What is Tenascin-X?

Tenascin X is a protein that binds together collagen. Tenascin X deficient EDS is a condition with features that can look similar to Classical EDS without the characteristic scarring. Tenascin X deficient EDS is a rare condition and testing has only recently become available in the UK. Therefore there is limited information available and many health professionals will not have seen someone with this diagnosis.

What are the main symptoms of Tenascin-X EDS?

Individuals with Tenascin-X EDS may have the following features:

- Hyperelastic skin
- Hypermobility joints, that may subluxate or dislocate
- Easy Bruising
- Normal Scarring

What causes Tenascin-X EDS?

Tenascin X deficient EDS is caused by alterations, also known as mutations, in the TNXB gene. This gene is the instruction for making the protein Tenascin X. The deficiency of Tenascin X causes changes to the connective tissues resulting in the clinical features seen in this condition. Inheritance is autosomal recessive.

What follow-up is recommended?

Evidence on management of Tenascin X deficient EDS is limited, on the basis of current information we would suggest:

- Individuals should have an echocardiogram (ultrasound scan) of their heart to check the heart valves. If anything unusual is found, treatment may be recommended by the heart specialist.
- If surgery is being considered, the surgeon must be aware of the diagnosis of Tenascin X deficient EDS and they may wish to seek further advice.
- A baseline pulmonary function test is suggested in adulthood.
- It is thought that individuals with Tenascin X deficient EDS may be at increased risk of diverticulitis of the colon. Abdominal pain or changes in bowel habit should be reported to the GP.

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**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