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Ehlers-Danlos Syndrome
A Loose and Unstable Story

By Nichole Harju PT, DPT, CAFAS

Objectives

- Participants will be able to independently list the 3 main types of Ehlers-Danlos Syndrome.
- Participants will be able to concisely summarize EDS in 3 sentences for patient education purposes.
- Participants will be able to identify 5 common objective findings during a typical assessment for hypermobility type EDS.
- Participants will be able to develop an appropriate treatment plan specific for EDS patients.
**EDS Definition**

An inherited group of connective tissue disorders that are caused by genetic mutations

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**EDS Etiology**

Mutations in genes or proteins

\[\downarrow\]

Altered collagen fibers

\[\downarrow\]

Altered tissues
EDS Prevalence

- Affects men and women of all ethnic backgrounds
- 1 in 2,500 – 1 in 5,000
- Most likely more common

EDS History

- 1967: First documented link between chronic musculoskeletal pain and joint hypermobility
- 1997: Villafranche criteria for 6 EDS subtypes
- 2017: New EDS criteria for 13 EDS subtypes and basic diagnostics
EDS Criteria

- New EDS criteria for subtypes and diagnostics debuted in March 2017
- Goals
  - Improve treatment
  - Improve research

EDS Diagnosis

- Officially diagnosed by doctor/geneticist
- Based on
  - Subjective reports
  - Objective findings
  - Molecular test
  - Family history
EDS Subtypes

- 13 subtypes
  - Classical EDS (cEDS)
  - Vascular EDS (vEDS)
  - Hypermobile EDS (hEDS)
  - Kyphoscoliotic EDS (kEDS)
  - Arthrochalasia EDS (aEDS)
  - Dermatosparaxis EDS (dEDS)
  - Classical-like EDS (clEDS)
  - Cardiac-valvular EDS (cvEDS)
  - Brittle cornea syndrome (BCS)
  - Spondylodysplastic EDS (spEDS)
  - Musculocontractural EDS (mcEDS)
  - Myopathic EDS (mEDS)
  - Peridontal EDS (pEDS)

EDS Subtypes

- Classical EDS (cEDS)
  - Generalized joint hypermobility (GJH)
  - Skin hyperextensibility
  - Atrophic scarring
EDS Subtypes

- Vascular EDS (vEDS)
  - Family history of vEDS with documented mutation
  - Arterial rupture at young age
  - Spontaneous sigmoid colon perforation
    - Without diverticular dx
  - Uterine rupture during 3rd trimester
    - Without previous C section
  - Carotid-cavernous sinus fistula

EDS Subtypes

- Kyphoscoliotic EDS (kEDS)
  - Congenital or early onset kyphoscoliosis
  - Congenital muscle hypotonia
  - GJH with dislocations/subluxations

Source: Matthew Dobbs
Science Daily
EDS Subtypes

- Arthrochalasia EDS (aEDS)
  - Congenital bilateral hip dislocation
  - Sever GJH with multiple dislocations/subluxations
  - Skin Hyperextensibility

- Dermatosparaxis EDS (dEDS)
  - Extreme skin fragility
  - Characteristic craniofacial features
  - + 1 other major criteria or 3 other minor criteria
EDS Subtypes

- Hypermobile EDS (hEDS)
  - No identified genetic cause → no molecular diagnostic test
  - Clinical presentation is on a spectrum
  - Most common
  - Autosomal dominant

EDS Subtypes

- Hypermobile EDS (hEDS) – Pre 2017 Criteria
  - Skin involvement - Hyperextensibility and/or smooth, velvety
  - Generalized joint hypermobility
  - Beighton Score ≥ 5
  - Recurring joint dislocations
  - Chronic joint/limb pain
  - Positive family history
Diagnosis

- Beighton Hypermobility Score
- Gold Standard for EDS and hypermobility diagnosis
- 5 passive range of motion movements
- Score 0-9

Beighton Score

- Pull little finger back beyond 90 degrees
- One point each side
Beighton Score

- Pull thumb back to touch forearm
- One point each side

Beighton Score

- Bend elbow backwards beyond 10 degrees
- One point each side
Beighton Score

- Bend knee backwards beyond 10 degrees
- One point each side

Beighton Score

- Lie hands flat on the floor while keeping knees straight and bending forward at the waist
- One point
hEDS Diagnostic Criteria - NEW

**CRITERION 1 - Generalized Joint Hypermobility**

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

If Brighton 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) stand on 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 knee ever dislocate more than once 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 (must be present):
- Unusually soft or velvety skin
- Mild skin hyperextensibility
- Unexplained severe distension or bruising at the back, groins, thighs, breasts and/or abdomen in adolescents, men or pre-pubescent women without a history of significant gain or loss of body fat or weight
- Bilateral pes planus of great toes
- Recurrent or multiple abdominal hernias
- Atlantoaxial instability involving at least two sites and without the formation of odynopרוגrosis and/or hemosideric scar seen in classical EDS
- Facial floor, rectus, and/or uterine prolapse in children, men or multiparous women without a history of marked obesity or another known predisposing medical condition
- Dental crowding and high or narrow palate
- Achondroplasia, as defined in one or more of the following:
  - [positive wrist sign], [positive thumb sign], [negative thumb sign], [negative wrist sign], [positive ulnar deviation] of each side
  - Armspan-to-height ratio < 0.85
  - Mitral valve prolapse: mild or greater based on strict echocardiographic criteria
  - Aortic root dilatation with Z-score ≥ 2

Feature B (optional):
- Positive family history; one or more first-degree relatives independently meeting the current criteria for EDS

Feature C (at least one):
- Musculoskeletal pain in two or more limbs, occurring daily for at least 3 months
- Chronic, widespread pain for > 3 months
- Recurrent joint dislocations or frank joint instability in the absence of trauma
hEDS Diagnostic Criteria - NEW

**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 EDS 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 EDS 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, Loes–Dietz syndrome, Marfan syndrome), and skeletal dysplasias (e.g. osteogenesis imperfecta). Exclusion of these considerations must be based upon history, physical examination, and/or molecular genetic testing, as indicated.

Nameology – Pre 2017

- Pre 2017
- Older classifications will appear in older literature
- Transition period

- Hypermobility Syndrome (HMS)
- Benign Joint Hypermobility Syndrome (BJHS)
- Joint Hypermobility Syndrome (JHS)
- EDS hypermobility type (EDS-HT)
Nameology – Post 2017

No more Joint Hypermobility Syndrome (JHS)
No more Hypermobility Syndrome (HMS)

EDS hypermobility type (hEDS)
Hypermobility Spectrum Disorders (HSD)

Does the name really matter?

YES  NO
hEDS Factors

- Spectrum of joint laxity
- Large variations in reported symptoms
- Can be an asset for athletes
- Affects all connective tissue

hEDS Patient Definition

1) An inherited disorder that affects all connective tissue in the body.
2) Where the connective tissue is stretched out like a loose rubber band instead of its normal tension.
3) Since connective tissue is part of tendons, ligaments, skin, eyes, gut, bones, and some organs.....there can be a lot of different symptoms.
Symptoms - hEDS

- Musculoskeletal Pain
  - Usually joint related (shoulders, back, hips, knees)
  - Headaches
  - Localized biomechanical overload with repetitive stress
  - Microtraumas due to joint instability
  - Altered movement patterns
    - Ex) gait – decreased lateral stability
  - Muscle tightness compensation
  - Modified neuromuscular activation patterns

- Cutaneous Factors
  - Skin hyperextensibility
  - Minor issues with wound healing
  - High risk of bruising – capillary fragility
Symptoms - hEDS

- Orthopedic Features
  - Frequent sprains, strains, dislocation and subluxations
  - Altered posture in weight bearing due to instability
  - Increased joint ROM

- Fatigue
  - Increased energy to stabilize
  - Poor sleep hygiene
Symptoms - hEDS

- Deconditioning
  - Muscle weakness
  - Decreased cardiovascular capacity
  - Activity avoidance

Symptoms - hEDS

- Neurological
  - Hyperalgesia
  - Upregulation of the CNS
Symptoms - hEDS

- Proprioception
  - Clumsiness
  - Limited balance
  - Poor posture

Altered Proprioception in hEDS

- Balance is based on sensory integration of vision, vestibular, and somatosensory input
- Excessive ligament stretching $\rightarrow$ joint instability
  $\rightarrow$ damage to surrounding proprioceptive receptors
- Skin hyperextensibility probably changes pressure input and tactile mechanoreceptors
Symptoms - hEDS

- Psychiatric
  - Anxiety related to activities
  - General anxiety
  - Depression
  - Schizophrenia
  - Autism spectrum

Associations with hEDS

- Cardiovascular/pulmonary abnormalities
  - ~25% mild mitral, tricuspid and aortic valve regurgitations
  - Poor pulmonary gas exchange
Associations with hEDS

- POTS – Postural orthostatic tachycardia syndrome
  - Collagen issues can affect blood vessels
  - Fainting
  - Temperature control issues

- Fibromyalgia
  - Muscle sensitivity
  - Trigger point pain

- Muscles
  - Weakness
  - Pain
  - Cramps
  - Ruptures
Associations with hEDS

- Rheumatic diseases
  - Rheumatoid arthritis
  - Psoriatic arthritis
  - Psoriasis
  - Ankylosing spondylitis
  - Systemic lupus erythematosus
  - Inflammatory eye disease

Undiagnosed Subtypes in hEDS

- Much variation between patients without a specific molecular profile

- Still a lot to be researched and classified for proper treatment
Assessment

Posture
Gait
Balance
Static Strength Testing
ROM
Flexibility
Agility/Coordination

Assessment - Posture

- Possible Observations
  - Forward head
  - Rounded shoulders
  - Scapular protraction
  - Increased lordosis
  - Knee hyperextension
  - Medial ankle roll in
  - Midfoot pronation
Assessment - Gait

- Possible Observations
  - Toe walking/flat foot
  - No hip rotation
  - No arm swing
  - Hip external rotation
  - Decreased push off
  - Falling motion
  - Labored gait
  - Wide/narrow BOS

Assessment - Balance

- Possible Observations
  - More hip strategy than ankle
  - Increased anterior tib use
  - Hip drop
  - Overuse of arms
  - Knee hyperextension
Assessment - MMT

- Possible Observations
  - Muscle compensations
  - Limited strength
  - Pain at joints

- Common Weakness
  - Parascap
  - Abdominals
  - Glutes
  - Hip rotators

Assessment - ROM

- Possible Observations
  - Excessive range
  - Usually major joints
  - Compensations
Assessment - Flexibility

- Possible Observations
  - Depends on patient
  - Usually decreased flexibility is from compensation not actual muscle tightness

- Common Tight Muscles
  - Upper traps
  - Paraspinals
  - Psoas
  - Gastrocs

Assessment - Agility

- Possible Observations
  - Decreased stability
  - Hip Adduction with jumping
  - Ankle medial collapse
  - Poor deceleration

Source: bretcontreras.com
Treatment

- Pain management
  - Medication
  - TENS
- Cognitive – behavioral therapy
- Lifestyle modifications
  - Energy management
  - School schedule changes
- Physical Therapy

Treatment - Physical Therapy

- General guidelines
  - NO PAIN
  - Start where patient is successful
  - May need to start with very small motions
  - Consistent cueing for posture during activities
  - Watch and correct compensations
  - Base POC on individual’s current status and needs
  - Celebrate the successes to help anxiety and depression
Treatment - Physical Therapy

- General guidelines
  - Work in function
  - Triplane motion in exercise
    - Sagittal, frontal and transverse
  - Refer to developmental sequence
    - Sitting, tall knee, half kneel, quadruped, standing, SLS

- Strengthening
- Aquatic therapy
- Proprioception/balance training
- Endurance training
- Stretching
- Bracing as needed
- EDUCATION
Strengthening

- May need to start with limited weightbearing depending on pain or compensations
  - Mat exercises/isometrics
  - Aquatic therapy
- Transition to weightbearing as soon as possible

Isometrics

- Can help activate shut down muscles
- May be the only thing to not cause pain at first

Examples

- Glute sets
- Quad sets
- Heel digs
- Ab sets
Mat Exercises

- Use when weight bearing causes pain or compensations
- Still incorporate triplane motion
- Good for HEP

Examples

- Clams
- Hook lying ABD/ADD
- Hamstring curls
- Bridges – triplane
- Crunches – straight and rotation
- Straight leg raises (forward, backward, lateral)
- Shoulder ROM with bands or hand weights
Stabilization

- BEST place to start
- Most important to train total body in weight bearing for function
- Uses tactile and verbal cues as needed for posture
- Need to have proper muscle activation before progressing
- Reference developmental sequence
- Also activates proprioceptors
- Include movement to be functional

Stabilization

**Activity**
- Single leg activities
- Tall kneel
- Half kneel
- Quadruped
- Staggered stance
- Sitting

**Surface**
- Floor
- Airex
- Bosu
- Reverse Bosu
- Rocker Board

**Movement**
- Rotational reaches
- Throwing
- Kicking
- Reaching low
- Body Blade
- Pulling Bands
- Rhythmic Stabilization
Stabilization

[Images of individuals performing stabilization exercises on a Bosu ball and using a bar]

Stabilization

[Images of individuals performing stabilization exercises on a Bosu ball and using a bar]
Dynamic Strengthening

- Transition to dynamic activities once patient has:
  - Awareness of correct posture
  - Proper muscle activation
- Can add resistance
- Don’t forget triplane!

Dynamic Strengthening

- Examples
  - Lateral crawling
  - Lunges – triplane
  - Squats
  - Planks
  - Ball bridges/crunches
  - Upper body strengthening
  - Penguin walks
Dynamic Strengthening

Dynamic Strengthening
Dynamic Strengthening

- Backward Crawling Video
- Rotational Plank Video
- Penguin Walk Video

Proprioception/Balance

- Start from success
- Can start in half or tall kneel to decrease compensation or pain
- Vary surfaces
- Add movement or games
Plyometrics

- Must have proper joint stabilization to start
- Incorporate before returning to sport
- Don’t forget triplane movements
- Can overlaps with endurance

Plyometrics

- Examples
  - Hops – triplane
  - Agility ladder
  - Jumping – triplane
  - Skaters
Endurance Training

- Start once joints are more stable
- Can be low or high impact
- Try activity circuits
  - Cardio and strength mixed for active recovery

Examples

- Jumping jacks
- High knees
- Lateral shuffles
- Grapevine
- Triplane jumping
- Mountain climbers
Stretching

- RARELY
- Muscle compensation vs. muscle shortness
  - Muscles tighten to stabilize joints in absence of strength
  - Don’t stretch what is already loose
- Muscle tightness associated with activity
- Establish stability before stretching
- Stretch only when needed

Stretching

Common compensations
- Forearms
- Quads
- Upper traps
- Hamstrings
- Gastrocs

Actual tightness associated with activity
- Gastroc
- Paraspinals
- Psoas
- Pecs
Other Treatments

- Aquatic therapy
  - Decreases stress on joint
  - Able to strengthen with less pain
  - Short term

Other Treatments

- Bracing/Orthoses
  - Provides stabilization
  - Use short term

- Kinesiotaping as an alternative
  - More flexible
  - Can activate muscles or stabilize joint
  - Patient can do as needed
Somatosensory Study

Goals
1) See the effect of somatosensory deficits on subjective visual vertical and postural stability
2) Impact of somatosensory orthoses on postural stability

- 6 hEDS and 6 age/sex matched controls
- Sitting, standing, lying on right side +/- visual information
- Postural control challenged on force platform
- +/- somatosensory orthoses

Somatosensory Study

- hEDS demonstrated more postural instability than controls (larger sway area)
- Postural instability was worse without vision (especially frontal)
- Orthoses improved postural stability especially in eyes closed and sagittal
Treatments to Be Aware of

- Surgery
- Long periods of inactivity
- Muscle relaxants
- Steroids
- Anti-epileptic drugs
- Anti-platelet drugs

Patient Education

- Most important
- Educate patient and parent
  - What the diagnosis is
  - Common issues - connect all the dots/symptoms
  - Hook the patient
  - Provide a HEP for every patient!
Patient Education

School/work

- Partial or modified day
- Stay in gym with modified activity if possible
- Use of isometrics for energy and pain reboots throughout the day
  - heel digs, quad sets, scap squeeze, TA sets, wall presses, cervical pushes

Patient Education

Lifestyle Recommendations

- Regular aerobic exercise
- Strengthening and proprioception exercises
- Weight control (BMI<25)
- Daily relaxation activities
- Avoid high impact sport/activities
- Avoid low temperatures
- Avoid prolonged sitting positions
Patient Education

Sleep
- No phone, TV or iPads an hour before bed
- Modify bed with pillows or towel roll for comfort

Energy Conservation
- Small bouts of cardio or strength to increase energy
- Activity management and planning

Patient Education

Energy conservation  (Parkwood Pacing Point System)
- Assign energy values to daily activities
- Map out day
- Correlate energy values with scheduled day
- Limit energy number for the day
- Include rest breaks
### Parkwood Pacing Points System

<table>
<thead>
<tr>
<th>Activity</th>
<th>Points</th>
</tr>
</thead>
<tbody>
<tr>
<td>Grocery shopping</td>
<td>5</td>
</tr>
<tr>
<td>Meal Prep: breakfast</td>
<td>0.5-1</td>
</tr>
<tr>
<td>Meal Prep: lunch</td>
<td>0.5-1</td>
</tr>
<tr>
<td>Meal Prep: simple supper</td>
<td>1-2</td>
</tr>
<tr>
<td>Meal Prep: complex supper</td>
<td>2-3</td>
</tr>
<tr>
<td>Cleaning: Vacuuming, dusting, bathrooms</td>
<td>1 per task</td>
</tr>
<tr>
<td>Talking on the phone (per 15 minutes)</td>
<td>2</td>
</tr>
<tr>
<td>Attending therapy: PT</td>
<td>3</td>
</tr>
<tr>
<td>Attending therapy: OT</td>
<td>2</td>
</tr>
<tr>
<td>Attending therapy: SW</td>
<td>2</td>
</tr>
<tr>
<td>Attending therapy: SLP</td>
<td>2</td>
</tr>
<tr>
<td>Driving as a passenger (short distance vs. long)</td>
<td>0.5-2</td>
</tr>
<tr>
<td>Driving as a driver (short distance vs. long)</td>
<td>1</td>
</tr>
<tr>
<td>Attending a doctor's appointment/dental appointment</td>
<td>3-4</td>
</tr>
<tr>
<td>Watching TV (per 30 minutes)</td>
<td>0.5</td>
</tr>
</tbody>
</table>

### Case Study - Kaylee

- 17 year old female with hEDS
  - Complaints: migraines, neck, shoulder, back, hips, knees, ankle and foot pain
  - Medical history: frequent injuries, POTs
Case Study - Kaylee

- Objective findings:
  - C3 R rotated and posteriorly
  - L1-3 posteriorly displaced
  - Extreme joint hypermobility
  - Significant muscle tone due to compensation: upper traps, paraspinals, gastrocs
- Significant weaknesses
  - Shoulder ER, shoulder flexion, poor scap stab, abdomen
  - Hip rotators, hip abd, glutes, hamstrings

Mat exercises
- Tall kneeling/stability
- Facilitated spine alignment
- Educated patient on kinesiotaping joints to decrease pain short term
- Short progressive bouts of cardio at home with strengthening (see included handout)
Case Study - Kaylee

- 22 visits from 12/6/16 – 8/18/17
- Results
  - Decreased pain overall (frequency and duration)
  - Lessened migraine intensity
  - 5/5 hip, glutes and lower extremity strength
  - 4+ 5/5 shoulder strength
  - More consistent upright posture with less compensations
  - Patient was able to participate in school and hike
- Progressed to a home program of strengthening and cardio

Case Study - Lucas

- 14 year old male with hEDS
  - Complaints: chronic pain (neck, back, knees, ankles), poor posture, chronic fatigue, difficulty attending school
  - Medical History: anxiety, POTs like symptoms, family history
Case Study - Lucas

- Objective Findings
  - Extreme joint hypermobility
  - Very poor posture with flared ribs
  - Significant muscle tone due to compensation: upper traps, pecs, paraspinals, gastrocs
  - Significant weaknesses
    - Shoulder ER, shoulder flexion, poor scap stab, abdomen
    - Hip rotators, hip abd, glutes, hamstrings

- Mat exercises -> stability -> dynamic -> plyometric -> endurance
- Core stability for rib stability and posture
- Pre-wrestling training
- School modification
  - Altered school day
  - Gym participation
  - Dyna disc
Case Study - Lucas

- Results
  - Significantly improved posture with less flaring
  - Almost 5/5 strength UE and LE
  - Able to tolerate most days of high school and gym
  - Participating in the wrestling team
- On and off for 3 years due to insurance issues
- Growth spurts
- Enabling parent

Case Study - Ryan

- 16 year old male
  - Complaints: Extreme fatigue, difficulty writing, holding a trumpet
  - Medical History: hEDS
Case Study - Ryan

- **Objective findings**
  - 6’4” with no muscle tone
  - Slouched posture
  - Full body significant weakness (UE, LE, Core)
  - Limited endurance

- **Stability -> plyometric -> cardio**
- Isometrics during school to boost energy
- Short strength + cardio HEP right after school to do homework
- Energy conservation
Case Study - Ryan

- 1x per week for about 11 months
- Results
  - 5/5 strength core and LE
  - Good posture and overall stability
  - Improved function at school
  - No longer needed naps after school
- Progressed to a HEP with cardio and strength

Case Study - Malea

- 32 year old female
  - Complaints: R shoulder, low back, arm, L hip, and R ankle pain, ankle instability causing falling
  - Medical History: cEDS and hEDS, CVID, orthostatic hypotension, dysautonomia, migraines
Case Study - Malea

- Objective findings:
  - Significant joint instability and excessive ROM
  - Poor posture: severe shoulder IR, slouched posture, knee hyperextension
  - Very tight muscles not in typical compensation patterns: forearms, scalp, quads, pecs, gastrocs

- Unable to do dynamic or stabilization activities
- Limited ability of mat exercises
- Currently doing isometrics, mat exercises and aquatic therapy
  - *Further testing
Questions?

Thank you!

References