Hypermobility Spectrum Disorders (HSD) and Hypermobile Ehlers-Danlos Syndrome (hEDS)

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Hypermobility Spectrum Disorder (HSD) is the most common systemic inherited connective tissue disorder in humans. This means that it runs in families. However, no specific gene has yet been identified, so genetic testing is not helpful at this time. We don't have good research showing how many people have symptomatic hypermobility, but it is believed to be 1-2% in the US.

System	**Health Issues** (Russek, 2019)
Bones & Joints	Frequent sprains, subluxations, and dislocations. Chronic joint pain, osteoarthritis. Scoliosis. Decreased bone density, increased fracture rate (controversial)
Soft Tissues	Tendinitis, bursitis, synovitis, tenosynovitis, fasciitis, or tendon ruptures. Trigger points, muscle spasm, muscle strain.
Autonomic Nervous System	Dysautonomia with orthostatic hypotension and/or postural orthostatic tachycardia syndrome (POTS) presenting with: rapid heart rate, syncope/presyncope (passing out or feeling like you will pass out), anxiety, chronic fatigue, sleep disorder, exercise intolerance, dependent edema, purpling skin, temperature dysregulation, "brain fog," and trouble concentrating. Raynaud syndrome.
Cardiovascular	Varicose veins. Mitral valve prolapse or aortic dilatation (not common). Rapid heart rate.
Neurological	Motor delay (in children). History of being clumsy or uncoordinated. Decreased body awareness and motor control deficits leading to clumsiness, frequent falls, trips, or bumping into things. Fibromyalgia/central sensitization, hyperalgesia (oversensitive nerves). Headaches, migraines, dizziness, ringing in the ears. Numbness/tingling and nerve compression disorders. Restless leg syndrome. Vulnerable to central nervous system problems that may lead to dystonia (abnormal muscle contractions), pseudo-seizures, difficulty thinking and loss of consciousness.
Cognitive	Anxiety and panic disorder. Memory or concentration problems ("brain fog"). Depression.
Gastrointestinal	Irritable bowel syndrome, constipation or diarrhea, bloating, abdominal pain, gastroparesis, food sensitivities. Gastroesophageal reflux, chronic gastritis, heartburn. Prolapsed rectum. Hernias (all types).
Skin	Hyperextensible skin. Slow healing or scarring, poor wound healing. Easy bruising.
Urogenital	Urinary incontinence. Prolapsed bladder or uterus. Frequent urinary tract infections. Dysmenorrhea, endometriosis, vulvodynia, pelvic pain, painful intercourse.
Immune	Mast cell activation syndrome (MCAS): hives, pruritus, flushing, chemical and environmental sensitivities, medication and food sensitivities, fatigue, trouble concentrating, migratory pain, excessive inflammatory response, trouble concentrating, anxiety.
Other	Insomnia, sleep disturbance, and debilitating chronic fatigue.

HSD complaints tend to fall in 3 phases, but each individual may have specific problem areas, or problems at any age/phase

Hypermobile phase	**Pain phase**	**Stiffness phase**
Hypermobile joints Clumsiness/motor delay Constipation/diarrhea Abdominal hernias	Chronic musculoskeletal pain Strains, sprains, dislocations Unrefreshing sleep and chronic fatigue Memory/cognitive problems Gastric reflux, abdominal pain Paresthesias, numbness, tingling Tachycardia (racing heart), anxiety, panic attacks Incontinence/urinary tract infections	Tendinosis/tendon rupture Chronic gastritis Stiffness • Castori et al, 2011, Tinkle et al, 2017

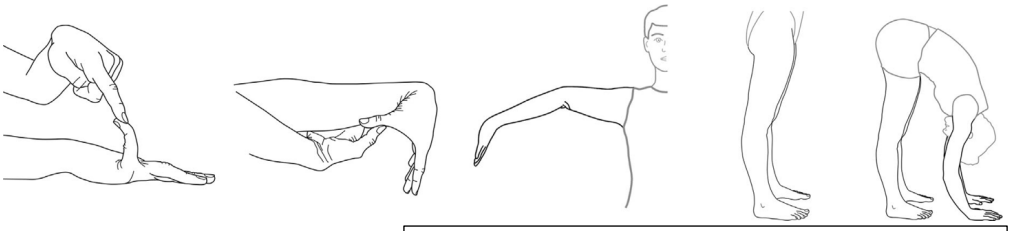
Diagnostic Criteria

- In 2017, researchers and clinicians redefined the diagnostic criteria to help identify possible genetic defects in the hypermobile type of EDS. They created a stricter set of diagnostic criteria for **hEDS** (see Ehlers-Danlos Society web page for details: https://www.ehlers-danlos.com/heds-diagnostic-checklist/)
- People who are hypermobile, have health problems related to hypermobility, and don't quite meet the criteria for hEDS are diagnosed with **HSD**. Initially, people thought that HSD was a milder form of hEDS, but we now know that is not true. HSD and hEDS can have similar severity, and may both affect multiple systems in the body (not just joints).
- **From a treatment perspective, HSD and hEDS are identical. There is no real benefit to distinguishing between them.**
- Prior names for hEDS/HSD include: "Hypermobility Syndrome," (HMS) or "Hypermobile Joint Syndrome" (HJS), "Joint Hypermobility Syndrome" (JHS) and "Benign Joint Hypermobility Syndrome" (BJHS). Although it won't kill you, it isn't benign!
- Hypermobility, laxity, and instability are different things. You can be hypermobile without having instability or symptoms.
  - Hypermobility refers to excessive range of motion at a joint (as demonstrated in the Beighton score).
  - Laxity refers to excessive looseness in a joint, such as wobble or too much accessory motion.
  - Instability refers to the inability to control movement at a joint. Instability occurs when the nerves and muscles are not trained to control movement, especially in mid-range. Exercises to train body awareness (proprioception) and motor control (activating the correct muscle at the correct time) can decrease instability. In contrast, being sedentary and not exercising can increase instability.
- The Beighton Score (next page) is the official measure of hypermobility. These are not the only important joints that may be hypermobile & problematic. Any joint may have excessive motion or instability. Other scales are now being studied.

Beighton Score (9 joints tested)

Cutoffs:

- 3-7 y/o: ♀≥6/9; ♂≥5/9
- 8-18 y/o: ♀≥5/9; ♂≥4/9
- 18-49 y/o: ≥5/9
- Over 50 y/o: ≥4/9



If you miss the Beighton cut-off by 1 point, you can add 1 point by answering Yes to ≥2/5 items on the "5-Item Questionnaire" shown in the box.

1. Can you now (or could you ever) place your hands flat on the floor without bending your knees?
2. Can you now (or could you ever) bend your thumb to touch your forearm?
3. As a child, did you amuse your friends by contorting your body into strange shapes or could you do the splits?
4. As a child or teenager, did your shoulder or kneecap dislocate on more than one occasion?
5. Do you consider yourself "double-jointed"?

Physical Therapy Evaluation

- Patients often have many complaints; focus on the ones that have the greatest impact on their lives.
- Identify tissues causing symptoms.
- Identify stressors affecting those tissues – this is critical! You need to fix the cause of symptoms, not just symptoms.
    1. Does poor posture, joint alignment, or gravity stress joints, muscles or nerves?
    2. Are poor coordination, poor motor control, or weak muscles leading to instability?
    3. What is causing painful muscle trigger points?
    4. Is there an imbalance between loose joints and tight muscles increasing instability?
    5. Is the patient fatigued, anxious, trouble sleeping? Do the Stand Test for POTS, if appropriate.

Physical Therapy Treatment Approach must be specific to each patient's needs. (Engelbert, 2017; Russek, 2019)

Education

- **Educate and empower the patient/family to manage the condition, since it cannot be 'cured'**
- Pain education & self-management: body mechanics, ergonomics, joint protection, TENS, topical medications, self-management of myofascial trigger points
- Body mechanics/ergonomics: using your body wisely, good posture, not stretching joints or straining muscles
- Orthotics, braces & splints if needed; canes, crutches, if condition is very involved
- Appropriate exercise/activity (in general, low impact, controlled activity; aquatic exercise, Pilates, Tai Chi etc. are ideal)
- Sleep hygiene & fatigue management (also see POTS and Graded Exercise Therapy)
- POTS self-management (for fatigue, exercise intolerance, anxiety, panic, difficulty concentrating, sleep disorder)
- Psychological & social wellness, including relaxation, mindfulness meditation, social contact
- Diet and fluid management – some people benefit from the Heidi Collins diet, FODMAP diet, or low histamine diet
- Other issues: GI dysfunction, MCAD, incontinence, pain with intimacy, etc.
- Refer to other professionals as appropriate: occupational therapist, nutritionist, psychologist, gastroenterologist, etc.

Exercise

- Begin with body position awareness (proprioception) motor control & coordination. Always start by moving correctly!
- Strengthening, careful not to stress joints; "start low, go slow" as tissues are fragile and strengthen slowly.
- Appropriate lengthening of tight muscles, careful to stabilize loose joints. We naturally stretch where we are already stretchiest.
- Cardiovascular conditioning using good body mechanics; "start low, go slow." Follow POTS guidelines if necessary
- "Graded Exercise Therapy" provides good guidelines for pacing and progressing exercise very slowly
- POTS-specific exercise for patients who have POTS: horizontal rather than upright, lower extremity first, etc.

Pain Management

- Pain management: learning about pain, cognitive behavioral strategies such as: pacing, stress management, avoiding negative thinking. Relaxation and physiological quieting, using mindfulness meditation, biofeedback, Tai Chi, yoga, etc.
- Self-care using exercise, TENS, braces/splints, topical analgesics, etc.
- Manual therapy (if/when appropriate): myofascial release, gentle joint realignment, etc.
- Address musculoskeletal injuries, such as subluxations, sprains, trigger points, tendinosis, impingement, etc.

Resources

- Dr. Russek provides (almost) weekly live lectures on many aspects of hEDS/HSD; recordings and slide hanouts at https://webspace.clarkson.edu/~lrussek/hsd.html. Many additional patient handouts are available at https://webspace.clarkson.edu/~lrussek/research.html.
- The Ehlers-Danlos Society web site: www.ehlers-danlos.com has excellent information on EDS for both patients and providers, guidance for school accommodations, and full text of the 18 professional publications on EDS from 2017.
- The Hypermobility Syndrome Association (HMSA): www.hypermobility.com has excellent information on hypermobility for both patients and providers, including an excellent section for children and adolescents. The School Toolkit (https://theschooltoolkit.org) has excellent school accommodation info.
- Another excellent handout: https://www.versusarthritis.org/media/24681/hypermobility-information-booklet-march2022.pdf
- POTS UK:(www.potsuk.com) and Dysautonomia International (http://www.dysautonomiainternational.org) have excellent information on POTS for both patients and providers.

- Russek LN, Stott P, Simmonds J. Recognizing and Effectively Managing Hypermobility-Related Conditions. Phys Ther. 2019;99(9):1189-200.
- Engelbert RHH, Juul-Kristensen B, Pacey V, De Wandele I, Smeenk S, Woinarosky N, et al. The Evidence-based rationale for physical therapy treatment of children, adolescents and adults diagnosed with joint hypermobility syndrome/hypermobile Ehlers Danlos Syndrome. Am J Med Genet C Semin Med Genet. 2017;175(1):158-67.

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