

I am Interested in the following:

- Membership of the Foundation
- Information about EDS
- Making a Donation to the EDNF

Please send the relevant information to the address below.

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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

■ What is Ehlers-Danlos Syndrome?

Ehlers-Danlos syndrome (EDS) is a group of heritable disorders of connective tissue often characterized by hyperextensible skin, hypermobile joints, easy bruisability of the skin and a bleeding diathesis (tendency). EDS is named for two physicians (Ehlers and Danlos) who described forms of the condition in the early 1900's. Ehlers-Danlos syndrome may be classified into six major types. These types are not gradations in severity, but represent distinct disorders that "run true" in a family. More detailed information on symptoms and management of EDS may be found in the leaflet "The Types of Ehlers-Danlos Syndrome".

■ What are Genes?

Genes are the smallest units of the hereditary material. They are like "blueprints" for directing normal growth and development. Hundreds of genes are packaged on chromosomes which are found in every cell of the body. Humans have 23 pairs of chromosomes in each cell. Twenty-two of the chromosomes are called autosomes. The 23rd pair of chromosomes determines the sex of the individual. An individual with two X chromosomes is female; an individual with one X and one Y chromosome is male. We receive half of our chromosomes (one from each pair) from each of our parents. Each gene in a pair can be functioning or non-functioning. A gene which is not functioning properly is a mutant gene (a change).

■ Inheritance Patterns of EDS

There are two different inheritance patterns of EDS. They are known as autosomal dominant and autosomal recessive. It is important to remember that regardless of the pattern of inheritance, we have no control over which genes we pass on to our children.

■ Autosomal Dominant

In Autosomal Dominant inheritance, only one member of the gene pair needs to be functioning improperly for an individual to be affected. When an affected person has children, there is a 50% chance with each pregnancy that the parent will pass on the gene to his/her children. Therefore there is a 50-50 chance that the child will be affected, regardless of the sex of the child. A person who does not carry the gene is not affected and can not pass the gene on to his/her children. EDS types that are inherited in the autosomal dominant fashion include the Classical Type, Hypermobility Type, Vascular Type, and Arthrochalasia Type.

■ Autosomal Recessive

In Autosomal Recessive inheritance, a person must carry both changed genes to be affected. A person who has one working gene and one non-working (changed) gene is said to be a gene carrier. When two people who carry the same recessive gene in common have children, there is a 25% chance that the child will have two working genes and be unaffected; a 50% chance that the

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child will be unaffected but will be a gene carrier like his/her parents; and a 25% chance that the child will carry both recessive genes and be affected. These percentage risks apply to each pregnancy. Types of EDS which are inherited as autosomal recessive are the Kyphoscoliosis Type and the Dermatosparaxis Type.

■ Genetic Counseling

Genetic counselors are medical professionals who are trained in medical genetics and counseling. They deal with the occurrence or risk of occurrence of a genetic disorder in a family. They most often work in conjunction with a medical geneticist (physician). They provide an accurate presentation of the facts to a patient who has been diagnosed or who is a known carrier of a genetic disorder. Most people find the genetic aspect of the disorder confusing.

A genetic counselor will help you sift through the facts to gain a better understanding of your inheritance pattern. They provide nondirective counseling to those who are considering starting a family to facilitate making an informed decision. They may act as a resource for educational information about a disorder, as a psychosocial counselor for families in crisis, as a link to support services/support groups and as a coordinator for patient care.

Laboratory confirmation of certain types of Ehlers-Danlos syndrome is possible. These include the Vascular, Kyphoscoliosis, Arthrochalasia and

Dermatosparaxis Types. If these types of EDS are suspected based on the clinical findings, the geneticist may want laboratory confirmation.

The genetic counselor may be the liaison between the patient and the genetic laboratory. In this role the counselor may be responsible for explaining the technical information to patients and other health care providers. The genetic counselor will most often be the person to explain how the testing will be done, the costs involved and the estimated length of time for test completion.

The tests involved are usually performed at highly specialized laboratories. Because they are often not the center where a person is being evaluated, the genetic counselor may also be responsible for gathering all pertinent medical information such as medical records, family history and consent forms. When lab results become available, the genetic counselor as well as the geneticist will meet with the patient or family to discuss the results and implications of the new findings.

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GENETICS AND INHERITANCE PATTERNS

in Ehlers-Danlos Syndrome