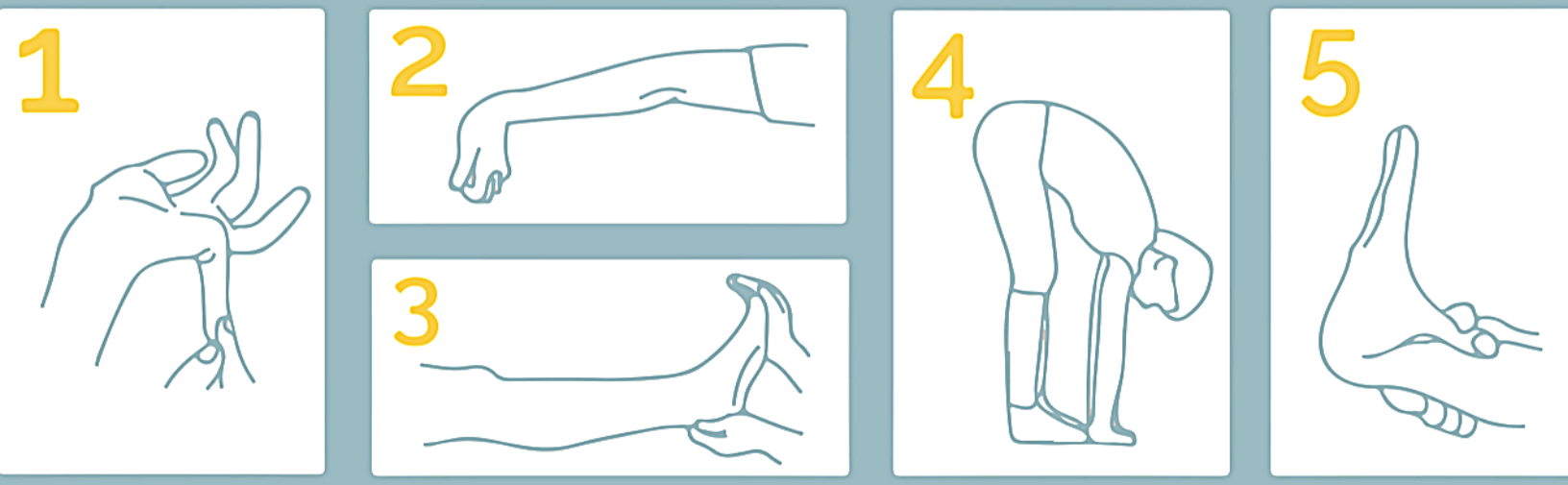
About Ehlers-Danlos Syndrome



BEIGHTON SCORE: A 9-point system used to quantify joint laxity and hypermobility.

Definition

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

There are many different types of EDS, including:

- classic
- hypermobile
- 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 Kyphscholiosis and Dermatorsparaxis types are autosomal recessive, meaning they are caused by mutations in both copies of a single gene.

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 knnow what they may be facing and allow the to become educated and proactive about the care they seek and receive.

Common Symptoms

- joint hypermobility (laxity)
- headaches
- abnormal scarring
- stretchy or fragile skin
- temporo-mandibular joint instability
- chronic pain
- easy bruising
- slow & poor wound healing
- cranio-cervical & cervical instability
- dysautonomia (POTs)

Diagnostic Tests

The diagnosis of Ehlers-Danlos Syndrome is best made by a clinical geneticist with experience inn diagnosing the hereditary disorders of connective tissue.

A comprehensive clinical evaluation is required, which includes:

- taking a 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.

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