

THE FACTS ABOUT EHLERS-DANLOS SYNDROME

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The Ehlers-Danlos National Foundation

(EDNF) is a 501(c)(3) non profit organization; founded in 1985 its purpose is to:

- Disseminate accurate information
- Provide a network of support and communication
- Foster and support research

The EDNF produces a range of educational media that is distributed free of charge to those who request it. Information leaflets, articles, multimedia programs, guides and newsletters are examples of the kind of programs that are available.

EDNF members are now able to communicate directly with each other through the interactive members area at www.ednf.org. With over 15,000 posts as of January 2004, EDNF members have built an extensive information repository on EDS and it is growing every day.

The EDNF currently has 36 local groups within the United States. By actively encouraging the development of such groups the Foundation is better able to meet the needs of communities at a local level.

Finally, with up to \$100,000 allocated for 2004, the EDNF is now directly funding research into the Ehlers-Danlos Syndrome.

To find out more about the Ehlers-Danlos National Foundation or to see if there is a local group in your area, please visit our easy to use web site at www.ednf.org or tear off the adjacent form and return it to us at the following address:

Ehlers-Danlos National Foundation
6399 Wilshire Boulevard #200
Los Angeles, CA 90048

Ehlers–Danlos syndrome (EDS) is a heterogeneous group of heritable connective tissue disorders, characterized by articular (joint) hypermobility, skin extensibility and tissue fragility. There are six major types of EDS. The different types of EDS are classified according to their manifestations of signs and symptoms. Each type of EDS is a distinct disorder that “runs true” in a family. This means that an individual with Vascular Type EDS will not have a child with Classical Type EDS.

Individuals with EDS have a defect in their connective tissue, the tissue which provides support to many body parts such as the skin, muscles and ligaments. The fragile skin and unstable joints found in EDS are the result of faulty collagen. Collagen is a protein which acts as a “glue” in the body, adding strength and elasticity to connective tissue.

■ Symptoms

Clinical manifestations of EDS are most often skin and joint related and may include:

Skin: soft velvet–like skin; variable skin hyper-extensibility; fragile skin that tears or bruises easily (bruising may be severe); severe scarring; slow and poor wound healing; development of molluscoid pseudo tumors (fleshy lesions associated with scars over pressure areas).

Joints: joint hypermobility; loose/unstable joints which are prone to frequent dislocations and/or subluxations; joint pain; hyperextensible joints (they

move beyond the joint’s normal range); early onset of osteoarthritis.

Miscellaneous/Less Common: chronic, early onset, debilitating musculoskeletal pain (usually associated with the Hypermobility Type); arterial/intestinal/uterine fragility or rupture (usually associated with the Vascular Type); Scoliosis at birth and scleral fragility (associated with the Kyphoscoliosis Type); poor muscle tone (associated with the Arthrochalasia Type); mitral valve prolapse; and gum disease.

■ Prevalence

At this time, research statistics of EDS show the prevalence as 1 in 5,000 to 1 in 10,000. It is known to affect both males and females of all racial and ethnic backgrounds.

■ Hereditary Patterns

The two known inheritance patterns for EDS include autosomal dominant and autosomal recessive. Specifics regarding genetic inheritance may be found by following the link below. Regardless of the inheritance pattern, we have no choice in which genes we pass on to our children.

■ How is EDS Diagnosed

Diagnosis of EDS is based upon clinical findings and upon the family history. Since many patients do not fit neatly into one of the specific types of EDS, a diagnosis is often delayed or overlooked. Specific diagnostic tests are available for some

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types of EDS in which there is a known biochemical defect. Sometimes, a physician may perform a skin biopsy to study the chemical makeup of the connective tissue. The biopsy involves removing a small piece of skin, under local anesthesia. Physicians who are able to diagnose EDS may include medical geneticists, pediatricians, rheumatologists, dermatologists and orthopaedists.

■ Treatment/Management of EDS

The gaping skin wounds, which are common in several types of EDS, should be approached with care. Proper repair of these wounds is necessary to prevent cosmetic disfigurement. Surgical procedures can be risky, as fragile tissues can unexpectedly tear. Suturing may present problems for the same reason. Excessive sun exposure should be avoided by the daily use of sunscreen. One should avoid activities that cause the joint to lock or overextend.

A physician may prescribe bracing to stabilize joints. Surgical repair of joints may be necessary at some time. Physicians may also consult a physical and/or occupational therapist to help strengthen muscles and to teach people how to properly use and preserve their joints. To decrease bruising and improve wound healing, some patients have responded to ascorbic acid (vitamin C). Always consult with your doctor before starting any treatment.

In general, medical intervention is limited to symptomatic therapy. Prior to pregnancy. Patients with EDS should have genetic counseling. Children with EDS should be provided with information about the disorder, so they can understand why contact sports and other physically stressful activities should be avoided. Children should be taught early on that demonstrating the unusual positions they can maintain due to Loose joints should not be done as this may cause early degeneration of the joints. Family members, teachers and friends should be provided with information about EDS so they can accept and assist the child as necessary.

■ Prognosis

The prognosis of EDS depends on the specific type. Life expectancy can be shortened with the Vascular Type of EDS due to the possibility of organ and vessel rupture. Life expectancy is usually not affected in the other types.

Reference:

Beighton, P., De Paepe, A., Steinmann, B., Tsipouras, P., & Wenstrup, R (1998). Ehlers-Danlos Syndromes: Revised Nosology, Villefranche, 1997. *American Journal of Medical Genetics*, 77, 31-37.

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