Too tall or too flexible?
When is it a connective tissue disorder?

Sharan Goobie, MD, MSc, FRCP C
Too tall or too flexible?
When is it a connective tissue disorder?

*Presenter Disclosure:*

Sharan Goobie has **no** potential for conflict of interest with this presentation.
Objectives

- To recognize features of a connective tissue disorder
- Review common connective tissue disorders
- Review health complications of these disorders
Connective Tissue

- Supports, anchors and connects various parts of the body
- Collagen fibres – provide strength
- Elastic fibres – provide stretch

- Abundant throughout the body:
  - Skin
  - Tendons/ligaments
  - Bone/ cartilage
  - Blood vessels
  - Organs
Hypermobility

4-13% of general population with have hypermobility not associated with disease

So when do we worry?

- Pain
- Joint dislocations
- Poor wound healing (wound dehiscence)
- Collagen-vascular disease (aneurysms)
- Family history of sudden death or aneurysm
Connective tissue disorder assessment

**Measurements:**

- Height (>97\textsuperscript{th}%)
- Arm span to height ratio (>1.05 abnormal)
- Reduced upper segment to lower segment ratio
- Middle finger length (>97\textsuperscript{th}% arachnodactyly)
Connective tissue disorder assessment

Other skeletal features:

- Scoliosis
- Pectus caranatum or excavatum
- Pes planus
- Hindfoot deformity
- Hypertelorism (wide spaced eyes)
- Cleft palate, bifid uvula
Connective tissue disorder assessment

Flexibility:
- Beighton scale
  \[ \geq 5/9 = \text{hypermobility} \]
- Skin extensibility
  (dorsum of hand or forearm)
Connective tissue disorders to know

- Ehlers Danlos syndrome (EDS)
  - Classical
  - Hypermobility
  - Vascular

- Marfan syndrome

- Loeys Dietz syndrome
Classical Ehlers Danlos Syndrome

- Joint hypermobility
- Hyperextensible skin
- Poor wound healing
- Atrophic scars

Medical implications:
- Pain
- Recurrent dislocations/subluxations
- Surgical /wound care recommendations
- Mitral valve prolapse
- Aortic root aneurysms (less common)
- Pregnancy complications
Hypermobility EDS

- Joint hypermobility
- Absence of skin fragility
- Chronic pain, fatigue
- Orthostatic hypotension

Often misdiagnosed as fibromyalgia, chronic fatigue, hyperchondriasis, psychiatric disease

Medical implications:
- Pain management and joint stability!
Vascular Ehlers Danlos syndrome

- Thin translucent skin
- Easy bruising
- Arterial fragility
- Intestinal fragility
- Uterine fragility

Medical implications:
- Vascular dissection
- Intestinal or organ rupture
- Pregnancy complications
- Unexplained sudden/severe pain should be considered a life threatening emergency
Marfan syndrome

Medical implications:
• Aortic root dilatation / aneurysm
• Lens subluxation
• Joint dislocations
Clinical manifestations of MFS in other organ systems were critically evaluated for their specificity and diagnostic utility based on expert opinion and the available literature. Several of the “minor” criteria from the old Ghent nosology were eliminated, but the most selective systemic features were included in the “systemic score.”

<table>
<thead>
<tr>
<th>Feature</th>
<th>Value</th>
<th>Click to include</th>
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</thead>
<tbody>
<tr>
<td>Wrist AND thumb sign</td>
<td>+</td>
<td>3</td>
</tr>
<tr>
<td>Wrist OR thumb sign</td>
<td>+</td>
<td></td>
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<tr>
<td>Pectus Carinatum Deformity</td>
<td>+</td>
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<tr>
<td>Pectus Excavatum or Chest Asymmetry</td>
<td>+</td>
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<tr>
<td>Hindfoot Deformity</td>
<td>+</td>
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<tr>
<td>Plain Flat Foot</td>
<td>+</td>
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<tr>
<td>Spontaneous Pneumothorax</td>
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<td>Dural Ectasia</td>
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<td>Pubic Acetabulum</td>
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<td>Scoliosis or Thoracolumbar Kyphosis</td>
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<tr>
<td>Reduced Elbow Extension</td>
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Loeys Dietz syndrome

Medical Implications:
Arterial dissections
Making a diagnosis

- Clinical diagnosis
- Genetic Testing

Guide management

- Recommendations for echo and vascular imaging
- Exercise / weight lifting restrictions
- Surgical precautions
- Pregnancy precautions
- Physical therapy / pain management
Some connective tissue disorders have no warning signs!

- Familial thoracic aneurysm and dissection syndromes (TAAD)

- Consider autopsy and banking DNA in patients with sudden death or known dissection

- Aneurysms in more than one generation (especially if no other predisposing factors) – refer to Medical Genetics
Summary:

- Joint hypermobility is common, most is benign
- Connective tissue disorders can have serious health implications, ie: vascular involvement
- Most are autosomal dominant – implications for other family members
- Follow surveillance guidelines
- Acknowledge physical and psychological aspects
- Know the features of connective tissue disorders and signs of a dissection – you could save a life!
Questions?