Joint hypermobility is common. It affects 1-4% of people and is often familial. Joint hypermobility varies in severity from simple joint hypermobility to Hypermobility Ehlers Danlos Syndrome (hEDS). Most people with joint hypermobility can be managed through their GP and do not need a genetics assessment.

Please see the attached information and referral guidelines for patients with joint hypermobility. A small proportion of people with joint hypermobility do need to be seen by a clinical geneticist for assessment and consideration of genetic testing. This group usually has additional distinctive clinical features or “red flags.” Usually an echocardiogram (to rule out dilatation of the ascending aorta or mitral valve prolapse) and an eye examination by an ophthalmologist or experienced optometrist (to rule out lens dislocation or high grade myopia) will be needed to complete the assessment.

Patients with hypermobility who do not meet these referral criteria can be assessed clinically for hypermobility-type Ehlers Danlos syndrome (hEDS) using the guidelines provided in the attached information sheet.

Genetic testing for hypermobility-type EDS is not helpful as causative single gene mutations have not been identified in the vast majority of people with this condition. People with hypermobility benefit from physical, psychological and pain-based therapies and utilisation of local allied health, pain and psychological services for management may be considered.

Referral to a genetics service is appropriate for any person fulfilling the referral criteria as such individuals may have a different connective tissue disorder, other than hEDS. Where applicable, we would welcome a referral with details of your assessment and any relevant investigation or specialist medical reports for these individuals.

If you have any questions, you can contact the Paediatric and Reproductive Genetics Unit on 8161 7375.
GP Fact Sheet: Genetic Testing
Joint Hypermobility / Ehlers Danlos Syndrome

Guidelines for Referring People with Joint Hypermobility to Genetics Services

1. Assess for joint hypermobility (using the Beighton score)
   - Significant hypermobility present
     - Beighton score is considered significant if it is:
       - ≥ 6 in pre-pubertal children and adolescents
       - ≥ 5 in pubertal men and women to age 50; or
       - ≥ 4 in men and women over age 50

2. Assess for additional features suggestive of a heritable connective tissue disorder (see “red flags”)

3. Arrange echocardiography and ophthalmology assessment

4. Features of classic / vascular / rare EDS
5. No concerning clinical features or red flags
6. Features of Marfan or Loeys-Dietz syndrome

Clinical Genetics referral is not likely to be useful. Refer to appropriate medical and allied health professionals

Refer to Paediatric and Reproductive Genetics unit for assessment and advice

For more information
Paediatric Reproductive Genetics Unit
Women’s and Children’s Hospital
72 King William Rd, North Adelaide SA 5006
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Reviewed: March 2023

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1. Assess for Joint Hypermobility using the Beighton Score:

<table>
<thead>
<tr>
<th>Feature</th>
<th>Points</th>
<th>Score</th>
</tr>
</thead>
<tbody>
<tr>
<td>LITTLE FINGERS: dorsiflexion beyond 90 degrees</td>
<td>1 point each finger</td>
<td></td>
</tr>
<tr>
<td>THUMBS: apposition to flexor aspect of the forearm</td>
<td>1 point each thumb</td>
<td></td>
</tr>
<tr>
<td>ELBOWS: hyperextension beyond 10 degrees</td>
<td>1 point each elbow</td>
<td></td>
</tr>
<tr>
<td>KNEES: hyperextension beyond 10 degrees</td>
<td>1 point each knee</td>
<td></td>
</tr>
<tr>
<td>SPINE: hyper-flexibility</td>
<td>1 point</td>
<td></td>
</tr>
</tbody>
</table>

The Beighton score is considered significant if it is:
- ≥6 in pre-pubertal children and adolescents; or
- ≥5 in pubertal men and women to age 50; or
- ≥4 men and women over the age of 50.
2. For individuals with significant joint hypermobility as assessed in Step 1: Assess for additional features suggestive of a heritable connective tissue disorder and “red flags” – see below

<table>
<thead>
<tr>
<th>System</th>
<th>“Red Flag” clinical features</th>
</tr>
</thead>
<tbody>
<tr>
<td>Skin</td>
<td>Widened, atrophic scars</td>
</tr>
<tr>
<td></td>
<td>Marked skin hyperextensibility</td>
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<tr>
<td></td>
<td>Skin fragility</td>
</tr>
<tr>
<td></td>
<td>Thin, translucent skin</td>
</tr>
<tr>
<td></td>
<td>Redundant, sagging skin</td>
</tr>
<tr>
<td></td>
<td>Easy bruising (spontaneous or with minimal trauma)</td>
</tr>
<tr>
<td>Body habitus</td>
<td>Marfanoid body habitus</td>
</tr>
<tr>
<td></td>
<td>Distinctive facial features (thin lips, prominent eyes, narrow nose)</td>
</tr>
<tr>
<td>Skeletal</td>
<td>Progressive and severe scoliosis / kyphosis</td>
</tr>
<tr>
<td></td>
<td>Congenital bilateral hip dislocation</td>
</tr>
<tr>
<td>Vascular</td>
<td>Aortic (or other arterial) aneurysm / rupture / dissection</td>
</tr>
<tr>
<td></td>
<td>Young onset thoracic aortic dilation</td>
</tr>
<tr>
<td></td>
<td>Unexplained extensive varicosities at young age (varicose veins)</td>
</tr>
<tr>
<td></td>
<td>Bicuspid aortic valve</td>
</tr>
<tr>
<td></td>
<td>Mitral valve prolapse with regurgitation</td>
</tr>
<tr>
<td>Ocular</td>
<td>Lens dislocation</td>
</tr>
<tr>
<td></td>
<td>High grade myopia</td>
</tr>
<tr>
<td>Oral</td>
<td>Severe periodontal disease</td>
</tr>
<tr>
<td></td>
<td>Lack of attached gingiva</td>
</tr>
<tr>
<td>Other</td>
<td>Spontaneous pneumothorax</td>
</tr>
<tr>
<td></td>
<td>Cleft palate / bifid uvuvla</td>
</tr>
<tr>
<td></td>
<td>Bowel perforation</td>
</tr>
<tr>
<td></td>
<td>Uterine or other organ rupture</td>
</tr>
<tr>
<td></td>
<td>Muscle or tendon rupture</td>
</tr>
<tr>
<td>Family history</td>
<td>Family history of “red flags”</td>
</tr>
<tr>
<td></td>
<td>Family history of sudden, unexplained death</td>
</tr>
</tbody>
</table>
3. Assess for the presence of other features that suggest hEDS (see diagnostic check sheet below)

4. Referral to a Genetics service is appropriate for any person with **significant joint hypermobility (as outlined in Step 1 and any personal or family history of features outlined in Step 2)**
GP Fact Sheet: Genetic Testing
Joint Hypermobility / Ehlers Danlos Syndrome

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

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

If Brighten 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 strie 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 (Wallier 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 B 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 which may also indicate 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, Loes- 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.

Diagnosis: _______________________

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GP Fact Sheet: Genetic Testing
Joint Hypermobility / Ehlers Danlos Syndrome

Resources for people with joint hypermobility

Online resources

- The Sydney Children’s Hospital Network fact sheet on hypermobility.

- Information about joint hypermobility from the UK National Health Service.
  - NHS - Joint Hypermobility Syndrome
  - NHS Inform - Joint Hypermobility

- Information about joint hypermobility from the American College of Rheumatology.
  - Hypermobility Syndrome

- Hypermobility Connect
  - [https://www.hypermobilityconnect.com/](https://www.hypermobilityconnect.com/)

- Hypermobility Syndromes Association
  - [https://www.hypermobility.org/](https://www.hypermobility.org/)

Online articles

  - Management of Syndromic Hypermobility: A Systematic Literature Review

  - Hypermobile Ehlers-Danlos Syndrome Clinical Description and History

  - Physical Therapy Treatment of Children, Adolescents, and Adults with Joint Hypermobility Syndrome/Hypermobile Ehlers Danlos Syndrome

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