JOINT HYPERMOBILITY OR HYPERMOBILE EHLERS-DANLOS SYNDROME INFORMATION SHEET

Thank you for referring your patient with Joint Hypermobility syndrome or Hypermobile Ehlers-Danlos syndrome or a family history of the same. You are requesting confirmation/exclusion of Hypermobile Ehlers-Danlos syndrome by genetic testing. We do not offer genetic testing for Hypermobility Ehlers Danlos syndrome as this is a clinical diagnosis, and the genetics are poorly understood.

Joint hypermobility is common in the general population and often familial. Only small proportion of people with hypermobility will require medical surveillance and genetic advice and they will usually have additional distinctive clinical features (see page 4)

Of those without additional features, some will have the relatively common ‘Ehlers Danlos syndrome (EDS) hypermobility type’. These individuals may have associated chronic pain and autonomic dysfunction. Currently there is no diagnostic genetic testing available for this condition and there is no specific treatment or surveillance offered by the Clinical Genetics service. Management of symptoms should be through referral to relevant medical specialists.

Unless there were some suspicious signs of either Classical or Vascular Ehlers Danlos syndrome (see checklist below) then we do not think that we need to see all cases with Hypermobility syndrome/Ehlers Danlos syndrome Type III (also see https://www.ehlers-danlos.com/heds-diagnostic-checklist) or with a family history of this condition:

- a) Extensive widened atrophic scars
- b) Significant sagging skin
- c) Premature aged appearance
- d) Significant kyphoscoliosis
- e) Personal/family history of organ rupture
- f) Young onset unexplained arterial dissection
- g) Hand and foot deformities
- h) Young age unexplained significant or extensive varicosities
- i) Recurrent large hernias
- j) Recurrent pneumothoraces
The manifestations of this type of EDS are predominantly rheumatological, gastroenterological or psychological. The condition can be associated with full or partial dislocation of joints, joint pains and sometimes with easy bruising on minimal trauma. It appears that irritable bowel syndrome and gastritis can be seen more commonly in some people with hypermobility, as can general fatigue. With this subtype of EDS there is no significant increase in risk for cardiovascular problems and cardiac screening is not indicated.

In general treatment is designed to alleviate symptoms and can include physiotherapy, psychological support for chronic fatigue and for pain management, pain medication tailored to symptoms, and appropriate therapy for any associated stomach or bowel problems. Low impact exercise is advisable.

We hope this assists you in appropriately managing your patient’s condition. We have cancelled the referral.
Genetic referrals for Ehlers Danlos Syndrome (EDS) hypermobility type [www.ehlers-danlos.com](http://www.ehlers-danlos.com)

See diagnostic criteria attached

Associated features of classical, vascular, kyphoscoliotic, arthrochalasia, dermatopraxis EDS?

(see next page for clinical features)

Associated features of Marfan syndrome spectrum?

(see next page for clinical features)

- YES
  - Clinical Genetics referral not indicated for EDS hypermobility type
  - Refer to appropriate medical specialist for management if required

- NO
  - Request echocardiography and ophthalmic assessment

- YES
  - Refer to Genetic Health Queensland for genetic advice
  - Refer to other specialists for evaluation of heart/eye/joint/skin findings

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<tr>
<th>Type</th>
<th>Major criteria</th>
<th>Minor criteria</th>
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<tbody>
<tr>
<td><strong>Classic (Type I/II)</strong> (Autosomal Dominant)</td>
<td>Skin hyperextensibility and widened atrophic scars (necessary criteria) Joint hypermobility</td>
<td>Easy bruising, Soft doughy skin, Skin fragility, Molluscoïd pseudotumours, Subcutaneous spheroids, Hernia, Epicanthal folds, Muscular hypotonia, Complications of joint hypermobility, Positive family history</td>
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<tr>
<td>Vascular (Type IV) (vEDS) (Autosomal Dominant)</td>
<td>Family history of vEDS Arterial/intestinal/uterine fragility or rupture Carotid-cavernous sinus fistula formation</td>
<td>Unusual bruising/unrelated to trauma, Thin, translucent skin, Characteristic facial appearance, Spontaneous pneumothorax, Acrogeria (premature ageing of hands and feet), Talipes equinovarus, Congenital hip dislocation, Early-onset varicose veins (&lt;30 and nulliparous if female), Hypermobility of small joints, Tendon and muscle rupture, Keratoconus, Gingival recession and fragility, Arteriovenous or carotid-cavernous sinus fistula, Pneumo (haemo)thorax, Positive family history, sudden death in close relative(s)</td>
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<td><strong>Kyphoscoliotic</strong> (Type VI) (Autosomal Recessive)</td>
<td>Congenital muscular hypotonia Congenital Kyphoscoliosis (both necessary) Generalised joint hypermobility with dislocations/subluxations</td>
<td>Tissue fragility, including atrophic scars, Easy bruising, Arterial rupture, Marfanoid habitus, Microcornea, Osteopenia</td>
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<td><strong>Arthrochalasia</strong> (Type VII A and B) (Autosomal Dominant)</td>
<td>Congenital bilateral hip dislocation (necessary criteria) Severe generalised joint hypermobility with recurrent subluxations/dislocations Skin hyperextensibility</td>
<td>Tissue fragility, including atrophic scars, Easy bruising, Muscular hypotonia, Kyphoscoliosis, Mild osteopenia</td>
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<tr>
<td><strong>Dermatosparaxis</strong> (Type VIIc) (Autosomal recessive)</td>
<td>Severe skin fragility Characteristic craniofacial features (both necessary) Sagging, redundant skin Excessive bruising</td>
<td>Soft, doughy skin texture, Premature rupture of membranes, Large herniae</td>
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**Marfan syndrome spectrum disorders**  ([www.marfan.org](http://www.marfan.org) and [www.loeysdietz.org](http://www.loeysdietz.org))

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<tr>
<th>Syndrome</th>
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<tr>
<td>Marfan syndrome (Autosomal Dominant)</td>
<td>Aortic root enlargement Ectopia lenti</td>
<td>Reduced upper segment / lower segment AND increased arm span/height ratios, arachnodactyly with positive thumb/wrist sign, pectus carinatum/excavatum/asymmetric, scoliosis or thoracolumbar kyphosis, hindfoot deformity, flat feet, pneumothorax, scoliosis or thoracolumbar kyphosis, skin striae, reduced elbow extension, myopia, mitral valve prolapse, dural ectasia</td>
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<tr>
<td>Loey-Dietz syndrome (Autosomal Dominant)</td>
<td>Aortic root aneurysm with dissection Generalized arterial tortuosity and aneurysms</td>
<td>Systemic features: arachnodactyly, pectus carinatum/excavatum/asymmetric, scoliosis, talipes equinovarus, soft and velvety skin, translucent skin, easy bruising, dural ectasia, highly arched palate/cleft palate, malar hypoplasia, micrognathia, retrognathia, hypertelorism, broad or bifid uvula</td>
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