Hypermobility Spectrum Disorder (HSD) & Hypermobile Ehlers Danlos Syndrome (hEDS)

Good Health Physical Therapy and Wellness
Goodhealthphysicaltherapy.com

You are Strong and Courageous

You are strong and courageous. How do we know? If you are reading this you have likely been facing one or many health challenges for some time. And you have been doing your best to live and function each day. That takes strength and courage. We are here to listen to you and to support you in your healing journey. In particular, we hope to support you being even more empowered to get your symptoms under control, to know the best ways to manage the health challenges and to get back to doing what you want to and need to be able to do for all the life you deserve. This is one of a series of patient education handouts – the Strong and Courageous Series – aimed at helping you to learn more about your condition, learn the controllable triggers of flare-ups, and learn the steps you can take to claim back your power and your ability to function.

The Spectrum

In our clinic, we see many patients with hypermobility spectrum disorders (HSD). Healthy joints exist in a balance between being stiff enough to be stable yet mobile enough to allow activity. Hypermobility spectrum disorders are all characterized by too much motion at the joints of the body often leading to instability and pain. The disorder can be with one to four joints, called a localized hypermobility, or in five or more joints which is referred to as generalized hypermobility. Generalized hypermobility is usually something a person is born with and often is inherited. Saying that hypermobility exists on a spectrum means that some cases are relatively mild with little pain and few complications and some cases more symptomatic. The more symptomatic forms of hypermobility disorder often result from faulty manufacture of connective tissue by the body – connective tissue disorders.

Ehlers Danlos Syndrome (EDS) a group of connective tissue disorders that are inherited and varied in effects. Generally, Ehlers Danlos Syndrome is characterized by joint hypermobility, hyper-stretchable skin and tissue fragility.

Connective Tissue

There are four major types of tissues which make up the human body: nerve tissue, muscle tissue, epithelial tissue (which covers and lines many organs such as the gut and is part of the skin) and connective tissue.

It is connective tissue we are most concerned with as we talk about hypermobility disorders. Connective tissue is everywhere in the body and serves many purposes. It supports, packages and compartmentalizes. The bowels, brain, spinal cord, nerves of the arms and legs and all muscles are wrapped with a connective tissue support web. Connective tissue holds cell types which are responsible for releasing chemicals to start the healing process including inflammation when there is an injury or an invasion by a foreign body or organism. Connective tissue holds fat cells which are themselves a type of

connective tissue and provide energy storage and insulation. In fact, technically, blood cells are a kind of connective tissue which carries nutrition and oxygen all over the body.

Collagen is a protein which makes up the main part of the structure of connective tissue. Normal collagen makes connective tissue strong and limits stretchiness. Healthy collagen does not break easily and heals well when it does. In hypermobility spectrum disorder and Ehlers Danlos Syndrome, faulty collagen makes the connective tissue weak, too stretchy, and easy to break. And then when this faulty tissue breaks it does not heal well.

Ehlers Danlos Syndrome

Between 2012 and 2017, an international consortium of doctors and scientists worked on a comprehensive classification system for the different types of EDS. They identified 13 types, including 12 classified types of EDS plus one non-classified type. Each of these have clear clinical diagnostic criteria. Twelve of the thirteen have identified genetic markers with available testing. One is thought to have a genetic link but does not yet have an identified genetic marker or a genetic test although a test is anticipated soon.

Here are the identified types of Ehlers Danlos with the abbreviations used to refer to them:

Classical, cEDS	Spondyloplastic, spEDS
Classical-like, clEDS	Musculocontractural, mcEDS
Cardiac-valvular, cvEDS	Mypathic, mEDS
Vascular, vEDS	Periodontal, pEDS
Arthrochalasia, aEDS	Hypermobile, hEDS
Dermatopraxis, dEDS	Unclassified AEBP1 mutation
Brittle cornea syndrome, BCS	

In this introductory paper, we will not delve into all of these types, but will focus on Hypermobile type (hEDS). The vast majority of cases, roughly 80- 90%, seen in the clinic are of this type. While hypermobile Ehlers Danlos Syndrome has very clear diagnostic criteria, it is the one that does not yet at this writing have a genetic test (although one is expected soon).

Symptoms and Associated Problems

As we said above, connective tissue is all over the body and penetrates every area. In fact, if some mad wizard suddenly made all of the other cell types in your body go away, there would still be a perfect three dimensional, see-through, form of your body made up just of connective tissue.

So, you might imagine that a connective tissue disorder, one which results in faulty production of collagen can affect almost every system of your body. Just about every patient we see with HSD or hEDS hurts in many places in their body, sometimes “everywhere” – mostly in muscles and joints. That said, it is common to have many other problem areas associated with hEDS including: skin, muscle, joint, digestive system, neurological system, eyes, reproductive and urinary systems plus some psychological complications.

We will not attempt to go into the full list of commonly associated problems here at this time. That list is long, and no one person has all of the possible problems, but a very short version could include: difficulty swallowing, chronic constipation, chronic diarrhea, problems urinating, bloating, nausea, hernias, joint dislocations, migraines and headaches, insomnia, jaw pain, anxiety, depression, eating

disorders, menstrual problems, infertility, pinched nerves, and a number of different kinds of chronic pain.

Diagnosis of hypermobile Ehlers Danlos (hEDS) and Hypermobility Spectrum Disorder (HSD)

As was said above, hypermobile Ehlers Danlos Syndrome (hEDS) is by far the most common type of Ehlers Danlos and is the only type which does not at this writing have a genetic test.

Diagnosis of hEDS is made based on three very specific levels of criteria. While your physical therapist can screen for many of the components of these criteria, the final diagnosis is based on exclusion of other disease types. This must be done by a medical provider.

Criteria 1 is a screening for generalized hypermobility and begins with a common screening test called the Beighton Test. See Table 1.

Criteria 2: This step screens for three areas of disease *features*, labeled "A, B, C". Two or more feature areas must be positive to qualify for EDS. See Table 2.

Criteria 3: This step looks at factors which could exclude the diagnosis of EDS. See Table 3.

Table 1
Criterion 1
Beighton Test

The test is scored from 0-9 points. The score is considered positive for generalized hypermobility if:

6 or greater for a prepubescent child,
5 or greater for men and women up to 50 years,
4 or greater for adults over 50.

- With palm on the table, can the patient pull their little finger up and back beyond 90 degrees? If yes, one point for each side.
- Can the patient pull their thumb and touch it to their forearm? If yes, one point for each side.
- When fully extended, does the patient's elbow go 10 degrees or more past straight? If yes, one point for each side.
- When fully extended, does the patient's knee go 10 degrees or more past straight? If yes, one point for each side.
- Keeping the knees straight and bending forward from the waist, can the patient place their palms on the floor. If yes, one point.
- Extra Points for Adults Based on History: If yes to two or more of these questions, add one point.
    - Can you now or could you ever place your hands on the floor without bending your knees?
    - Can you now or could you ever bend your thumb back to touch your forearm?
    - As a child, did you amuse your friends by contorting your body in to strange shapes or could you do the splits?
    - As a teenager, did your shoulder or kneecap dislocate on more than one occasion?
    - Do you consider yourself "double jointed"?

Table 2

Criterion 2: Features. Two or more features must be positive for Hypermobile EDS

Features A (must have five or more to be counted as positive for this feature)

- Unusually soft or velvety skin
- Mildly excessive skin stretchability
- Unexplained streaks and red marks (stretch marks) in the back, groin, thighs, breasts without a history of major weight loss of gain.
- Piezogenic papules of both heels; small whitish raised lumps on the heels usually where shoes rub
- Recurrent or multiple hernias
- Sunken or dented scars (atrophic) of at least two sites. Note: scars that look like cigarette paper or have a yellowish-brown pigment due the presence of a blood pigment (hemosiderin) may indicate a different type of EDS.
- Pelvic floor, rectal or uterine prolapse (collapse or falling out) without predisposing medical conditions such as multiple child births or severe obesity
- Dental crowding and high or narrow palate
- Abnormally long and slender. (Arachnodactyly, "spider fingers"). This item is positive if one or both of the following tests are positive. (1) Have the patient grasp around their opposite wrist with their thumb and pinky finger. If the patient's thumb nail overlaps the nail of the pinky finger, this is a positive "Wrist Sign" also called Walker sign. (2) With the thumb folded across the hand, the patient's thumb tip extends past the border of the hand on the pinky side. This is called "Thumb Sign" or Steinberg test.
- Arm span to height ratio greater than or equal to 1.05. Note: with arms held straight out to the sides from the shoulder measure the distance from the tip of the long finger to long finger. Divide by the patient's height. Divide by the patient's height. If greater than or equal to 1.05, this is a positive.
- Mitral valve prolapse -- diagnosed by a physician
- Aortic root dilatation with Z-score greater than +2 – diagnosed by a physician

Feature B

- One or more immediate family members who meet the criteria for hEDS.

Feature C (must have at least one to be counted as positive for this feature)

- Pain in bones, joints or muscles or two or more limbs occurring daily for at least three months.
- Chronic widespread pain for three or more months.
- Recurrent joint dislocations or joint instability, which is obvious to examiner, in the absence of trauma.

Table 3

Criterion 3. Ruling out other conditions. All must be met for the criterion to be positive.

- No unusual skin fragility. When present, consider other types of EDS.
- Ruled out other medical inherited or acquired connective tissue disorders such as lupus or rheumatoid arthritis
- Ruled by examination, history, lab test and/ or genetic tests, other conditions that may make connective tissue too mobile and muscles too soft. Examples of other conditions can be other types of EDS, Marfans Syndrome and neuromuscular conditions.

All three criteria levels must be positive for a diagnosis of Ehlers Danlos Syndrome to be made. If some criteria are positive but not enough, it is likely that the patient will be diagnosed with hypermobility spectrum disorder (HSD). A diagnosis of HSD is not better and does not mean that the patient is having an easier time than EDS. Every case is individual.

Now that You Have a Diagnosis

With many of our patients who have finally been diagnosed with EDS or HSD, there is often real relief to know that all or almost all of the varied problems they have struggled with for years can be traced back to one central explanation. One survey done several years ago of patients with Ehlers Danlos syndrome found that the average time to diagnosis of the condition was 20 years. In that time, many patients have suffered a great deal of medical trauma -- practitioners who did not believe them and sometimes were rude or abusive, and too often, invasive and ultimately unnecessary procedures. If this is you, we hope you will take a moment to breathe and to honor yourself for all that you have been through.

When the diagnosis of HSD or hEDS or another form of EDS has been made, it is important to pause to know some important things:

- this is a long-term condition that does not have a cure but which has good ways to manage
- there are answers to the worst of your symptoms and many of the lesser ones too
- you are not alone – millions of people across the world also the same diagnosis as you
- you are strong and courageous and there is great reason for hope.

Treatment

The course ahead for treatment will be very individual and based on your case. That said, we have found a number of common principles that are part of a successful strategy.

Principle 1: Build Your Team. We have mentioned this before, but in all cases where patients have reached a successful level of living with their condition, they have built a team – a resource network of family, friends, others with the same diagnosis and healthcare providers.

We like to imagine this as being like building your own house and acting as your own general contractor. You get to choose. You are not a passive consumer of healthcare. And, once you have educated yourself, you will hire a health team to act as your partners on the journey.

Depending on what conditions you are dealing with, a successful health team will require one to several of the following: a primary care medical or osteopathic or naturopathic physician (always needed), orthopedist, cardiologist, neurologist, allergist or immunologist, gastrointestinal specialist, pain management specialist, physical therapist, occupational therapist, speech pathologist, pelvic health specialist, mental health provider.

And how do you decide on what partners you bring on to your team? First, you are looking for knowledge. In the case of HSD and hEDS or EDS, this means that the provider has some knowledge of and experience with these conditions – the more the better. Membership in an EDS support group can be helpful in finding these providers. Second, the provider does a reasonably good job of listening to you and engaging with what you are telling them. (Remember they are human too and often subject to many pressures from the healthcare system beyond their control, so give them some latitude if you can.) Third, you feel you can trust them. This becomes especially important at times in which the provider has to nudge you or perhaps give you information you do not like.

Principle 2: Deal with the worst first. This might seem silly to say, but we find that patients are usually dealing with one to several worst problems at a time. It is important to focus and take them one-by-one or at least a few at a time. If you try to fix everything at once, it will be very easy to lose focus and never feel like you are gaining.

Principle 3: Educate Yourself. We have mentioned this before, but the more you know the better able you will be to problem solve and also to direct your care. Please join the Oregon Area Ehlers Danlos Support Group on Facebook if you have not already done so. Also visit the website of the Ehlers Danlos Association: **https://www.ehlers-danlos.com/** .

Principle 4: Joint Protection. As someone with HSD or hEDS, your joints are too mobile. As you progress through your physical therapy program, your therapist will help you to learn how to protect your joints and teach plus practice the safest ways of using your body. Here are a few guidelines in advance:

- Take special care of the smallest joints.
    - In your hands, this means being careful not to pound on things with your hands and to be very careful about bearing weight through your hands. Some patients find protective supports helpful, especially at night, your therapist will guide you.
    - In your feet, supportive shoes, especially those that give good shock absorption, are very important.
- Lift with care. Lift from your legs rather than bending at the waist. Keep the load close to you. Avoid twisting. Make notes below and ask your therapist to help you analyze and problem solve difficult situations.
- Sit with care. So many of the problems we see with our HSD and hEDS patients are made worse by sustained computer and cell phone postures which stress the joints. Consider having a friend,

child or housemate take some candid profile pictures of you on your phone so we can review your work station set-up with you. See the homework below.

Principle 5: Strengthening and Stretching will be your friends: We do know that many of our patients have had physical therapists who were rough or who pushed them to do things which hurt their body. Now that we do know what we are dealing with, we can, we hope, take a wiser, slower, more gentle and gradual approach.

Having generalized hypermobility does not mean that you cannot or do not have some muscles that are too tight. When we find this, gentle stretching exercises can be your friend. And on the other side of this, we know from experience that gentle strengthening to keep the joints as stable as possible will be a long-term program. We will do everything possible to keep the number of exercises down to the most important ones and to work in a way to keep pain to minimal or none. Oh, and fun … have fun as much as possible.

Final Note

There are two conditions which are so often associated with HSD and hEDS that we want you to be aware of them even if you do not have them.

Postural orthostatic tachycardia syndrome, POTS, is a condition in which the involuntary nerves which control the opening and closing of blood vessels and heart rate and blood pressure become out of balance because of the connective tissue problems. The most common symptom of this is dizziness and/ or faintness and/ or racing heart rate with prolonged standing. This may come and go in mild cases. If this sounds like something that has troubled you, especially if we have not talked about it with you, please bring it up with your therapist.

Mast Cell Activation Syndrome (or Disorder), MCAS or MCAD. There are small and important cells in the connective tissue of the body which send out chemical signals to tell the body that there has been a wound or a trauma or an infection. These are called Mast Cells and they start the process of healing, especially the inflammatory process which is natural and necessary and important. Sometimes though, these cells become too active when there is a connective tissue problem. The result can be many different types of inflammatory problems all over the body. If there are many different conditions that all seem joined by inflammation, it may be important to screen and recommend that you see your primary physician. MCAS is normally well treated by some simple and conservative medications.

Welcome to Good Health Physical therapy and Wellness.

- **Homework: Your Common Postures**
  Make a list of the activities and especially the postures which most commonly make you hurt. Bring this for your therapist. If you can recreate these postures such as how you lift, or how you sleep, or sitting at your computer or texting on your phone, have someone take some pictures of you doing the activity. Note: Candid is better. And please do not photograph toward an open window – this just makes the pictures too dark.
  Thanks. See you next visit!

Notes: