**Joint Hypermobility**

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**Pathophysiology**

- Unclear
- Common Hypotheses:
  - Biomechanical
  - Localized biomechanical overload during activity
  - Joint pain/instability → microtrauma, compensation for overload in other MSK areas
  - proprioception altered
  - Deconditioning
  - Generalized hyperalgesia
  - Up-regulation of central pain processes
  - Decreased responsiveness to lidocaine and opioids

**Joint Hypermobility Syndrome**

- Increased range of motion beyond its normal range (recognizing that range of motion is to be compared to sibling or other) 
- 30%-40% general population
- Not clear if Joint Hypermobility Syndrome and Ehlers-Danlos Hypermobility type are the same
- Can be associated with musculoskeletal pain
- Easier/more frequent joint dislocations
- Probable early degenerative arthritis
- May also have muscular pain
- Important to recognize
  - Reassurance to patient - no evidence for more serious disease (e.g., cancer)
  - Give a name/reason to the patient for what they are feeling - learn there is more about it and avoid causing anxiety
  - Prevent unnecessary laboratory tests or X-rays
  - Rarely, identify a more serious cause of joint hypermobility

**Screening Questions**

- Questionnaire
  - 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 kneecap or shoulder dislocate on more than 1 occasion?
  - Do you consider yourself 'double-jointed'?

- ≥ 2 are positive, Sensitivity 84%, Specificity 80%

**Diagnosis**

- Beighton Score
  - Place hands flat on floor
  - Hyperextend elbow > 10°
  - Hyperextend knee > 10°
  - Bend thumb to forearm
  - Dorsiflex pinkie > 90°

1 point for each
9 points possible

**Brighton Criteria**

- Major
  - Beighton score of ≥ 4
  - Arthralgia for ≥ 3 months in ≥ 4 joints
- Minor
  - Beighton score of ≤ 4
  - Arthralgia in ≥ 4 joints, back pain/loose/pulled/strained
  - ≥ 1 soft tissue problems
  - marfanoid habitus (tall and slender)
  - Skin: striae, hyperextensibility, thin skin, abit scarring
  - Eye: drooping eyelid, myopia, antimonogalous slant
  - Venous veins, hernia or uterine/rectal prolapse

- Diagnosis
  - ≥ 2 major
  - ≥ 1 major and a minor
  - ≥ 4 minor
  - 2 minor if affected 9th degree relative

- Exclude Marfan’s and EDS criteria

- Caveat: Criteria designed for epidemiology research
  - Gives organized approach to pt
Differential Diagnosis
- EDS classic types
- Marfan’s Syndrome
- Osteogenesis Imperfecta
- Others
  - Loey-Dietz
  - Arterial Tortuosity Syndrome
  - Lateral Meningocele Syndrome

Ehler’s Danlos Types
- **Classic**: Skin hyperextensibility w widened atrophic scars and joint hypermobility
- **Vascular**: Arterial, intestinal, uterine fragility with (catastrophic) rupture
- **Kyphoscoliotic**: Scoliosis at birth w scleral fragility joint hypermobility
- **Arthrochalasia**: Recurrent, severe joint subluxations w severe joint hypermobility
- **Dermatosparaxis**: Severe skin fragility

Ehler’s-Danlos Hypermobility Type (III)
- Joint Hypermobility
- Soft, thin stretchy skin with thin scars
- Family history may be present
- Systemic involvement
  - Musculoskeletal pain (often labeled fibromyalgia)
  - Nonanatomic
  - Post-exertional worsening
  - Autonomic Dysfunction
  - Postural tachycardia syndrome (POTS) – most common
  - Gastrointestinal dysmotility
  - Constipation
  - Rectoceles
  - GI overlap with GU
  - Nausea, abdominal pain, reflux, bloating

Osteogenesis Imperfecta
- Joint hypermobility
- Susceptibility to bone fractures
- Blue sclera
- Opalescent dentine
- Sensorineural deafness

Marfan’s Syndrome
- Hereditary connective tissue disease
- Autosomal dominant
- FBN1 gene mutations (encoding fibrillin 1 protein)
- Manifestations
  - Cardiovascular – aortic dilatation or aneurysm
  - Skeletal – joint hypermobility + Marfanoid Habitus
  - Ocular – ectopia lentis
  - Others
- Marfanoid Habitus:
  - Tall, thin
  - Arachnodactyly
  - Dolichostenomelia: arm span: height ratio ≥ 1.03

Marfan’s Syndrome (MFS): Diagnostic Criteria
- (+) Family History
  - Ectopia Lentis (+) = MFS
  - **Systemic** ≥ 7 = MFS
  - Aortic diameter ≥ (mm)
    - > 20 years, Z ≥ 2 = MFS
    - < 20 years, Z ≥ 3 = MFS
- (-) Family History
  - Ao (Z ≥ 2) + EL = MFS
  - Ao (Z ≥ 2) + FBN1 = MFS
  - Ao (Z ≥ 2) + Systemic (≥ 7) = MFS
  - EL + FBN1 = MFS

  - Z age<40 years = [measured aortic root diameter - (0.97 + 1.12 × BSA)] / 0.24
  - Z age>40 years = [measured aortic root diameter - (1.92 + 0.74 × BSA)] / 0.37
Scoring of Systemic Features

- Thumb and wrist sign – 3
- Wrist or thumb sign – 1
- Pectus carinatum – 2
- Hind foot deformity – 2
- Pneumothorax – 2
- Dural ectasia – 2
- Protrusio acetabuli – 2
- Reduced upper segment/lower segment and increased arm/length ratio and no severe scoliosis – 1
- Scoliosis or thoracic kyphosis – 1
- Reduced elbow extension – 1
- Facial features (1/3) – (dolicocephaly, enophthalmos, down-slanting palpebral fissures, malar hypoplasia, retrognathia)
- Sinus stenosis – 1
- Myopia > 3 diopters – 1
- Mitral valve prolapse – 1
- Pectus excavatum or chest asymmetry – 1
- Plain pes planus – 1

Others: Uncommon to Rare

- Loeys-Dietz: autosomal dominant
- Aortic aneurysm
- Hypertelorism
- Bifid uvula or cleft palate
- Arterial Tortuosity Syndrome: Autosomal dominant
- Tortuosity and elongation of large and medium-sized arteries
- Tendency for aneurysm formation
- Lateral meningocele Syndrome
- Widespread spinal lateral meningeal cysts protruding through intervertebral spaces

Approach to a patient

- Physical Therapy
- Avoidance of activity causing dislocations
- Occupational therapy (if finger laxity present)
- Cognitive Behavioral Therapy
- Family history, suspected other disease (e.g. Marfan's, Osteogenesis Imperfecta) – referral to pediatric genetics clinic.

Take Home Points

- Joint mobility is a continuum
- Joint hypermobility syndrome: chronic joint pain (and more)
- Diagnose with Beighton and Brighton scores
- Look for other causes
  - EDS
  - Marfan's
  - Osteogenesis Imperfecta
- Therapy: Physical therapy (and others)
- Refer if suspicion for other causes is high

Questions?

- References