What is Hypermobility?

Hypermobility joints are joints that move further than the usual range, taking into account someone’s age, gender and ethnic background. Many individuals have one or several Hypermobility joints and factors such as bone shape and muscle tone can increase the range of movement of a joint. For some this is not associated with any difficulties. The diagnosis of EDS-HT is made when there are other problems associated with hypermobility.

What are the main symptoms of EDS-HT?

There can be considerable variability in the condition, even within the same family. Some people have joint hypermobility but do not have any other symptoms. Others can be more severely affected. Individuals with EDS-HT may have the following features:

- Joint hypermobility with the joints having a wider range of movement than usual.
- Loose, unstable joints that can lead to dislocations and subluxations.
- Joint pain and fatigue.
- Easy bruising.
- Gastrointestinal dysfunction.
- POTS (postural orthostatic tachycardia syndrome) causing fast heart rate, dizziness and fainting.
- Mitral valve prolapse, a heart valve abnormality which is usually only mild in EDS-HT.
- Uterine, rectal or bladder prolapse.
- Urinary dysfunction.
- Varicose veins.

What causes EDS-HT?

The exact cause(s) of EDS-HT is unknown. The features of HEDS suggest that there is a problem with connective tissues and possibly collagen. The condition appears to be inherited which suggests that there is a genetic cause. It is likely that there is an alteration in a gene, or several genes, containing the instructions for making connective tissue. This results in the connective tissue being less effective.

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What is Ehlers-Danlos syndrome (EDS)?

EDS is an officially recognised, multisystemic, inherited connective tissue disorder. Mutations of certain genes cause abnormal connective tissue synthesis, resulting in abnormal fragile, hyper-extensible tissue. Main symptoms include; easy bruising, atrophic (depressed) scarring and delayed wound healing. The syndrome is named after Danish and French dermatologists, Edvard Ehlers and Henri-Alexandre Danlos. Edvard Ehlers first described the syndrome as a separate entity in 1901.

There are currently 6 main types of EDS:
- the arthrochalasia type
- the classic type
- the dermatosparaxis type
- the hypermobility type
- the kyphoscoliosis type
- the vascular type

Other forms of the condition may exist, but they have been reported only in single families or are not well characterized.

Some forms of EDS, notably the vascular and kyphoscoliosis types, can involve serious and potentially life-threatening complications. Blood vessels can tear (rupture) unpredictably, causing internal bleeding, stroke, and shock. The vascular type of EDS is also associated with an increased risk of organ rupture, including tearing of the intestine and rupture of the uterus (womb) during pregnancy. People with the kyphoscoliosis form of EDS experience severe, progressive curvature of the spine that can interfere with breathing.

How common is EDS?

<table>
<thead>
<tr>
<th>Major Type</th>
<th>Incidence</th>
<th>Inheritance</th>
<th>Causative Genes</th>
</tr>
</thead>
<tbody>
<tr>
<td>Classical</td>
<td>1/20,000</td>
<td>AD</td>
<td>COL5A1/COL5A2</td>
</tr>
<tr>
<td>Hypermobility</td>
<td>1/5,000</td>
<td>AD</td>
<td>- UNKNOWN GENE-</td>
</tr>
<tr>
<td>Vascular</td>
<td>1/50,000</td>
<td>AD</td>
<td>COL3A1</td>
</tr>
<tr>
<td>Kyphoscoliotic</td>
<td>1/100,000</td>
<td>AR</td>
<td>PLOD1</td>
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<tr>
<td>Arthrochalasia</td>
<td>30</td>
<td>AD</td>
<td>COL1A1/COL1A1</td>
</tr>
<tr>
<td>Dermatosparatic</td>
<td>8</td>
<td>AR</td>
<td>ADAMTS2</td>
</tr>
</tbody>
</table>

(AD) Autosomal Dominant inheritance — Condition develops even if one abnormal gene is inherited
(AR) Autosomal Recessive inheritance — 2 copies of the abnormal gene need to be inherited for the condition to develop