Hypermobile Ehlers-Danlos Syndrome in Children

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Today’s Presentation

- Overview of hypermobile EDS and new criteria in children
- Presenting symptoms and signs
- Management recommendations
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EDS, General Facts in Children

- Hereditary disorders of connective tissue
- Most due to defects in collagen but not all
- Most types have skin involvement/fragility and joint laxity
- Can have involvement of cardiovascular system
- Can be progressive and unpredictable
- Likely more common than 1:5000
- Most types autosomal dominant
EDS is Autosomal Dominant

New Criteria for Hypermobile EDS

- New criteria
  - Based more on adults than children
  - Very strict in order to help find the gene

- Three major requirements
  - Hypermobility: higher score if prepubertal
  - Systemic signs: many may not yet have developed or be to the maximum required
  - Musculoskeletal signs: many joints may not yet be involved
  - Family history is the same
Beighton Score

- Based on exam of some major joints
- Joint hypermobility? 1 point per item out of a total of 9
- Hypermobility = Beighton score at least 5/9 post puberty and at least 6/9 prepubertally

http://hypermobility.org/help-advice/hypermobility-syndromes/beighton-score
Beighton Score out of 9

Passive dorsiflexion 5th finger beyond 90 degrees? Right _ Left _ Total _
Passive apposition of thumbs to flexor forearm? Right _ Left _ Total _
Hyperextension of elbows beyond 10 degrees? Right _ Left _ Total _
Hyperextension of knees beyond 10 degrees? Right _ Left _ Total _
Forward flexion of trunk, knees extended, palms flat on floor? Point _
Total points _____

Can get an extra point if 2 of: if used to be able to do these, if there has been double jointedness, if can do tricks and/or had dislocations in the past.
Tricks with Joints
Systemic Signs - 5 or more needed. Some easier in children

- Unusually soft, velvety skin
- Unexplained striae
- Dental crowding and high narrow palate
Systemic Signs - some difficult to reach full requirement in children

- Unusually soft, velvety skin
- Unexplained striae
- Dental crowding and high narrow palate
- Skin hyperextensibility more than 1.5 cm on forearm
- Bilateral piezogenic papules
- Two or more atrophic scars
Systemic Signs - some much less common and unlikely in EDS

- Unusually soft, velvety skin
- Unexplained striae
- Dental crowding and high narrow palate
- Skin hyperextensibility more than 1.5 cm on forearm
- Bilateral piezogenic papules of heels
- Two or more atrophic scars
- Long arms
- Long fingers
- Recurrent or multiple abdominal hernias
Systemic Signs - some rare in children

- Pelvic floor, rectal or uterine prolapse
- Mitral valve prolapse
- Aortic root dilatation

- Thus very difficult to get to a systemic score of 5 or more!
- Since the new criteria: 50% HM EDS now hypermobile spectrum disorder
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A review of 192 EDS patients seen
- Female to male ratio is 62% to 38% or 1.6:1
- HM EDS to other (classical mostly) is 85% to 15%
- Age range 7 months - 21 years
- Race/Ethnicity: White 84%, Black 6%, Asian 2%, Latino 3%, Biracial 5%

Referrals
- Made by 82% Pediatrics, 11% Orthopedics, 7% Other
- Reasons for referral
  - 64% hypermobility, rule out EDS
  - 20% rule out Marfan syndrome
  - 16% other (i.e. HDCT, OI, etc)
Presenting Symptoms for Possible EDS

- Pain in more than one joint
- Recurrent pain in a joint
- Excessive pain after injuring a joint
- Hypermobility of joints
- Frequent subluxations
- Several dislocations
- Dizziness and/or fatigue
Musculoskeletal Complications

- Pain in joints
- Flexible joints
- Easily dislocated joints, 33% in our group
- Fractures, 18% in our group
Joint Findings

- Flat feet
- Tricks with joints
- Hypermobility
Skin Findings

- Stretchy skin
- Stretchy ears
- Cutis marmorata
- Soft, velvety skin
- Visible veins
Skin Complications

- Unusual scars
- Abnormal scarring 33%
- Stretch marks
- Easy bruising 28%
Other Findings

- Piezogenic papules
- Winged scapulae
- Pectus excavatum
- Scoliosis
GI Complaints

- Frequent abdominal pain
- Constipation 16% of our group
- Recurrent diarrhea
- Nausea, vomiting
- Poor bowel motility and dysfunction, 21% of our group
- Gastroesophageal reflux, 6% of our group
- Mast cell dysfunction, 4% of our group
Cardiac Complaints

- Dizziness when standing up
- Frequent light-headedness
- Feeling like about to faint
- Fainting
Cardiac Involvement

- Dilation of aortic root, 3% in our group
- Repairable before complications if found
- Recent study showed increased size of aortic root in 3% but with rare dilation at 15 years of age or more
- Risk for valvular involvement, especially mitral valve prolapse, 1.6% of our group

http://mjr.barnabasheart.org/
Postural Orthostatic Tachycardia Syndrome (POTS)

- In 13% of our group
- Orthostatic symptoms: dizziness, lightheadedness, weakness, change in vision, palpitations
- Children: increase in HR by 30-40 bpm within 10 min of standing up or children less than 13 years HR above 130
- Dysautonomia, 0.2% of our group
Neurologic Complaints

- Frequent fatigue
- Brain fog, confusion
- Decreased energy
- Need for increased sleep
- Hypotonia, 12% of our group
- Headaches, 20% of our group
- Migraines, 12% of our group
Brain and Spine

- Craniocervical junction abnormality, less than 1% of our group
- Chiari type I malformation, 3% of our group
- Full evaluation for this: upright brain MRI, upright cervical spine MRI in flexion and extension, and CT scan of the upper spine in neutral, rotation to the right, rotation to the left (specific centers specialize in upright MRI)
Behavior and Development

- Unrelated to EDS
- Behavior
  - ADHD, 14% of our group
  - Anxiety, 13% of our group
  - Depression, 7% of our group
- Autism spectrum disorder, 5% of our group
- Development
  - Delays/intellectual disability, 12% of our group
  - Learning disability, 4% of our group
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EDS, Joint Management

- **Activities:**
  - Curtail high impact, aerobic sports
  - Highly recommend swimming and other lower impact sports/exercises

- **Joints:**
  - Recommend physical therapy for core and limbs, consider warm water
  - Myofascial trigger point massage
  - Support for painful joints but not total immobilization
  - NSAIDs
Joint Management, cont.

- Feet and legs
  - Physical therapy
  - Foot support (eg AFO’s)
  - Continue exercising!

- Hands and arms
  - Splints for acute issues
  - Pen/pencil accommodations from OT
  - Ring splints for extra support

- School: Modified PE, 504 plan, self-regulation, accommodations
  - Wheelchair if needed, 3% in our group
EDS, Pain Management in Pediatrics

- Not well studied and needs to be
- Overall recommendations
  - Physical therapy, especially warm water
  - Regular strengthening of muscles, minimize contact sports and encourage swimming
  - Use Epsom salts in bath or foot soaks
  - Myofascial trigger point release
- Referral to PM&R physicians or Pain Clinic
- Medications: Analgesics, no narcotics, consider topical approach (eg Lidocaine, Indomethicin, Diclofenac); no compound creams below 15 years
- Use accommodations in school, handicapped parking for malls, be sensible
EDS, Cardiac Management

- Echocardiogram:
  - Initially in all children to assess aortic root size
  - Repeat in 3-5 years if normal through late teens
  - Refer to pediatric cardiology if abnormal

- POTS treatment:
  - Hydration plus electrolytes
  - Rising up slowly
  - Compression stockings
  - Exercises
EDS, GI Management

- Constipation, motility issues
  - Consider medications
  - Gastric emptying study if indicated
  - Refer to pediatric GI if severe

- Mast cell dysfunction
  - Tryptase level at time of involvement
  - Consider eliminating histamines from diet and/or identifying trigger
  - Medications if indicated
  - Refer to pediatric immunology if severe
EDS, Neurologic Management

- Chiari malformation/Craniocervical instability
  - Cervical collar
  - Cervical traction and/or surgical decompression can provide neurologic improvement

- Fatigue, poor sleep
  - Regular bedtime
  - No oversleeping
  - No caffeine
  - Special pillows
EDS, Behavior Management

- Validate that pain and symptoms are real
- Provide supportive reassurance
- Obtain school accommodations
- Consider family counseling and/or behavior counseling
- Mindfulness-based stress reduction
  - Yoga
  - Meditation
HM EDS and Current Studies

- Search for the genetic cause of HM EDS
- Ongoing evaluation to assess mitochondrial function; fatigue may be related to this
- Survey study GBMC and JHH to assess quality of life in the pediatric population with non-vascular EDS
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