What is Arthrochalasia EDS?

This form of EDS characterised by generalised joint hypermobility and recurrent subluxations or joint displacements, in particular congenital bilateral hip displacements. Arthrochalasia EDS has an autosomal dominant inheritance pattern, meaning that there is a 50% chance of passing it to offspring. Arthrochalasia EDS is extremely rare, with only 30 cases reported worldwide.

What are the main symptoms of Arthrochalasia EDS?

Individuals with Arthrochalasia EDS may have the following features:

- Severe generalised joint hypermobility with recurrent subluxations
- Congenital bilateral hip displacement
- Hyper extensive skin
- Tissue fragility, atrophic scarring
- Weak muscle tone
- Kyphoscoliosis
- Short stature

What causes Arthrochalasia EDS?

Arthrochalasia EDS is a genetic condition caused by having an alteration, also known as a mutation in the COL1A1 and COL1A2 genes which create Type I Collagen. This mutation leads to deficient processing of the amino-terminal end of the pro-α1 (I) and pro-α2 (I) chains which make up procollagen, the precursor to collagen. This genetic change interferes with the assembly, structure and processing of type I collagen, which ultimately affects the skin, bones and tendons.

Additional Information:

- Congenital hip dislocation has been present in all biochemically proven individuals.
- Short stature is not a manifestation, unless it is a complication of severe kyphoscoliosis and/or hip dislocation.
- Larsen syndrome should be considered in the differential diagnosis.

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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>
</tr>
<tr>
<td>Arthrochalasia</td>
<td>30</td>
<td>AD</td>
<td>COL1A1/COL1A1</td>
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<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