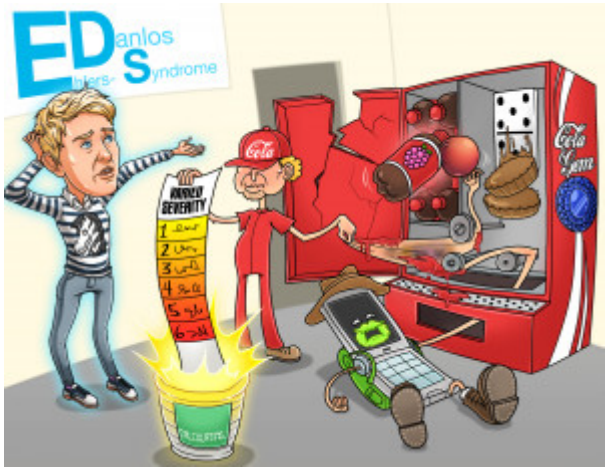
Ehlers-Danlos Syndrome Disease

Ehlers-Danlos syndrome is a connective tissue disorder caused from defective collagen synthesis. It leads to hyperextensible skin, a tendency to bleed an joint hypermobility. There are six different representations of this disease which vary in specific symptoms, inheritence patterns and severity.



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Pathophysiology

Faulty Collagen Synthesis
Broken Cola-gem Machine

Ehlers-Danlos syndrome is caused by a variety of gene mutations. These mutations lead to faulty collagen synthesis by altering the structure, production, or processing of collagen (or proteins interacting with collagen).

Autosomal Dominant or Recessive
Domino and Recessive-chocolate

The inheritance pattern for Ehlers-Danlos syndrome varies for each type and may be inherited in an autosomal-dominant or autosomal-recessive pattern.

Various Severities
Varied Severity quote

There are six main/six clinically relevant types of Ehlers-Danlos, and they are each different in their combination of clinical effects, inheritance patterns, and gene mutations. Hypermobility type is the most common. The classical type has joint and skin symptoms and is caused by a mutation in type V collagen. Vascular type causes vascular and organ rupturing and is due to a deficiency in type III collagen.

Signs and Symptoms

Hypermobile Joints
Flexible Hiker-mobile

In hypermobility type Ehlers-Danlos, patients have joint hypermobility that can manifest as joint instability and chronic musculoskeletal pain. They experience frequent joint dislocations and subluxations.

Hyperextensible Skin
Hiker-with-extensible Skin

In classical Ehlers-Danlos, there is a defect in type V collagen as well as type I. Patients have skin involvement, showing hyperextensible and elastic skin.

Easy Bruising/Bleeding
Bruising and Bleeding

In the vascular type presentation of Ehlers-Danlos, patients have a defect in type III collagen synthesis. These patients have a propensity to bleed subcutaneously because blood vessels are prone to tearing, causing bruising without trauma (ecchymoses).

Berry (Saccular) Aneurysm
Bulging-aneurysm

Patients with vascular type Ehlers-Danlos have a defect in type III collagen synthesis, leading to fragile organs and blood vessels, which are prone to rupturing. Patients are at high risk of developing aneurysms and organ ruptures.

Considerations

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Brighton Criteria

Bright-tin

This criteria suggests that to diagnose Ehlers-Danlos syndrome, patients must have two major criteria, one major and two minor criteria, or four minor criteria. Major criteria include a Brighton score of >4/9 and arthralgias for longer than three months in four or more joints. Some of the minor criteria include marfanoid habitus, abnormal skin, soft tissue rheumatism, and subluxation/dislocation in one or more joints.

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