Ehlers-Danlos Syndrome and the Hypermobile Patient: Understanding and Treating Musculoskeletal, Systemic, and Biopsychosocial Effects

Heather A Purdin, MS, PT, CMPT
Good Health Physical Therapy & Wellness
Portland, OR

Heather Purdin, M.S., P.T., C.M.P.T.

- Graduate of Duke University 1995 with BS in BioPsychoSocial Psychology, Health Psych, and Neuropsych
- Honors Thesis on Pain Behaviors in Children
- Master’s Degree in Physical Therapy Duke University 1997
- Special Initiatives award for “enhancing awareness of cultural diversity in our program and profession”
- Private practice owner with mission to provide holistic care. Large portion of patients have EDS and chronic pain
- Personal experience living with HEDS
- President of the Oregon Area Ehlers-Danlos Support Group
- Manual Therapy Certificate through NAIOMT
- Voted among “Portland's Top Doctors and Nurses 2015” Portland Monthly Magazine
- Attended Ehlers-Danlos National Foundation conferences and Doctor’s Conference last 3 years
- Personal interaction with Portland, OR area EDS experts
Learning Objectives

• After completing this session, participants will be able to:
  • Examine prevalence and common presentation of EDS
  • Contrast EDS with similar conditions, performing differential diagnosis using evidence-based tests
  • Formulate appropriate interventions including lifestyle, physical, functional and activity modification
  • Formulate specific exercises guidelines that will not flare patients with EDS

Outline of Content

• Definition and diagnosis of EDS & subtypes
• Common comorbidities
• Biopsychosocial approach
• Interview & physical exam
• Evaluation considerations
• Management
  • Patient education and self-management training
  • Therapeutic exercise/neuromuscular re-education
  • Manual therapy
  • Pain management
Overview of EDS

- Inherited connective tissue disorders affecting collagen
- All 6 main types have hypermobile joints and skin involvement
- Many have chronic, widespread pain and multi-system involvement

Types of EDS: Primary Differences

- **Hypermobility** (III): loose joints, joint pain. Most common.
- **Classical** (I & II): velvety, stretchy, fragile skin. Common.
- **Vascular** (IV): possible arterial/organ rupture. Most serious.
- **Kyphoscoliosis**: joint laxity, muscle hypotonia, developmental delay. Severe functional loss over time.
- **Arthrochalasia** (VII): congenital hip dislocation, lax joints
- **Dermatosparaxis**: Severe skin fragility & bruising
Hypermobility Spectrum Disorders (HSD)

- Symptoms tend to change over time
- Rate of change varies by patient
- Hypermobility $\rightarrow$ Pain $\rightarrow$ Stiffness

- Generalized Joint Hypermobility (GJH) – not necessarily symptomatic
- Localized Hypermobility Spectrum Disorder (L-HSD) - symptoms
- Peripheral Hypermobility Spectrum Disorder (P-HSD) - symptoms
- Generalized joint Hypermobility Spectrum Disorder (G-HSD) – symptoms
- Historical Hypermobility Spectrum Disorder (H-HSD) – no longer hypermobile
- Hypermobile Ehlers-Danlos Syndrome (hEDS) – specific criteria

Prevalence of EDS

- Overall prevalence in US: 1-3%
- 80-90% of all EDS is h-EDS
- hEDS in musculoskeletal healthcare: 30-55%
- hEDS is probably the most common systemic inherited connective tissue disorder in humans
- Affects ~10 million people in the U.S.

1. Connelley et al, 2015; Clark & Simmonds, 2011
2. Tinkle, et al, 2017
Common Signs & Symptoms

- **Musculoskeletal**: joint hypermobility, subluxations/dislocations, sprains, muscle spasm, TMD, flat feet, finger deformities, arthralgia, myalgia, osteoporosis, fractures, pain
- **Integumentary**: soft stretchy skin, easy bruising, atrophic scarring, poor wound healing, frequent hernias
- **Cardiovascular**: dysautonomia, postural orthostatic tachycardia syndrome (POTS), *mitral valve prolapse*
- **Other**: prolapse, gastritis, IBS, incontinence, developmental delay, poor coordination, anxiety, *organ rupture, aneurisms*
  - Castori et al, 2011; Columbi et al, 2015; Tinkle et al, 2017

hEDS Transformation of Symptoms Throughout the Life Span

- **Hypermobile phase**
  - Hypermobile joints
  - Clumsiness/motor delay
  - Constipation/diarrhea
  - Abdominal hernias
- **Pain phase**
  - Chronic fatigue
  - Unrefreshing sleep
  - Chronic back pain
  - Chronic muscle pain/cramps
  - Strains, sprains
  - Dislocations
- **Pain + phase**
  - Memory/cognitive problems
  - Gastric reflux
  - Recurrent abdominal pain
  - Paresthesias
  - Tachycardia
  - Incontinence/UTI
- **Stiffness phase**
  - Tendonosis/tendon rupture
  - Chronic gastritis
  - Stiffness
    - Castori et al, 2011
    - Tinkle et al, 2017
Major Comorbidities

- Dysautonomia: POTS, thermoregulation, gut, sexual dysfunction
- Mast Cell Activation Disorder: systemic inflammation
- Gastrointestinal disorders: GERD, IBS, malabsorption syndrome
- Chronic pain: fibromyalgia, myofascial pain, OA
  - Tinkle et al, 2017

Dysautonomia

- Postural orthostatic tachycardia syndrome (POTS)
- Orthostatic hypotension
- Secondary S&S: fatigue, dizziness, fainting, syncope, memory and concentration problems
- May look like anxiety or panic disorder
  - Tinkle et al, 2017
POTS Criteria

- Heart rate increases ≥30 bpm from supine to standing (10 min)
- Symptoms worsen with standing and improved with recumbence
- Symptoms last ≥ 6 mo
- Absence of other overt cause of orthostatic symptoms or tachycardia (e.g., active bleeding, acute dehydration, medications) Raj et al, 2013

Mast Cell Activation Disorder

- Flushing, pruritis/itching, hypotension, asthma, diarrhea, abdominal bloating, cramping
- Food sensitivities
  - Tinkle et al, 2017
We must treat this Vicious Cycle Holistically!

Gastrointestinal Disorders

• Present in 33-75% of those with hEDS
• Gastroesophageal reflux, heartburn
• Bloating, abdominal pain
• Irritable bowel, constipation/gastroparesis, diarrhea
• Abdominal hernias, rectal prolapse
• Dysphagia?
  • Tinkle et al, 2017
Psychosocial Issues

• Depression, anxiety, affective disorder, low self-confidence, negative thinking, hopelessness
• Chronic pain
• Prior life trauma (physical, emotional, sexual)
• Low quality of life
  • Tinkle et al, 2017

Diagnostic Criteria

Hypermobile Ehlers-Danlos Syndrome
Ehlers-Danlos Syndrome, Hypermobile Type, or Type III
Joint Hypermobility Syndrome
An Evolving Diagnosis

- Villafranche was used mostly by geneticists for children: hEDS
- Brighton was used mostly by rheumatologists for adults: JHS
- New criteria...

2017 International Classification of the Ehlers-Danlos Syndromes

3 Criteria to meet hEDS (must meet all 1-3)
   1. Generalized joint hypermobility (GJH): Beighton score = or > 5/9
   2. Features of a Connective Tissue Disorder (must have 2)
      A) Systemic Manifestations (must have 5 or more)
      B) Family History
      C) Musculoskeletal complications (must have 1 or more)
   3. Exclusion criteria (can’t have something else that explains sx)
Beighton

Beighton score
• 2- Touch thumb to forearm
• 2- Bend pinky back >90°
• 2- Elbow hyperextension >10°
• 2- Knee hyperextension >10°
• 1- Touch palms to floor, knees straight
≥5/9 puberty to 50 y.o. (Brighton used ≥4/9)
≥6/9 Kids
≥4/9 over 50 y.o.

Juul-Kristensen, 2017

Beighton

• does have good reproducibility
• Valid with adult > 16 y.o.
• has some standards but not as objective as it needs to be (ie: do we need to use a goniometer or just eyeball?)
• adjustments for age - higher for children, lower for older adults
• Currently no adjustment allowed for injury/trauma or for other joints (shoulder, hip, back)
• past history of laxity (5 point questionnaire) – if 2 or more questions + get 1 point on Beighton
5 point questionnaire (if > or = 2 yes, get 1 more point on Beighton)

• Can you now or could you ever place your hands flat on the flor 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?”

2. Features of a Heritable Connective Tissue Disorder (must have 2 or more of the 3 subcategories A, B, C)

A. Systemic Manifestations (must have 5 or more)
- unusually soft or velvety skin
- mild skin hyperextensibility (1.5 cm back of hand)
- unexplained straiae (stretch marks)
- hernias: recurrent or multiple abdominal (not hiatal hernia)
- atrophic scarring involving at least 2 sites
- pelvic floor, rectal, and/or uterine prolapse
- Dental crowding AND high/narrow palate
- bilateral piezogenic papules of heel
- Arachnodactyly with bilat Steinberg signs (wrists) and/or Walker signs (thumbs)
- Arm span to height ratio >/= 1.05
- Mitral valve prolapse mild or greater based on echocardiographic criteria
- Aortic root dilatation with Z-score > +2
Piezogenic Papules of the heel

2. Features of a Heritable Connective Tissue Disorder continued

B. Family History of a 1st degree relative independently meeting diagnostic criteria
C. Musculoskeletal complications (must have 1)
1) pain in ≥ or equal to 2 limbs, recurring daily for at least 3 months
2) chronic widespread pain for ≥ or equal to 3 months
3) recurrent joint dislocations or medical joint instability
   o 3 or more atraumatic dislocations of same joint OR 2 or more dislocations of 2 different joints at different times
   o Medical confirmation of atraumatic joint instability in ≥ 2 sites

3. Absence of Exclusion Criteria (don’t have something else)
   o Other heritable or acquired connective tissue disorder (Lupus or RA ruled out usually by Rheumatology)
   o Neuromuscular Disorders which may cause joint hypermobility by means of hypotonia OR connective tissue laxity (ex: mitochondrial disorder causing hypotonia ruled out usu. by Geneticist)
   o Unusual Skin Fragility – should prompt consideration of other types of EDS (rule out by Geneticist)
Evaluation

<table>
<thead>
<tr>
<th>Pain Source</th>
<th>Quality</th>
<th>Medications/Referral</th>
<th>Exercise</th>
<th>Other Intervention</th>
</tr>
</thead>
<tbody>
<tr>
<td>Neuropsych</td>
<td>Whole body pain, difficult to localize</td>
<td>SNRI's, triptylines, LDN &lt;br&gt; &lt;em&gt;Primary MD, Psych&lt;/em&gt;</td>
<td>Cardio Mindful Movement External Focus</td>
<td>Meditation Breathing</td>
</tr>
<tr>
<td>Inflammatory</td>
<td>Burning in broad area not dermatomal, bruised</td>
<td>Anti-inflammatories, Allergy meds, MCAD &lt;br&gt; &lt;em&gt;Primary MD, Allergy&lt;/em&gt;</td>
<td>Pool Proprioceptive Input Mindful Movement</td>
<td>Modalities, Tool assisted Scraping, Dietry guidance</td>
</tr>
<tr>
<td>Mechanical</td>
<td>localized, occurs with certain motions</td>
<td>NSAIDs &lt;br&gt; &lt;em&gt;Primary MD&lt;/em&gt;</td>
<td>Strength/Mobility balance t/o kinetic chain Proprioception</td>
<td>Bracing, taping Ergonomics training Setting muscles</td>
</tr>
<tr>
<td>Nerve</td>
<td>Burning in a line, searing</td>
<td>Gabapentin, Lyrica, SNRI's, LDN, Triptylines &lt;br&gt; &lt;em&gt;Primary MD, Neuro&lt;/em&gt;</td>
<td>Nerve flossing Mindful Movement Cardio External Focus</td>
<td>Posture training to address entrapped nerves, positions of slack</td>
</tr>
</tbody>
</table>

<em>Castori et al 2012</em>
Sources of Pain

- Don’t just look for symptomatic tissue – find the cause of tissue symptoms
- Chronic pain - consider
  - Childhood trauma
  - Stress
  - Psychosocial factors

Patient Management
Biopsychosocial model:

• Bio/Mechanics = PT but we spill into psycho and social aspects
• Discharge goal is to teach neutral joint mechanics at all joints for all activities.
• Where to start? Is it the loosest or the tightest link that is the biggest problem? Beware of the tight one!
• Start at the biggest complaint & relieve the most pain quickly to get buy in and if this isn’t the tightest or loosest link then go there next.

Key Components of PT Program

• Patient education
• Assistive devices
• Exercise
• Manual therapy
• Pain management
  • Engelbert et al, Clinical Guidelines, 2017
Patient Education

- Body mechanics/ergonomics
  - Postural and ergonomic hygiene especially during sleep, at school, and at workplace
- Appropriate exercise/activity and pacing
- POTS self-management
- Sleep hygiene (“Sleep is King”)
- Diet and fluid management (“The Gut is Queen”)
- Psychological & social wellness
  - Relaxation

Body Mechanics & Ergonomics

- Postural and ergonomic hygiene at school/work
- Sit to stand?
- Sleeping posture, surface, support
- Engaging muscles before moving (compensating with cognitive activation)
- Assistive devices (e.g., pens, tools, etc.)
Most Common differences biomechanically

- Shoulder setting
  - Cue up instead of down
  - Ball back in socket, not just scapulae – cue upside down “G”
- Hip posterior seating
  - No hip flexor use without glut use or
  - Alter position of exercise to assist stability/posterior seating such as in long sitting
- Ankle protection
  - Bed sheets can sublux talus anteriorly
  - Sitting on ankles
- Patellofemoral instability
  - Typical issues Osgood Schlatter’s, tracking issues
  - Craniocervical instability – always screen if treating neck
    - Often just long hold into mobilization direction is enough to normalize
- Rib setting
  - ½ inhale before lifting
  - If depressed ribs, inhale with activity
  - If elevated ribs, exhale with activity

Bracing and Taping

- Multidirectional instability of the
  - Hips, Shoulders, SI, Knees, Ribs, Fingers
- Give patient resources to self manage painful areas as they arise
- Teach patient to brace for the activity
- Benefit may be from proprioceptive feedback
- Compression garments
Manual Therapy – soft tissue

- Re-align collagen fibers and release cross fibers through myofascial release
- Reset Autonomic NS
- Reduce guarding in muscles and fascia – indirect before direct techniques
  - Unwinding, Functional Indirect, Strain-counterstrain with caution to avoid end range at unstable areas
  - Tool assisted scraping

Manual Therapy - Joints

- Correct alignment with Muscle energy technique, specific mobilization, long hold mobs
- Extreme caution with Grade V manipulation - be very specific and not beyond strength of tissues, (similar to pregnancy) and caution re: stretch reflex activation
- Specialized training (NAIOMT, etc)
- DC using Activator, Atlas Orthogonal Technique
Appropriate Exercise/Activity

- Promote:
  - Regular, aerobic fitness
  - Fitness support with strengthening, gentle stretching, and proprioception exercises
    - Consider Qigong, Tai Chi as multipurpose exercise
  - Exercise through full range (Pacey et al, 2014)
  - Weight control (BMI < 25)

Appropriate Exercise/Activity

- Avoid:
  - Showing off bendy tricks (take a picture)
  - High impact sports/activities
  - Prolonged sitting positions and prolonged recumbency
  - Sudden head-up postural change
  - Excessive weight lifting/carrying, joint distraction
Strength exercise progression

Easier

- More proprioceptive feedback – tactile/visual/verbal feedback, use wall or floor, machines
- Chronic Pain – unstable platform, flowing motion, alternate agonist/antagonist or contract/relax and stretch
- Joint stability – i.e. isolate leg or arm movements without spine movement, or isolate shoulder stabilizers
- External focus exercises -lasers
- Visualization with ex – Qigong
- Mid range
- Short duration
- Slow
- Symmetrical/bilateral
- RPE measure starts 4/10

Harder

- Less input as with free weights, unstable platforms, not against wall or floor, less coaching
- Chronic Pain- longer holds, isometrics, longer sets, stretch later in session
- Coordinate arm and leg and core combined movements with load
- Internal focus exercises
- Tai Chi – putting several movements together, Sports
- Full Range
- Longer duration
- Fast
- Unilateral/asymmetrical
- 7.5/10 ultimate goal

Do No Harm

- Caution with exercise bands that get harder at end range
- Caution for nerve entrapment sites and repetitive loading
- Keep in mind tensile strength of tissue varies with level of activity, menstrual cycle, inflammatory state, age and will limit max loads
- Neuro re-education: what was once automatic needs cognitive compensation (think before moving)
- Body mechanics – look before you cue
- Avoid mechanical pain whenever possible by engaging stabilizers
- Slow progression to allow histological change in tissues
- Agree on how long pain is allowed to flair after (2 hours? 4?) – adjust Dose
POTS Self-Management

- Pts need to be aware of their autonomic systems
- Increased isotonic fluid intake (H₂O + salt)
- Decrease orthostatic intolerance
  - Change positions slowly; isometric exercises before movement
  - Avoid hot environment, large meals, alcohol, vasodilator meds, sympathomimetic meds
- Manage fatigue
  - Lie down, legs elevated

  • Raj et al, 2013; Benarroch et al, 2012

POTS Self-Management

- Progress exercises gradually
  - Start horizontal, progress to vertical
  - Start with compression garments
- Focus on aerobics and LE resistance
- Children’s Hospital of Philadelphia (CHOP) protocol described in Exercise section
  http://standinguptopots.org/fnuwifh289ry298fhizewf/misc/30-chop-modified-dallas-pots-exercise-program
Sleep Hygiene

• Exercise during the day
• Avoid large meals close to bedtime
• Avoid caffeine and nicotine close to bedtime
• Practice relaxation techniques, avoid stress
• Have a regular bedtime, bedtime routine
  • Avoid computers, TV, phone, etc. (esp blue light)
• Associate bed with sleep  Hakim et al, 2017

Dietary Advice

• Avoid large meals (especially of refined carbohydrates)
• Avoid hard foods intake and excessive jaw movements (ice, gums, etc.)
• Avoid bladder irritant foods (e.g., coffee and citrus products)
• Avoid nicotine and alcohol intake
• Abdominal Massage and Microcurrent? for Gastroparesis
• Heidi Collins Diet 2015 – supplements for poor absorption
Other Lifestyle Changes

- Early management of TMD
- Promote lubrication during sexual intercourse (women)
- Encourage use of compression garments, bracing, assistive devices as needed

Women’s/Men’s Health Issues

- Uterine, bladder prolapse, rectal prolapse is common
- Incontinence – pessaries are braces to support bladder/rectum
- If bladder weirdness, urge incontinence, consider tethered cord
- Rectocele worsens due to constipation and straining, leads to increase in inflammation in gut and infection of bladder, yeast infections
- Teach proper toileting techniques – squat position to relax pelvic floor, big belly, deep breathing
- Pelvic pain associated with involuntary guarding to gain stability of pelvis, organs
- Chronic inflammation and infection can lead to interstitial cystitis (MCAD of the bladder/urethra), vulvodynia
- Erectile Dysfunction associated with low blood pressure and vascular insufficiency
- Hyperarousal associated with malfunctioning Autonomic NS
Psychological:

• Understanding pain reduces pain perception
• Realizing that many health issues are linked by one common cause reduces worry
• Cognitive Behavioral Therapy
  • Pain is depressing (chemically)
  • Coping skills
  • Self-efficacy

PTs learn psychology for a reason – Russek et al, 2015

• Pacing (time based not task based)
• Explain pain
• Meditation and pleasant imagery
• Mirror Box therapy
• Distraction and sense of humor
• Make it fun! You and your patient will benefit!
• Active listening skills, paraphrasing
• Identify and challenge negative thoughts
• Exacerbation management
• Self care
Relaxation Training

• Yoga, Thai Chi, Chi Gong, Ai Chi, Aqua aerobics
• Diaphragmatic breathing
• Meditation (e.g., mindfulness, guided relaxation, visualization)
• Biofeedback (e.g., heart-rate variability)

Social:

• Goals to get out and be active again
• Exercise/rehab can be a fun, social outing – socialize!
• Exercise classes
• Support groups
  • Fibromyalgia support group
    • portlandfibrocfs.com
  • Ehlers Danlos Support Group
    • www.oreds.org
    • www.ehlers-danlos.com International Organization
    • www.inspire.com International EDS Online Support
Variables Affecting Outcomes

- Stress level
- Ability to tolerate medications
- Finances, access to adequate care
- Body Type
- Support System
- Early Access to Preventative Care (before FMS/sensitization)
- Coping Skills
- Luck/life events

Set realistic Goals

- Patient avoids disability/reverses disability
- Fewer “bad” days
- Self treatment strategies reduce need for medical intervention
- Increased Sense of Control over Illness
Other Providers on the Team

• MD – but who is in charge?
  • Primary care, Pain Doc, Physiatry, Rheumatologist, Geneticist, Orthopedist, Gynecologist, Cardiologist, Ophthalmologist, Psychiatrist?
  • Caution for Quacks
• Massage: Bowen technique, Neuro integrative Therapy, Myofascial release
• Psychologist
• Reiki, Acupuncture, other Naturopathic Rx
• Spiritual and Religious

It’s Not Just in Your Head – *Explain Pain*

It is very rare that I have a patient that is lying about their pain. You must assume the complaints are legitimate and problem solve to discover their cause.

- Iris Wolfe, PT
References

- Butler, D and Moseley, L. Explain Pain
- Cincinnati Children’s Hospital 2014 Identification and Management of Pediatric Joint Hypermobility- In children and adolescents aged 4 to 21 years old. Evidence-Based Care Guideline for Management of Pediatric Joint Hypermobility, Cincinnati Children’s Hospital Medical Center, Guideline 43: 1-22
- Clark & Simmonds 2011
- Columbi et al, 2015.
- Connelley et al, 2015

References Con’t

- Hakim, Keer and Grahame Hypermobility, Fibromyalgia and Chronic Pain 2010
- Knight, Isobel with Hakim, A A Guide to Living with Hypermobility Syndrome: Bending without Breaking 2010
- Pocinki, Alan G, MD, PLLC Joint Hypermobility and Joint Hypermobility Syndrome
References Con’t


References Con’t

• www.ehlers-danlos.com
• www.inspire.com
• www.oreds.org
• Facebook: Official Oregon Area Ehlers-Danlos Support Group, Fibromyalgia Support Group Portland
• http://prettyill.com
http://medicalzebras.com
http://ehlers-danlos.org (UK)
http://ehlersdanlosnetwork.org
http://murraywoodfoundation.org
http://www.reumatologia-dr-bravo.cl (CL)
• www.dinet.org  Dysautonomia information
• Mobilisation of the Nervous System – NOI group course
• North American Institute or Orthopaedic and Manual Therapy (NAIOMT) courses

Heather Purdin, PT
4475 SW Scholls Ferry Rd, Suite 258
Portland, OR 97225
Ph: 503-292-5882
www.goodhealthphysicaltherapy.com
heather@goodhealthphysicaltherapy.com
Questions?

Hypermobile EDS is a Heterogeneous Syndrome with varying presentations and intensities. Any body system that relies on collagen is suspect.
Diagnosing Classical EDS

Major criteria required:

1st Major Criteria – must have BOTH of the following:

☐ Skin hyperextensibility
☐ Atrophic scarring

2nd Major criteria: Generalized joint hypermobility

Beighton Test

Please check the following items you are able to do:

☐ Touch right thumb to bottom of forearm
☐ Touch left thumb to bottom of forearm
☐ Bend right pinky back >90°
☐ Bend left pinky back >90°
☐ Straighten right elbow so it bends back >10°
☐ Straighten left elbow so it bends back >10°
☐ Straighten right knee so it bends back >10°
☐ Straighten left knee so it bends back >10°
☐ Touch the floor while keeping knees straight

Add up all items you checked.

Total score: ___/9

*if Beighton score is 1 point below diagnosis score, then complete 5 point questions (5PQ) for past medical history that may affect joint hypermobility

5 Point Questionnaire

Please check for yes to the following:

☐ 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"?

Total items selected: ________ If yes to ≥ 2 items and Beighton score is 1 point below score needed for age group, then positive for generalized joint hypermobility.

If yes to both major criteria, then can diagnose EDS Classical Type

If yes to first major criteria, but no joint hypermobility, then need ≥ 3 minor criteria:

Heather Purdin, PT, Good Health Physical Therapy & Wellness 2017
Minor criteria

☐ Easy bruising - can occur anywhere on the body including unusual sites, shins stained with hemosiderin from previous bruises
☐ soft doughy skin - skin texture is a subjective assessment
☐ skin fragility
☐ molloscoid pseudotumours - over pressure points - elbow, knees, fingers (swelling)
☐ subcutaneous spheroids - small hard bodies frequently mobile and palpable on forearms and shins. May be calcified and detectable on x-ray
☐ hernia or history
☐ epicathal folds in eyes - often seen in childhood and may see in adulthood-fold in nasal part of eye
☐ family hx of 1st deg relative who meets clinical criteria

Total items selected: _______

If yes to ≥ 3 minor criteria + first 2 major criteria, then can diagnose EDS Classical Type
**Diagnosing Hypermobile EDS**
Must meet all 3 criterion 1-3 (defined below)

1. Generalized joint hypermobility
2. Features of connective tissue disorder: systemic, family history, musculoskeletal
3. Absence of exclusion criteria

**Criterion 1. Generalized joint hypermobility**

**Beighton Test**

Please check the following items you are able to do:

- Touch right thumb to bottom of forearm
- Touch left thumb to bottom of forearm
- Bend right pinky back >90°
- Bend left pinky back >90°
- Straighten right elbow so it bends back >10°
- Straighten left elbow so it bends back >10°
- Straighten right knee so it bends back >10°
- Straighten left knee so it bends back >10°
- Touch the floor while keeping knees straight

Add up all items you checked.

| Total score: | /9 |

Check your age group:

- pre-pubescent (score ≥ 6 = hypermobile)
- pubescent and ≤ 50 years old (score ≥ 5 = hypermobile)
- 50 years old (score ≥ 4 = hypermobile)

*if Beighton score is 1 point below diagnosis score, then complete 5 point questions (5PQ) for past medical history that may affect joint hypermobility

**5 Point Questionnaire**

Please check for yes to the following:

- 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"?

Total items selected: If yes to ≥ 2 items and Beighton score is 1 point below score needed for age group, then positive for Category 1.
Criterion 2. Features of connective tissue disorder

A. Systemic manifestations

B. Family history

C. Musculoskeletal complications

Feature A: Systemic manifestations
Please check for yes:
- unusually soft or velvety skin
- mild skin hyperextensibility
- unexplained striae (stretch marks) on the back, groins, thighs, breasts, and/or abdomen in adolescents, men or prepubertal women without history of significant changes in weight
- recurrent or multiple abdominal hernias (umbilical, inguinal, crural, not hiatal)
- atrophic scarring involving at least 2 sites without papyraceous and/or hemosideric scars
- bilateral piezogenic papules of heel
- pelvic floor, rectal, and/or uterine prolapse in children, men, or women who have not birthed children, without history of morbid obesity or other predisposing medical condition
- Dental crowding and high/narrow palate
- Arachnodactyly with bilateral Steinberg signs (wrists) and/or Walker signs (thumbs)
- Arm span-to-height ≥1.05
- Mitral valve prolapse mild or greater based on echocardiographic criteria
- Aortic root dilatation with Z-score ≥+

Total score:__________ (score ≥ 5, then positive for Feature A)

Feature B: Family history
- One or more 1st degree relatives independently meets diagnostic criteria for hEDS

Feature C: Musculoskeletal complications
- pain ≥ 2 limbs, recurring daily for at least 3 months
- chronic widespread pain for ≥ 3 months
- recurrent joint dislocations or medical joint instability
  - ≥ 3 atraumatic dislocations in the same joint or ≥ 2 atraumatic dislocations in 2 different joints at different times
  - Medical confirmation of atraumatic joint instability ≥ 2 sites

Total score:__________ (score ≥ 1, then positive Feature C)

If yes to ≥ 2 features, then positive for Criterion 2.
**Criterion 3. Exclusion criteria (don't have something else)**

- other heritable or acquired connective tissue disorder (lupus or RA)
- neuromuscular disorders, which may cause joint hypermobility by means of hypotonia
  OR connective tissue laxity (ex: mitochondrial disorder causing hypotonia)
- unusual skin fragility which should prompt consideration of other types of EDS

**Must meet all three items to be positive for Criterion 3.**

---

**Criterion 1, 2, and 3 must all be present to diagnose hEDS**

**Treatment**

Physical therapy for:

- Strengthening muscles around loose joints to improve movement and stability, posture, movement patterning, movement mechanics, adaptive ideas, releasing.
- Releasing tense tissues that pull joints out of alignment
- Assisting joints back into alignment with manual therapy
- Teaching safe movement patterns, strategies, and activity modifications to prevent injury and reduce pain
- Recommendations for garments to improve blood pressure, dizziness
- Exercise recommendations

Equipment for:

- Bracing: ankles, knees, SI joint, fingers, wrists, and any other problem areas
- Shoe inserts, arch supports, metatarsal pads for foot pain and alignment
MCAD: Mast Cell Activation Disorder
- mast cells are part of inflammatory process
- can contribute to joint pain because there are too many activated mast cells in the body
- can affect organ systems
- may not show up in lab tests as abnormal

References:
   https://www.youtube.com/watch?v=ktFdr-9rpIM
   https://www.youtube.com/watch?v=ee4Ps5bTZKQ
   Abstract
   Mast cell activation disease comprises disorders characterized by accumulation of genetically altered mast cells and/or abnormal release of these cells' mediators, affecting functions in potentially every organ system, often without causing abnormalities in routine laboratory or radiologic testing. In most cases of mast cell activation disease diagnosis is possible by relatively non-invasive investigation. Effective therapy often consists simply of antihistamines and mast cell membrane-stabilising compounds supplemented with medications targeted at specific symptoms and complications. Mast cell activation disease is now appreciated to likely be considerably prevalent and thus should be considered routinely in the differential diagnosis of patients with chronic multisystem polymorbidity or patients with whom a definitively diagnosed major illness does not well account for the entirety of the patient's presentation.
Do you experience any of the following? (check for yes):

<table>
<thead>
<tr>
<th>Abdominal symptoms (stomach/gut)</th>
<th>Respiratory symptoms (lungs/breathing)</th>
</tr>
</thead>
<tbody>
<tr>
<td>☐ pain</td>
<td>☐ cough</td>
</tr>
<tr>
<td>☐ cramping and bloating</td>
<td>☐ asthma-like symptoms</td>
</tr>
<tr>
<td>☐ diarrhea and/or constipation</td>
<td>☐ dyspnea (shortness of breath)</td>
</tr>
<tr>
<td>☐ nausea</td>
<td>☐ rhinitis (stuffy nose)</td>
</tr>
<tr>
<td>☐ heliobacter pylori-negative gastritis</td>
<td>☐ sinusitis (sinus infections, pressure)</td>
</tr>
<tr>
<td>☐ malabsorption</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Eyes</th>
<th>Liver</th>
</tr>
</thead>
<tbody>
<tr>
<td>☐ conjunctivitis (pink eye)</td>
<td>☐ splenomegaly (large spleen)</td>
</tr>
<tr>
<td>☐ difficulty focusing</td>
<td>☐ hyperbilirubinemia (too much bilirubin in blood)</td>
</tr>
<tr>
<td></td>
<td>☐ high cholesterol</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Cardiovascular (heart)</th>
<th>Neurologic (brain, nerves)</th>
</tr>
</thead>
<tbody>
<tr>
<td>☐ tachycardia (high heart rate)</td>
<td>☐ headache</td>
</tr>
<tr>
<td>☐ Abnormal blood pressure</td>
<td>☐ neuropathic pain (burning pain)</td>
</tr>
<tr>
<td>☐ fainting</td>
<td>☐ difficulty concentrating</td>
</tr>
<tr>
<td>☐ hot flash</td>
<td>☐ forgetfulness</td>
</tr>
<tr>
<td></td>
<td>☐ anxiety</td>
</tr>
<tr>
<td></td>
<td>☐ difficulty sleeping</td>
</tr>
<tr>
<td></td>
<td>☐ vertigo (dizziness)</td>
</tr>
<tr>
<td></td>
<td>☐ tinnitus (ringing or buzzing in ears)</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Cutaneous (skin)</th>
<th>Blood</th>
</tr>
</thead>
<tbody>
<tr>
<td>☐ urticaria pigmentosa (darker or red areas)</td>
<td>☐ abnormal bleeding</td>
</tr>
<tr>
<td>☐ hives</td>
<td>☐ splenomegaly (enlarged spleen)</td>
</tr>
<tr>
<td>☐ itching</td>
<td>☐ lymphadenopathy (affects lymph nodes)</td>
</tr>
</tbody>
</table>

| ☐ Interstitial cystitis (painful bladder syndrome) | |
| Musculoskeletal (muscles, bones, soft tissues, joints) | Constitutional (Other/multiple body systems) |
| ☐ muscle pain                        | ☐ fatigue, general tiredness             |
| ☐ joint pain/arthritis               | ☐ asthenia (abnormal lack of energy and weakness) |
| ☐ osteoporosis/osteopenia (decreased bone density) | ☐ fever                                 |
|                                  | ☐ environmental sensitivity              |

Diagnostic Tests
1. Treat with medications (under treatment section) and see if symptoms improve.
2. Biopsy with staining using CD2, CD25, CD30, and CD117 stains at the site of symptoms to identify presence of chemicals made by Mast Cells (CD117 identifies presence of activated Mast Cells themselves). *Gold Standard
3. 24-hour urine sample to test for presence of histamines, prostaglandin D2, and 11β prostaglandin f2α
4. Serum Tryptase 2-4 hours into a flare up and frequent false negative

MCAD diagnosis if:
- Episodic symptoms consistent with mast cell mediator release affecting ≥2 organ symptoms.
- A decrease or resolution of symptoms with anti-mediator therapy (positive diagnostic test #1).
- Evidence of an elevation in a validated urinary/serum marker of mast cell activation, particularly during a symptomatic period (positive diagnostic test #3).
- Primary and secondary disorders of mast cell activation ruled out.
  - mastocytosis, pheochromocytoma, low thyroid or low testosterone, eosinophilic disorders

Treatment with **mast cell stabilizers**:

<table>
<thead>
<tr>
<th>Medication</th>
<th>Dosage/Instructions</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cromolyn sodium</td>
<td>200mg/10mL, 2x/day before meals, best for GI symptoms</td>
</tr>
<tr>
<td>Ketotifen</td>
<td>1-4mg PM, 1-2mg AM, start at night because it might cause drowsiness, best for global joint pain</td>
</tr>
</tbody>
</table>

with **histamine blockers**

<table>
<thead>
<tr>
<th>Blocker Type</th>
<th>Medications</th>
</tr>
</thead>
<tbody>
<tr>
<td>H1 blockers</td>
<td>Zyrtec, Claritin</td>
</tr>
<tr>
<td>H2 blockers</td>
<td>Zantac</td>
</tr>
<tr>
<td>Leukotriene blockers</td>
<td>Singulair, Accolate</td>
</tr>
<tr>
<td>H3 blockers</td>
<td>For brain fog and nerve pain, digestive symptoms. Low dose amitriptyline (drowsy), protriptyline (stimulating)</td>
</tr>
</tbody>
</table>

Avoid triggers
- Dietary intake, sympathetic nervous system activation (stress, fear, adrenaline triggers), environmental (sudden temperature change, extreme temperatures, pressure, etc)

Food intake
- See dietary recommendations section following pages
POTS: Postural Orthostatic Tachycardia Syndrome

Due to decreased blood flow to the brain, it can affect cognition, memory, and attention. It is often misdiagnosed with panic disorders and anxiety. Other conditions that may cause POTS include: Ehlers Danlos Syndrome, Mast Cell Activation Disorder, Diabetes, Autoimmune diseases, Chiari Malformation, and others.

Check for ‘yes’ if you have experienced any of the following:

- Drop in blood pressure upon standing
- Low blood volume
- High levels of norepinephrine due to increased sympathetic nervous system activity
- Fatigue
- Headaches
- Lightheadedness
- Fainting
- Heart palpitations
- Exercise intolerance (difficulty exercising)
- Nausea
- Shortness of breath (dyspnea)
- Coldness or pain in arms and legs
- Reddish or purple color in legs upon standing

Orthostatic hypotension/POTS test

Date:
Side:

<table>
<thead>
<tr>
<th></th>
<th>Supine 5 mins</th>
<th>Standing &lt;1 min</th>
<th>Standing 3 mins</th>
<th>Standing 10 mins</th>
</tr>
</thead>
<tbody>
<tr>
<td>Blood pressure (mmHg)</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Heart rate (bpm)</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Patient symptoms</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Interpretation:
A drop in systolic BP of ≥20 mm Hg, or in diastolic BP of ≥10 mm Hg, or experiencing lightheadedness or dizziness is considered abnormal. An increase in HR ≥30 bpm is considered abnormal and indicative of POTS in the absence of orthostatic hypotension.

Reference:
**Treatment**

<table>
<thead>
<tr>
<th>Sleep</th>
<th>At least 8 hrs/night</th>
</tr>
</thead>
<tbody>
<tr>
<td>Avoid inflammatory triggers</td>
<td>Dietary intake, stress, sympathetic nervous system activation</td>
</tr>
<tr>
<td>Drink coffee for low energy</td>
<td>Be careful about increased fluid loss from urination</td>
</tr>
</tbody>
</table>
| Intake more salt and electrolytes | Add electrolytes to water with products like Endure and Nuun. If drink 64oz, try ½ with these electrolytes.  
|                                | Add more salt to diet. |
| Compression garments           | TED hose socks, compression garments, abdominal brace |
| Exercise                       | Increases circulation, blood flow, blood pressure. *be sure to stretch after each exercise at least 5 seconds.  
|                                | Exercise at 50% effort to begin and within pain free range of motion. Work up to 80% range of motion and effort (since 100% is into hypermobility)  
|                                | If fatigued start with exercises laying down and work up to standing but if energetic then start standing and work toward laying down |
| Reclining                      | Positions to bring feet up, especially above heart, so gravity assists with blood flowing up towards heart and head |
| Medication                     | Low dose beta blockers. Fluidrocortisone for low BP, midodrine for low BP, dramamine if in fight or flight. |
| Transfers                      | Slow transitions from laying to standing |