What is Classical Ehlers-Danlos syndrome (EDS)?

Classical EDS is a type of EDS, often caused by faulty collagen V. Classical EDS can be very variable even within the same family. It is a rare condition and therefore many health professionals will not have seen someone with this diagnosis.

What are the main symptoms of Classical EDS?

Individuals with Classical EDS may have the following features:

- Joint hypermobility.
- Loose, unstable joints that can lead to dislocations and subluxations.
- Stretchy (hyperextensible) and fragile skin which can split easily.
- Smooth, velvety skin that bruises easily.
- Wounds can be slow to heal and leave distinctive widened scars.
- Fragile and extensible tissues can also result in hernias, prolapse and cervical insufficiency.

What causes Classical EDS?

Classical EDS is a genetic condition caused by a mutation in the COL5A1 or COL5A2 genes. These genes are the instructions for making collagen type V. When either gene is altered it causes a lack or deficiency of this collagen. This leads to disordered packing of collagen fibres making the connective tissue less effective, particularly in the skin and joints.

Is there anything an individual with Classical EDS should avoid doing?

It is recommended that people with Classical EDS avoid contact sports, such as rugby, football or boxing, to minimise the possibility of skin splitting and damage to joint ligaments. However, it is important to maintain a healthy lifestyle so gentle exercise, such as walking, cycling or swimming may be beneficial.

Contact Info:

Address: PO Box 748, Borehamwood WD6 9HU
Tel: 0208 736 5604
Freephone Helpline number: 0800 907 8518
Website: www.ehlers-danlos.org
Ehlers-Danlos syndrome

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