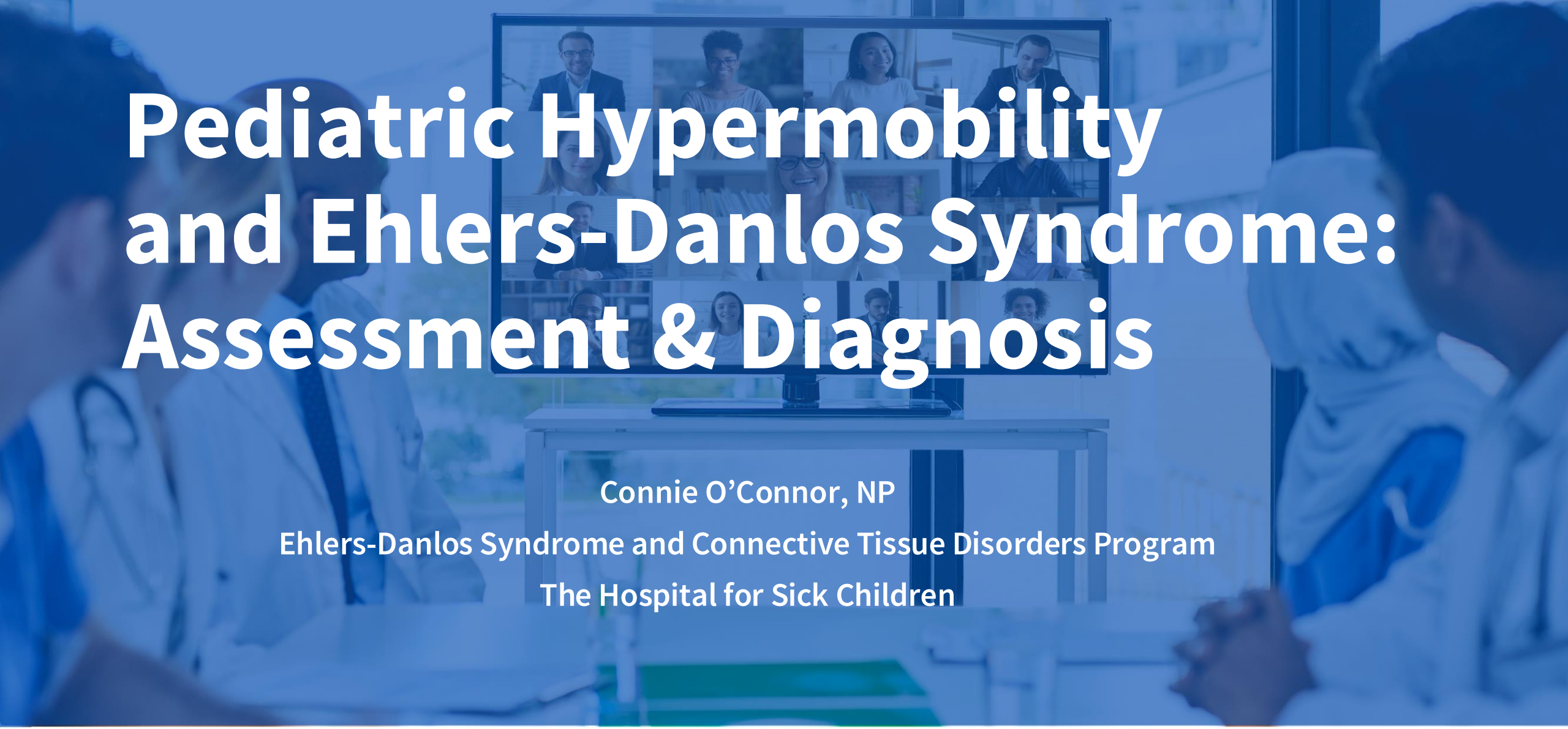

Pediatric Hypermobility and Ehlers-Danlos Syndrome: Assessment & Diagnosis

Connie O'Connor, NP

Ehlers-Danlos Syndrome and Connective Tissue Disorders Program

The Hospital for Sick Children

Project ECHO®

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Declaration of Disclosure

I have no actual or potential conflict of interest in relation to this program.

I assume responsibility for ensuring the scientific validity, objectivity, and completeness of the content of my presentation.

Learning Objectives

Understand	The normal range of joint mobility in the pediatric population
Identify	Patients with generalized joint hypermobility(GJH) using objective and accurate measurement
Review and discuss	The 2023 framework for the assessment of pediatric hypermobility disorders

Ehlers-Danlos Syndromes (EDS)

Group of 13 heritable connective tissue disorders associated with joint hypermobility, skin hyperextensibility, and tissue fragility

Each type of EDS has its own set of features and distinct diagnostic criteria

12 types of EDS have known associated genes and can be confirmed with molecular testing if clinical features are present

Currently, the genetic cause of hypermobile type EDS (hEDS) is unknown

Pediatric (Hyper)mobility

Hypermobility is a **normal** finding in infants and young children that gradually decreases with age, tapering off at puberty

While most childhood hypermobility is benign, hypermobility of a joint may increase the incidence of joint injury, dislocation, and pain

Diagnosis of Hypermobility

Hypermobility cannot be objectively measured before 5 years of age

Validated tools should be used to assess and document hypermobility

Hypermobility occurs along a spectrum and is not always associated with a specific syndrome or disease

Generalized Joint Hypermobility (GJH)

- Foundational component of many diagnoses
- Not concerning if in isolation
- **Beighton Score**

Beighton Score

- Objective and validated measurement that scores joint hypermobility out of 9 points:
  - Knees
  - Elbows
  - Spine
  - Wrists
  - 5th fingers at the metacarpophalangeal joint (MCP)

- A score of **6/9** for pre-pubertal children and **5/9** for post-pubertal adolescents is considered indicative of generalized joint hypermobility beyond the range that is typically found in the general population

CLINICAL MANEUVER	UNABLE TO PERFORM (0 POINTS)	ABLE TO PERFORM (1 POINT)
Apposition of thumb to forearm		
Right	0	1
Left	0	1
Extension of fifth finger beyond 90 degrees		
Right	0	1
Left	0	1
Extension of elbow beyond 10 degrees		
Right	0	1
Left	0	1
Extension of knee beyond 10 degrees		
Right	0	1
Left	0	1
Forward flexion of trunk, legs straight, palms touching floor	0	1
Total Beighton Score (sum of points for each maneuver)	0 to 9 points	

The Ehlers Danlos Society

THE BEIGHTON SCORING SYSTEM

Measuring joint hypermobility

A positive Beighton score is any score **greater than or equal to:**

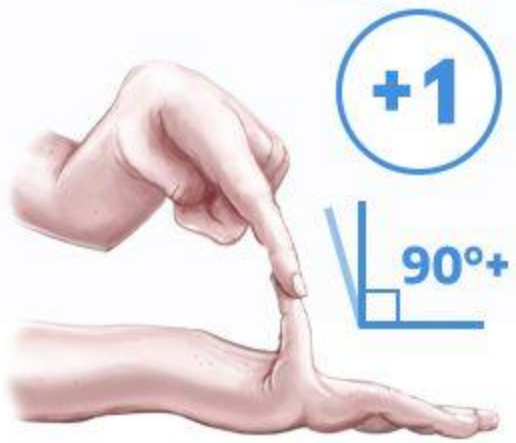
- **6/9** points in **children before puberty**
- **5/9** points from **puberty up to the age of 50**
- **4/9** points for those **50 years of age and over**

A. 5th FINGER / PINKIES

Test **both sides:** Rest palm of the hand and forearm on a **flat surface** with the 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.



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.

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.

D. KNEES

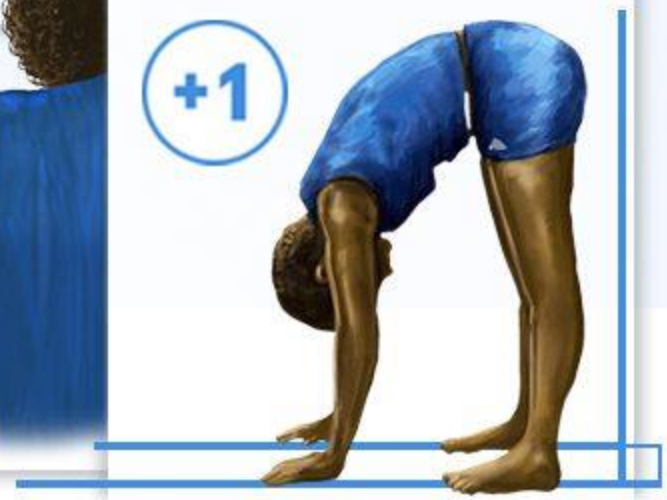
Test **both sides:** While standing, with knees locked (bent backwards as far as possible), does the lower part of either leg to extend more than 10 degrees forward?

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

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.**



Common issues in Performing the Beighton Score

- Improper positioning giving the false appearance of hypermobility
- Allowing for significant effort or force to be applied to achieve points
- Overestimation of degree of hyperextension of knees and elbows

Beighton Score: Take home points!

- Ensure focused assessment of individual joints with proper positioning
- Goniometer is needed to assess multiple items but is not practical for many clinical settings
- If appears borderline, note that the score is an estimate and do not provide a firm diagnosis to patients and caregivers

2017 Diagnostic Criteria for Hypermobile Type EDS (hEDS)

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 women* 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

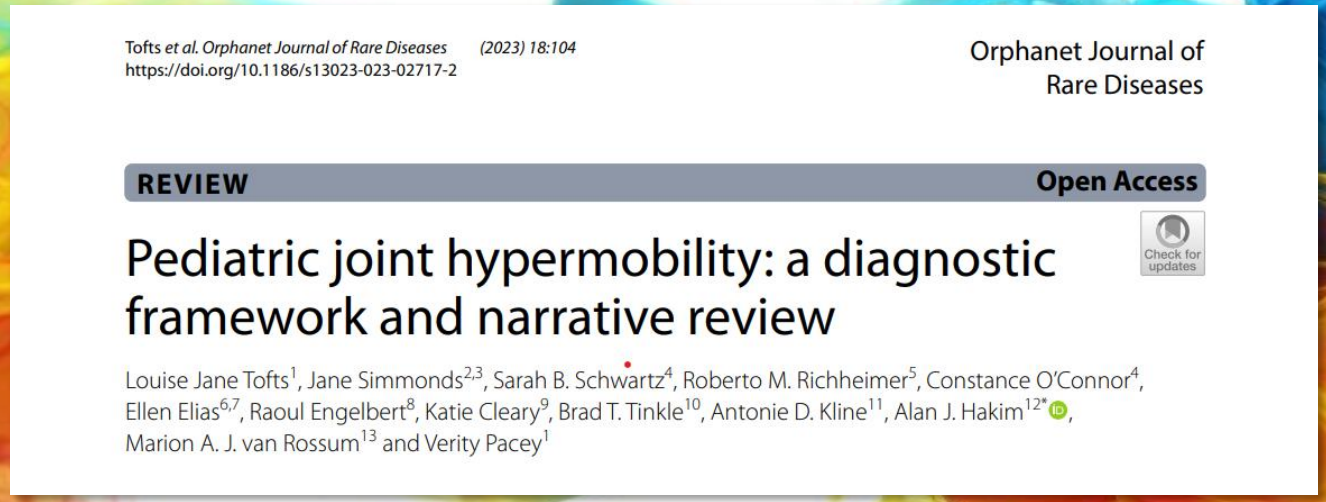
CRITERION 3 – All of the following prerequisites MUST be met

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.

Limitations of 2017 hEDS Diagnostic Criteria in Pediatrics

- Hypermobility changes over time in childhood and adolescence
- Historical questions DO NOT apply to children, adolescents, and young adults
- Feature A criteria often not present in younger patients
- Feature B criteria should apply to parents only
- Feature C criteria often has not advanced to minimal degree required to achieve in younger patients

Pediatric Joint Hypermobility Diagnostic Framework



Tofts *et al. Orphanet Journal of Rare Diseases* (2023) 18:104
https://doi.org/10.1186/s13023-023-02717-2

Orphanet Journal of Rare Diseases

REVIEW Open Access

Pediatric joint hypermobility: a diagnostic framework and narrative review

Louise Jane Tofts[1], Jane Simmonds[2,3], Sarah B. Schwartz[4], Roberto M. Richheimer[5], Constance O'Connor[4], Ellen Elias[6,7], Raoul Engelbert[8], Katie Cleary[9], Brad T. Tinkle[10], Antonie D. Kline[11], Alan J. Hakim[12*], Marion A. J. van Rossum[13] and Verity Pacey[1]

Evolution of clinical presentation and recategorization over time is expected

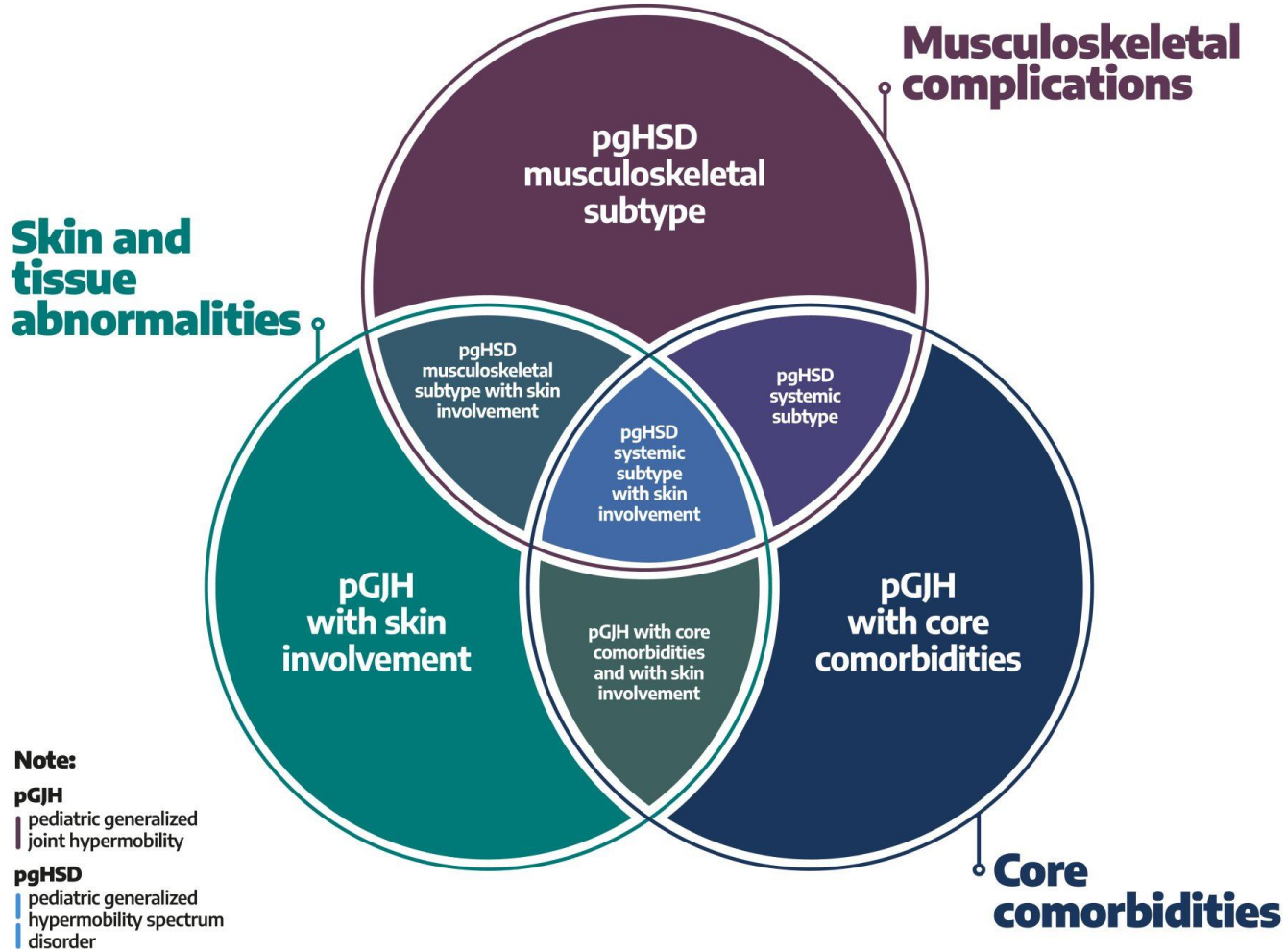
Recommendation: **No diagnosis of hEDS before biological maturity**

Key components

- Skin and tissue abnormalities
- Musculoskeletal complications
- Core comorbidities

The Ehlers Danlos Society.

EVALUATING CHILDREN & ADOLESCENTS WITH GENERALIZED JOINT HYPERMOBILITY



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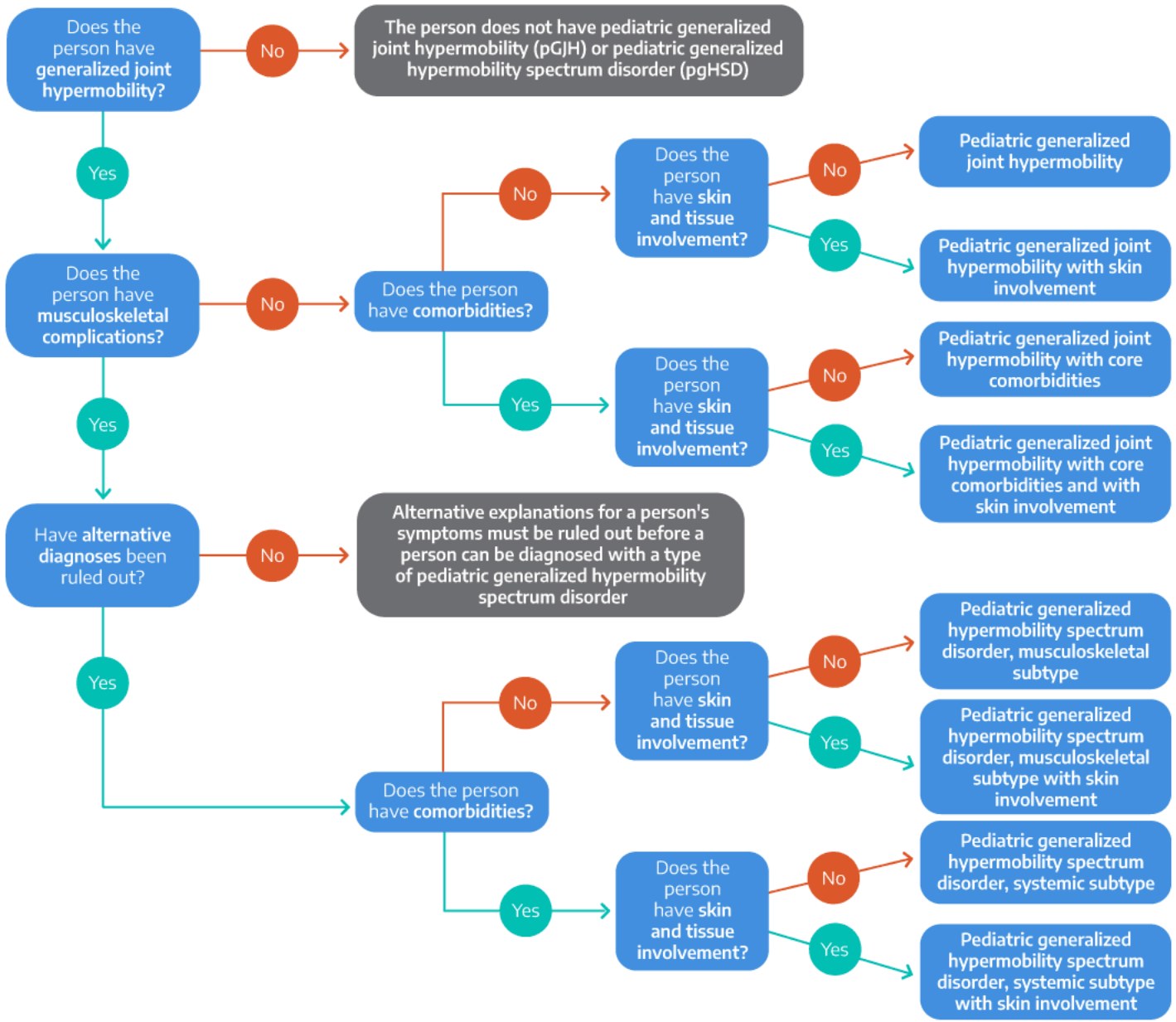
PEDIATRIC DIAGNOSTIC FLOWCHART



Diagnostic Criteria for Pediatric Joint Hypermobility

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

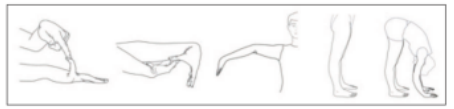
Diagnostic Criteria for Paediatric Joint Hypermobility

This diagnostic checklist is to support doctors to diagnose paediatric joint hypermobility and hypermobility spectrum disorder

Distributed by The Ehlers Danlos Society.

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

Children From 5 Years Of Age Until Biological Maturity



L☐R☐ L☐R☐ L☐R☐ L☐R☐ ☐

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

Prerequisites:

1. This framework can only be used after exclusion of other Ehlers Danlos subtypes, heritable disorders of connective tissue, syndromic conditions, chromosomal microdeletions, skeletal dysplasia's, or neuromuscular disorders. From biological maturity or the 18th birthday, whichever is earlier, the 2017 Adult criteria should be used.

2. No genetic cause for hEDS has been identified at the time of publication of the checklist. In the future disease-causing genetic mutations may be identified in hEDS. In that scenario, if a child has a biological parent with an hEDS diagnosis and a confirmed disease-causing genetic mutation and the child also has the same mutation with GJH then the hEDS diagnosis should be used.

Pediatric Diagnostic Categories

Pediatric Generalized Joint Hypermobility [pGJH] Subtypes	Musculoskeletal Complications	Skin and Tissue Abnormalities	Core Comorbidities
Pediatric generalized joint hypermobility	Absent	Absent	Absent
Pediatric generalized joint hypermobility with skin involvement	Absent	Present	Absent
Pediatric generalized joint hypermobility with core comorbidities	Absent	Absent	Present
Pediatric generalized joint hypermobility with core comorbidities and with skin involvement	Absent	Present	Present

Pediatric Generalized Hypermobility Spectrum Disorder [pgHSD] Subtypes	Musculoskeletal Complications	Skin and Tissue Abnormalities	Core Comorbidities
Pediatric generalized hypermobility spectrum disorder, musculoskeletal subtype	Present	Absent	Absent
Pediatric generalized hypermobility spectrum disorder, musculoskeletal subtype with skin involvement	Present	Present	Absent
Pediatric generalized hypermobility spectrum disorder, systemic subtype	Present	Absent	Present
Pediatric generalized hypermobility spectrum disorder, systemic subtype with skin involvement	Present	Present	Present

Emerging co-morbidities

- Autism Spectrum Disorder
- ADHD
- Developmental discoordination disorder

Summary

- Accurate assessment of hypermobility in children and adolescents can help to guide care and education
- Diagnostic framework developed to improve clinical care and research
  - Informed by best available evidence and expert opinion
  - Ongoing evaluation
- Similar to the expected evolution of clinical presentation in children and adolescents, this framework will evolve over time.
- **No diagnosis of hEDS in children**

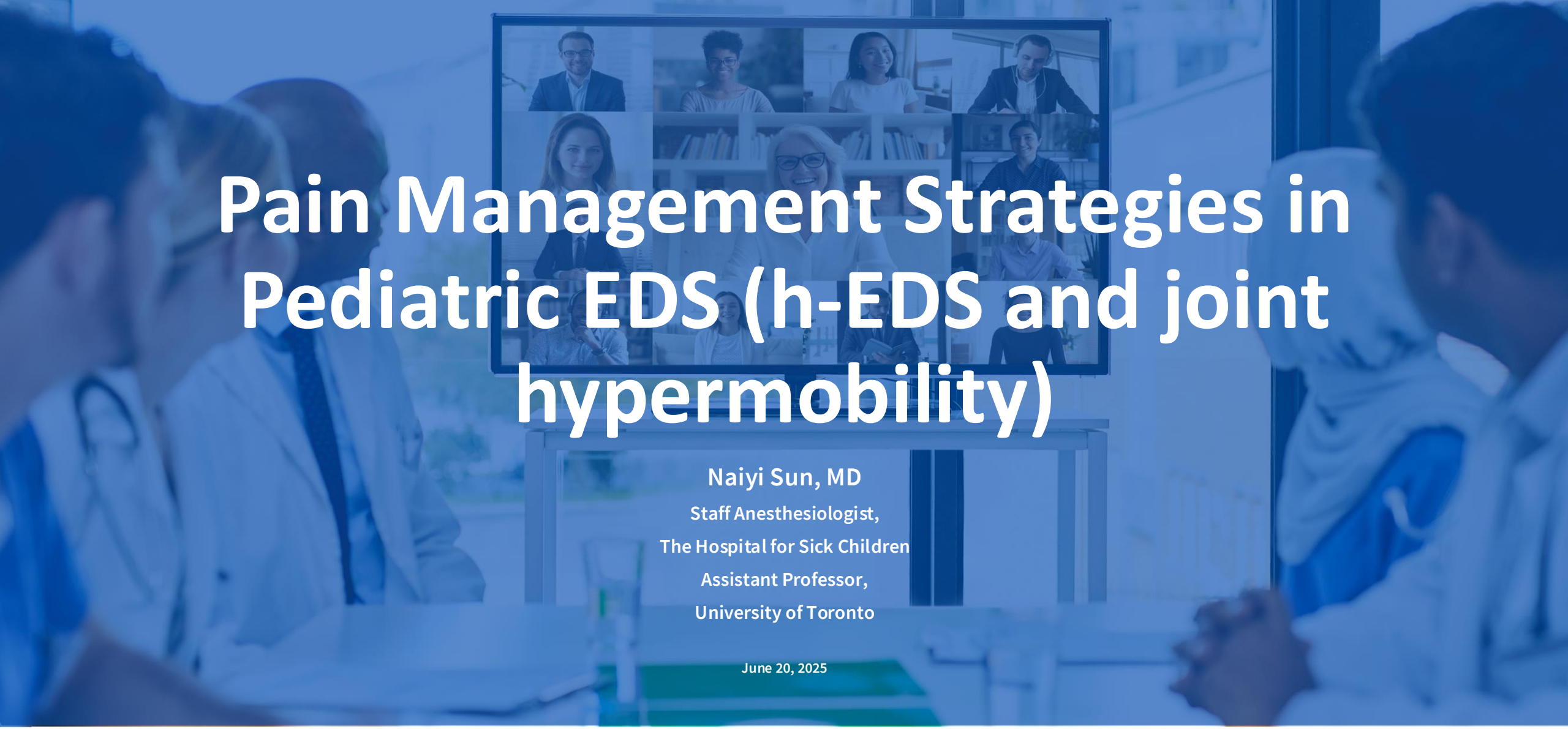
Resources and Supporting Information

- 2023 Diagnostic Framework for Pediatric Joint Hypermobility - The Ehlers Danlos Society

https://www.ehlers-danlos.com/diagnosis/new-diagnostic-framework-for-pediatric-joint-hypermobility-v2/

- Pediatric joint hypermobility: a diagnostic framework and narrative review | Orphanet Journal of Rare Diseases | Full Text

https://ojrd.biomedcentral.com/articles/10.1186/s13023-023-02717-2



Pain Management Strategies in Pediatric EDS (h-EDS and joint hypermobility)

Naiyi Sun, MD

Staff Anesthesiologist,

The Hospital for Sick Children

Assistant Professor,

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June 20, 2025

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Faculty/Presenter Disclosure

- Faculty: Naiyi Sun
- Relationships with commercial interests:
  - None
- This program has received financial support from the Ministry of Health (MOH)

Clinical Diagnosis of Hypermobile EDS:

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

Pain is a Common Symptom

Adults	Pediatrics	Most common
• up to 90%	• 30-56%	• MSK symptoms

In Children:

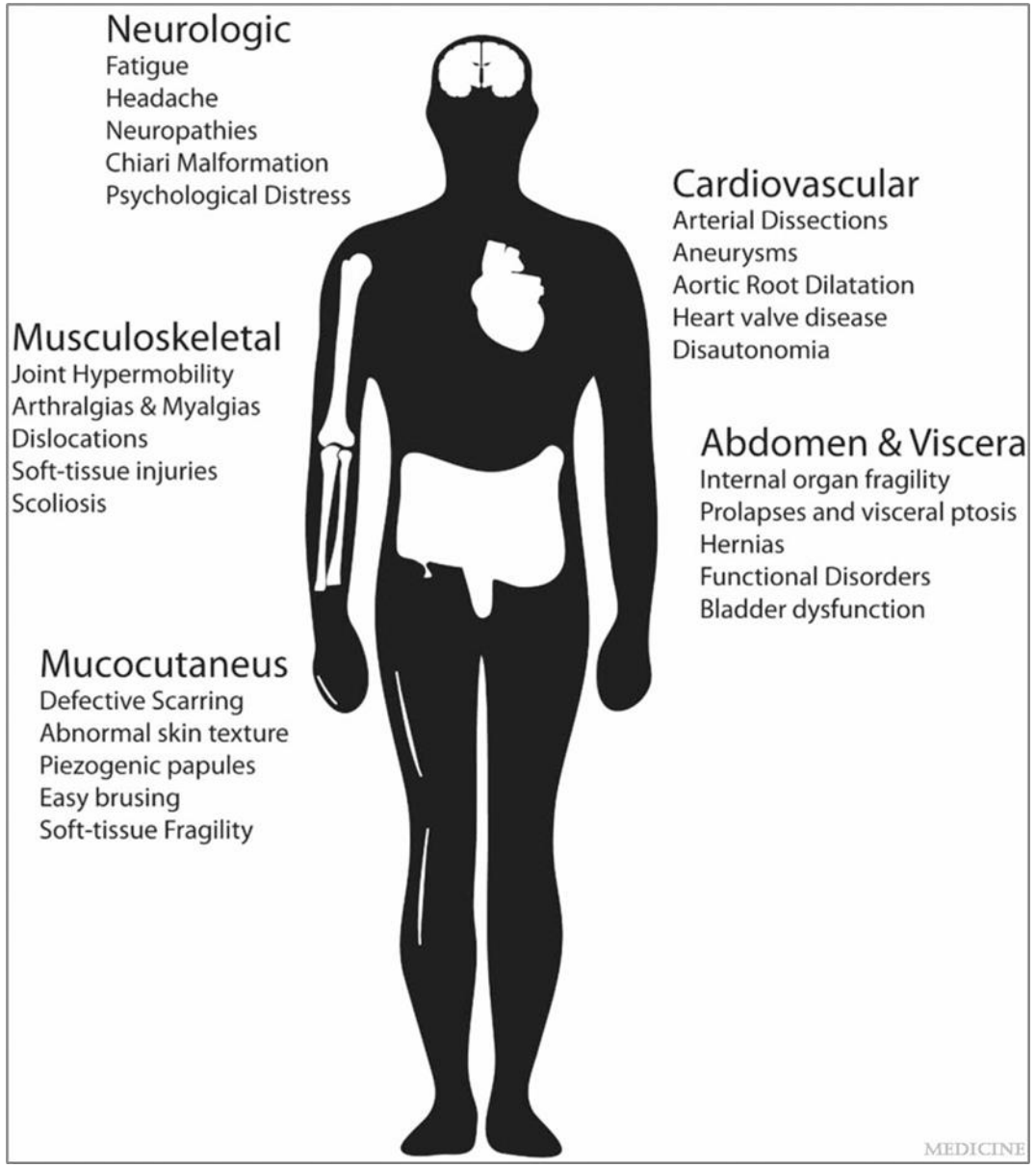
Study with 34 youth with h-EDS (8-17 y.o.)	Retrospective study in 205 children (6-19 years) with EDS (5,6% h-EDS)
• Pain intensity average of 5.5/10 (NRS) • Widespread pain index score: 7 locations (SD 5) and symptom severity score of 5. • Sites of pain: most frequently lower back, neck, shoulders	• Most common pain locations: knee (43.4%), back (32,2%) and shoulder (31.2%) pain

Types of Pain in hEDS

Manifestations	Incidence
Generalized body pain	90%
Soft tissue pain	90%
Dislocations	78%
Joint Pain	Elbow (43%), Shoulders (80%), Hands (70%), Knees (71%), Spine (67%)
Fatigue	55-95%
Neuropathic Pain	68%
Headaches	75%
GI Pain	85.7%
TMJ Pain	71.4%
Dysmenorrhea	73.1%
Vulvodynia	42%

Chopra P et al. Pain management in the Ehlers-Danlos syndromes. Am J Med Genet C Semin Med Genet. 2017 Mar;175(1):212-219.

Systemic comorbidities in EDS



Zhou Z, Rewari A, Shanthanna H. Management of chronic pain in Ehlers-Danlos syndrome: Two case reports and a review of literature. Medicine (Baltimore). 2018 Nov;97(45):e13115.

Types of Pain:

Nociceptive pain

Pain from actual or potential damage to non-neural tissue.

Neuropathic pain

Pain that arises from a lesion or disease of somatosensory nervous system.

Nociplastic pain:

Pain from a dysfunction of the nervous system.

Etiology

- Etiology poorly understood – combination of theories?
  - Vicious circle of repetitive soft tissue microtrauma
  - Generalized hyperalgesia – central sensitisation
  - Neuropathic pain: nerve compression, axonal neuropathy, nerve (sub)luxations, spinal cord compression, small fibre neuropathy
  - Fear of injury and pain (kinesiophobia) – related anxiety and depression

Young Age

Joint hypermobility

Multiple trauma

Dislocations

Predominantly Nociceptive Pain

Widespread MSK Pain

Arthralgias/Myalgias

Enthesopathies

Non-articular pain

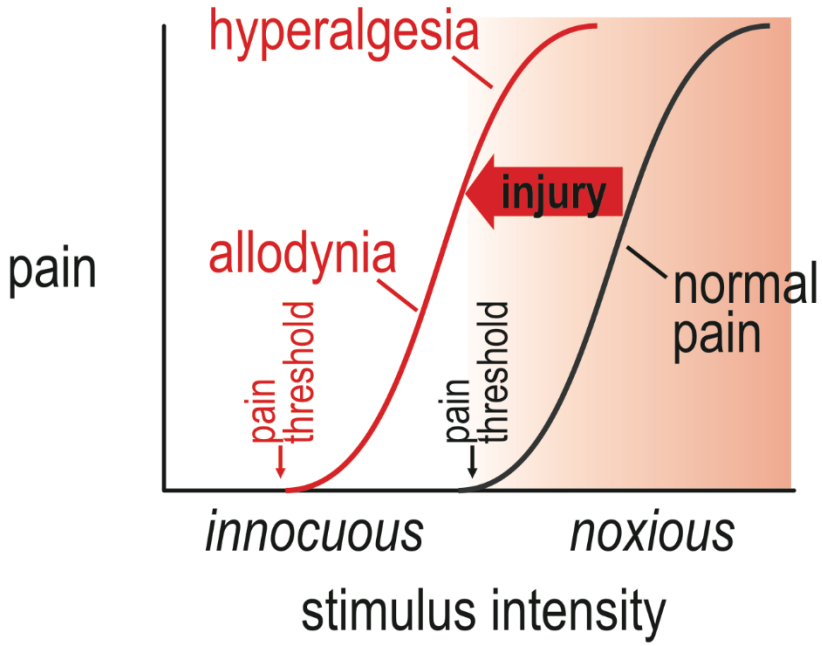
Neuropathic pain

Peripheral paresthesia

Signs of central sensitization

Central Sensitization

- Previous inputs which were below threshold now reach threshold and initiate action potential.
  - Stimuli that generally do not provoke pain can produce pain (allodynia).
  - Stimuli that normally provoke pain can produce pain of a higher intensity (hyperalgesia).
- Increase in spontaneous activity.
- Enlargement of the area in periphery where stimulus will activate neurons.



Multidisciplinary Pediatric Pain Treatment:

Pharmacological
- WHO ladder
- Multimodal treatment

PATIENT

Physical
- Ice/heat
- Core stabilizing
- Joint stabilizing
- Proprioception enhancing
- Posture re-education
- TENS

Psychological
- Pain Education
- Distraction
- Relaxation
- CBT, mindfulness, imagery
- ACT,
- Motivational interviewing

Pharmacological Treatments

Acute Pain Treatments:

- Acetaminophen
- NSAIDs
- Opioids (short term)
- Muscle relaxant (for acute muscle spasms)
- Topical treatments

Chronic Pain Treatments

- Acetaminophen
- NSAIDS
- Anticonvulsants
- Antidepressants
- Topical Medications
- Nerve blocks

GENERAL PRINCIPLES OF CHRONIC PAIN MANAGEMENT

Balanced Analgesia - More than one class of analgesic or adjuvant each working in a different way = better pain relief with fewer side effects

Start low and go slow – titration is critical to limiting side effects

Trial and error approach – inter-individual variation in response to treatment.

Set realistic expectations: improve function with minimal side effects, and reduce pain.

Physiotherapy Management of Pediatric Hypermobility and Ehlers-Danlos Syndrome

Jessie Young

Registered Physiotherapist

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The Hospital for Sick Children

Toronto, Canada

Project ECHO®

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Declaration of disclosure

- Nothing to disclose

Acknowledgement

- Brianne Burton
  Registered PT
  EDS/CTD Program
  The Hospital for Sick Children

Learning objectives

1

Identify
Physiotherapy (PT) principles based on personal clinical experience in a pediatric setting

2

Discuss
Components of PT treatment for children and adolescents with symptomatic joint hypermobility

3

Understand
Real examples of PT exercises that address symptomatic joint hypermobility in the pediatric population

When to consider PT

Asymptomatic Hypermobility

vs.

Symptomatic Hypermobility

Personal clinical experience

"My pain is all over. I feel it in my hips, thighs, knees, ankles, feet, back..."

"I used to love basketball but since my shoulders started popping out I can't play anymore."

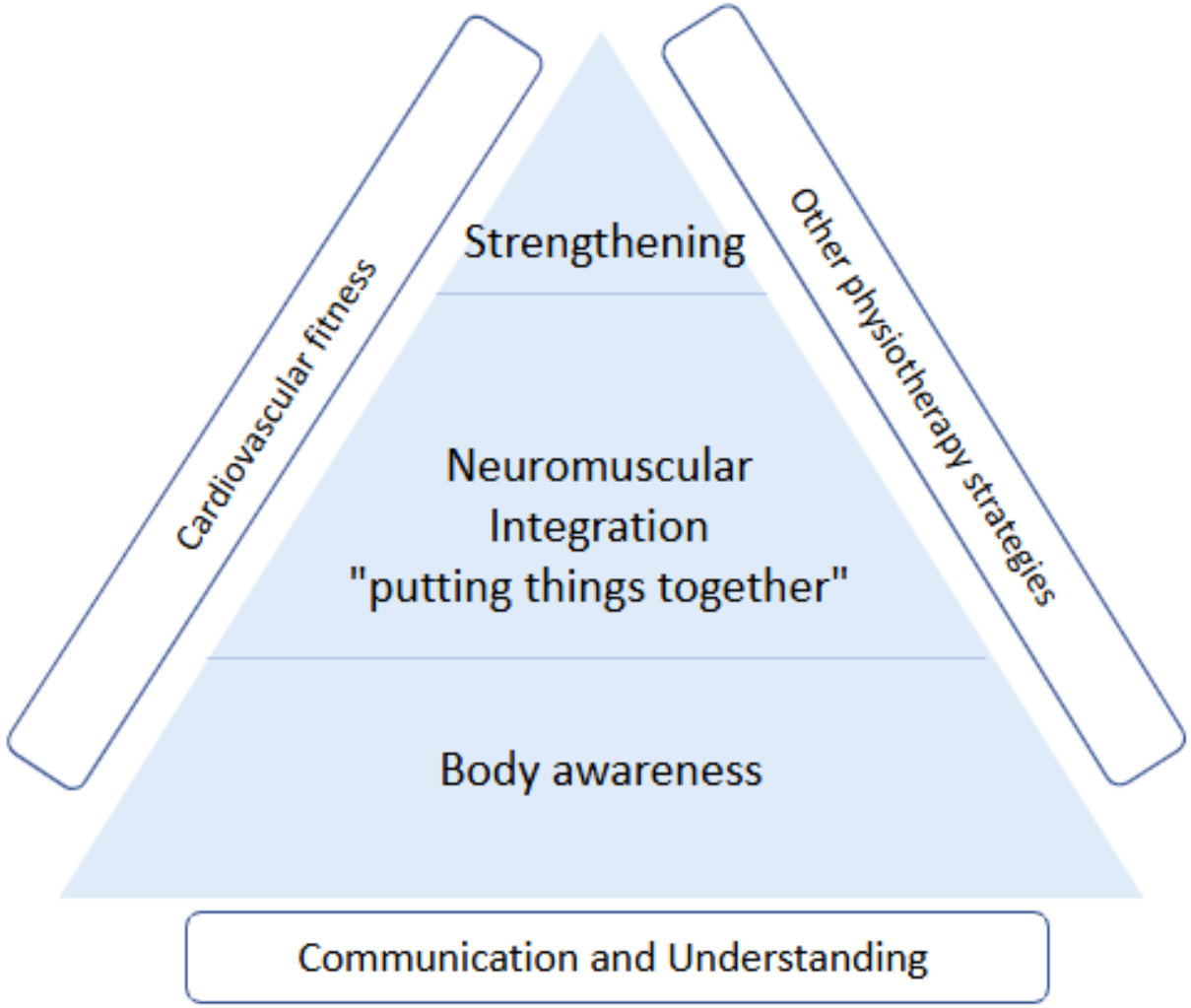
"When I have joint pain I just try to ignore it and tough it out."

"My child is very clumsy and falls down often. I am worried they will get seriously hurt."

"I need a cane to walk for longer than 30 minutes."

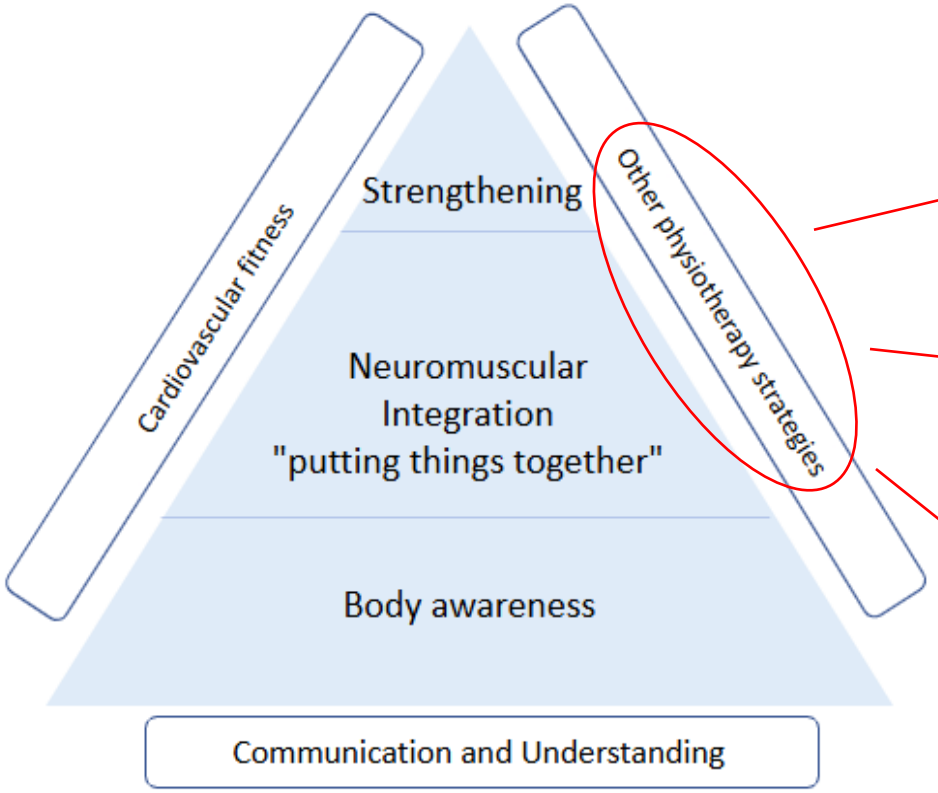
"I've tried physio before but I never really did my exercises at home."

PT Treatment: Elements to consider



PT Treatment: Elements to consider

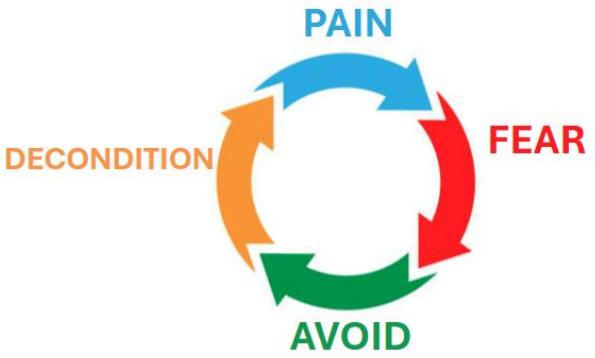
FOR CHILDREN AND ADOLESCENTS



Involve patient in decision making and goal identification

Relate exercises to meaningful activities. Make it fun!

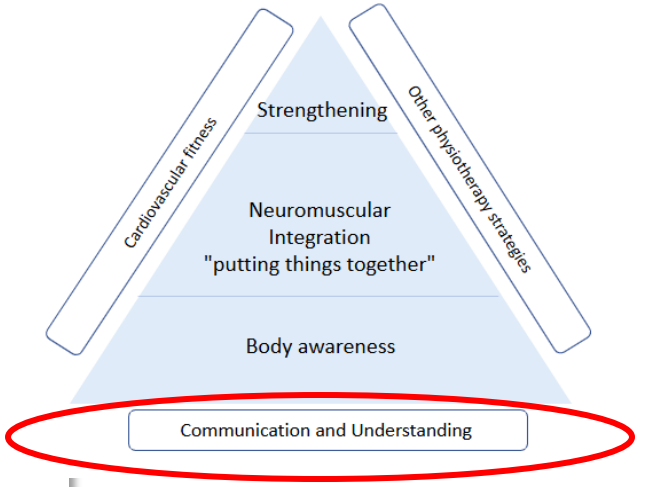
Eliminate fear of movement. Get moving!



Communication and Understanding

- Communication is key
- What is causing joint or muscle pain?
- Can this be managed or prevented?
- What are your goals?
  - Keep up with peers
  - Resume participation in meaningful physical activities
  - Reduce pain
  - Prevent injuries

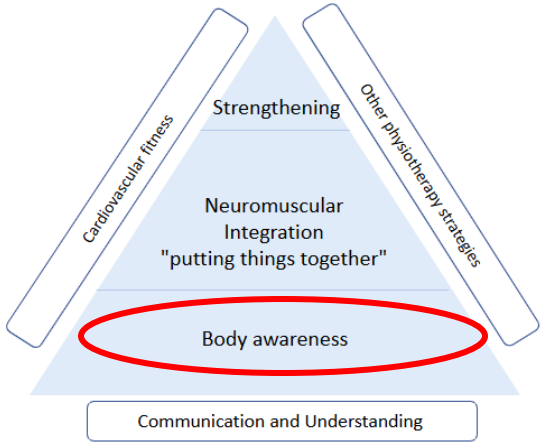
….And what can you do to reach your goals?



Body Awareness (Proprioception)

- Definition: Proprioception refers to one's sense of their body position in space

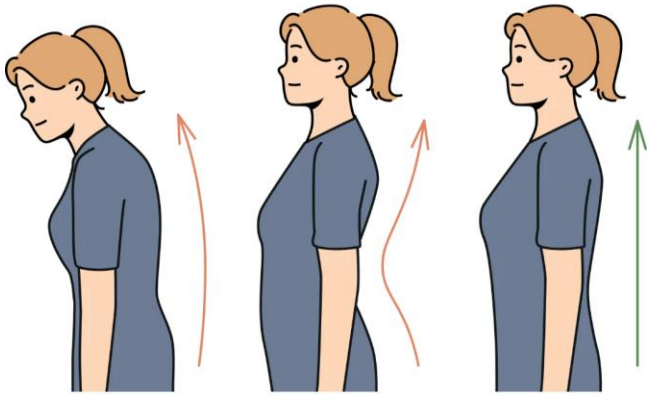
- According to the research,
  - Symptomatic joint hypermobility → decreased proprioceptive acuity
  - PT treatment → increase proprioceptive acuity + decrease pain

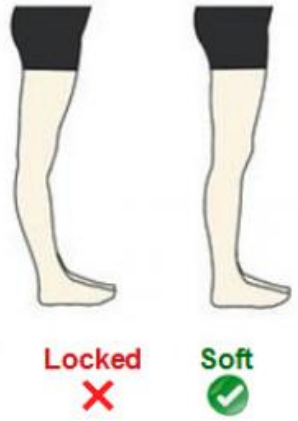


[Schubert-Hajlmarsson et al., 2012; Smith et al., 2013; Fatoye et al., 2009; Sahin et al., 2008; Ferrell 2004]

Basic proprioception training: Examples

Resting Posture

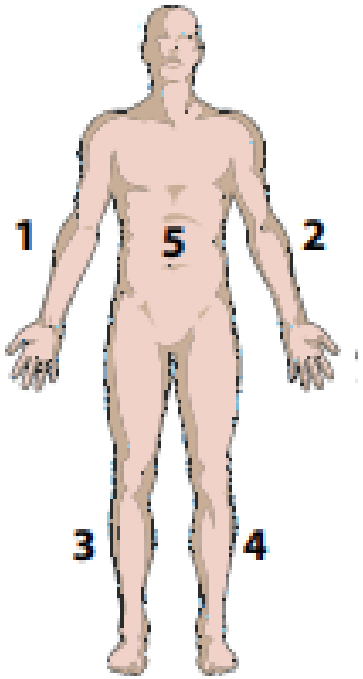




Muscle Tension Education

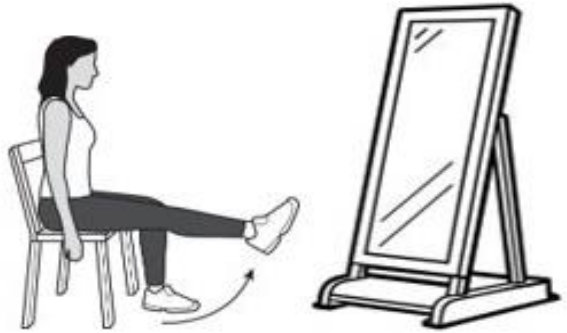
Contract / Relax

Grading (100% vs. 50%)



[Modified from psychologytools.com]

Visual and Sensory Feedback



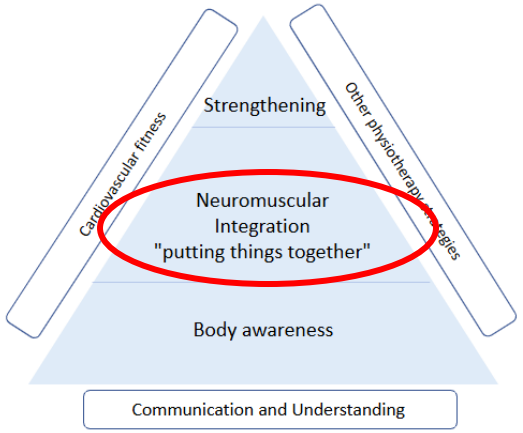
Balance

Proprioception aids and cues: External Supports

Barss et al 2018; Ehrstrom et al 2018; Zamporri et al 2018; Dupuy et al 2017*

Neuromuscular integration: "Putting things together"

- Definition: Neuromuscular integration describes the ability of the nervous system and the muscular system to work together to produce coordinated movement
- This requires having good body awareness as a foundation
- You need to be able to activate the right muscles at the right time
- Movements are more complex, functional, transferable



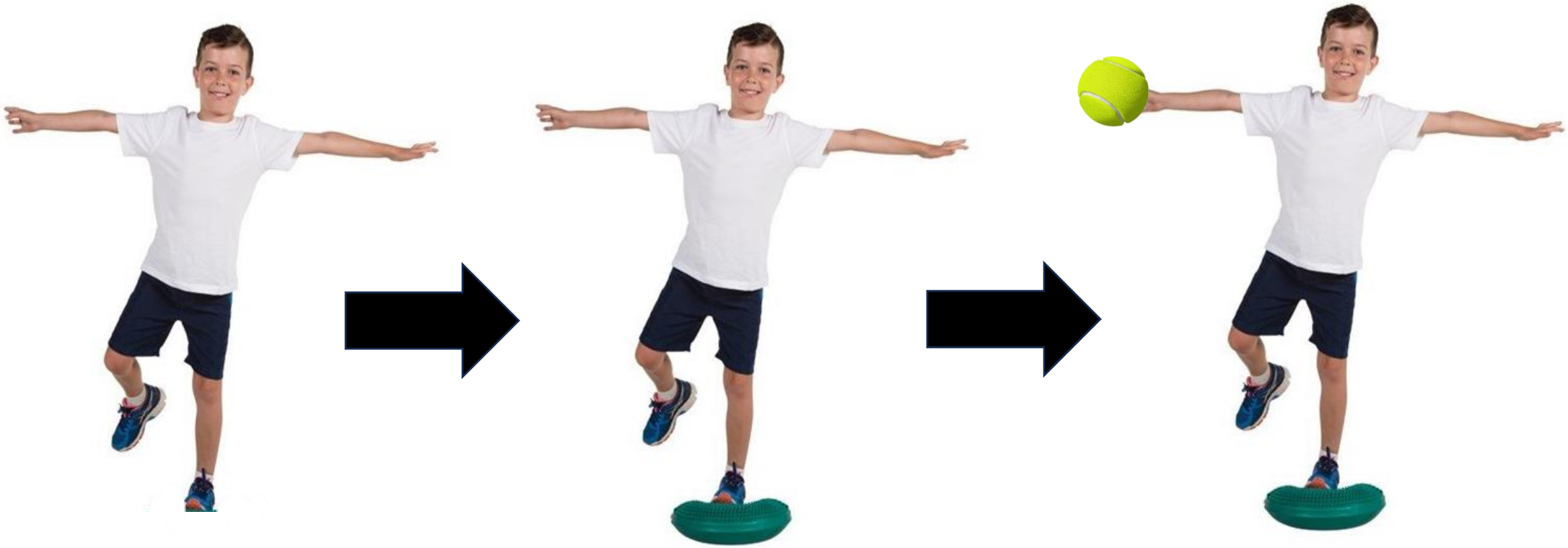
Neuromuscular training: Example

Object Manipulation

Balance and Coordination

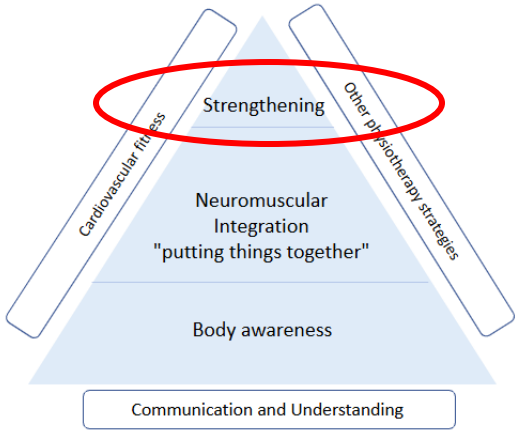
Sensory feedback

Neuromuscular training: Example



Strengthening

- Strengthening muscles that cross a joint will increase the joint's stability and improve your control of movement
- You need to have a strong base to move off from before strengthening distal muscles (core strength)
- To get the best results from strength training you must first address body awareness (proprioception) and neuromuscular integration

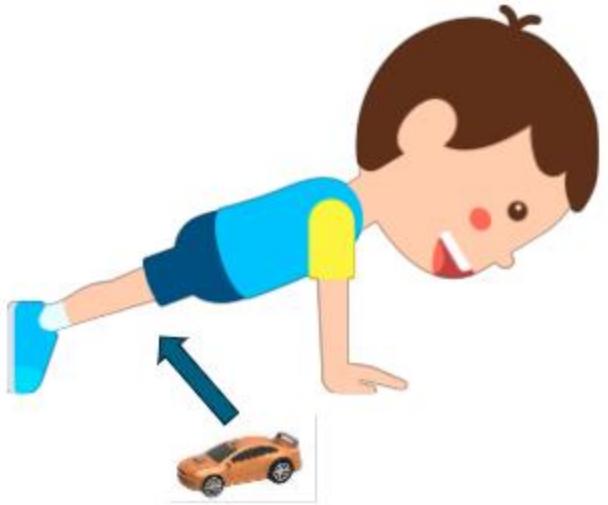


Strength training: Examples

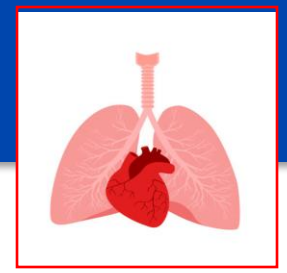
Body weight resistance

TheraBand resistance

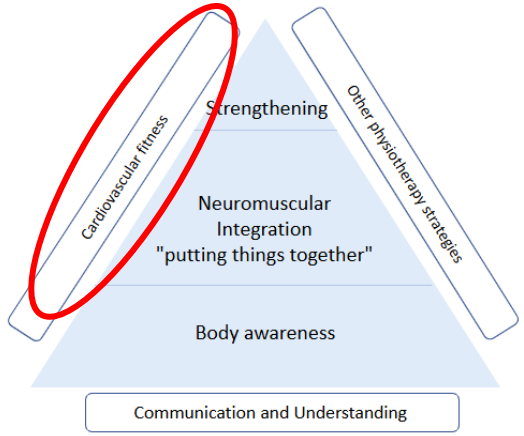
Core strengthening



Endurance training



- Definitions:
  - **Cardiovascular "Cardio" endurance** is the ability of the heart and lungs to supply the body with oxygen, to be able to perform physical exercise for longer
  - **Muscular endurance** is the ability of a muscle or group of muscles to remain active (contract) for longer



- *Goal*: last longer in desired physical activities

- *Key*: decrease intensity , increase time

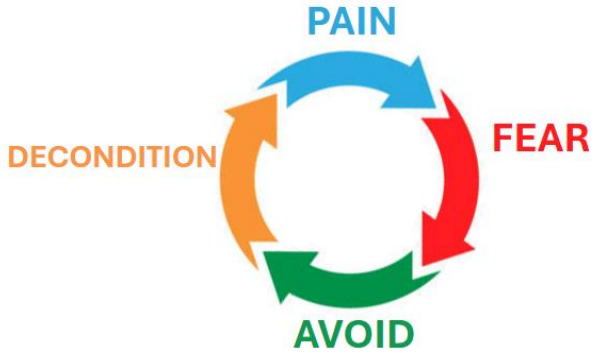
- Symptomatic joint pain

  → activity avoidance

  → deconditioning of both cardiovascular and muscular endurance

  → walk less far, stand less long, etc.

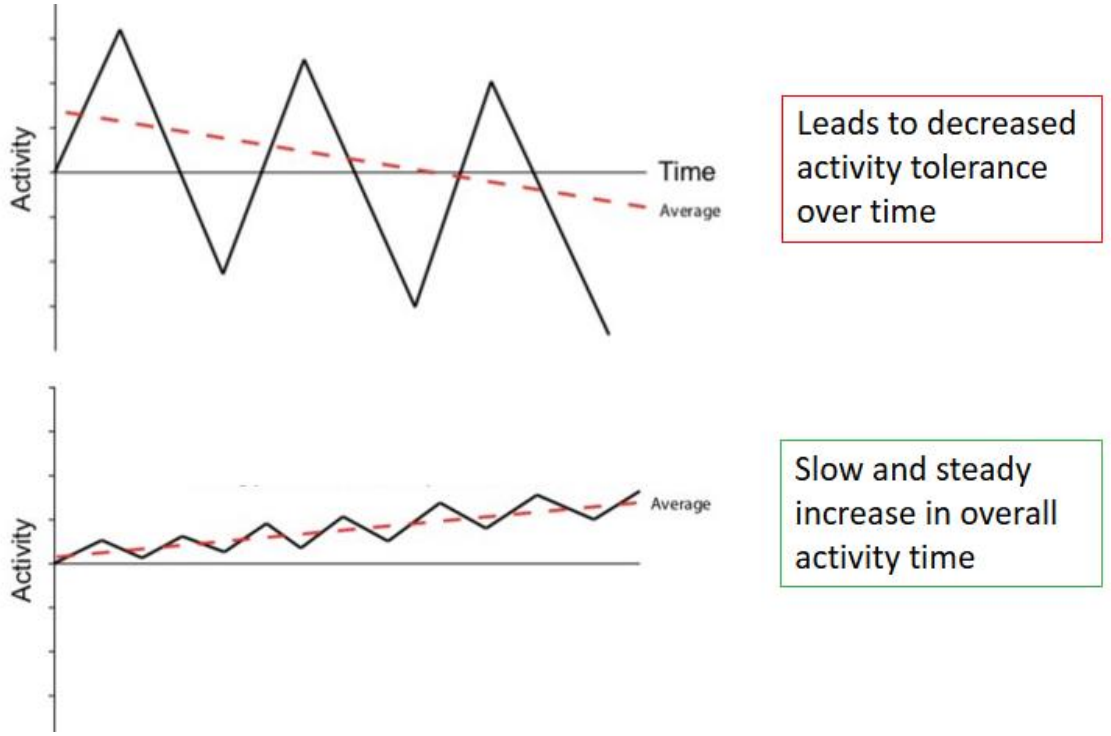
  → can't participate in desired activities



Endurance training: Examples

Low impact activities

Pacing

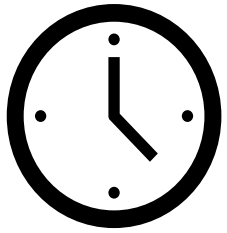


[Modified from psychologytools.com]

Diving Deeper: Hypermobility and Muscular Endurance

Joints are looser
so muscles need to stay
"on" more regularly
to maintain posture
or control movement.

Muscles that are "on" for
more of the time will
fatigue more easily



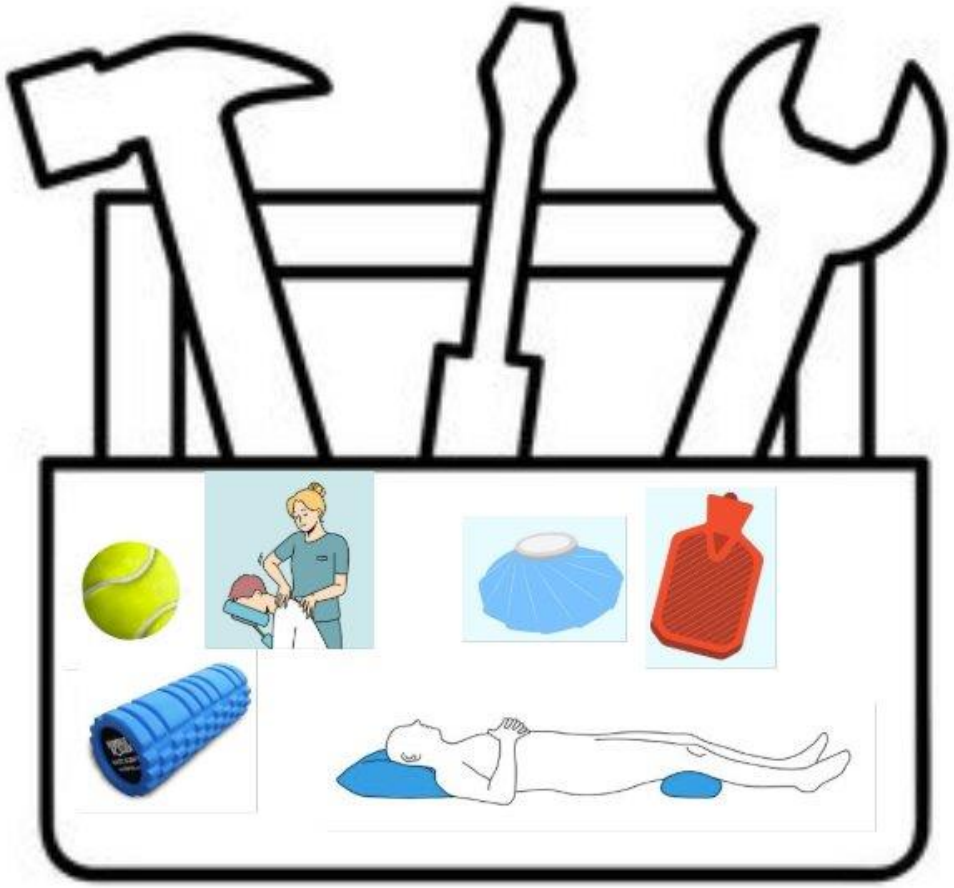
The elasticity of muscles
comes from collagen and
elastin, proteins that there
are less of in EDS and
hypermobility disorders.

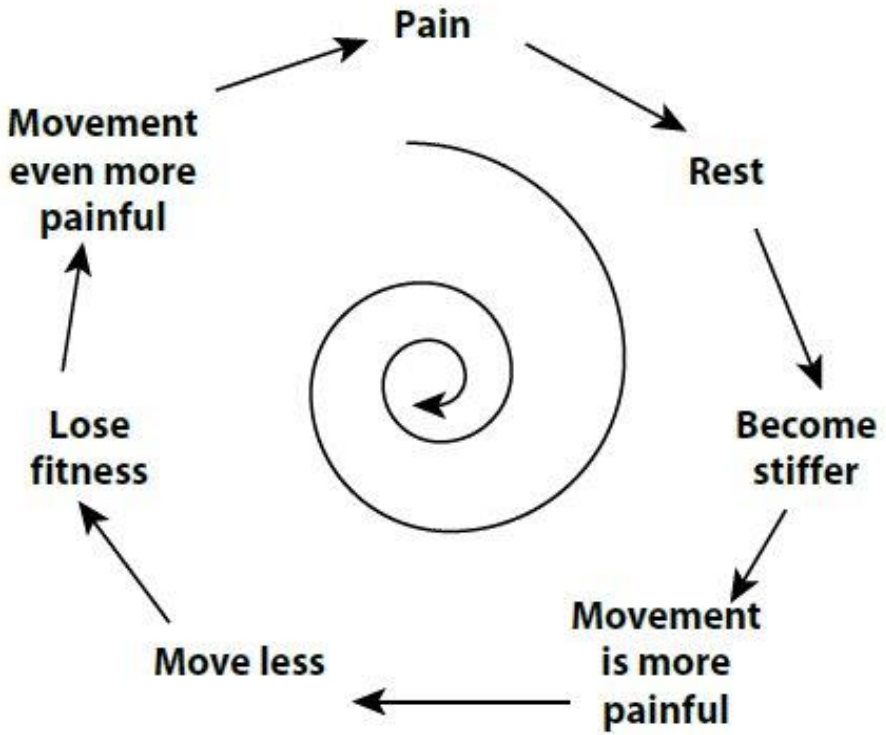
Less natural recoil results in
greater energy demand on muscles
and therefore they
fatigue more easily

Very flexible people may rely
more on their range of motion
than their strength for basic
resting posture.

Example: Locking out knees to stand
If your leg muscles aren't trained to
help you stand, they will
fatigue more easily when the
muscles are challenged

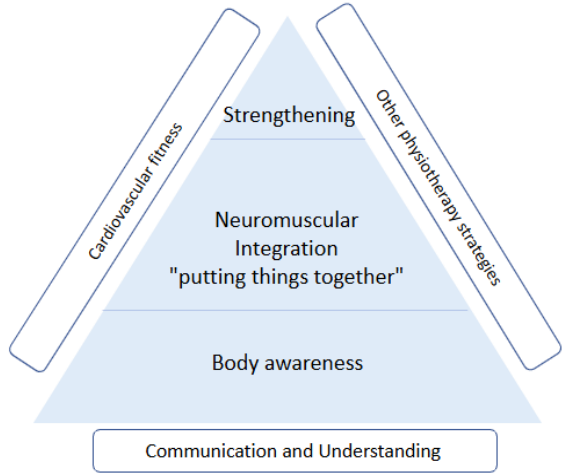
Pain management education





[Modified from psychologytools.com]

Summary



- Start simple: body awareness, posture, balance
- Increase complexity of exercises over time
- Strength training for increased joint stability *internally*
- Braces, K-tape, other supports for increased joint stability *externally*
- Endurance training: low impact and pace yourself
- Manage even low levels of pain to decrease sedentary time
- Make exercise fun, engaging, easily replicable for children!

Thank You

- Questions?