Dermatosparaxis Ehlers-Danlos syndrome

What is Dermatosparaxis EDS?

Dermatosparaxis EDS is a form of EDS characterised by severe fragility of the skin. People with this form of EDS often have saggy, redundant skin with a soft, doughy texture and are easily bruised. Dermatosparaxis EDS has an autosomal recessive inheritance pattern and can be diagnosed via skin biopsy.

What are the main symptoms of Dermatosparaxis EDS?

Individuals with Dermatosparaxis EDS may have the following features:

- Saggy, redundant skin
- Severely fragile skin
- Soft, doughy texture to the skin
- Easy bruising
- Premature rupturing of fetal membranes
- Large hernias (umbilical, inguinal)

What causes Dermatosparaxis EDS?

Dermatosparaxis EDS is a genetic condition caused by having an alteration, also known as a mutation, in a gene called ADAMTS2. This gene is the instruction for making the enzyme procollagen I N-terminal peptidase. This enzyme is needed to form procollagen, the precursor to collagen. When this gene is altered it reduces or stops the activity of the enzyme and affects procollagen processing. This results in a ‘ribbon-like’ assembly of collagen fibres which weakens the connective tissue and causes the symptoms associated.

Additional Information:

- Skin fragility and bruising are substantial.
- Wound healing is not impaired and the scars are not atrophic.
- Redundancy of the facial skin results in an appearance resembling cutis laxa (another rare connective tissue disorder); however, bruising and skin fragility are not manifestations of cutis laxa.
- The name was taken from a similar phenotype and biochemical defect previously recognised in cattle, sheep and other animals.
- The number of patients reported is small and the phenotypic spectrum might expand.

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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