Ehlers-Danlos Syndrome

**Definitions**

Ehlers-Danlos Syndrome (EDS) is a hereditary connective tissue disorder. It is caused by genetic changes that affect the connective tissue that stabilize and support the joints and organs throughout the body.

There are many different types of EDS, including the following:

- **classic**
- **hypermobile**  
  *most common*
- **vascular**
- **kyphoscoliosis**
- **arthrochalasia**
- **dermatosparaxis**

**CAUSES**

- EDS is caused by mutations in the genes that build connective tissue. Different types of EDS are caused by mutations in different genes
- hypermobile, classic, vascular, and arthrochalasia types are autosomal dominant, meaning they are caused by a mutation in one copy of a particular gene
- the kyphoscoliosis and dermatosparaxis types are autosomal recessive, meaning they are caused by mutations in both copies of a single gene

A "Beighton score" can be calculated to help assess hypermobility. Patients obtain a certain number of points during an evaluation from a physician based on the tests above.

**Common Findings**

- joint hypermobility (laxity)
- soft, stretchy or fragile skin
- abnormal scarring
- slow & poor wound healing
- tempo-mandibular joint instability and pain
- dysautonomia, including POTS and gastrointestinal symptoms

**DIAGNOSTIC TESTS**

- the diagnosis of Ehlers-Danlos is best made by a clinical geneticist with experience in diagnosing the hereditary disorders of connective tissue. A comprehensive clinical evaluation is required, including taking a complete medical history
- evaluation of family history
- clinical evaluation to test EDS related physical signs.
- depending on EDS-type, genetic testing may be done
  The gene or genes causing the hypermobile type are not yet known

Patients with EDS often spend years, if not decades, in search of a diagnosis. Because the manifestations are often multiple and complex, a unifying diagnosis may be elusive.

**TREATMENT**

Currently, there is no cure for EDS, although there are many treatments that may improve symptoms.

Diagnosis allows the patient and their families to know what they may be facing and allow them to become educated and proactive about the care they seek and receive.