Hypermobility Syndromes

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A little about me...

- Physical Medicine & Rehabilitation/Physiatry
- Trained at University of Pittsburgh Medical Center
- Private practice Chicago
- "Nonoperative orthopedics"
 - PT management
 - US diagnosis, injections
 - Corticosteroid injections
 - PRP, prolotherapy
 - Spine injections
 - etc

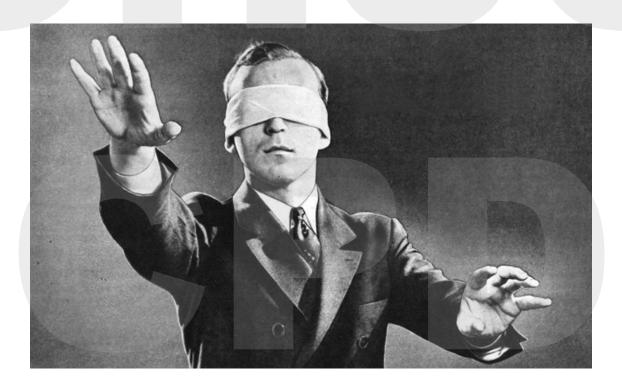


No disclosures



Well, one thing to disclose

Low evidence zone





Why discuss it?

- Underdiagnosed syndrome
- Online forums, support groups
- "Medicalization"
- We don't talk about it much



- Hypermobility syndromes
- Epidemiology
- Diagnosis
- General Complaints
- MSK Complaints
- General Treatment Principles
- Take Home Points



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Hypermobility Syndromes^{1,2}

- Classical Ehlers Danlos Syndrome (EDS)
 - Poor wound healing, "papyraceous" skin
 - Bruising
 - Joint hypermobility
 - AD type V collagen defect
- Hypermobile type (hEDS) or hypermobile type (h
 - Mild skin involvement "velvety
 - Unclear genetics AD likely
 - Disordered collagen
 - Female predominant
 - Most common

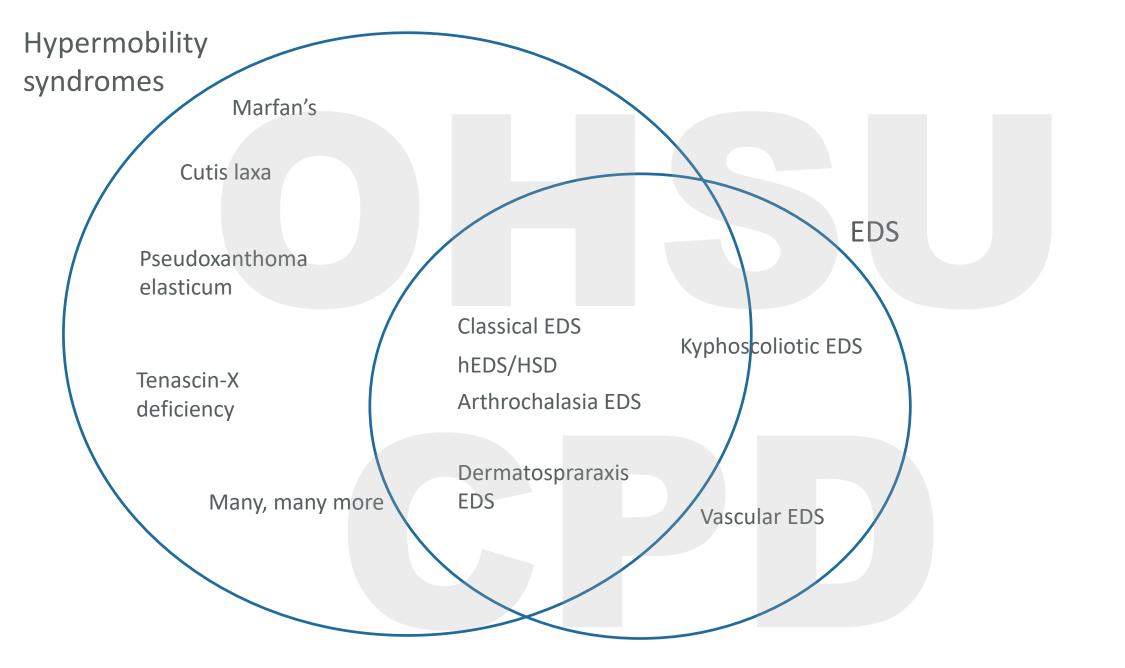




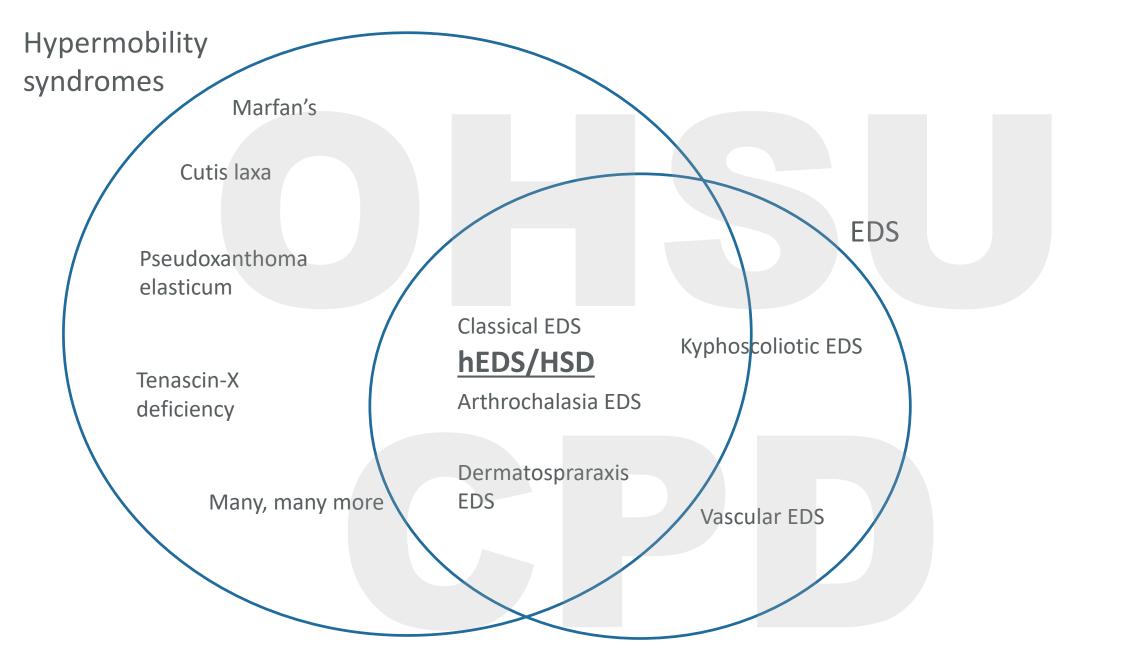
Hypermobility Syndromes^{1,2}

- Vascular type EDS
 - Dangerous, bowel and artery rupture
 - Skin/joint less severe
 - Bruising
 - AD, type III collagen defect
- Kyphoscoliotic type EDS
 - Ocular globe fragility
 - Severe scoliosis
 - Skin/joint laxity
 - AR











Hypermobile type EDS Hypermobility spectrum disorders

Familial Articular Hypermobility Generalized joint hypermobility Joint hypermobility syndrome Hypermobile type EDS



Variable Expression²⁶

- Poorly understood pathogenesis, component of pleiotrophy
- Modifiers strongly related to MSK features
 - Sex
 - Mechanical forces
 - Lifestyle
 - Job
 - Accidents
- "Secondary effects mediated by joint hypermobility"



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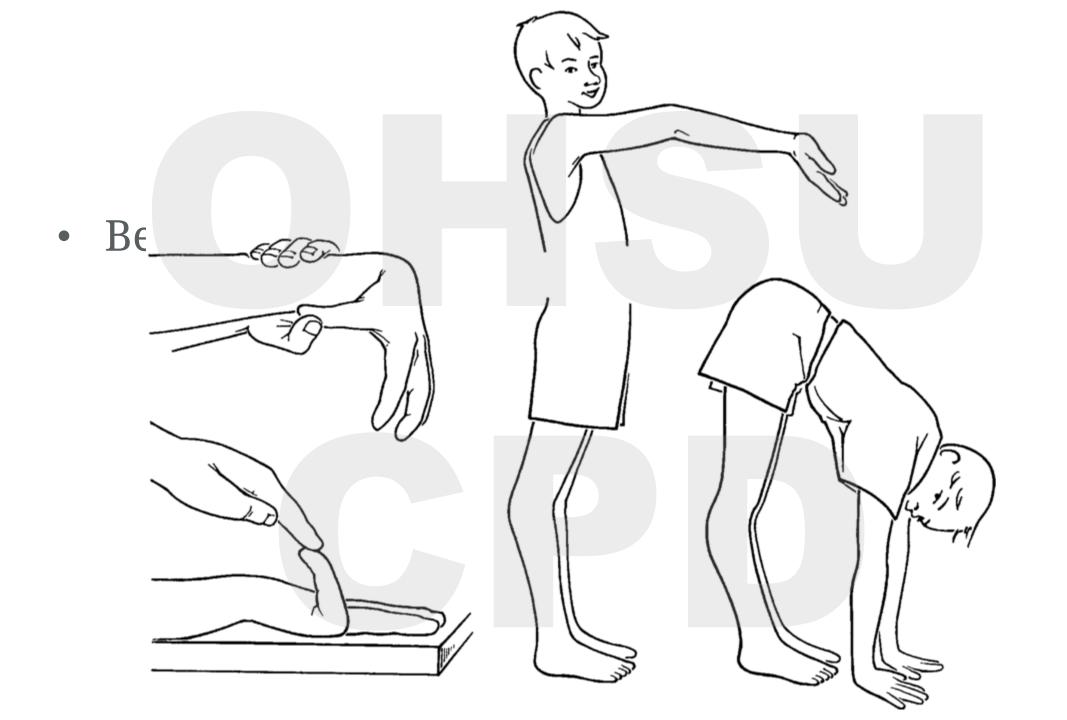
hEDS Epidemiology^{3,4}

- Overall hypermobility 10-20% population
 - Children/adolescents
 - Female
 - Asian
 - West African
 - African American > Caucasian
- Hypermobility + pain (HSD) = \sim 3% in UK population
 - Estimated 1/500 overall
- Reduced prevalence hypermobility in older
 - ?scarring
 - ?age related



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



in reconstruction of the same	un discipinies to be	. doic to diagnose 205	-	Society:
Patient name:	DOB:	DOV:	Evaluator:	
The clinical diagnosis of hypermobile EDS	needs the simultaneo	ous presence of all crite	ria, 1 and 2 and 3.	
CRITERION 1 – Generalized Joint Hy	permobility			
One of the following selected: \$6\$ pre-pubertal children and adolescented \$5\$ pubertal men* and women* to age 5 \$4\$ men* and women* over the age of 5	0 Beighton S	icore:/9	LAN	16 8
If Beighton Score is one point below age- an □ Can you now (or could you ever) place you □ Can you now (or could you ever) bend you □ As a child, did you amuse your friends b □ As a child or teenager, did your shoulde □ Do you consider yourself "double jointe	our hands flat on the floo our thumb to touch your y contorting your body i r or kneecap dislocate or	or without bending your k r forearm? into strange shapes or cou	rnees? uld you do the splits?	et criterion:
CRITERION 2 – Two or more of the f	ollowing features (A, B, or C) must be pr	esent	
Feature A (five must be present) ☐ Unusually soft or velvety skin ☐ Mild skin hyperextensibility ☐ Unexplained striae distensae or rubae a without a history of significant gain or lead to the silateral piezogenic papules of the heel ☐ Recurrent or multiple abdominal hernia ☐ Atrophic scarring involving at least two seems of the pelvic floor, rectal, and/or uterine prolappredisposing medical condition ☐ Dental crowding and high or narrow pale of the pelvic floor, rectal, and/or uterine prolappredisposing medical condition ☐ Dental crowding and high or narrow pale of the pelvic floor, rectal, and/or uterine prolapse of the prolapse of the pelvic floor, rectal, and/or uterine prolapse of the pelvic floor, rectal, and/or uterine prolapse of the pelvic floor, rectal, and/or uterine prolapse of the pelvic floor, and the pe	oss of body fat or weight (s) iites and without the form use in children, men or no ate ore of the following: oth sides, (ii) positive thu	mation of truly papyraceou ulliparous women without umb sign (Steinberg sign) o	us and/or hemosideric scars as t a history of morbid obesity o	seen in classical EDS
Feature B Positive family history; one or more firs	t-degree relatives indep	endently meeting the cur	rent criteria for hEDS	
Feature C (must have at least one) ☐ Musculoskeletal pain in two or more lim ☐ Chronic, widespread pain for ≥3 months ☐ Recurrent joint dislocations or frank join	bs, recurring daily for at	t least 3 months		
CRITERION 3 - All of the following p	rerequisites MUST	be met		
Absence of unusual skin fragility, w				
 Exclusion of other heritable and acc with an acquired CTD (e.g. Lupus, R Criterion 2. Feature C of Criterion 2 	heumatoid Arthritis, etc	.), additional diagnosis of	hEDS requires meeting both F	Features A and B of
 Exclusion of alternative diagnoses of Alternative diagnoses and diagnost other hereditary disorders of the co dysplasias (e.g. osteogenesis imper molecular genetic testing, as indica 	tic categories include, but innective tissue (e.g. oth fecta). Exclusion of thes	ut are not limited to, neuro ner types of EDS, Loeys-Di	omuscular disorders (e.g. Beth etz syndrome, Marfan syndro	elem myopathy), ome), and skeletal





CRITERION 1 - Generalized Joint Hypermobility

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- □ ≥6 pre-pubertal children and adolescents
- □ ≥5 pubertal men* and women* to age 50
- □ ≥4 men* and women* over the age of 50





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

- Absence of unusual skin fragility, which should prompt consideration of other types of EDS
- 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.



- Hypermobility syndromes
- Epidemiology
- Diagnosis
- General Complaints
- MSK Complaints
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- Take Home Points



General Complaints^{2, 6, 7}

- Joint instability
 - Subluxation/dislocation (96.3%)
 - Joint pain (100%)
 - ITB syndrome, snapping
 - TMJ
 - Tendinopathy (25.9%)
- ?Early OA
 - Conflicting data
- ?Reduced bone density
 - Conflicting data



General Complaints^{2, 6, 8, 9,10}

- Fatigue, sleep disturbance (84%)
- Headache
- Myofascial pain
- Impaired proprioception
 - Fear of falling/increased incidence of falls (95% of adults with JHS/hEDS)



General Complaints^{2, 6, 8, 11}

- Psychiatric issues common (chicken/egg)
- Neuropathic pain without electrodiagnostic findings
 - ?Stretch
- Chronic pain
 - Severity >> expected from imaging
 - Repetitive microtrauma
 - Combo of central sensitization, impaired proprioception, weakness, kinesiophobia
 - Psych issues play a role



General Complaints^{2, 6, 12}

- Autonomic dysfunction
 - POTS, atypical chest pain
 - 88% dizzy/lightheaded, 40% presyncope,
 27% standing intolerance, 43% palpitations,
 100% 5+ orthostatic symptoms >6 months
- Cardiovascular
 - No clear associations/concerns



General Complaints^{6, 13}

- GI
 - Functional bowel disorders (up to 67%)
 - GERD, gastritis, delayed emptying
 - Refractory
- OB/gyn
 - Short labor/delivery, PROM, postpartum hemorrhage
 - Pelvic pain
 - Pain during pregnancy
 - Increased dysautonomia during pregnancy



Epidemiology Diagnosis

- General Complaints
- MSK Complaints
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- Take Home Points



MSK Complications²⁶

- Macrotrauma
 - Dislocation, subluxation, soft tissue injury
 - Damage to muscle, ligament, tendon, synovium, cartilage
 - Leads to acute pain, loss of function, needs acute treatment
- Microtrauma
 - Subtle/silent injury
 - May predispose to persistent or recurrent pain
 - ?OA



Shoulder

MSK Complications¹⁴

- **Multidirectional instability
- Reduced proprioception
- Altered neuromuscular control of glenohumeral joint AND scapula
- **Poor dynamic control -> increased translation
 - Inferior instability with abduction



Shoulder

MSK Complications¹⁴

- Primary direction of instability common
- Diagnosis
 - Sulcus sign
 - Rule out cervical pathology
- Management
 - PT, but poor evidence
 - Rockwood (old), Derby (too intense), Watse program
 - Surgery: goal to reduce capsular volume
 - Inferior capsular shift and capsular plication
 - Thermal capsulorrhaphy (worst)



Hand/Wrist MSK Complications¹⁵

- Laxity at CMC -> ?OA
- Notable for:
 - Trapeziometacarpal arthritis (16%)
 - Sublux (66%)
 - Dislocation (29%)
- Not as commonly involved as other joints



MSK Complications^{16, 17, 18}

- Greater trochanteric pain syndrome
 - Likely due to IT band dysfunction
- Impingement common
- Extreme ROM -> FAI without pincer/cam
 - Microinstability exacerbates
- Mainstay: PT
- Surgical:
 - Plication, labral repair/debridement, femoral resections, psoas tenotomies



Knee

MSK Complications¹⁵

- Associated with ACL injury
 - Knee hyperextension -> 5x greater risk than matched controls
 - Laxity remains after ACL repair
- Rarely radiographic changes
- Poorly associated with OA
- Patients still very commonly complain of pain
- Mainstay: PT

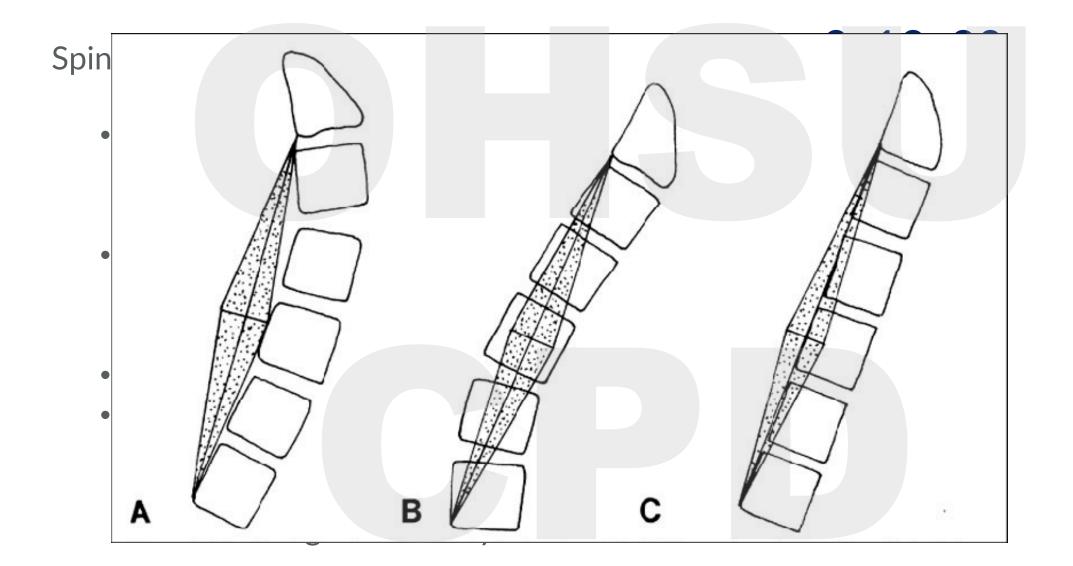


Ankle

MSK Complications¹⁵

- Midfoot laxity in ~93%
- Up to 2x risk of sprain
 - Midfoot collapse in gait -> instability -> strain
- Mainstay: PT
- Surgery: consider autograft or allograft for stabilization







Spine

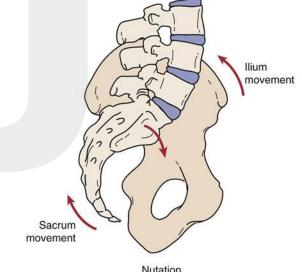
MSK Complications^{9, 19, 20}

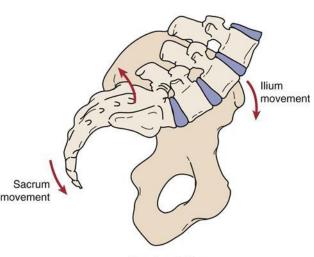
- Lumbar spine
 - Less associated
- Increased compression fractures in EDS (classical and hEDS)
 - Similar DEXA findings with control
- Treatment for all of spine:
 - PT for stability, positioning
 - Bracing as needed
 - Trigger avoidance
 - Surgery: for AAI and myelopathy/radiculopathy, but unknown adjacent level disease occurrence



Sacroiliac MSK Complications^{21,27}

- Imbalance/asymmetry -> pain
 - With motion only
- Increased laxity in pregnancy
 - Consider bracing
- Treatment
 - Manipulation/modalities, strengthening, motor control
 - Correct adaptive changes
 - Establish HEP
 - Prolotherapy? Steroids?





Headache

MSK Complications²³

- Multifactorial
 - Migraine, mixed chronic headache most common
 - Intracranial and peripheral vessel involvement
 - Dysautonomia
 - Cervical MSK issues (overuse, facet)
 - TMJ
 - ?CSF leak
 - Chiari 1 malformation
 - Injury susceptibility
- Treatment: investigate and pursue source; single treatment failure common



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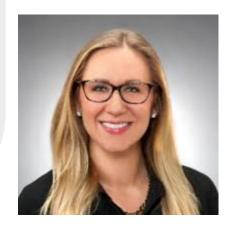
General Treatment Principles^{6, 10, 24}

- PT is essential
 - Modalities
 - Alexander technique
 - Low resistance, graduated increase
 - Closed chain; avoid open chain
 - Can use hypermobility
 - Proprioception/balance work
 - Pelvic floor therapy
- Exercise
 - Low evidence, unclear what kind is best
 - Appears to help



PT perspective

- **Isometrics** to strengthen without putting pressure on joints
- Mirror feedback for **posture and training of body awareness** (often these patients use compensatory strategies to stand, walk, etc that actually lead to more pain and damage one example is standing with locked knees)
- Mirror and therapist feedback during basic movement patterns (getting out of chair, reaching for something) to demonstrate potentially damaging habits. We use something called **Alexander Technique**, but also just teach **core stability and concepts of posture**)
- **Core stability** exercises that do not put pressure on joints (swiss ball activities, yoga, tai chi)
- **Positioning** interventions (supportive pillows for sleep, lumbar support when sitting, ergonomics, etc)
- Pacing concepts, listening to the body, flare management and developing of a plan for when pain is higher than baseline
- Coping strategies surrounding the management of chronic conditions, acceptance, active versus passive coping, meditation and relaxation (often these patient have some weak, and some excessively tight muscles)



Megan Driscoll



General Treatment Principles^{6, 10, 24}

- Bracing prn
- Medication think layers
 - NSAIDs/COX-2, topicals, muscle relaxers, neuropathics
 - Magnesium oral/topical for spasm
 - Opioids last resort
- Injections
 - Corticosteroid, glucosamine/hyaluronic acid, prolotherapy
 - ?Sclerotic agents
- Surgery expect mixed results
- Autonomic dysfunction
 - Greatly increase H2O and salt intake
 - LE strengthening, behavioral modification
 - Fludricortisone, midodrine



Take Home Points²⁵

- Psychiatric, neurologic, autonomic, AND musculoskeletal
- High impact on HRQoL
- High likelihood of seeking care
- High utilization, high medication use
- Pain rarely improves greatly
 - Function sees the gains
- Recognition is key
- Multidisciplinary team is essential for long term management



Questions?



Thank You!



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