A Genetic Connective Tissue Disorder

What is Ehlers-Danlos Syndrome?

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The Ehlers-Danlos syndromes are inherited in the genes that are passed from parents to offspring. They are categorized according to the form of genetic transmission into different types with many features differing between patients in any given type. The fragile skin and loose joints and tissue fragility is often a result of abnormal genes that produce abnormal proteins that confer an inherited frailty of collagen (the normal protein "glue" of our tissues).

In 2001, researchers discovered a new form of Ehlers-Danlos syndrome that is caused by an inherited abnormality in a protein other than collagen that also normally plays a role in binding together the cells of our tissues (including the skin, tendons, muscle, and blood vessels). Abnormalities in this protein, called tenascin, also lead to a form of Ehlers-Danlos syndrome. Researchers suspect that tenascin could play a role in regulating the normal distribution of collagen in the connective tissues of the body

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There are six major types of EDS. The different types of EDS are classified according to their manifestations of signs and symptoms.

Classical Type

Hypermobility Type

Vascular Type

Kyphoscoliosis Type

Arthrochalasia Type

Dermatosparaxis Type

Tenascin-X Deficient Type

web site by ~ Lynn Sanders

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How are Ehlers-Danlos syndromes diagnosed?

The diagnosis of Ehlers-Danlos syndrome is based upon the clinical findings of the patient and the family history. For some types of Ehlers-Danlos syndrome, a skin biopsy to determine the chemical makeup of the connective tissue can help to suggest the diagnosis.

How are Ehlers-Danlos syndromes treated?

Ehlers-Danlos syndromes are treated according to the particular manifestations present in a given individual.

Skin protection (from injury of trauma and sun, etc.) is critical. Wounds must be tended with great care and infections treated and prevented. Suturing can be difficult as the skin can be extremely fragile.

Joint injury must be avoided. Occasionally, bracing may be necessary to maintain joint stability. Exercises that strengthen the muscles that support the joints can help to minimize joint injury. Contact sports and activities involving joint impact should be avoided.

Role of Smooth Muscle Cells In Vascular Ehlers-Danlos Syndrome

Dr. Dianna M. Milewicz MD PhD
Dr. Amy Reid MD PhD

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Ehlers Danlos Syndrome Network C.A.R.E.S. Foundation we are currently funding our first EDS research project. So far we have given $75,000 towards this research. Every dollar that we raise for research, only goes for EDS research. Everyone who helps run this foundation are all volunteers.

No paid staff...
We are working hard so we can be
"One step closer to a cure!"

Connective Tissue Disorders Clinical and Molecular Manifestations of Heritable Disorders of Connective Tissue

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If you would like to donate for our EDS Research Fund
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Dr. Hal Dietz

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