Ehlers-Danlos Syndrome Types

Ehlers-Danlos Syndrome (EDS) refers to a group of genetic mutations that lead to a defect in collagen synthesis. The most common type of EDS is hypermobility type (formerly type 3 EDS), which is characterized by joint instability, where there is a deficiency in type-III collagen. Classic EDS, formerly referred to as types I and II, is a defect in type V collagen, which affects the joints and skin. Vascular type involves a defect in type III collagen and is characterized by vascular defects and organ rupture. Classic signs and symptoms of EDS include hypermobile joints, hyperextensible skin, easy bruising and bleeding, and saccular aneurysms. EDS may be inherited in an autosomal-dominant or autosomal-recessive fashion. Diagnosis is based off the Brighton Criteria. Treatment is generally supportive; some patients may benefit from Vitamin C supplementation to improve bruising.

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Types

Types 1 and 2 - Classical
(1) Wand with (2) Tutu
Classical Type (formerly Types 1 and 2) features include fragile, velvety and hyperelastic skin as well as delayed wound healing and joint hypermobility. It involves a defect in Type V collagen, and is inherited in an autosomal dominant pattern.

Type V Collagen Mutation
Mutant Cola-gem machine with (5) Hand
Classical Type (formerly Types 1 and 2) features include fragile, velvety and hyperelastic skin as well as delayed wound healing and joint hypermobility. It involves a defect in Type V collagen, and is inherited in an autosomal dominant pattern.

Joints and Skin
Joints and Skin Stretched
Classical Type (formerly Types 1 and 2) features include fragile, velvety and hyperelastic skin as well as delayed wound healing and joint hypermobility. It involves a defect in Type V collagen, and is inherited in an autosomal dominant pattern.

Type 3 - Hypermobility
(3) Tree Hiker-mobile phone
Hypermobility Type (formerly Type 3) is the most common variant of EDS. Patients present with unstable and dislocatable joints.

Most Common
#1 Foam finger
Hyper mobility Type Ehlers-Dalos (formerly Type 3) is the most common variant of EDS.

Tenascin X Deficiency
Tennis X Racket with Down-arrow
Patients with type 3 Ehlers-Danlos syndrome develop the hypermobility variant of this disease. Here, the glycoprotein Tenascin-X, which is found in connective tissues, joints and muscles, is absent. Because of this, patients develop hypermobile joints, and are more prone to dislocations and subluxations.

Joint Instability
Instability
In the hypermobility variant of Ehlers-Danlos syndrome, patients have less severe skin manifestations, and more frequent joint pathologies. The hallmarks of this type of disease are joint instability, characterized by dislocations and subluxations (with or without trauma), along with chronic musculoskeletal pain.

Type 4 - Vascular
Vascular man with (4) Fork
Vascular Type (formerly Type 4) is due to a defect in Type III collagen, and is characterized by translucent skin with visible vessels, vascular and organ rupture (i.e. vertebral artery dissection), and easy bruising.

Type III Collagen Mutation

Mutant Cola-gem with (3) Tree

The vascular variant of Ehlers-Danlos syndrome occurs when there is a defect in type-III collagen synthesis. Type-III collagen is an important protein that is found in extensible connective tissues, such as skin and the vascular system.

Vascular and Organ Rupture

Ruptured organs

In this variant of Ehlers-Danlos syndrome, blood vessels and organs are fragile and prone to tearing, especially when an infection is present. These are important complications of this disease to be familiar with.