Why Does Hypermobility Matter?

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Who Am I?

- Professor Emeritus, Physical Therapy, Clarkson University.
- Staff PT, St. Lawrence Health System, Potsdam NY.
  - Clinical specialties: hypermobility, chronic pain, fibromyalgia, headaches, temporomandibular disorders
  - I treat through Lawrence Ave Physical Rehab office, 315-261-5460
- Facilitator of the North America Allied Health Professionals ECHO
- Member of:
  - The Allied Health Working Group of the International Consortium of Ehlers-Danlos Syndromes and Hypermobility Spectrum Disorders
  - Past member of The National Academy of Sciences, Engineering and Medicine Committee on Selected Heritable Connective Tissue Disorders and Disability.
- Author of many articles related to HSD/hEDS
- Author of "Chronic Pain" chapter in *Physical Rehabilitation* textbook for PT students
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I do not have any conflicts of interest to report

Objectives

By the end of this presentation, participants will be able to:

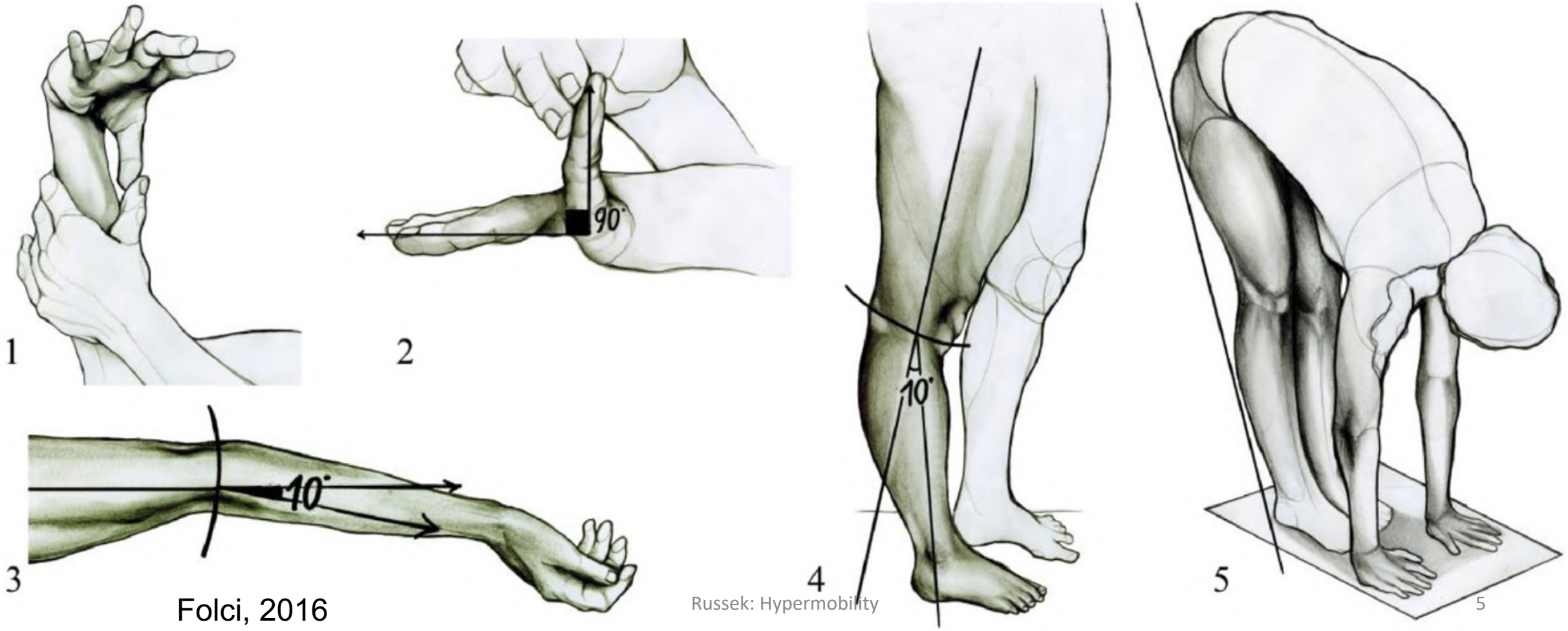
1. Recognize patients with Hypermobility Spectrum Disorders/ hypermobile Ehlers Danlos Syndrome (HSD/hEDS)
2. Describe why it is important to recognize HSD/hEDS and the common comorbidities of Postural Orthostatic Tachycardia Syndrome (POTS) and Mast Cell Activation Disorder (MCAD)
3. Describe general treatment approaches for HSD/hEDS
4. List several precautions due to HSD/hEDS, POTS or MCAD

NASEM Guide Statement on Delayed Diagnosis of Heritable Disorders of Connective Tissues (HDCT)

- *"Delayed or misdiagnosis of individuals with HDCTs can result in*
  - *inappropriate medical interventions;*
  - *inability to accurately assess the risks and benefits associated with medical procedures;*
  - *inability to access necessary reasonable accommodations at work or school;*
  - *family stress and dysfunction;*
  - *stress associated with unexplained and repeated evidence of trauma, leading to inappropriate suspicion of child abuse;*
  - *inappropriate assessments and incorrect diagnoses; and*
  - *mistrust of health care providers and negative expectations for future health care encounters"*
- https://nap.nationalacademies.org/catalog/26431/selected-heritable-disorders-of-connective-tissue-and-disability (page 296)

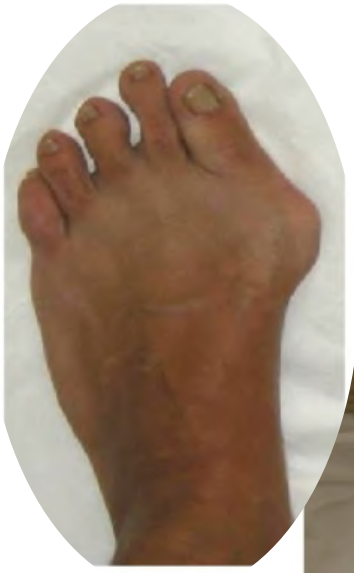
- Delayed diagnosis increases pain, disability and burden of the disorder (Kalisch, 2019; Adib, 2005)

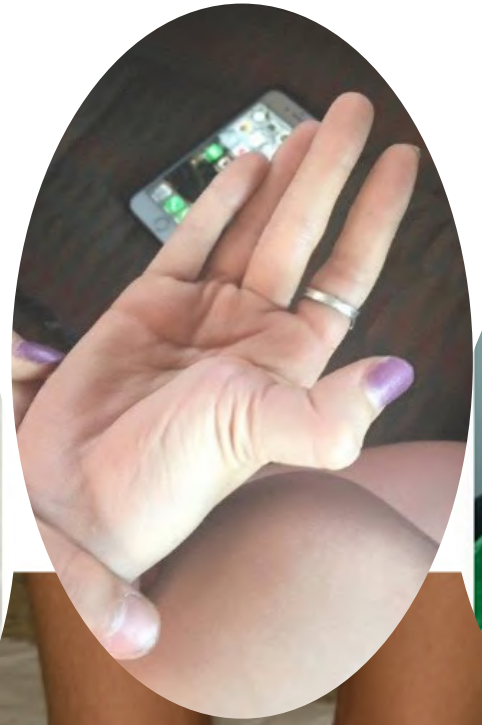
Does This Matter to YOU?

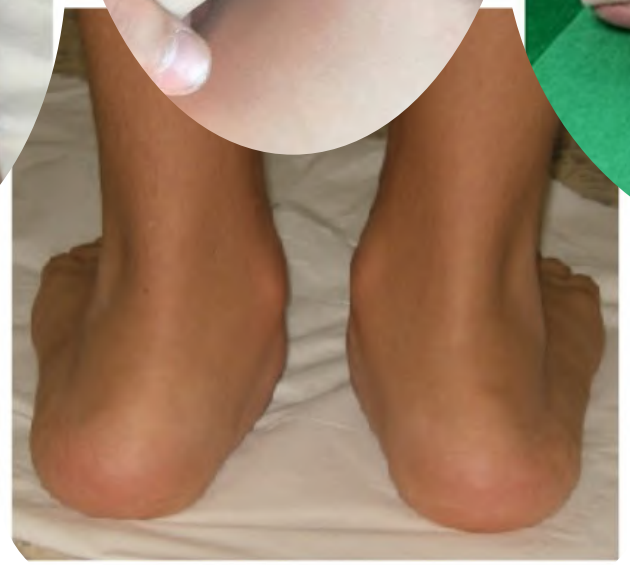


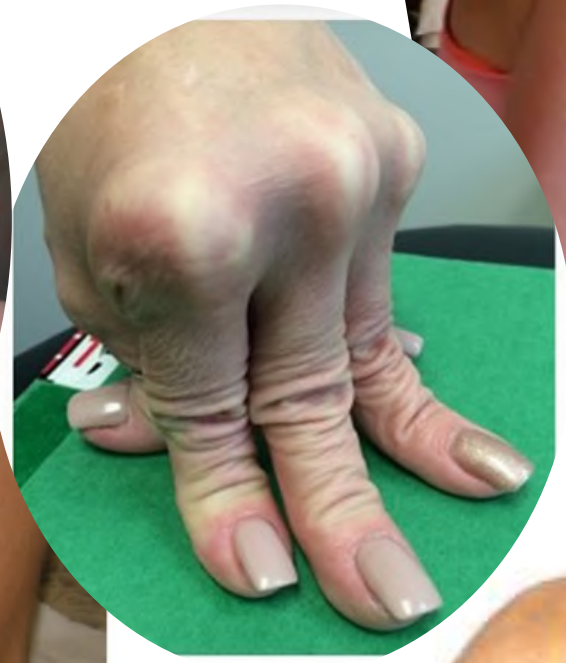
Folci, 2016

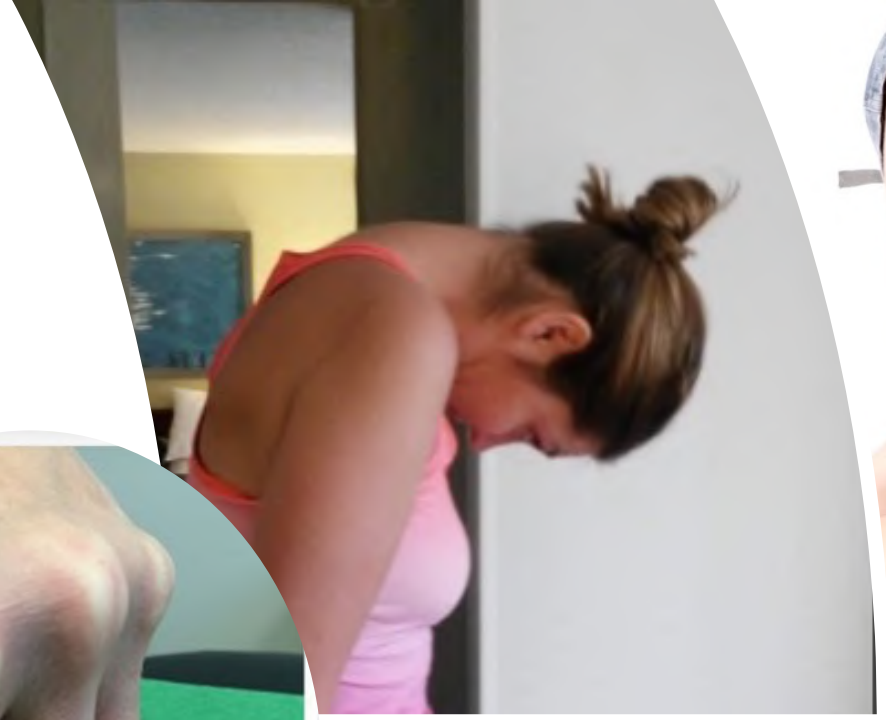
Does This Look Familiar?





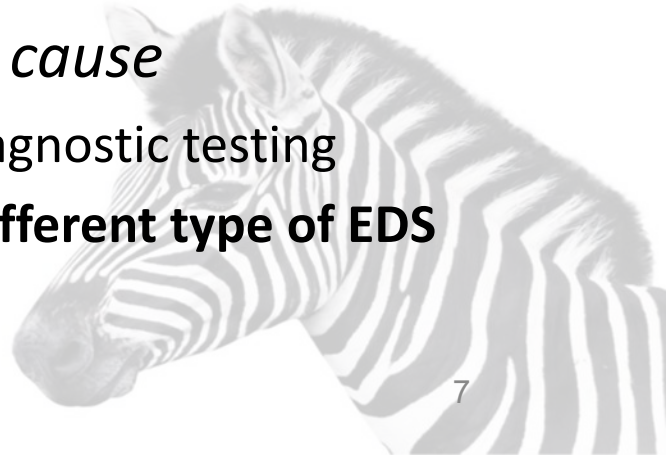






Ehlers-Danlos Syndrome (EDS)

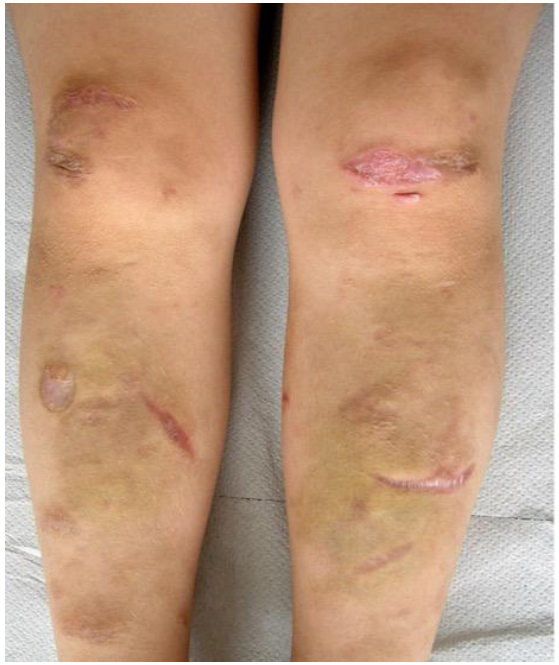
- Generalized joint hypermobility (GJH): asymptomatic widespread hypermobility
- Ehlers-Danlos syndrome (EDS): Inherited connective tissue disorders
  - 13 subtypes defined in 2017 classification (Malfait, 2017)
- **Hypermobile EDS (hEDS) and Hypermobility Spectrum Disorders (HSD)**
  - HSD/hEDS is the most common subtype (~90% of EDS)
  - hEDS/HSD is the only EDS subtype with *no known genetic cause*
    - Diagnosis is clinical – based on signs & symptoms rather than diagnostic testing
    - **Genetic testing is NOT generally helpful unless you suspect a different type of EDS**

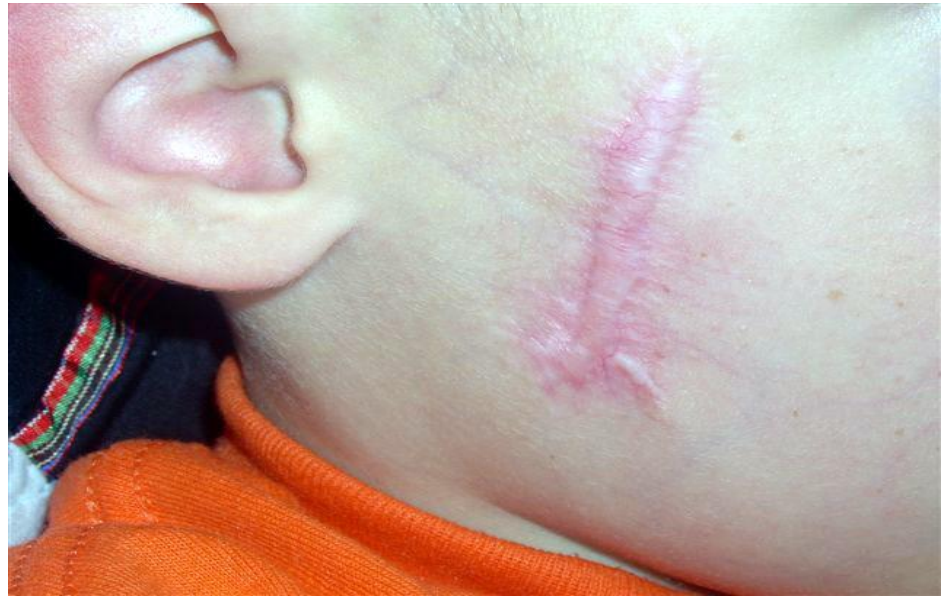


Other Types of EDS

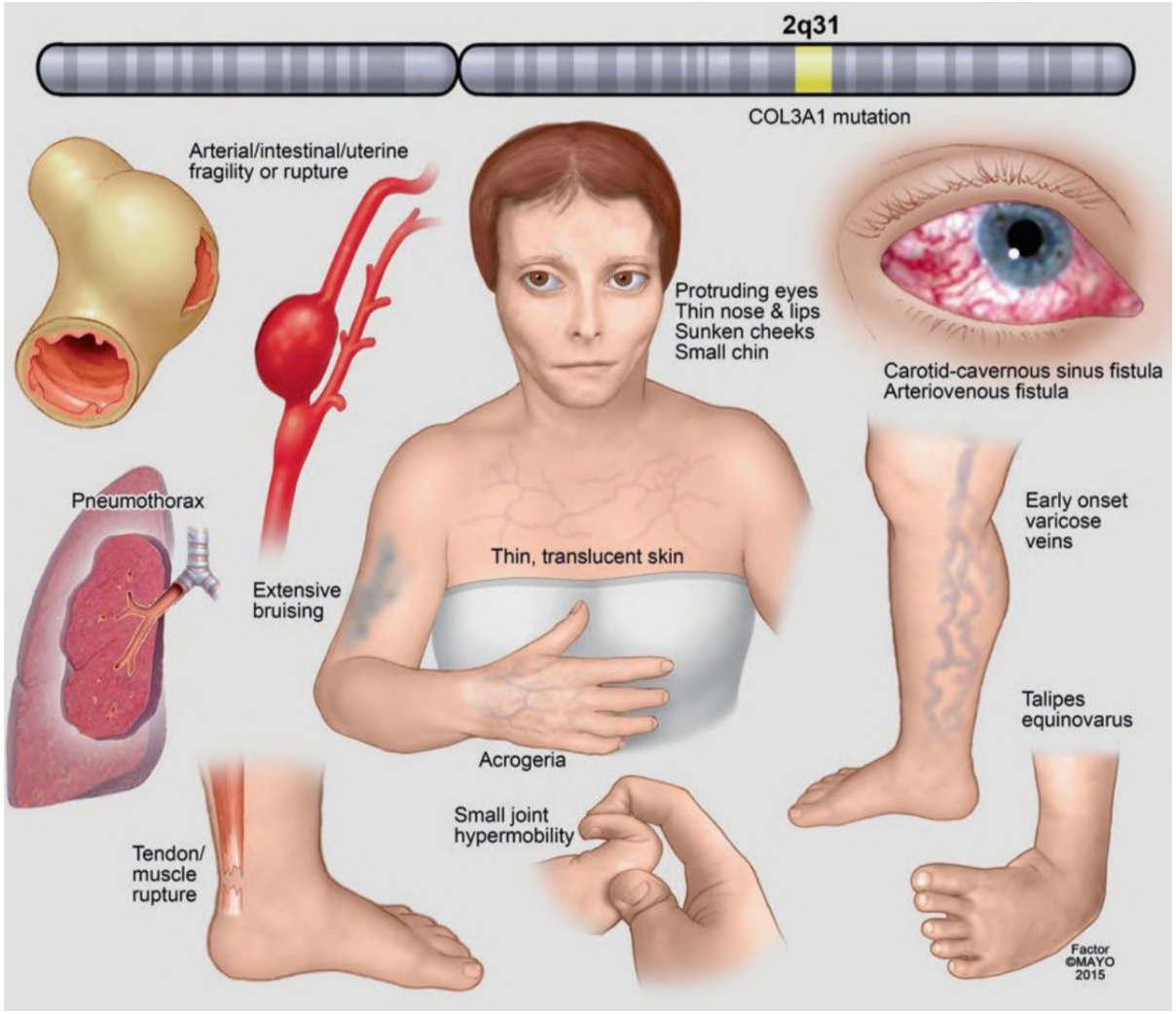
- Classical (old names Type I & II): Velvety, stretchy, fragile skin.
  - Second most common (after hypermobile).
- Vascular (old name Type IV): Possible arterial/organ rupture.
  - Very rare (1/50,000-1/200,000), but most serious.
- 10 Other sub-types, quite rare. More info at:
  - https://www.ehlers-danlos.com/eds-types/

Classical EDS





Recognizing Vascular EDS



- Very rare (<0.002%), but don't want to overlook it
- In childhood, may present with excessive bleeding and bruising, very translucent skin, varicose veins.
- Personal or family Hx of vascular or organ rupture
  - Sudden death of family member <20 y/o
  - But only 50% have family Hx
- Hypermobility of small joints
  - Born with dislocated hip or talipes
- Premature aged appearance of hands

Byers, 2017

https://twitter.com/drkeithsiau/status/1761810953157407171

What is Connective Tissue (CT)?

- Hypermobility is due to abnormal collagen, a fiber that gives structure and strength to connective tissue
- Connective tissue connects, surrounds, and supports most structures in the body
  - Ligaments, tendons, joint capsules, fascia, cartilage
  - CT exists inside and around muscles and nerves to support and protect
  - CT surrounds, protects, and supports organs: dura (brain and spinal cord), pericardium (heart), mesentery (gut), pleura (lungs)
  - Blood vessels and lymphatic vessels
  - Bones
- Hypermobile joints are just the most visible abnormality

PRIMEVIEW

THE EHLERS–DANLOS SYNDROMES

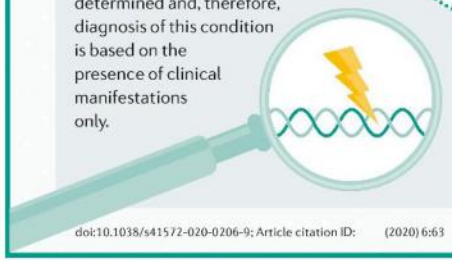
nature reviews disease primers

For the Primer, visit doi:10.1038/s41572-020-0194-9

The Ehlers–Danlos syndromes (EDS) are a group of connective tissue disorders caused by alterations in genes encoding fibrillar collagens, collagen modifying or collagen-processing enzymes, or enzymes that modify glycosaminoglycans within the extracellular matrix (ECM).

DIAGNOSIS

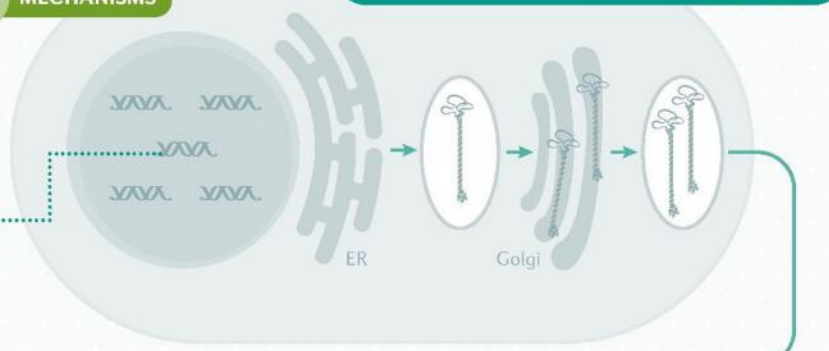
The symptoms of EDS are diverse and differ between subtypes. Symptoms that are found in multiple EDS subtypes include joint hypermobility, soft and hyperextensible skin, poor wound healing, pain and easy bruising. Vascular and musculoskeletal symptoms are present in some subtypes. Diagnostic work-up comprises clinical examination, followed by genetic testing in individuals who fulfil the clinical criteria for an EDS subtype. Genetic testing can include targeted analysis in those with a family history of EDS caused by a known genetic variant or, more frequently, next-generation sequencing using multi-gene panels. Genetic diagnosis should lead to family testing to enable detection of EDS in family members and, for patients with a recessive form of EDS, carrier testing in their partners to evaluate the risk of transmission to offspring. Of note, the genetic cause of hypermobile EDS has not been determined and, therefore, diagnosis of this condition is based on the presence of clinical manifestations only.

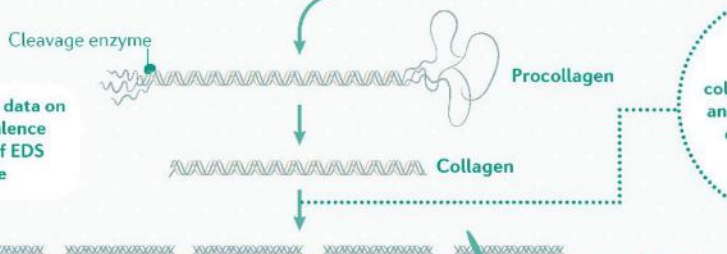


MECHANISMS

Most types of EDS are inherited in an autosomal dominant manner, although some cases can arise owing to de novo mutations or are inherited in a recessive manner

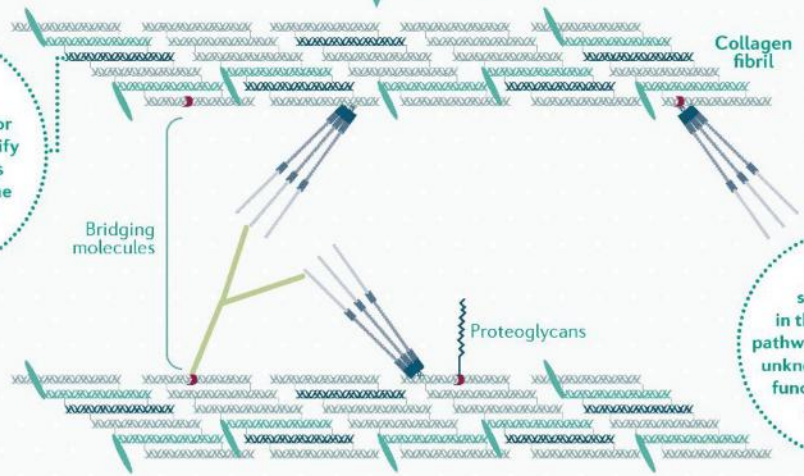
Variants in genes encoding the fibrillar procollagens I, III and V, or in genes encoding enzymes responsible for cleaving procollagen to mature collagen, lead to collagen fibrils with an abnormal structure or composition





Accurate data on the prevalence and incidence of EDS are not available

Variants in genes involved in collagen crosslinking and collagen folding causes impaired collagen crosslinking



Variants in genes encoding ECM bridging molecules or in enzymes that modify ECM proteoglycans can interfere with the organization of collagen fibrils

Other variants, such as those in the complement pathway and in genes of unknown intracellular function, can cause rarer forms of EDS

MANAGEMENT

All patients with EDS should receive multi-disciplinary care and, if available, be part of a patient advocacy community. The precise treatment depends upon the subtype of EDS and its manifestations. Physiotherapy is essential for patients with musculoskeletal alterations. Helmets and/or skin protection, or joint protection, braces or splints can be used to reduce the risk of injury in patients with skin fragility or joint hypermobility. In addition, low-resistance exercise (such as walking or swimming) can improve joint stability, although exercise that place considerable strain on the joints (such as gymnastics or weight lifting) should be avoided. Monitoring for cardiovascular alterations using non-invasive procedures is recommended in patients at risk of adverse cardiovascular events.

OUTLOOK

Despite improvements in genetic testing, some forms of EDS (hypermobile EDS) and some patients with other forms of EDS have no identified genetic cause, which can hinder diagnosis of these conditions. Large-scale international studies are underway to address this issue. Genotype–phenotype correlations for EDS are only starting to emerge; additional correlations may be identified by ongoing research.

doi:10.1038/s41572-020-0206-9; Article citation ID: (2020) 6:63

Written by Louise Adams; designed by Laura Marshall

Pathophysiology (for reference)

https://www.nature.com/articles/s41572-020-0206-9

How Common is Hypermobility?

- hEDS/HSD is the most common systemic inherited connective tissue disorder in humans. (Tinkle, 2017)

- Overall prevalence of symptomatic HSD in US: 1-3%
- hEDS/HSD in healthcare: (Simmonds, 2022; To, 2017)
  - 37% of adult patients in rheumatology clinics
  - 39% of adult patients in pain clinics
  - 30% of adult patients in primary care clinics
  - 21% of pediatric PT patients

THE BEIGHTON SCORING SYSTEM
Measuring joint hypermobility

The Ehlers Danlos Society

A. 5th FINGER / 'PINKIES'

Test **both sides**: Rest palm of the hand and forearm a **flat surface** with palm side down and fingers out straight.

Can the **fifth finger** be bent/lifted upwards at the knuckle to go back **beyond 90 degrees?**

If yes, add **one point** for each hand.

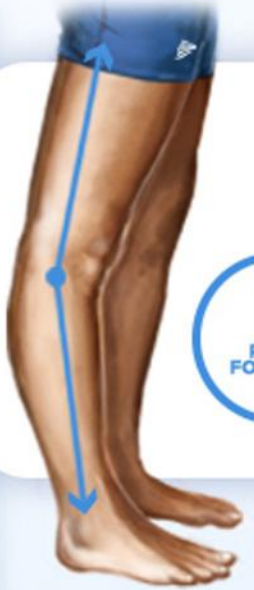
1 POINT FOR EACH HAND

B. THUMBS

Test **both sides**: With the arm out straight, the palm facing down, and the wrist then fully bent downward, can the thumb be pushed back to touch the forearm?

If yes, add **one point** for each thumb.

1 POINT FOR EACH THUMB



1 POINT FOR EACH LEG

D. KNEES

Test **both sides**: While standing, with knees locked (bent backwards as far as possible), does the lower part of either leg extend **more than 10 degrees forward?**

If yes, add **one point** for each side.

C. ELBOWS

Test **both sides**: With arms outstretched and palms facing upwards, does the elbow extend (bend too far) upwards **more than an extra 10 degrees** beyond a normal outstretched position?

If yes, add **one point** for each side.

1 POINT FOR EACH ARM

E. SPINE

Bend forward, can you place the palms of your hands **flat on the floor in front of your feet without bending your knees?**

If yes, **add one point.**

1 POINT

Used with permission from the Ehlers-Danlos Society, https://www.ehlers-danlos.com/assessing-joint-hypermobility/

Diagnosis: Adults 2017 hEDS Criteria

hEDS was defined for the purpose of seeking a genetic marker for hypermobile EDS

hEDS Must meet <u>all 3 criteria</u>:

1. Generalized joint hypermobility
2. Two of the following:
   - A. ≥5/12 Features of inherited connective tissue disorder
   - B. Family history
   - C. Musculoskeletal pain > 3 months, or dislocations
3. Absence of exclusion criteria

   (there isn't another good explanation)

- These replace both Brighton and Villefranche Criteria for JHS, HMS, EDS-HT, EDS-type III
- **These hEDS diagnostic criteria are problematic for several reasons, and do not alter conservative care**

The International Consortium on Ehlers-Danlos Syndromes & Related Disorders
In Association with The Ehlers-Danlos Society

Diagnostic Criteria for Hypermobile Ehlers-Danlos Syndrome (hEDS)
This diagnostic checklist is for doctors across all disciplines to be able to diagnose EDS

Distributed by The Ehlers Danlos Society.

Patient name: _____ DOB: _____ DOV: _____ Evaluator: _____

The clinical diagnosis of hypermobile EDS needs the simultaneous presence of all criteria, 1 ***and*** 2 ***and*** 3.

CRITERION 1 – Generalized Joint Hypermobility

One of the following selected:
- ☐ ≥6 pre-pubertal children and adolescents
- ☐ ≥5 pubertal men and woman to age 50
- ☐ ≥4 men and women over the age of 50

Beighton Score: _____/9

If Beighton Score is one point below age- and sex-specific cut off, two or more of the following must also be selected to meet criterion:
- ☐ Can you now (or could you ever) place your hands flat on the floor without bending your knees?
- ☐ Can you now (or could you ever) bend your thumb to touch your forearm?
- ☐ As a child, did you amuse your friends by contorting your body into strange shapes or could you do the splits?
- ☐ As a child or teenager, did your shoulder or kneecap dislocate on more than one occasion?
- ☐ Do you consider yourself "double jointed"?

CRITERION 2 – Two or more of the following features (A, B, or C) must be present

Feature A (five must be present)
- ☐ Unusually soft or velvety skin
- ☐ Mild skin hyperextensibility
- ☐ Unexplained striae distensae or rubae at the back, groins, thighs, breasts and/or abdomen in adolescents, men or pre-pubertal women without a history of significant gain or loss of body fat or weight
- ☐ Bilateral piezogenic papules of the heel
- ☐ Recurrent or multiple abdominal hernia(s)
- ☐ Atrophic scarring involving at least two sites and without the formation of truly papyraceous and/or hemosideric scars as seen in classical EDS
- ☐ Pelvic floor, rectal, and/or uterine prolapse in children, men or nulliparous women without a history of morbid obesity or other known predisposing medical condition
- ☐ Dental crowding and high or narrow palate
- ☐ Arachnodactyly, as defined in one or more of the following: (i) positive wrist sign (Walker sign) on both sides, (ii) positive thumb sign (Steinberg sign) on both sides
- ☐ Arm span-to-height ratio ≥1.05
- ☐ Mitral valve prolapse (MVP) mild or greater based on strict echocardiographic criteria
- ☐ Aortic root dilatation with Z-score >+2

Feature A total: _____/12

Feature B
- ☐ Positive family history; one or more first-degree relatives independently meeting the current criteria for hEDS

Feature C (must have at least one)
- ☐ Musculoskeletal pain in two or more limbs, recurring daily for at least 3 months
- ☐ Chronic, widespread pain for ≥3 months
- ☐ Recurrent joint dislocations or frank joint instability, in the absence of trauma

CRITERION 3 – All of the following prerequisites MUST be met

1. Absence of unusual skin fragility, which should prompt consideration of other types of EDS
2. Exclusion of other heritable and acquired connective tissue disorders, including autoimmune rheumatologic conditions. In patients with an acquired CTD (*e.g.* Lupus, Rheumatoid Arthritis, etc.), additional diagnosis of hEDS requires meeting both Features A and B of Criterion 2. Feature C of Criterion 2 (chronic pain and/or instability) cannot be counted toward a diagnosis of hEDS in this situation.
3. Exclusion of alternative diagnoses that may also include joint hypermobility by means of hypotonia and/or connective tissue laxity. Alternative diagnoses and diagnostic categories include, but are not limited to, neuromuscular disorders (*e.g.* Bethlem myopathy), other hereditary disorders of the connective tissue (e.g. other types of EDS, Loeys-Dietz syndrome, Marfan syndrome), and skeletal dysplasias (*e.g.* osteogenesis imperfecta). Exclusion of these considerations may be based upon history, physical examination, and/or molecular genetic testing, as indicated.

Diagnosis: _____

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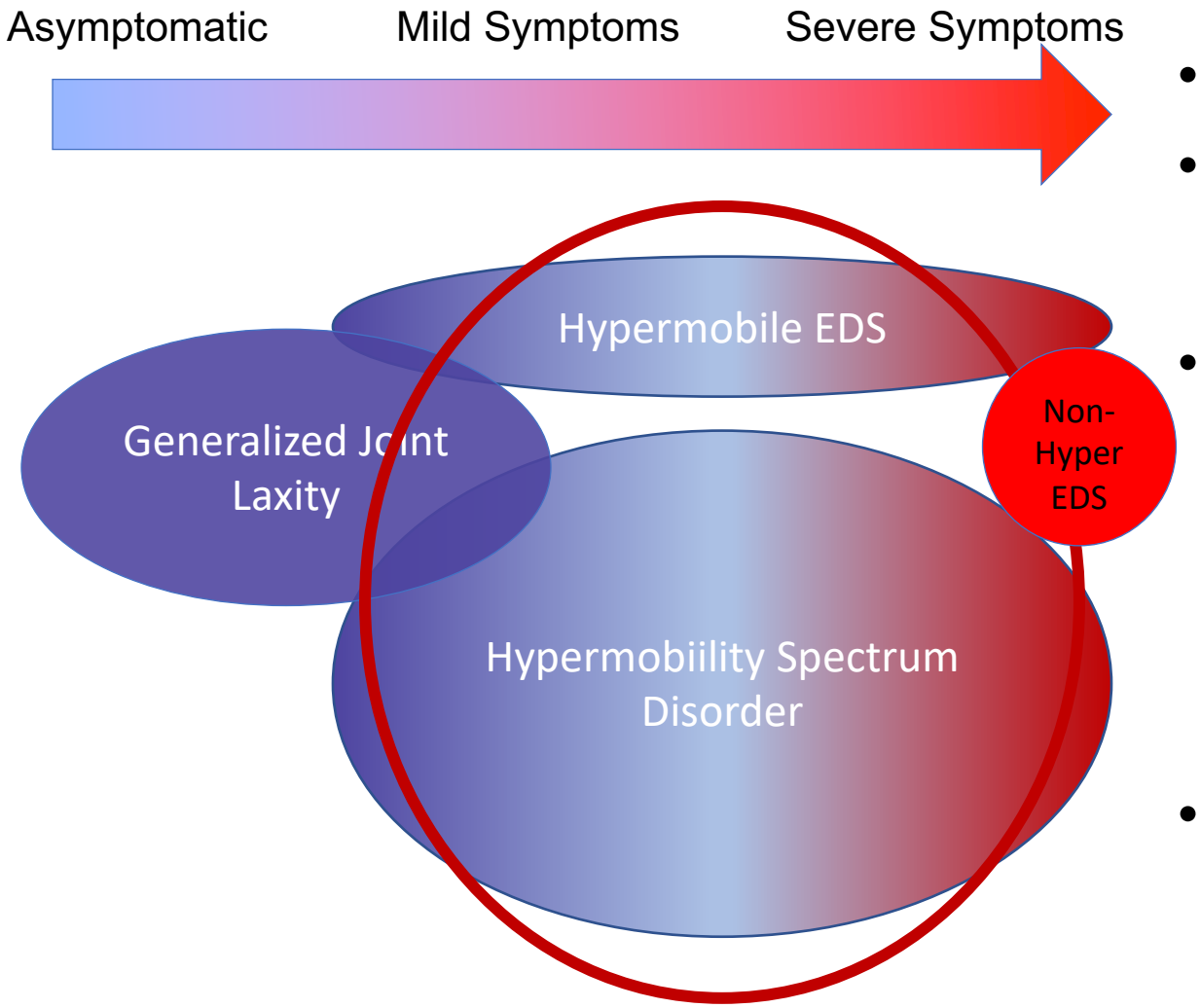
Diagnostic checklist in handout and at https://www.ehlers-danlos.com/heds-diagnostic-checklist/

HSD vs. hEDS



- hEDS if all checklist criteria are met
- HSD for "*all individuals who present with complaints and/or life quality limitations because of joint hypermobility*" (Castori, 2017)
- Pain, symptoms, & disability are similar for both groups – one is not worse than the other

(Copetti, 2019; Aubry-Rozier, 2021)

  - Patients who are hypermobile and have problems related to their hypermobility, have HSD/hEDS
- So… using the hEDS diagnostic criteria is not very helpful for patient care.

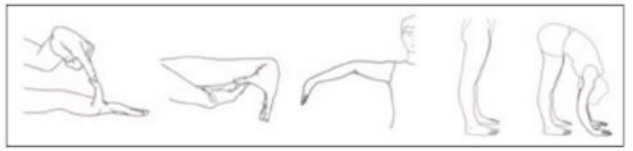
2023 Joint Hypermobility Criteria for Children

Five key questions to ask:

1. Generalized joint hypermobility
   a. Beighton Score ≥ 6
2. Skin and Tissue Abnormalities
   a. 3 out of 6 criteria → skin involvement subtype
3. Musculoskeletal complications
   a. 2 out of 3 criteria → HSD
4. Co-morbidities
   a. Yes → co-morbidities subtype
5. Excluding other conditions
   a. Patient's symptoms caused by other conditions

(Tofts, 2023)

Children From 5 Years Of Age Until Biological Maturity



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Beighton Score: _____/9
Must be a minimum of 6

Skin and Tissue Abnormalities

- ☐ Unusually Soft Skin – unusually soft and/or velvety skin
- ☐ Mild Skin extensibility
- ☐ Unexplained striae distensae or rubae at the back, groin, thighs, breasts and/or abdomen without a history of significant gain or loss of body fat or weight
- ☐ Atrophic scarring involving at least 1 site and without the formation of truly papyraceous and/or haemosideric scars as seen in classical EDS
- ☐ Bilateral piezogenic papules in the heel
- ☐ Recurrent hernia, or hernia in more than one site (excludes congenital umbilical hernia)

Score: _____/6
Must be a minimum of 3

Musculoskeletal Complications

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- ☐ Episodic Activity related pain not meeting the chronic pain frequency and duration criteria
- ☐ Recurrent joint dislocations, or recurrent subluxations in the absence of trauma, and/or frank joint subluxation on physical exam in more than one joint (excludes radial head <2yrs)
- ☐ Soft tissue injuries – One major (needing surgical repair) and/or current multiple minor tendon, and/or ligament tears

Score: _____/3
Must be a minimum of 2

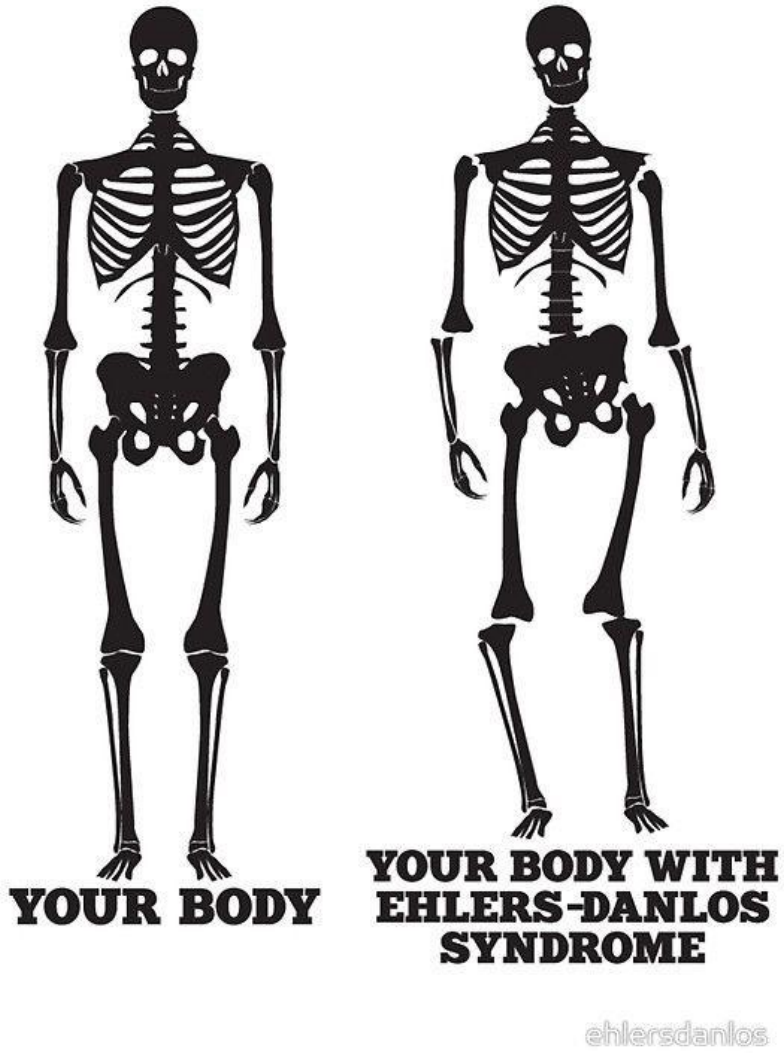
Co-Morbidities

- ☐ Chronic primary pain
- ☐ Chronic fatigue
- ☐ Functional GI disorders
- ☐ Functional bladder disorders
- ☐ Primary dysautonomia
- ☐ Anxiety

Any number causing distressor disability?
Y / N

Clinical Presentation & Common Comorbidities

“The Terrible Trifecta”

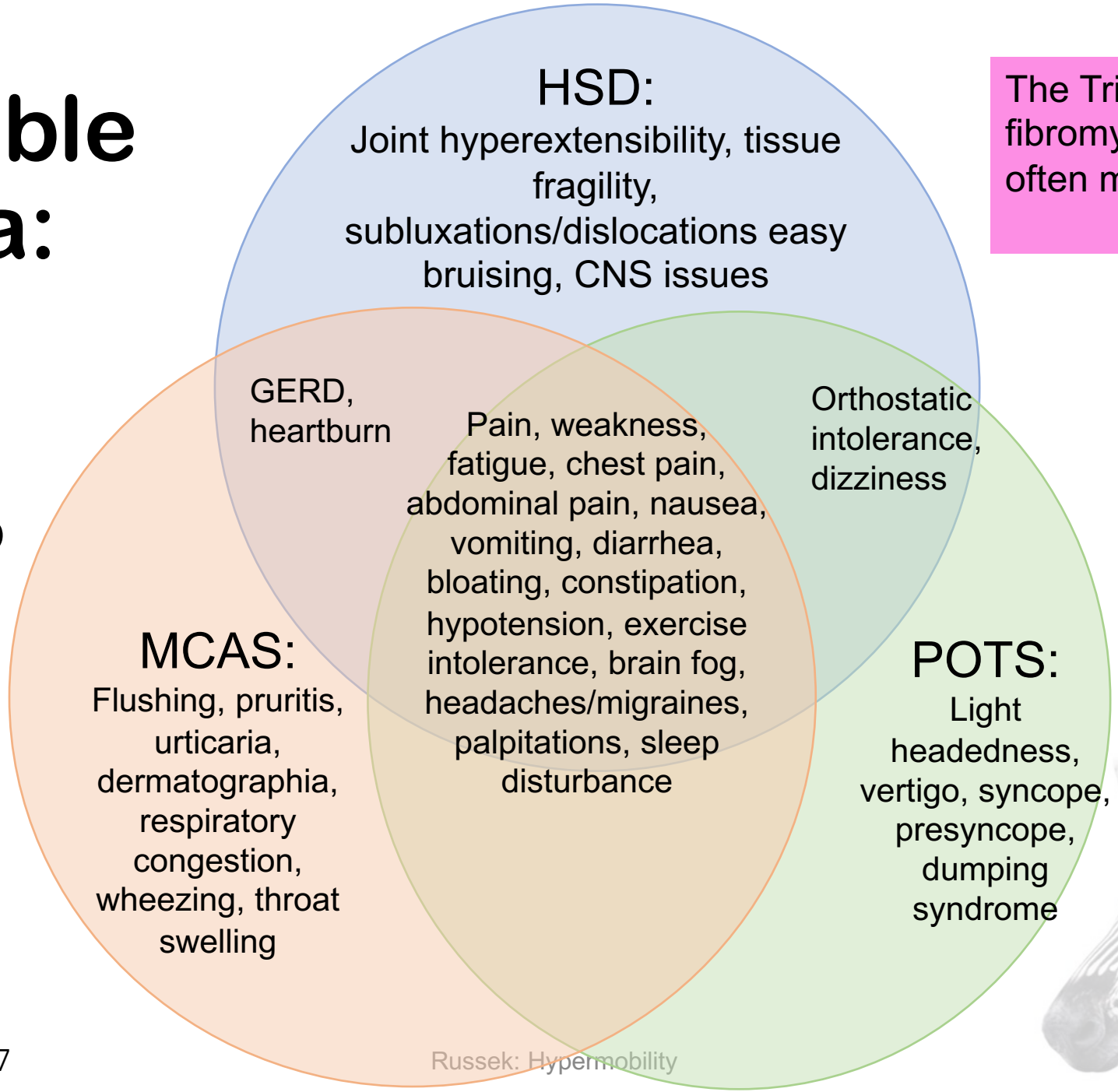


Presentation & Comorbidities

- **Musculoskeletal**: joint instability, joint pain, soft tissue injury, muscle spasm/trigger points, scoliosis, decreased bone density
- **Gastrointestinal**: GERD, IBS with diarrhea and/or constipation, painful bloating, gastroparesis, nausea/vomiting, prolapse, hernias
- **Autonomic nervous system**: dysautonomia (POTS or orthostatic intolerance), fatigue, dizziness, Raynaud's, exercise & heat intolerance
- **Neurological:** proprioceptive deficits, interoceptive deficits, clumsiness, headache, migraine, nerve compression, central sensitization, cervico-medullary syndrome, Chiari, Tarlov cysts, CSF leaks, tethered cord
- **Cardiopulmonary**: dysfunctional breathing, bleeding disorders, varicose veins, mitral valve prolapse, pelvic congestion
- **Mental health**: anxiety, panic disorder, depression, brain fog.
- **Neurodevelopmental:** developmental co-ordination disorder, motor delay, autistic spectrum disorder, attention deficit hyperactivity disorder
- **Immunologic:** MCAS, rashes, hives, chemical and food sensitivities, excessive inflammatory response, persistent fatigue, GI dysfunction, migraine
- **Dermatologic:** hyperextensible skin, fragile skin, poor wound healing, easy bruising
- **Urogenital:** incontinence, UTI, interstitial cystitis, dysmenorrhea, pelvic pain, vulvodynia, painful intercourse, prolapse
- **Multisystemic:** chronic fatigue, sleep disorder, brain fog

Russek, 2019; Simmons, 2022

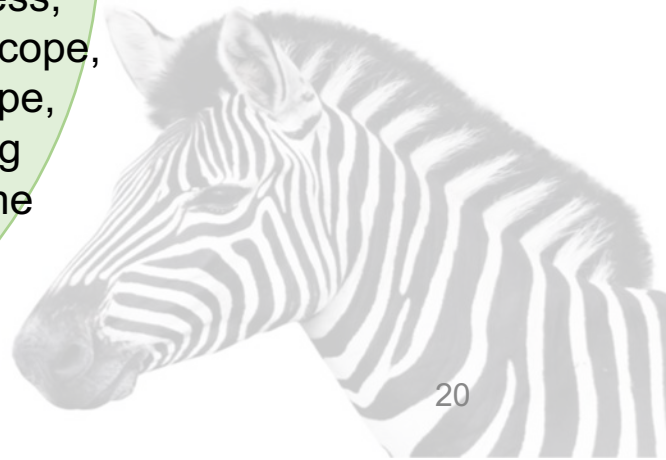
The Terrible Trifecta: HSD POTS MCAS



The Trifecta looks a lot like fibromyalgia, and is probably often misdiagnosed as FM (Russek, 2018)

Mast Cell Activation Syndrome/Disorder

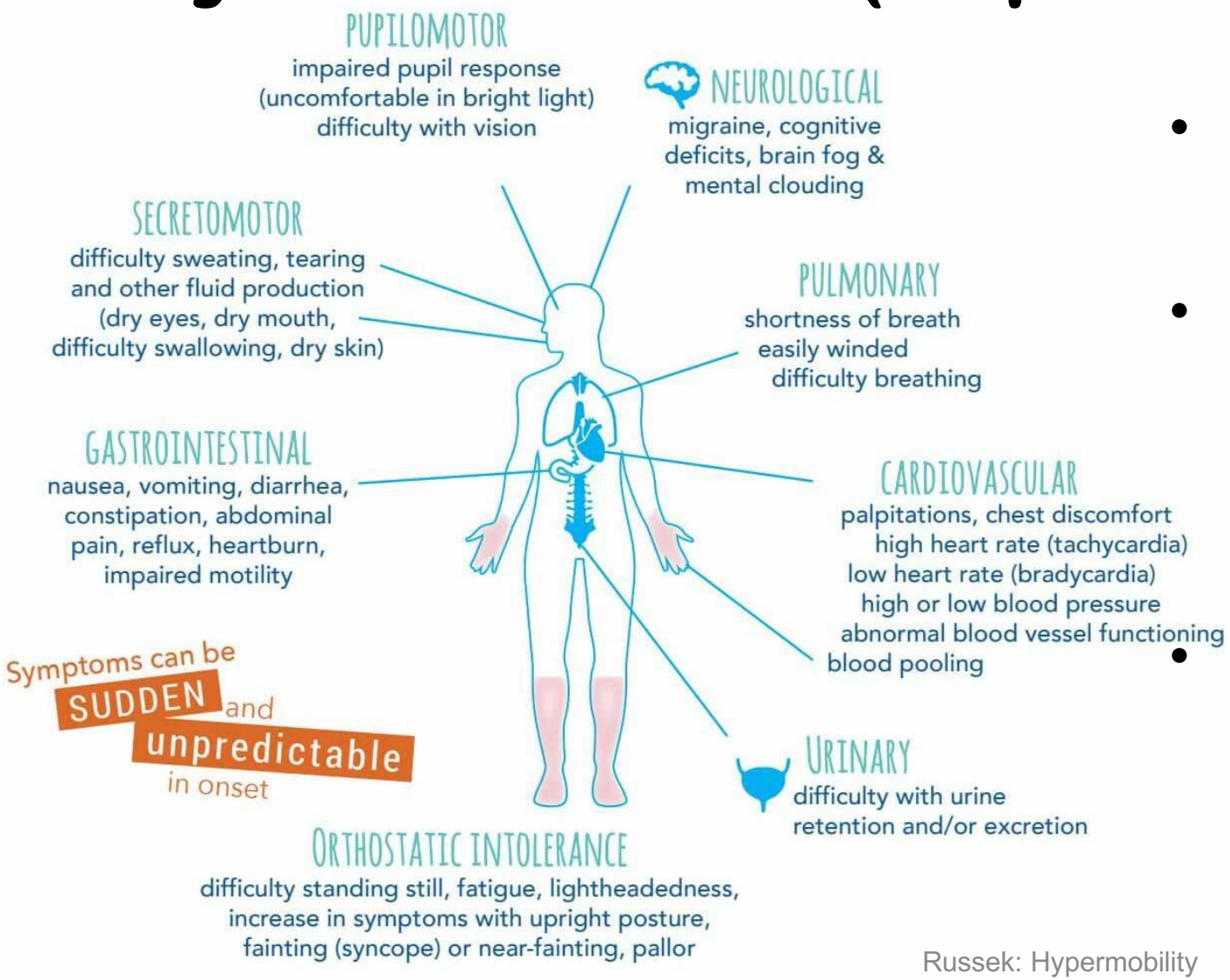
Postural Orthostatic Tachycardia Syndrome



Seneviratne, 2017; Hakim, 2017

Dysautonomia (especially POTS)

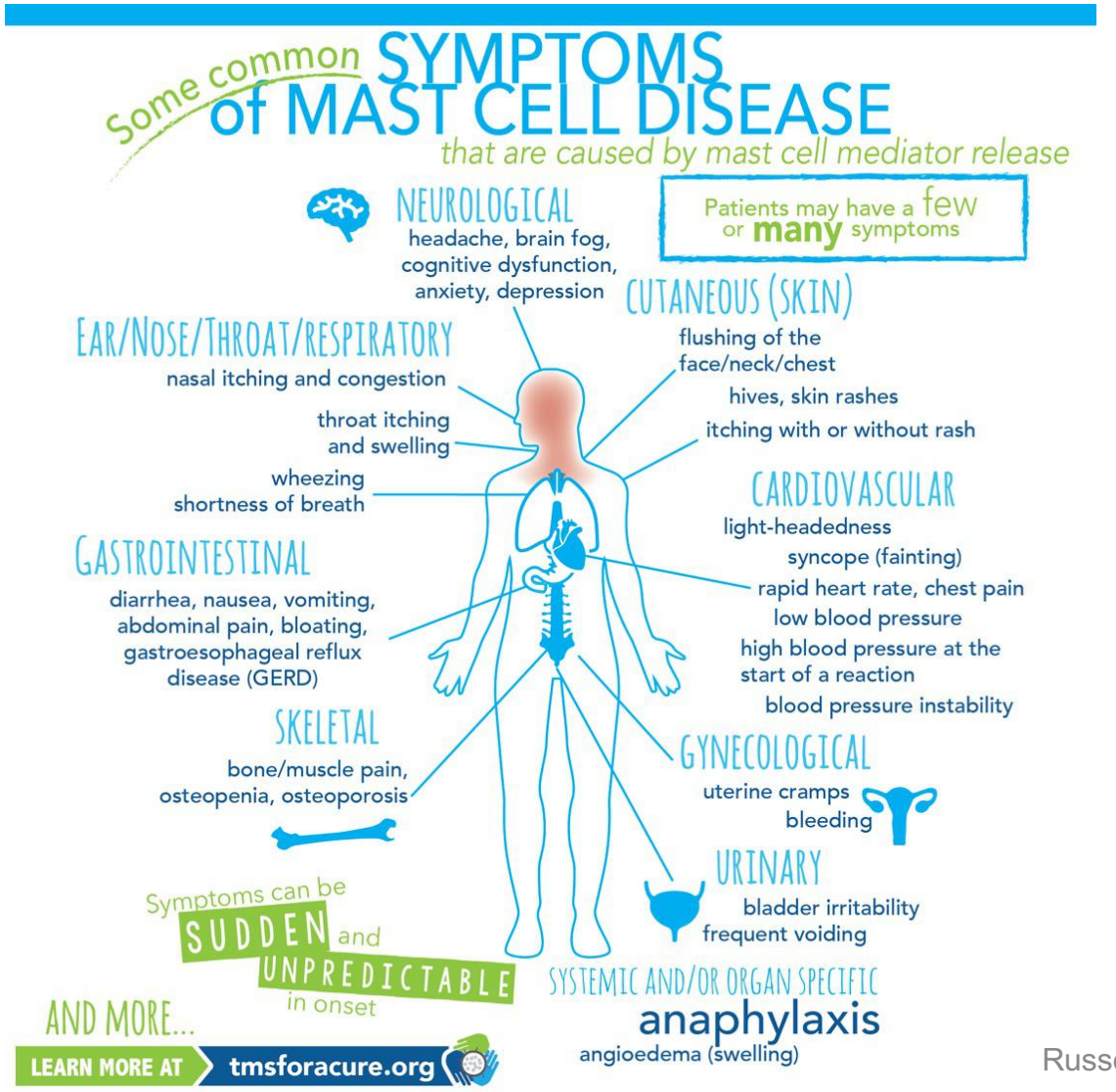
Overview of POTS



- HSD is a risk factor
  - Female:male ratio is 5:1
  - 62% of kids with POTS have HSD/hEDS
- Common triggers:
  - Puberty
  - Concussion
  - COVID
  - Deconditioning
  - Sleep disorders
- Different types of POTS: hypovolemic, neuropathic, MCAS, hyperadrenergic

Boris, 2021; Chen, 2020; Zhang, 2020; Bryarly, 2019

Mast Cell Activation Syndrome (MCAS)



Lab testing has very, very high false negative rates

- Possibly 17% of people in US
- Diagnosis

1. Allergy symptoms and other symptoms in two or more organ systems that keep coming back or are chronic
2. Higher than normal levels of tryptase, histamine, or prostaglandins in your blood
3. Getting better after using antihistamine medications or other drugs that block chemicals released by mast cells

- Can also use “Validated MCAS Questionnaire” (Afrin, 2014; Afrin, 2016)

Other Common Comorbidities

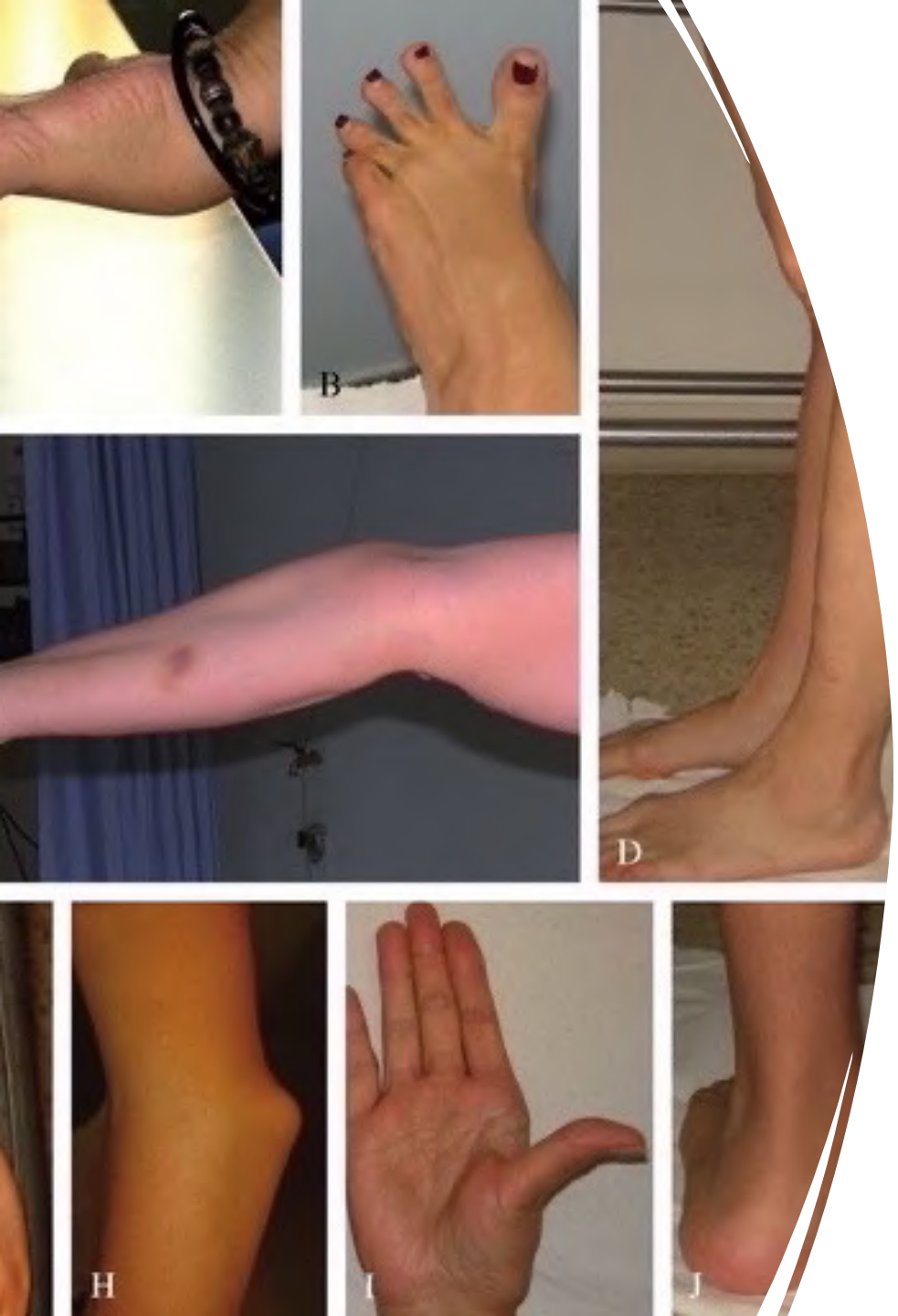
- Sensitive nervous system > pain sensitivity, dysautonomia
- Chronic fatigue and sleep problems
- Gastrointestinal problems
- Neurological issues
- Psychiatric/psychological problems, including neurodivergence and gender dysphoria

Hakim, 2017; Blajwajs, 2023; Lam, 2023; Henderson, 2017; Russek, 2022; Jones, 2022; Bulbena-Cabré, 2021

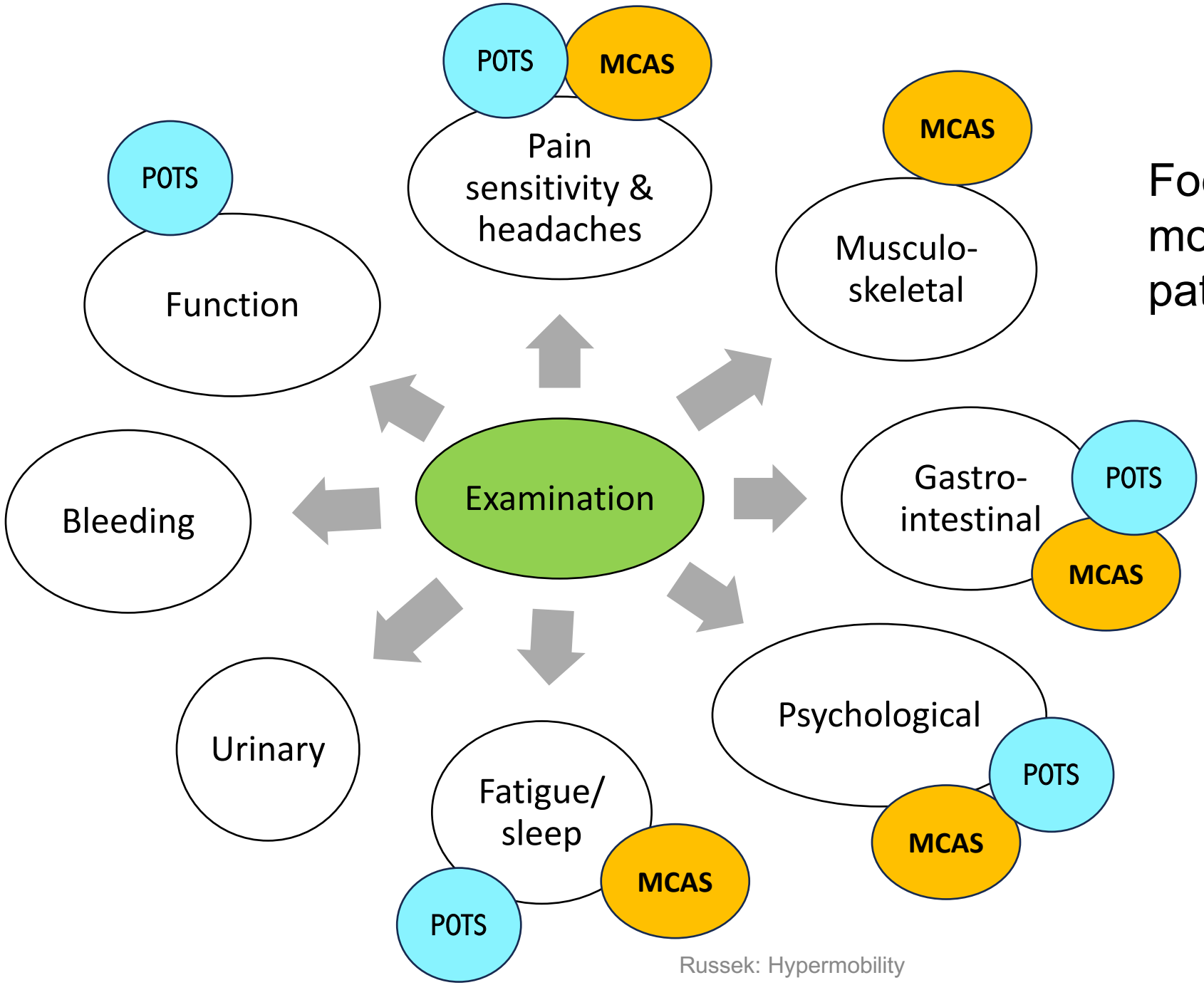
Bleeding Disorders

- 62% of EDS (mixed types) patients had bleeding disorders
  - Easy bruising
  - Hematomas
  - Menorrhagia
  - Oral bleeding, minor wounds, nose bleeds
  - Excessive surgical bleeding, post partum bleeding
  - GI bleeding, NSAID bleeding, hematuria, hemarthrosis
- International Society of Thrombosis and Haemostasis bleeding assessment tool (ISTH-BAT)
  - Scores >5 are abnormal in adult women
- 14% of patients with EDS reported menorrhagia that was life threatening or required surgery

Kumskova, 2022



Examination of Patients with HSD



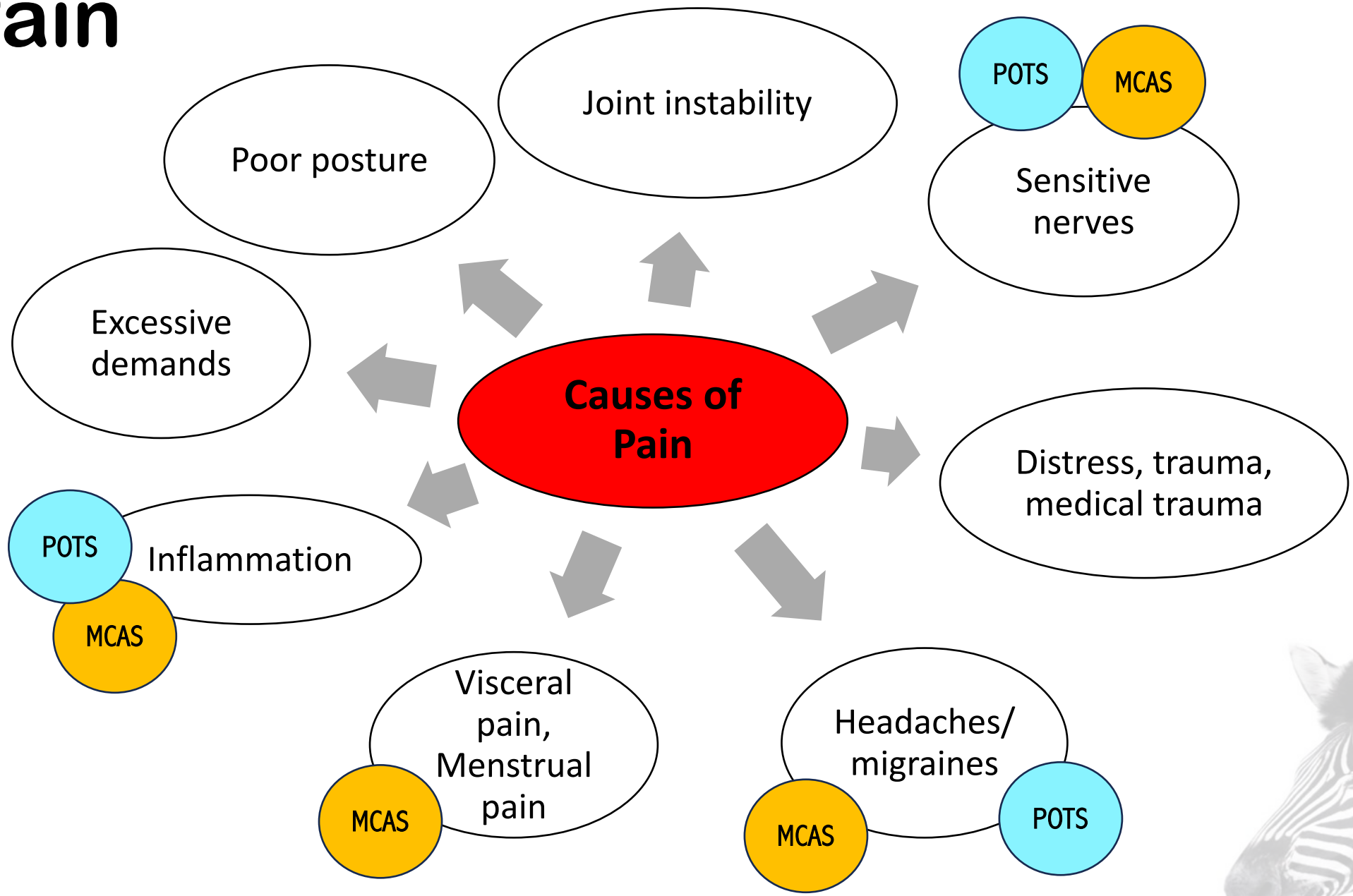
Focus on what is most important to the patient/family

Hypermobility, Laxity, and Instability

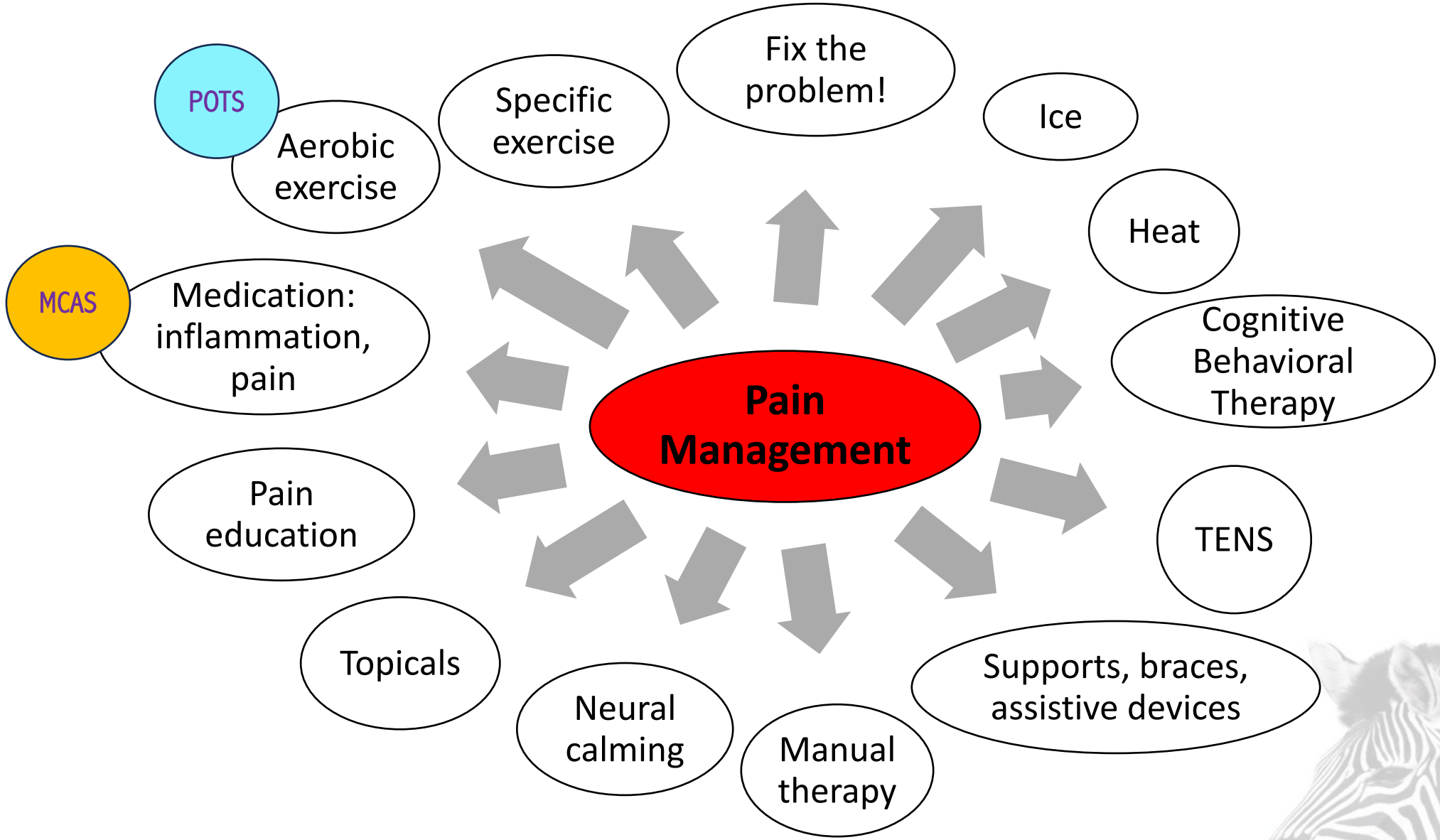
- Many people with GJH do not experience problems
- Definitions:
  - **Hypermobility:** increased physiological motion
  - **Laxity:** excessive accessory motion
  - **Instability:** lack of motor control, perceived sense of the joint slipping out of place, inability to control motion at joints
- Being hypermobile is not necessarily a problem
  - Being unstable often is a problem
  - **Physical examination should focus on motor control, not just hypermobility**

(Nicholson, 2022b)

Pain



Daylor, 2023a; Junghans-Rutelonis , 2019; Revivo, 2019; Nicholson, 2022



Junghans-Rutelonis , 2019; Revivo, 2019; Nicholson, 2022; Daylor, 2023b

(Physical Therapy) Management of HSD/hEDS

Russek: Hypermobility
https://www.physio.co.uk/what-we-treat/musculoskeletal/symptoms/hypermobility.php

Stratified Management of Hypermobility

SIMPLE/ EARLY

Episode of acute musculoskeletal injury, sprains, dislocation, subluxations, enthesopathies

SIMPLE/ EARLY

Ice, electrotherapy, tape, support, exercise (inc. proprioception & motor control), screen, educate; rehabilitate and prevent recurrence

INTERMEDIATE

Recurrent episodes, series of episodes at different sites, deconditioning, some central/ peripheral sensitization, mild to moderate systemic conditions

INTERMEDIATE

Standard PT approaches have temporary effect, no effect or exacerbate.
Need to address causative factors.
More focus on functional restoration

COMPLEX/ LONG TERM

Chronic, longstanding, severe, unremitting pain with profound deconditioning, comorbidities, CNS involvement, and disability

COMPLEX/ LONG TERM

Multi-disciplinary management program using functional and cognitive behavioral approaches and assistive devices

Modified from model proposed by Jane Simmonds, DProf, MCSP, MACP, SFHEA, Univ. Coll. London


MUSIC FES
TICKET PASS
Hypermobility
Self-Care
Toolbox

Patient Education

Educate and empower the patient/family

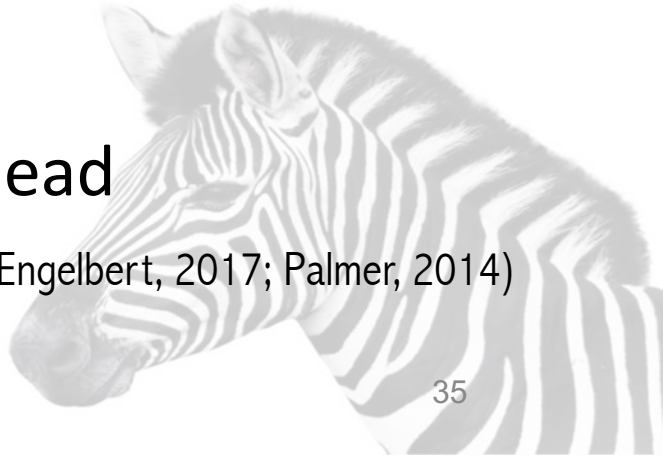
- Posture, joint protection, body mechanics, ergonomics
  - Orthotics, braces, & splints if/when needed
- Self-care, injury prevention, pain self-management
  - Problem-solving, braces, heat/ice, TENS, topicals, physiological quieting
- Appropriate exercise/activity
- Sleep hygiene & positioning; fatigue management, breathing
- POTS self-management and exercise progression
- Advice regarding impact of diet on POTS and MCAD
- Other issues: GI dysfunction, MCAD, incontinence, etc.
- Psychological & social wellness and coping

(Engelbert, 2017; Chopra, 2017; Russek, 2019; Simmonds, 2022)

Exercise Prescription: What's Different

- Teach body awareness
- Start movements from good alignment
- Make sure every movement is done correctly, with control
- Emphasize motor control & coordination
- "Start Low, Go Slow"
- POTS-specific exercise
  - Horizontal positioning, pumping blood to the heart/head

(Engelbert, 2017; Palmer, 2014)

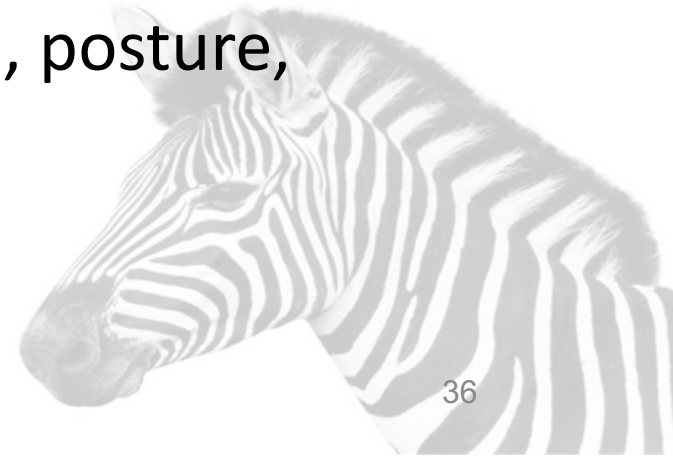


Exercise: Do No Harm!

- Pts with HSD often have negative past experiences with PT

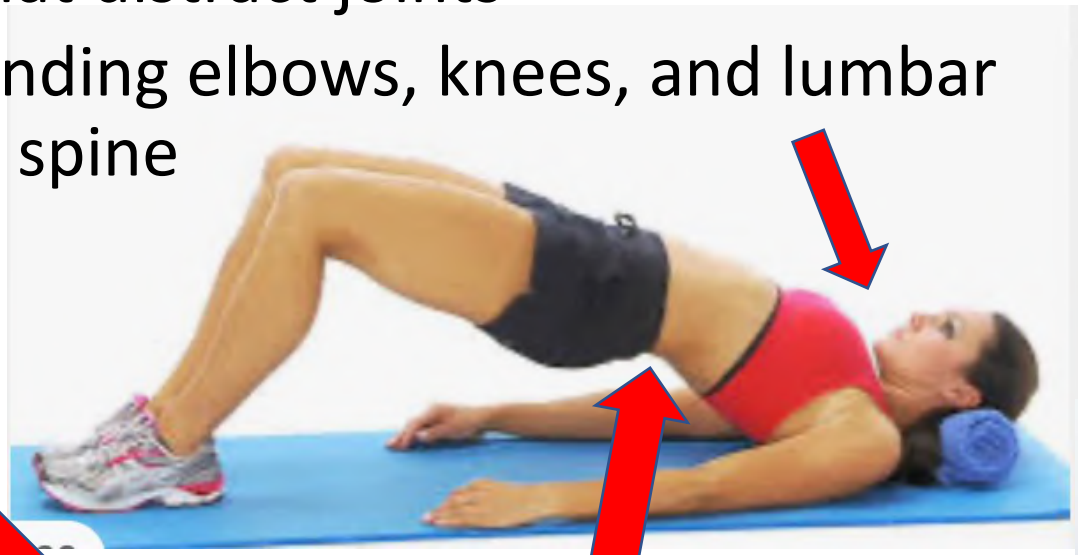
(Bovet, 2016; Simmonds, 2019)

- Avoid mechanical pain due to excessive tissue stress
  - With poor proprioception & motor control, patients do exercises incorrectly
  - Tissues often start weaker than in deconditioned non-HSD patients
  - Tensile strength changes with activity, age, inflammatory state, menses
  - Slow progression to allow for slower histological changes: “Start low, go slow”
- Pay attention to other joints, nerves, excessive tension, posture, orthostatic position



Don’t Stress Other Joints!

- Be careful with:
  - Gripping weights or resistance bands
  - Weight on extended wrists
  - Weights that distract joints
  - Hyperextending elbows, knees, and lumbar or cervical spine



Manual Therapy: Do No Harm!

- Gentle soft tissue manual therapy is often appropriate and helpful
- Very light/controlled joint mobilization may be appropriate and helpful
- But…
  - Do not over-mobilize! Especially the neck!
  - Do not manipulate unless you really understand HSD and know what you are doing
- Treat cautiously, as you would a pregnant or post-partum patient
- If the patient gets worse every time you touch them – work on central sensitization

Russek, 2019; Simmonds, 2022

Medical Management of HSD

Multidisciplinary Management

- Multidisciplinary management is ideal, but not often available
  - Cognitive behavioral approaches
  - Fear-avoidance education/management, relaxation training
  - Exercise (proprioception, core stability, strength, aerobic and hydrotherapy) with home exercise program
  - Education of parents

(Revivo, 2019; Nicholson, 2022; Van Meulenbroek, 2020)

Medications

- Little definitive research evidence for medications
- Limit NSAIDs to use for true inflammation
  - NSAIDs may slow tissue healing, aggravate GI & MCAS symptoms
- Tricyclics, anti-seizure, SNRI meds for neuropathic pain
- Topical analgesics and anti-inflammatory medications
- Acetaminophen
- Low dose naltrexone (LDN) seems to be helpful (Daylor, 2023b)
  - It is a mast cell stabilizer in the brain
- Cautions:
  - Opiates for short term use only
  - Muscle relaxers and Botox may aggravate instability

Chopra, 2017; Tinkle, 2017

Surgical Precautions

- Orthopedic surgery (e.g. rotator cuff or ACL repair) is only effective 34% of the time in patients with hEDS; this is 50% as often as non-hypermobile pts.
  - Therefore, it is important that conservative management (with an hEDS knowledgeable provider) be fully explored before resorting to surgery.
- Orthopedic, spinal, and GI surgeries in hEDS are more likely to have complications.
- Surgeons should take hEDS into account when planning surgery.
- Vascular EDS has much higher risk of complications. Typical complications include arterial perforation or tears, bowel perforation or tears, recurrent hernias or increased bleeding.

Rombaut, 2011; Yonko, 2021; Homere, 2020; Chi, 2023; Kulas Søborg, 2017; Burcharth, 2012)

Handout for patients to share with surgeons and anesthesiologists

Surgical and Anesthetic Precautions:

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

The main feature of HSD/hEDS is ***laxity of connective tissue***, including skin, ligaments, blood vessels and nerves. This can cause ***potentially fatal problems*** for these patients when unconscious, and/or having surgery.

BEWARE THE UNCONSCIOUS PATIENT!	***In the unconscious HSD/hEDS patient, a little force may displace any joint.*** ***Treat unconscious HSD/hEDS patients with full spinal stabilization*** as if they have a spinal injury. If you don't, then you may cause one! ***Use NO traction on limbs.*** ***Use extreme care with the chest:*** the ribs easily dislocate front or back.
BEWARE THE LARYNGOSCOPE!	***Use extreme gentleness, with minimal, if any, anterior traction on the laryngoscope. The jaw may dislocate*** on one or both sides. Manipulation of the laryngoscope can also damage the cricopharyngeal muscle and its nerves, the esophagus and the cervical spine.
BEWARE NECK MOTION!	***Keep patient's head in neutral position throughout.*** Movement of unstable subcranial joints may cause spinal cord damage during incautious patient handling during anesthesia. Consider a soft collar.
LOCAL ANESTHESIA	HSD/hEDS patients are often resistant to local anesthetics: ***they may need much larger doses than other patients, and these may need to be repeated during a procedure.*** Ropivacaine may work better than lidocaine or bupivacaine.
SURGICAL TECHNIQUE	Use minimal force when cutting or moving tissues. Cut blood vessels may contract poorly: **electrocautery is appropriate**. Tissue healing may be prolonged. **Close layers without tension using slowly-absorbable or non-absorbable sutures**. Reinforce them with steri-strips etc. as appropriate.
BLEEDING & BRUISING	These are due to fragile small blood vessels, not an intrinsic blood disorder, so **elaborate clotting tests are rarely indicated**. Be alert for slowly-accumulating, deep hematomas.
POST-OPERATIVE PAIN	Painful polyneuropathy is common in HSD/hEDS. Post-operative pain may be more severe and more prolonged than normal. **Be liberal with analgesics**.
CARDIO-VASCULAR INSTABILITY	HSD/hEDS patients are subject to hypotension and/or tachycardia due to low blood volume, and defective venoconstriction. ***Liberal IV fluids usually can address this.***
GI DYSFUNCTION	Poor GI motility is routine in HSD/hEDS, worse after surgery. **Minimize constipating agents, and use laxatives pre-emptively**. Consider pro-motility agents.
CARDIAC RESCUCITATION	Some HSD/hEDS patients have **loose costosternal joints,** sometimes palpably displaced. For them, **chest compressions could in theory be very dangerous, causing rib detachments, a flail chest and even heart or lung puncture** by freed anterior ribs. There is no consensus on whether cardiac resuscitation should include chest compressions in patients with clear evidence of rib displacements.

Alan Spanos, MD, (919) 967-2927, alan.spanos@yahoo.com.

This document is online at *www.AlanSpanosMD.com*. It was updated March 2019.

For more information, see the Ehlers Danlos Society at ehlers-danlos.com.

- This handout should be at (but isn't at the moment):
- https://edswellness.org/wp-content/uploads/2019/07/7-Surgical-Anesthetic-Precautions.pdf
- Chronic Pain Partners has an excellent patient handout at: https://www.chronicpainpartners.com/wp-content/uploads/2023/02/surgery-prep-meeting-your-eds-hospital-stay-needs.pdf
- I have a more technical handout for providers:
- https://webspace.clarkson.edu/~lrussek/docs/hypermobility/SurgeryHSD.pdf

Summary

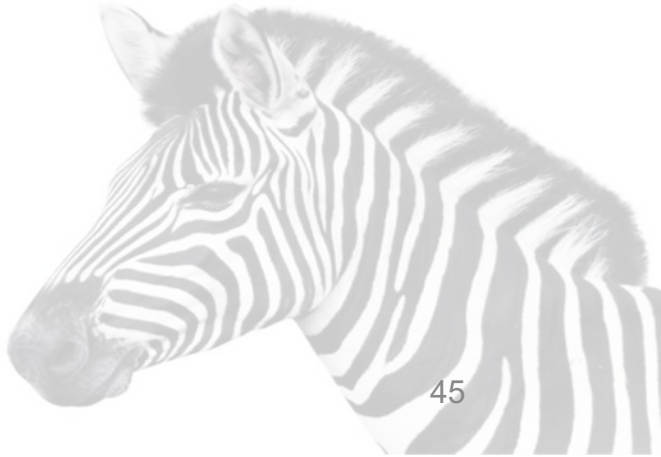
- Look for HSD when things don't make sense
  - ***"When you can't connect the issues, think connective tissues"***
- Their bleeding issues may be due to different reasons
- HSD impairments may affect all systems in the body, not just joints
- Each patient with HSD is an individual – there is no standard Rx for everyone.
- This is a chronic condition and patients need to learn self-management strategies
- LISTEN to patients and BELIEVE what they say!
  - Medical traumatization is real (Halverson, 2023)

Handouts Available

- https://webspace.clarkson.edu/~lrussek/research.html

Available on my website

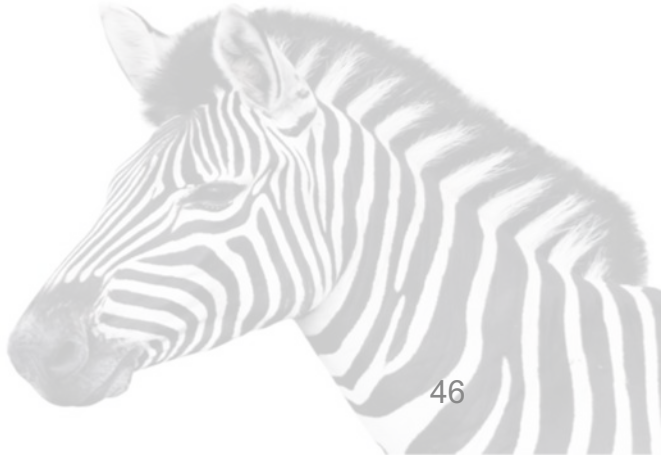
- General Information
  - Overview of Hypermobility Spectrum Disorder
  - HSD in children and adolescents
  - Checklist of physical therapy treatment approaches for HSD/hEDS.
  - Surgical precautions for people with HSD/hEDS.
  - List of HSD and POTS disability/accommodations resources.
- Self-Care Strategies
- Pain Management
- Exercise
- Managing POTS and MCAS



Hypermobility Lecture Series Schedule

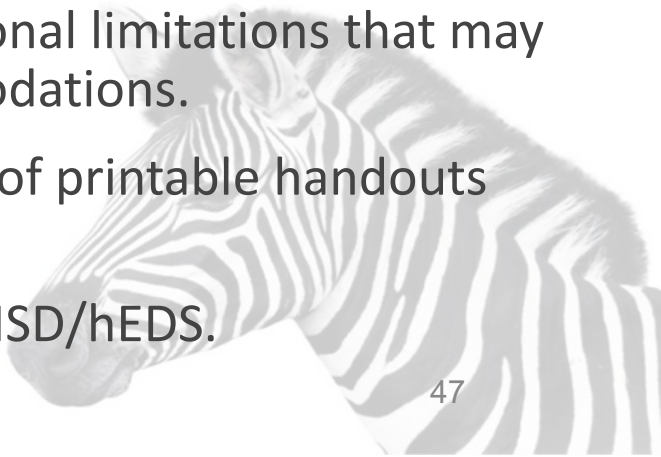
https://webspace.clarkson.edu/~lrussek/hsd.html

- HSD 101: Basics of HSD/hEDS and self-care
- HSD 102: POTS and POTS self-care, basics of MCAS
- HSD 103: Pain management in HSD/hEDS
- HSD 104: Safe exercise selection and progression with HSD/hEDS
- HSD 105: Posture and joint protection
- HSD 106: Gut issues in HSD/hEDS, POTS, MCAS
- HSD 107: Fatigue in HSD/hEDS and POTS
- HSD 108: Headaches, migraines, & TMJ pain associated with HSD, POTS and MCAS
- HSD 109: Breathing disorders in HSD
- HSD 110: Lumbar instability
- HSD 111: Conservative management of cervical instability
- HSD 112: The vagus nerve
- HSD 113: The role of fascia
- HSD 114: Hospitalization with HSD, POTS, MCAD



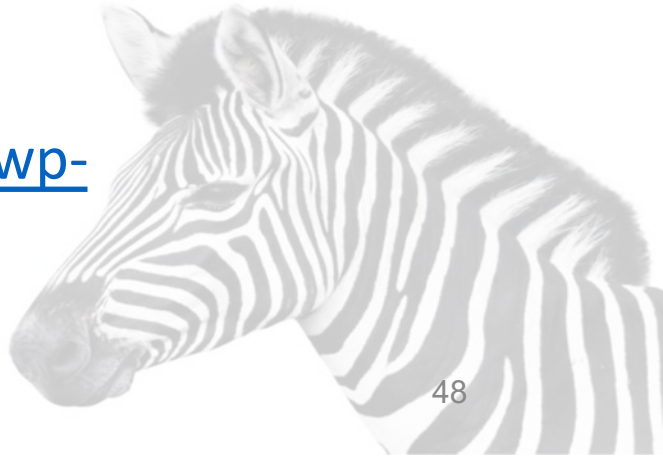
Resources for Hypermobility

- The Ehlers-Danlos Society, Has many excellent resources for all forms of EDS. It also has a list of EDS healthcare providers, and support groups in the US. Based in the US.
  - **Conferences and on-line workshops: https://www.ehlers-danlos.com/upcoming-events/**
  - **On-line training programs: https://www.ehlers-danlos.com/echo/**
- The Ehlers-Danlos Syndrome Toolkit. Information about diagnosis, management and available resources intended for primary care and other health providers.
- "Hypermobility Syndromes" booklet for patients. 27 pages discussing all aspects of hypermobility.
- Hypermobility Syndromes Association (HMSA).
- NASEM Report on HSD/EDS and disability. The National Academy of Science, Engineering and Medicine report that I helped create is a comprehensive overview of functional limitations that may lead to disability people with HSD/EDS. It also lists many potential accommodations.
- School Toolkit for HSD/EDS. Great ideas to help kids succeed in school. Lots of printable handouts to bring to school to facilitate communication and understanding
- ***Taming the Zebra***, book by P. Stott and H. Purdin, about systemic issues in HSD/hEDS.



Resources for POTS

- **Good primary care overview:** Seeley MC, Lau DH, Gallagher C. Postural Orthostatic Tachycardia Syndrome: Diagnosis and Management Guide for Nurses. *Nursing: Research and Reviews*. 2023;13:41-49.
- **Thorough review:** Vernino S, Bourne KM, Stiles LE, et al. Postural orthostatic tachycardia syndrome (POTS): State of the science and clinical care from a 2019 National Institutes of Health Expert Consensus Meeting - Part 1. *Auton Neurosci*. Nov 2021;235:102828.
- **Dysautonomia International** has great patient info: http://www.dysautonomiainternational.org
- **NASA Lean Test instructions:** https://batemanhornecenter.org/wp-content/uploads/2016/09/NASA-Lean-Test-Instructions-1.pdf



Resources for MCAS

- Afrin LB, Butterfield JH, Raithel M, Molderings GJ. Often seen, rarely recognized: mast cell activation disease--a guide to diagnosis and therapeutic options. *Ann Med*. 2016;48(3):190-201. *Good overview.*
- Afrin L, Molderings GJ. A concise, practical guide to diagnostic assessment for mast cell activation disease. *World J Hematol*. 2014;3(1):1-17. *Includes validated questionnaire to identify MCAS signs and symptoms*
- Information for both patients and health care providers:
  - https://www.tmsforacure.org
  - https://www.mastzellaktivierung.info/en/introduction.html
- Summary for health care providers: https://tmsforacure.org/wp-content/uploads/Mast_Cell_Disease_Primer_Slides_TMS_09.20.2021_Final.pdf
- Medication recommendations, starting with OTC H1 and H2 inhibitors
  - https://tmsforacure.org/treatments/medications-treat-mast-cell-diseases/

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Thank You!