that most components of CBT are not studied head-to-head. Instead, the components of CBT are studied in a package, making it unclear that CBT is the best package or if there is a more powerful technique out there.

Dr. Lynch-Jordan introduced 5 different relaxation techniques for EDSers to look into and hopefully try.

1. **Diaphragmatic breathing**: promotes a parasympathetic response (reduced blood pressure, muscle tension, heart rate, etc.)
2. **Progressive muscle relaxation**: reduces muscle tension and promotes body awareness
3. **Autogenic relaxation**: parallels meditation techniques and focuses on desired autogenic responses. “My arms are warm and heavy,” said repeatedly.
4. **Imagery/Visualization**: pleasant mental images aimed to distract away from pain or distress
5. **Mindfulness meditation**: meditation with a focus on a calm awareness of the present moment and acceptance without judgment of bodily sensations and emotions

Two types of activity patterns for behavioral activation and regulation are common & equally problematic:

- Cycle 1: Under-exertion
    - Fear of pain, avoidance, disuse & deconditioning, and the presence of disability.
- Cycle 2: Over-exertion
    - Unhealthily high levels of activity, task persistence, and the presence of disability.

According to McCracken et al., 2007, there are 4 types of activity patterns:

- **Avoiders**: less activity, greater functional disability, greater anxiety
- **Doers**: more up time, high activity no avoidance, most functional (but at what cost?); less anxious, more acceptance
- **Medium Cyclers**: moderate levels of activity & avoidance, between the Doers and the Extreme Cyclers.
- **Extreme Cyclers**: high levels of activity & avoidance, greatest level of disability & anxiety

The goal of cognitive modification is to reduce catastrophic thinking about pain. For example, when one thinks, "Physical therapy will hurt me," emotions of "fear & apprehension," develop leading to physical symptoms of "muscle tension," & an increased physiological arousal (jangled nerves affecting signals). All of this will then result in the individual taking action to "guard, brace, & avoid" which may in turn cause the flare up that individual is so fearful of.

Methods of Cognitive Modification include:

- Identifying negative beliefs & attitudes: Black & white thinking; fortune telling. For example, "I cannot function when I'm in pain" … "My health is hopeless" … "I'm never going to be able to cope with pain."
- Creating calm, supportive self-statements: For example, "My flare up won't last forever" … "I can get through this" … "There are still good things in life."
- Examining worries: For example, "In 5 years, will I remember (or care) about this worry?" … "Do I know for sure it will be as bad as I anticipate?" … "What is the worst that can happen?"

Regular practice of these techniques promotes continued re-training of physiology, serves as a preventive mechanism, and prepares for effective use during flare-ups. It is important to prepare for potential flare-ups. It is recommended to engage in problem solving skills in anticipation so that an individual's disability doesn't become extreme.

Some additional techniques include biofeedback, hypnosis, and yoga. Biofeedback was developed in the 1960s. It was previously believed that people were unable to

gain voluntary control of certain body processes. So they began investigating the "average" person's ability to control autonomic responses (i.e. heart rate, respiration, blood pressure, muscle tension, & peripheral blood flow). They found that most people do not have interoceptive awareness. Biofeedback is most commonly used for migraine or tension-type headaches. The best effects are found as part of combination therapy (either with relaxation training or CBT packages). Unfortunately, there is no evidence for the purely physiological model of biofeedback success. It has been difficult to clearly establish criteria for "acquired physiological control." According to Dr. Lynch-Jordan, biofeedback is helpful for many pain problems, even those without direct contribution of muscle tension (i.e. recurrent abdominal pain). The hypnotic process involves induction, which is the initial suggestion for changes in behavior or perception (i.e. for focused attention and/or relaxation). Specific suggestions are made for alterations in how pain is viewed or experienced. Suggestion that the benefits experienced during the session will last beyond the session, for minutes, hours, or even for days, months and years. Certain individuals high on hypnotizability and imagery vividness tend to benefit most from all relaxation based techniques; techniques are more similar than different. There have been several randomized control trials for yoga. A limitation to yoga is a poor ability to construct a placebo yoga group that takes into account interpersonal attention and exercise. Yoga for children has been shown to be effective for reducing disability, and mood problems for kids with IBS.

Sleep Disorders in Ehlers-Danlos Syndrome

Presented By: Alan G. Pocinki, M.D.

The autonomic nervous system (ANS) regulates all body processes, including sleep. ANS dysfunction is very common in EDS and other hypermobility syndromes, and underlies many of their symptoms. The most common type of sleep disorder seen in the hypermobility syndromes appears to have an autonomic basis.

The sympathetic nervous system is responsible for our "fight or flight" response. It is our accelerator. The parasympathetic nervous system is responsible for our "rest and digest" response. It is our brakes.

Autonomic instability rides on the concept of an adrenaline reserve. The central paradox explains as the lower the adrenaline reserves are, the more exaggerated your stress response will be, which causes your body to overrespond to minor stresses. This overresponse often triggers an overcorrection, then an overresponse, and a cycle forms.

Non-restorative sleep in EDS results in frequent arousals and awakenings resulting in little to no deep sleep for the EDSer.

Another paradox exists in heart rate variability. The lower sympathetic activity is, the greater the heart variability…OR…the more exhausted you get, the more "depleted" your energy reserves, the more exaggerated the heart rate fluctuations will be. The more your heart rate fluctuates, the more disrupted your sleep (not to mention daytime activities). The more disrupted your sleep, the more exhausted you get – a nasty vicious cycle.

Sleep "Misperception" is another paradox that resides amongst many EDSers. Many EDSers report that they "sleep fine." "I'm a great sleeper. I can fall asleep any time, anywhere." But when they are asked if they feel rested when they get up, they respond with, "No, I never feel rested" … "I wake up feeling like I haven't slept" … "I don't think I know what feeling rested would feel like." Sleep "Misperception is not just a problem amongst EDSers, about 90% of people with sleep apnea are not aware of it.

So in order to treat autonomic dysfunction, you need to get better sleep by addressing underlying problems. These problems may include: pain, fatigue, dehydration, low blood sugar, and emotional stresses. Better sleep will help replenish reserves, but doing all these other things will also help you sleep better: adequate pain control, not pushing through fatigue (taking breaks), adequate salt and fluid intake, avoiding hypoglycemia, and minimizing emotional stresses.

When treating sleep disorders you don't want to overlook the basics like practicing good sleep hygiene, having a comfortable mattress to sleep on, sleeping in a dark & quiet room, elevating the head of the bed (if lightheaded during the day), and treating sleep apnea, limb movement only if significant. After addressing the basics, if you are still battling a sleep disorder, a complex medication "regimen" is often required. Unfortunately, finding the right combination can be a frustrating trial and error process. A home sleep monitor may actually be helpful (www.myzeo.com). Beta and alpha blockers, Clonidine and Guanfacine, block extra adrenaline. Benzodiazepines and SSRI's offset extra adrenaline. Analgesics, muscle relaxants, Neurontin, and Lyrica have been shown to effectively reduce pain. And Trazodone, Amytryptiline, and Doxepin help to increase deep sleep. Antidepressants are not usually used to treat sleep disorders but can affect sleep. Note to use "sleeping pills" sparingly.

Beta Blockers

Start Propranolol with 10 mg at bedtime. Increase by 10 mg every 4-5 days until fewer awakenings, side effects, or no further benefit. Switch to long-acting if needed. Take some earlier to offset "second wind." Often need smaller daytime dose as well.

Start Metoprolol with half a 25 mg tablet (metoprolol tartrate). Increase by half a tablet every 4-5 days. Add long-acting (metoprolol succinate) if needed.

Start Nadolol with 20 mg. Increase by 20 mg every 4-5 days. Add smaller AM dose if needed for daytime symptoms. Nadolol is safe in asthma (Bystolic is also safe in asthma, but must take once daily).

Start Carvedilol with 3.125 mg. Increase by one tablet every 4-5 days. Add smaller AM dose if needed for daytime symptoms.

Start Clonidine with 0.1 mg at bedtime. Increase by 0.1 mg no sooner than one week. No more than 0.3 mg. Usually lasts about 6 hours.

Guanfacine is very similar to clonidine but lasts longer. Recently remarketed as Intuniv for ADD.

Alpha Blockers

Prazosin is the best studied and shown to reduce nightmares in PTSD, where "a hypersensitivity to adrenaline triggered many of their nightmares." In a VA study, 75-80% of PTSD patients stopped having nightmares. The usual dose is 5 mg. Prazosin can worsen orthostatic intolerance. Not clear if a combination of alpha and beta blockers (e.g. carvedilol) are as effective, but probably not.

Benzodiazepines

Benzodiazepines all have beneficial properties: sedative; anti-anxiety; muscle relaxant; anti-movement, anticonvulsant; and anti-adrenaline. Some potential problems of benzodiazepines include: impair cognition, motor performance; depress mood, respiration; cause or worsen fatigue; tolerance; dependence; withdrawal. Some common benzodiazepines include:

- Clonazepam (Klonopin): longest-lasting, most likely to have residual effects; also effective for restless leg, PLMS
- Diazepam (Valium): Typically lasts about 8 hours; probably best muscle relaxant
- Temazepam (Restoril): Typically lasts about 7 hours; Capsule limits dosage adjustment

- Lorazepam (Ativan): Typically lasts about 6 hours; metabolized differently (less variability, interactions)

Analgesics

Anti-inflammatories include NSAID's (Naproxen, Meloxicam, Celebrex) and Prednisone.

Tramadol is a short and long acting analgesic

Narcotics are available as short and long acting and comes in patches as well like Fentanyl and Butrans

Cymbalta and Savella are analgesics

Gabapentin (Neurontin) and Lyrica are also used as analgesics

Lidoderm, Flector, Voltaren Gel, and Pennsaid are also very commonly used analgesics.

Muscle Relaxants

Cyclobenzaprine is shown to improve sleep quality in fibromyalgia. Cyclobenzaprine has analgesic, sedative, and muscle relaxant properties.

Soma is less sedating, and has been reported to have more of an analgesic effect, especially with narcotics.

Skelaxin is less sedating, some can tolerate daytime doses.

Tizanidine is more sedating, and has a high margin of safety.

Baclofen is potent, and is used for severe painful spasms only.

Other Agents

Trazodone is probably most effective at increasing deep sleep. Low dose ranges fro 40-150 mg. Most people take 50 mg.

Amitryptiline also increases deep sleep, especially with pain. Start amitryptiline at 10 mg. Most people take 20-40 mg.

Doxepin enhances sleep more at lower doses. Doxepin is available in a 10 mg tablet, liquid or as Silenor 3mg, 6mg.

Sleeping Pills

Zolpidem is available as short and long acting. It doesn't reduce arousals or improve sleep architecture. Zolpidem usually lasts about 5 hours; ER (extended-release) lasts about 7 hours.

Lunesta doesn't reduce arousals or improve sleep architecture either. Occasionally it helps with sleep onset and maintenance (until other medications become effective). Lunesta usually lasts about 7 hours.

Zaleplon is good for sleep onset, especially for getting back to sleep. It lasts 2-3 hours, with no cognitive impairment.

Melatonin/Rozerem is most helpful for Circadian problems (the evening "second wind").

Antidepressants

SSRI's often cause shallower sleep and more dreams. Prozac is seen as being the worst of the SSRI's and Lexapro the best. It is recommended to use the lowest effective dose. You may want to consider liquid formulations.

Cymbalta causes the patient to be sleep neutral if taken in the morning.

Tricyclics generally improve sleep, but often cause daytime sedation.

Wellbutrin impairs sleep if taken late in the day. So it is recommended that you either take Wellbutrin XL once daily early in the day or consider taking Wellbutrin SR twice a day in the morning.

Remeron generally improves sleep, and can cause weight gain.

The most common type of sleep disorder seen in the hypermobility syndromes appears to be characterized by excessive heart rate variability at night. Medications to suppress, offset, or block this excess activity are effective in improving sleep, measured both by polysomnography and symptoms. Improving sleep and minimizing daytime stresses helps to replenish autonomic balance and also helps improve sleep, which in turn improves daytime function, which in turn improves circadian rhythms and sleep, which is how you get better!

EDSers United
Stay Strong
Stay Connected

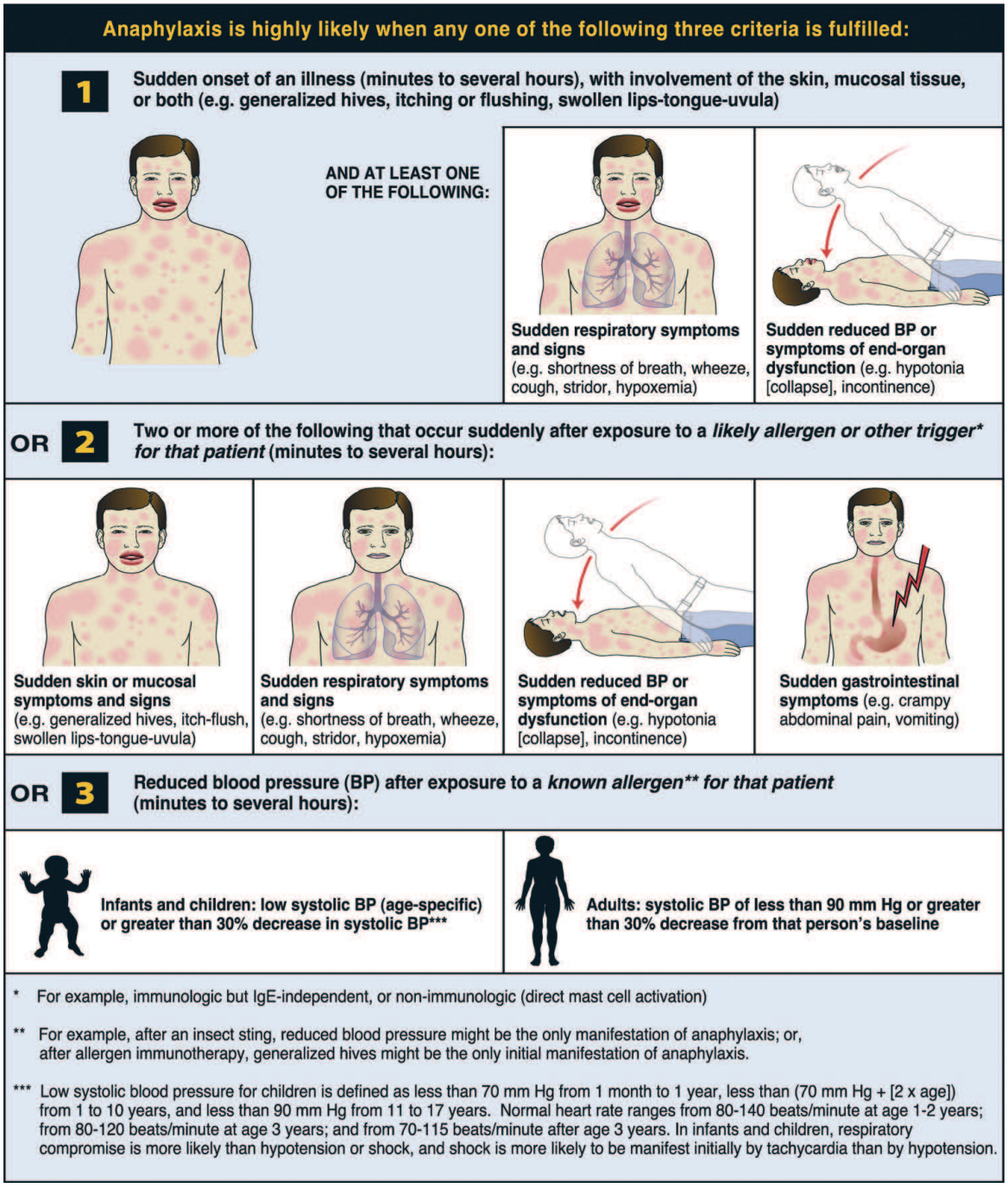
Mast Cell Disorders

Presented By: Andrew M. Smith, MD, MS

Anaphylaxis is defined traditionally as a systemic, immediate hypersensitivity reaction caused by immunoglobulin E (IgE) – mediated immunologic release of mediators from mast cells and basophils. The World Allergy Organization defines anaphylaxis as a severe, life threatening, generalized or systemic hypersensitivity reaction involving two or more organ systems.

Anaphylaxis Symptoms

Signs & Symptoms	Percentage
Cutaneous	**>90%**
• Urticaria/Angioedema	85-90%
• Flushing	45-55%
Respiratory	**40-60%**
• Dyspnea/Wheezing	45-50%
• Rhinitis	15-20%
Dizziness/Syncope	**30-35%**
Hypotension	**30-35%**
Abdominal	**25-30%**
• Nausea/Vomiting	
• Diarrhea	
Miscellaneous	
• Headache	5-8%
• Substernal pain	4-6%
• Seizure	1-2%



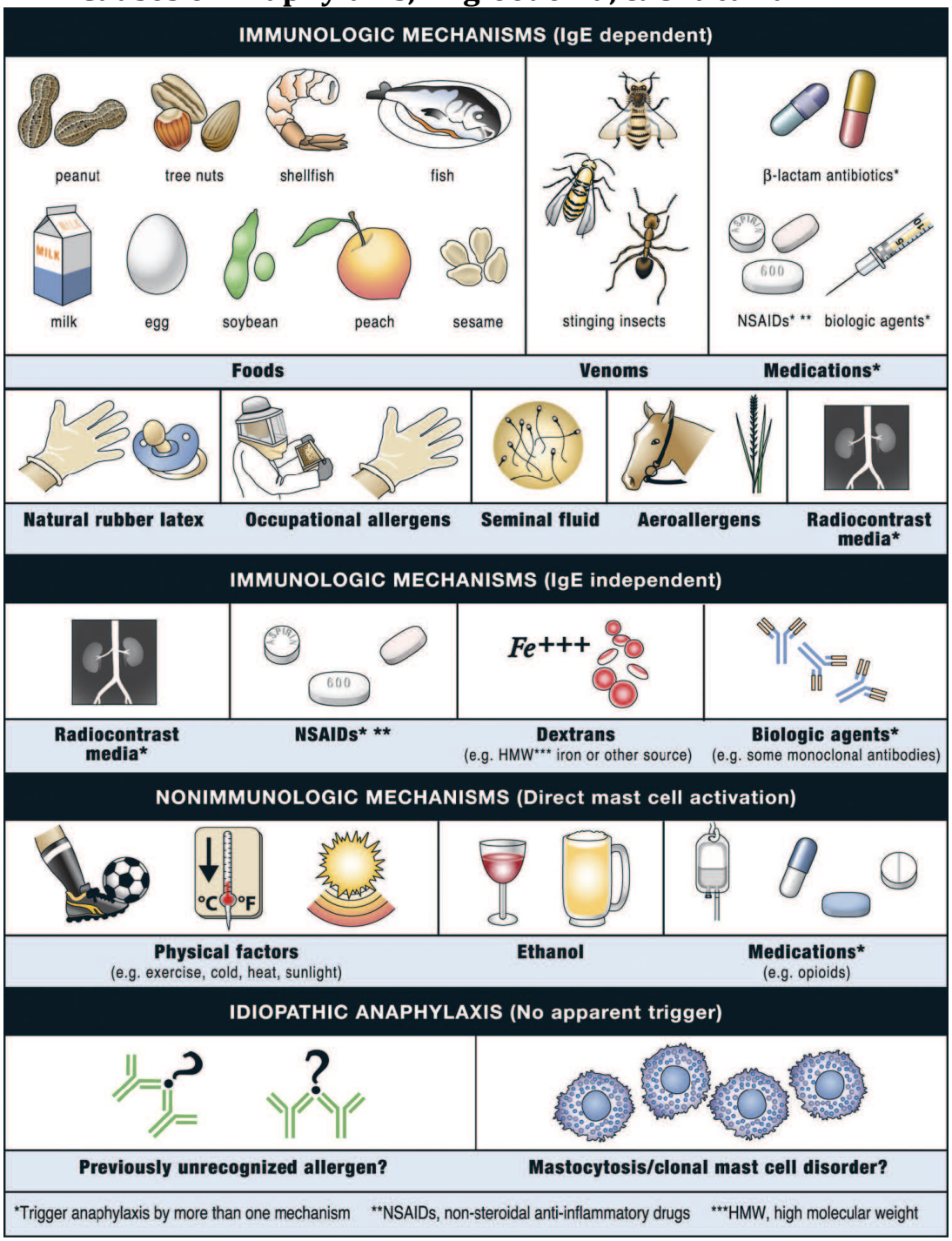
Angioedema is defined as swelling in the deep skin tissue. Fewer sensory nerves are involved in this reaction so there is little to no itch involved. It may be described as painful or burning. Generally asymmetric and short-lived swelling occurs. Angioedema may occur in isolation, and accompanied by urticaria, or as a component of anaphylaxis; and can be life-threatening. Angioedema can in the

larynx, on skin and mucous membranes and in the bowel wall. Angioedema on the larynx can develop rapidly. Early symptoms include hoarse voice, throat tightness, and difficulty swallowing. Angioedema on skin and mucous membranes can cause mild pain and warmth, as well as a burning sensation; and resolves without leaving residual markings on the skin. Angioedema throughout the bowel wall can result in colicky abdominal pain, sometimes nausea, vomiting, and/or diarrhea. Bowel wall edema is often visualized by abdominal CT or ultrasound.

Urticaria (hives) are itchy, red raised lesions that turn pale when pressed, indicating the presence of enlarged blood vessels and fluid. Hives can last from minutes to hours and resolve without discoloration/scarring.

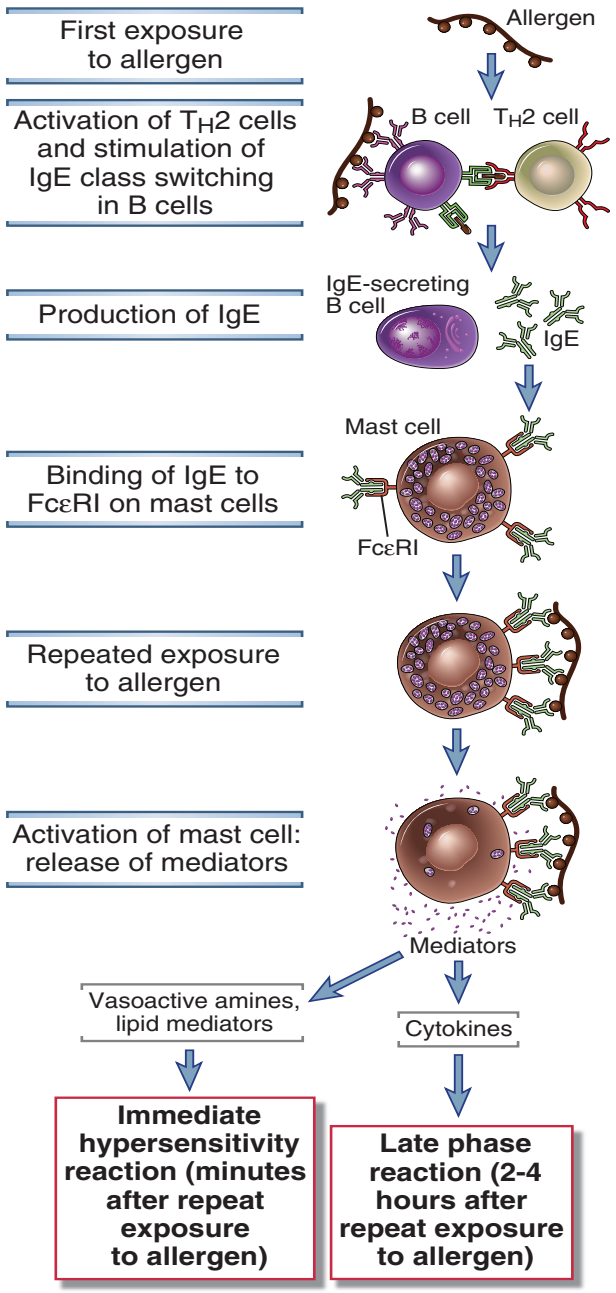
Approximately 1% of the population is dispensed outpatient epinephrine.

Causes of Anaphylaxis, Angioedema, & Urticaria



Mast cells fight off worm and bacterial infections. Mast cell dysfunction causes anaphylaxis, systemic mastocytosis, mast cell activation syndrome (MCAS), and MCAS and postural tachycardia syndrome (POTS).

Mechanisms of Anaphylaxis



Systemic mastocytosis includes excess growth of mast cells and accumulation in 1 or more organs. 93% of cases are due to a mutation of the c-kit gene (KIT D816V). Symptoms of systemic mastocytosis are due to mast cell mediator release. Systemic mastocytosis symptoms include abdominal pain, diarrhea, nausea/vomiting, itching/flushing, fixed skin lesions, fast heart rate, loss of consciousness, anaphylaxis, headache, confusion, bone pain, weakness, fatigue malaise, shortness of breath, and nasal symptoms. In order to diagnose systemic mastocytosis, the presence of at least 1 major and 1 minor or 3 minor criteria in bone marrow or other organ must be found.

- Major
  - Excessive mast cells on bone marrow biopsy
- Minor
  - Excessive atypical mast cells
  - C-KIT D816V mutation
  - Mast cells with surface markers (CD2, CD25, CD117)
  - Serum tryptase > 20ng/mL

A neuroendocrine evaluation should be conducted in order to rule out pheochomocytoma, CIPoma, and carcinoid syndrome. A multispecialty approach is likely necessary. Some physicians may prescribe H1 Antihistamines, H2 Antihistamines, Epinephrine, Cromolyn, LTRA, or Steriods in order to block mast cell related symptoms. Chemotherapy is an option only in severe systemic mastocytosis cases. It is important to avoid mastocytosis triggers like: alcohol, spicy foods, NSAIDs, Narcotics, intense exercise, stinging insects, stress.

Mast Cell Activation Syndrome (MCAS) is a newly recognized disorder with no excessive growth of mast cells. Symptoms are due to mast cell mediator release. MCAS can happen in any age group. Symptoms of MCAS include: abdominal pain, dermatographism, flushing, headache, poor concentration and memory, diarrhea, nose and eye symptoms, asthma, and anaphylaxis. In order to diagnose MCAS, a serum total tryptase, a 24-hour urine for histamine, N-methylhistamine, or prostaglandin D2 laboratory evaluation can be conducted. It is important to rule out other causes, including systemic mastocytosis (via bone marrow biopsy).

Postural Tachycardia Syndrome (POTS) is a rare, disabling condition where the patient experiences high heart rates with standing. POTS is more common in young females. POTS has an unclear cause. It may be a result of poor nerve signaling (neuropathic), and/or excessive adrenal activation (hyperadrenergic). For patients with MCAS and POTS, it is important to avoid the following triggers: exercise, prolonged standing, heat intolerances, and emotional stress.

Treatment options:

- **H1-Antihistamines** (Cetirizine, Doxepin, Cyproheptadine): decreases itch, flush, urticaria, sneezing, and rhinorrhea; doesn't prevent or relieve obstruction to airflow or hypotension/shock
- **H2-Antihistamines** (Ranitidine)
- **Glucocorticoids:** switch off transcription of activated genes that encode pro-inflammatory proteins; onset of action takes several hours; used to prevent and relieve mast cell related symptoms
- **Leukotriene Modifying Agents** (LTMA): Montelukast, Zafirlukast, Zileuton
- **Cromones** (Cromolyn): mast cell stabilizer for GI symptoms only

Mast cell disorders are relatively rare but severe. Patient history is extremely important. Extensive evaluation and a multispecialty approach may be necessary to rule out all probable causes.