EHLERS-DANLOS-Syndrom

... Involuntary Acrobats

Information for those affected and those interested

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Doctors and patients in discussion

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Contents

Dear reader,

Look at an acrobat and it's hard not to be impressed by their extraordinary power and capacity. Stamina, discipline and agility are what we see as the foundation of prowess and are all attributes that deserve admiration.

Sometimes, however, it is an illness that is concealed behind these "acrobatic abilities" rather than athletic capacity. The person in question becomes an acrobat against his will. Ehlers-Danlos syndrome is a rare hereditary connective tissue disorder which causes the breakdown of the body's collagen composition.

These inherent "abilities" are therefore not to be regarded as positive characteristics; in fact, they bring about a range of health problems. The symptoms of EDS are varied; for instance, it can lead to hyperelastic skin that can easily be damaged or to the rupture of internal organs and vessels.

Ehlers-Danlos syndrome is to a great extent inescapable. EDS is a rare illness and is therefore largely neglected by scientific research. Diagnosis is difficult as some other connective tissue disorders (for example Marfan syndrome) bear a resemblance to EDS. EDS is therefore often not recognised until very late. Therapeutic measures mainly apply only to the treatment of symptoms.

All of this makes it even more important to raise awareness of Ehlers-Danlos syndrome. It is above all children, adolescents and young adults as well as their parents and guardians in schools and preschools who are the focus of attention. Although they cannot be avoided, potential concomitant diseases/consequences of the illness can certainly be alleviated in terms of symptoms if EDS is dealt with appropriately. And even adult victims of EDS require more support in dealing with the illness.

This brochure is intended to provide you with basic information about EDS, reveal causes and symptoms and, by means of a concise, standardised questionnaire, support the recognition and identification of EDS. Furthermore, existing therapeutic alternatives will be highlighted and insights into day-to-day life with the illness will be given.

Yours,
Barbara Kleffmann
Chairwoman of the Ehlers-Danlos Syndrome Self Help e.V.

Basic Information

Ehlers-Danlos syndrome, or EDS for short, is a rare illness.

The definition of a rare illness is:
When less than one in 2,000 people is affected by a disorder, it is defined as rare.

In the year 2000, around 400 people were faced with the diagnosis of EDS. According to estimates, around 2,500 to 5,000 people in Germany may have EDS.

EDS is a connective tissue disorder that brings about the breakdown of the body's collagen composition (= collagen biosynthesis), a so-called structural protein of the connective tissue.

Protein molecules that form a sort of structural material in tissue and cells and give them their elasticity and stability are referred to as structural proteins. In the case of EDS, the interconnection of these structural proteins, and respectively the collagen, is affected in an adverse way.

Connective tissue
The connective tissue – aka supporting tissue – has important functions within the human body. To associate the connective tissue solely with potential cellulite (orange peel skin or weakness of the connective tissue) would be to totally underestimate its complex significance.

In the case of EDS, weakness of the connective tissue along with painful symptoms means that the protection of vessels and, above all, nerves that are close to the surface of the skin is inhibited. This leads to neurological irritations and neuropathies. The result is often chronic pain, for instance something similar to a polyneuropathy or fibromyalgia.

Connective tissue envelops organs, muscles, bones, blood vessels and nerves. It stabilises the individual organs and provides tendons and ligaments with efficacy. Connective tissue is even important for the cornea as it helps with its transparency.

Moreover, the connective tissue serves as padding, as a water reservoir and as fat storage. Likewise, the connective tissue transmits nerve impulses, regulates the supply of nutrients and our body temperature as well as the removal of cell excretion.

Causes

The cause of EDS is genetic mutation. EDS is thus a hereditary disease.

Genes are the hereditary disposition that is handed down through replication in offspring. They form the foundation of each individual person.
Various somatic cells exist in our bodies; the nuclei in these cells contain the information of these genes on so-called chromosomes.

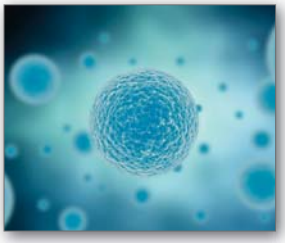
Human cells normally comprise 23 chromosome pairs.
A set of "duplicates" of the chromosomes of the mother and those of the father are passed on each time.

If a gene is changed (mutated), its function can be impaired. As a result, a disorder can arise because this gene no longer transmits all the information to the human organism correctly.

In most cases, EDS is inherited in an autosomal dominant manner.
This means that if one parent has the disease, EDS can be inherited. Therefore the disorder can be inherited either from the mother or the father.
The risk of the child inheriting the disorder is around 50%.

Good to know:

- EDS is a hereditary disease.
- Not all children of victims of EDS necessarily inherit EDS.
- EDS can also occur as a new mutation, ie. even when none of the parents suffer from EDS, it can develop through a genetic mutation (in the egg or the sperm cell).
- It is sometimes the case that a generation of EDS offspring exhibits only weak, unnoticed symptoms. It is nevertheless still possible that the disease will be passed down to the next generation.



Different Types of EDS

Ehlers-Danlos syndrome was delineated at length at the beginning of the twentieth century by doctors Edward Ehlers and Henri-Alexandre Danlos, who then ultimately named the disorder after themselves.

Records show, however, that even Hippocrates had concerned himself with the "Disease of effusion and haemorrhaging".

Initially, ten different types of EDS were defined in 1988. Based on further scientific findings, this definition was then revised in 1997 and the Villefranche classification was published, whereby six relevant forms of EDS are distinguished.

This classification improves the distinction between EDS and other similar connective tissue disorders (see following chapter, 'Diagnostics') and simplifies the diagnostic possibilities.

The triggering genetic mutations have since been able to be identified with most types of EDS. Only the causal gene in the case of the hypermobility type as well as with exotic variants of EDS are yet to be verified.

The most common types of EDS are the hypermobility and the classic types.

Currently the definite classification of older patients who weren't recognised as having EDS until late on is proving to be problematic. Furthermore, new types are always being recorded. Therefore, the research will not be concluded for a long time. The classification of types is thus a rough guideline; many symptoms are represented irrespective of type and thus often complicate definite clinical allocation.

"Villefranche" EDS classification

Classic type	
Type	Type I and II (symptoms of Type I are less distinct than Type II)
Symptoms	Extremely hyperelastic and easily damaged skin Tendency towards bruising Slow wound healing with wide scar formation Laxity of the joints Involvement of inner organs and vessels

Hypermobility type

Type	Type III
Symptoms	Little involvement of the skin, velvety skin
	Distinct laxity of the joints
	Frequent dislocation of the joints
	Tendinitis, slipped discs, chronic muscle and joint pains, in part with inflammation and swelling
	Deformation of vertebrae and extremities

The overlapping symptoms of the hypermobility type of EDS and the distinct Hypermobility syndrome (HMS) are currently being scientifically explored.

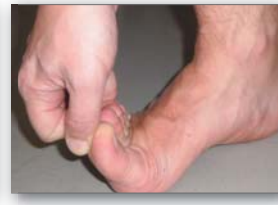
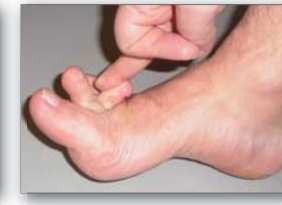
Hypermobility syndrome is a development malfunction of the connective tissue that occurs in early childhood. The symptoms are frequently confused with rheumatic illnesses, the so-called Soft Tissue Rheumatism and the fibromyalgia that resides there. Likewise, the symptoms are very similar to those of EDS.

Vascular type

Type	Type IV
Symptoms	Thin, translucent skin with easily visible blood vessels
	Distinct tendency towards bruising
	Laxity of small joints
	Involvement of the eyes
	Involvement of inner organs

Kyphoscoliosis type

Type	Type VI
Symptoms	Moderate to severe hyperelasticity of the skin
	Bad wound healing with wide scarring
	Severe laxity of the joints
	Sideways distortion of the vertebrae (scoliosis)
	Involvement of the eyes
	Involvement of the inner organs

Arthrochalasia type

Type	Type VII A / B
Symptoms	Little to moderate hyperelasticity of the skin
	Flexible, thin skin
	Bad wound healing with wide scarring
	Distinct laxity of the joints
	Frequent dislocation of the hips

Dermatosparaxis type

Type	Type VII C
Symptoms	Very limp (sagging) skin
	Distinct laxity of the joints
	Involvement of inner organs

Further EDS types

The further hitherto established types of EDS can be labelled "exotic", as they arise very rarely.

Known types include EDS type V / x-linked, EDS type VIII / periodontitis type, EDS type XI / familial hypermobility syndrome, EDS type X / fibronectin-deficient and progeroid EDS.

Heredity

As already described in the 'Causes' chapter, most types of EDS are inherited in an autosomal dominant manner. In other words, only one parent exhibiting the genetic mutation will suffice.
This applies to types I, II, III, IV and VII.

Types VI and VIIC are, however, inherited in an autosomal recessive manner. In other words, both parents must have the genetic defect in order to be able to pass it on to their offspring.



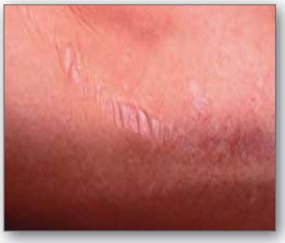
Symptoms

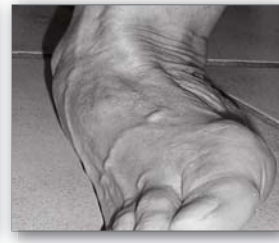
The symptoms of Ehlers-Danlos syndrome are very varied, which explains the preceding classification.
In addition, symptoms can be far more pronounced in some cases than others. They differ in frequency of occurrence and not all highlighted symptoms occur with every sufferer of EDS in equal measure. Likewise, and this is not so uncommon, so-called overlaps within classifications arise. These mixed types and their repercussions on the disease overall complicate matters further.

This fact often leads to a long-lasting quest for the patient from doctor to doctor until a diagnosis can perhaps be made. Moreover, due to the various different health problems that can arise, a variety of medical specialists (eg. orthopaedists, rheumatologists, cardiologists, internists, dentists, ophthalmologists, neurologists and so on) are consulted, who each see and treat only one angle of the (EDS) symptomatology, which impedes the conclusion of the actual cause of the symptoms and their treatment.

In addition, existing EDS symptoms can bring about derivative afflictions, or so-called secondary symptoms. Incidentally, some of the typical – more common – symptoms of EDS are brought on once again:

- Hyperelasticity, over-ductility/-extensibility of the joints
- Abnormal ruptures, easily damaged skin
- Delayed wound healing and formation of scars, bruising
- Malposition and deformation of the joints and vertebrae
- Mutation of cardiac valves and large blood vessels (for instance, vascular dilation, enlarged cardiac vessels)
- Inner and outer visceral fractures, eg. inguinal hernia





Diagnostics

The formation of a suspicion of EDS is crucial for a fast and clear EDS diagnosis. The suspicion of EDS arises as a result of existing symptoms. If a suspicion exists, a human genetic examination can be carried out. Biochemical evidence of specific enzyme defects is recommended. In addition, a skin biopsy, which can be performed under local anaesthetic above the elbow, can check the chemical structure of the collagen in the skin. This way, the EDS type can normally be detected by an electron microscopy laboratory and molecular biology. Meanwhile there is another more gentle option for diagnosis which will make skin biopsies redundant in the future.

Further studies can take place by means of blood and urine examinations, as the circumstances require. Nevertheless, although there is every indication that this will work, there are always cases in which a 100% diagnosis cannot be made.

A prenatal diagnosis (prior to birth) is possible.

Beighton Test
In support of the diagnosis of such illnesses, an internationally acknowledged 9 point programme has been developed for Hypermobility syndrome (see Classification, Hypermobility type) on the basis of which the magnitude of the laxity of the joints can be established. Please note that this test can only give a possible indication of EDS if further symptoms other than those relating to Hypermobility syndrome are also present.

Test	Points
• Palms of the hands can be placed flat on the floor when knees are outstretched in standing position (gap between fingers and ground <0)	1
• Elbow joint can be stretched backwards by at least 10°	1 per side
• Thumb can be placed on forearm	1 per side
• Little finger can be pulled backwards 90	1 per side
• Knee joint can be stretched backwards by at least 10°	1 per side

0 – 2 points = not hypermobile
3 – 4 points = moderately hypermobile
5 – 9 points = generalised hypermobility

Symptom Questionnaire

- in terms of Hypermobility syndrome, comparable with EDS type III -

- I am tall and thin
- I have relatively long arms and legs
- I have relatively long fingers
- I have a crooked spine
- I can put my shoulder blades in the "angels' wings position"
- I can overstretch my elbow and knee joints
- I have "X" or "O" legs
- I have flat feet or splay feet
- I can crack my joints
- Even as a child I was double-jointed
- I also have double-jointed parents/siblings/children
- I have dislocated a joint at least once
- I have a tendency towards tendinitis
- My skin is pale, dry; appears thin/translucent
- I have stretch-marks
- I hurt myself easily
- I bruise easily
- I bleed more strongly and for longer when I injure myself
- Scars become wide and ugly
- I break bones quite easily
- I suffer from shrinking of the gums and easily lose teeth
- I suffer from gastritis
- I have a diaphragmatic hernia
- I suffer from stomach cramps in general
- I have had an inguinal hernia
- I am very sensitive to the cold
- I suffer from varicose veins/haemorrhoids
- I suffer from low blood pressure
- I sometimes feel my heart beating quickly
- One of my pulmonary alveoli has once burst
- I urinate involuntarily
- I suffer from descensus/prolapse of the uterus
- I have had a miscarriage
- I get tired very quickly
- I am anxious and panic easily
- I have trouble sleeping or sleep very lightly
- I am sensitive to changes in the weather

Source: Prof. Dr. Günther Haberhauer, Internal Medicine Consult, Rheumatology and Metabolic Diseases, Vienna

If the answer to more than five questions is 'yes', this could indicate a Hypermobility syndrome or EDS. A diagnosis can, however, only be made by a doctor.

Differential diagnosis

As already explained, EDS displays similarities with other connective tissue disorders. Within the diagnosis it must therefore be clarified whether or not the symptoms at hand are caused by one of the following illnesses. Marfan syndrome, Osteogenesis imperfecta and tendency to bleed are particularly likely to be mistaken for EDS.

Other such illnesses are connective tissue disorders such as chondrodysplasia, epidermolysis bullos and Alport syndrome.

It is also important to clarify that the illness in question is not rheumatoid arthritis, fibromyalgia, multiple sclerosis, hypermobility syndrome or growing pains.

Therapeutic Measures

Since the illness EDS is based on a genetic defect, no cure or immediate therapy of the cause is currently possible.

Therapy therefore involves the treatment of the symptoms that arise in each case. Since EDS patients often suffer with pain, this may be attending pain therapy led by a pain therapist.

Effects on the cardiovascular system must be monitored and handled accordingly. Orthopaedic aids can also be of use.

As the vascular EDS – type IV – can be distinguished from the other EDS types by the easy damaging of the skin, and consequently the possibility of a rupture of the inner organs exists, regular medical check-ups should be carried out.

There is no prevention in the classic sense as EDS develops differently in every patient. Eventually the deceleration of certain characteristics may however ensue.

A human genetic consultation is recommended.



Preventative Measures

Even children and adolescents are athletically active and like to gauge their strength at one point. Minor injuries are thus common.
That is why it is particularly important for children affected by EDS, as well as those who care for them in school for instance, to convey an understanding of the illness, while at the same time avoiding taking their pleasure in sporty activity – where this is possible – away from them.

Listed below you will find some additional steps that are indeed equally applicable to both young and adult EDS patients:

- Lifelong, moderate training in support of stability, strength and balance – with minimal involvement of the joints, using little strength and avoiding over-stretching – use joint guards when needed
- Avoid contact sports
- Possibly take part in some gentle training, eg. Tai Chi, aqua aerobics, swimming, Pilates, Yoga, Feldenkrais, ergotherapy
- Avoid injuries; use bandages and taping on the joints when needed; avoid adhesive bandages as these can lead to cracks in the skin; tension-free wound closure; use blister plasters on wide scars; immediately apply cold compress in event of haemorrhaging;
- Use in-soles as the circumstances require
- Use ergonomic aids, eg. around the house, for school or to increase com fort in bed
- Go for regular eye check-ups (retina)
- Go for regular dental check-ups; take preventative medicines – it could be the effect of the local anaesthetic is reduced
- Go for regular heart check-ups
- If needed, go for regular ultrasound check-ups (inner organs)
- Avoid obesity; smaller meals recommended
- Manual medicine / osteopathy is recommended in most cases
- Always have your emergency ID to hand

Living with EDS

It is natural to be filled with uncertainty while waiting for a diagnosis. It is often the case that physical afflictions are downplayed simply because they haven't been assessed. This state is strenuous and crushing.

Once the diagnosis is made, it is initially a relief because the symptoms can now be given a name. For many of those affected by EDS, however, uncertainty soon sets in once again due to the realisation that the illness cannot be cured, the symptoms are very varied and medical knowledge of EDS is still somewhat confined.

In this situation it is very helpful to get in contact with other people affected by EDS. Especially with rare illnesses, there is a development of communication which helps combat this uncertainty in dealing with the illness itself. Consultations with doctors can be reinforced by self help and personal knowledge of EDS expanded.

Of course the day-to-day limitations are sometimes more and sometimes less encumbering. When mobility diminishes, depression takes over, pains become restricting and peaceful sleep becomes a thing of the past, it becomes quite hard to get by.

Once again, actively dealing with the illness itself as well as communication with other EDS sufferers can be a good source of motivation; this way you know you're not suffering alone.

"My dream is to live in a world where every doctor makes their patients – especially those who are chronically ill – aware of the issue of self help, as we see ourselves as mediators between doctors and patients," says Barbara Kleffmann, Chairwoman of the Ehlers-Danlos Self Help e.V.

"Also, strengthened interdisciplinary exchange between doctors would not only be helpful, but also of great importance as a rare illness such as EDS cannot be managed by just one specific consultant."



Self Help – Ehlers-Danlos Self Help e.V.

In 2006, a small group of EDS sufferers had the vision to share their experiences and knowledge with those in a similar situation. From this emerged the Ehlers-Danlos Self Help e.V., a nationwide, active and acknowledged self help association.

Our aims:

- Recognition of the illness EDS under the Disabilities Act
- Expansion of the cooperation and reinforcement of the exchange of information between doctors, clinics, therapists and patients, on both a national and a European scale
- Support for EDS research
- Support for the construction of treatment centres for those affected
- Support for the set-up and expansion of therapy and rehabilitation measures
- Public work in nurseries and schools
- Expansion of medical advisory committees

Our activities:

- Continuous expansion and revision of flyers and brochures (reference texts)
- Emergency ID (3 languages)
- Findings folders
- Symptom catalogue inquiry form (14 page) / self-diagnosis forms
- Advanced training / education
- Technical lectures for those affected and members, as well as those who are simply interested
- Support of existing self help groups and establishment of new groups
- Exchange of experiences / group meetings / weekend seminars with those affected by EDS, members, those interested and advisors
- Attendance of conventions and selected health fairs
- Wide-ranging public work
- Internet presence

Contact
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EHLERS DANLOS Selbsthilfe e.V.

Additional Literature / Internet

Literature

Das Ehlers-Danlos-Syndrom – Eine interdisziplinäre Herausforderung
PD Dr. Andreas K. Luttkus, De Gruyter Verlag, 2011, ISBN 978-3-11-024955-2
Only paper-back books about Ehlers-Danlos syndrome are currently available in Germany.
However, this book is aimed specifically at doctors and therapists and is thus very academically structured.

Joint Hypermobility Handbook – A Guide for the Issues & Management of Ehlers-Danlos Syndrome Hypermobility Type and the Hypermobility Syndrome
Brad T. Tinkle, MD, PhD, Left Praw Press, 2010, ISBN 978-0-9825771-5-8
English literature on EDS is readily available. The above book is highly recommended and is also comprehensible for patients.

Gelenküberbeweglichkeit – Ursachen, Formen, Therapie
Dr. Med Günther Haberhauer, Dr. Med Marin Skoumal, Verlaghaus der Arzte, ISBN 10:3-901488-79-0
In this book, HMS, Marfan and EDS are explored among others.

Internet

www.achse-online.de
The Alliance of Rare Chronic Illnesses e.V. (ACHSE) represents the interests of rare chronic illnesses in Germany. The association forms a network for rare illnesses with its 90+ member organisations. The main task of ACHSE is to give people with rare illnesses the opportunity to be heard in public and to advocate their interests regarding politics and the health care system.

www.euroridis.org
Euroridis is a non-governmental, patient-led alliance of organisations and individuals that concerns itself with the aim of improving the quality of life of all people in Europe living with a rare illness.

www.orpha.net/national/DE-DE
Orphanet is an internet portal for information about rare illnesses and orphan drugs (pharmaceuticals for rare illnesses). Examples of services offered are: a directory of orphan drugs in varying stages of development, a directory of ongoing studies, registries, networks, newsletters, Centres for Rare Illnesses, etc. All patient organisations of rare illnesses can be found on the website www.orpha.net.

With the kind assistance of

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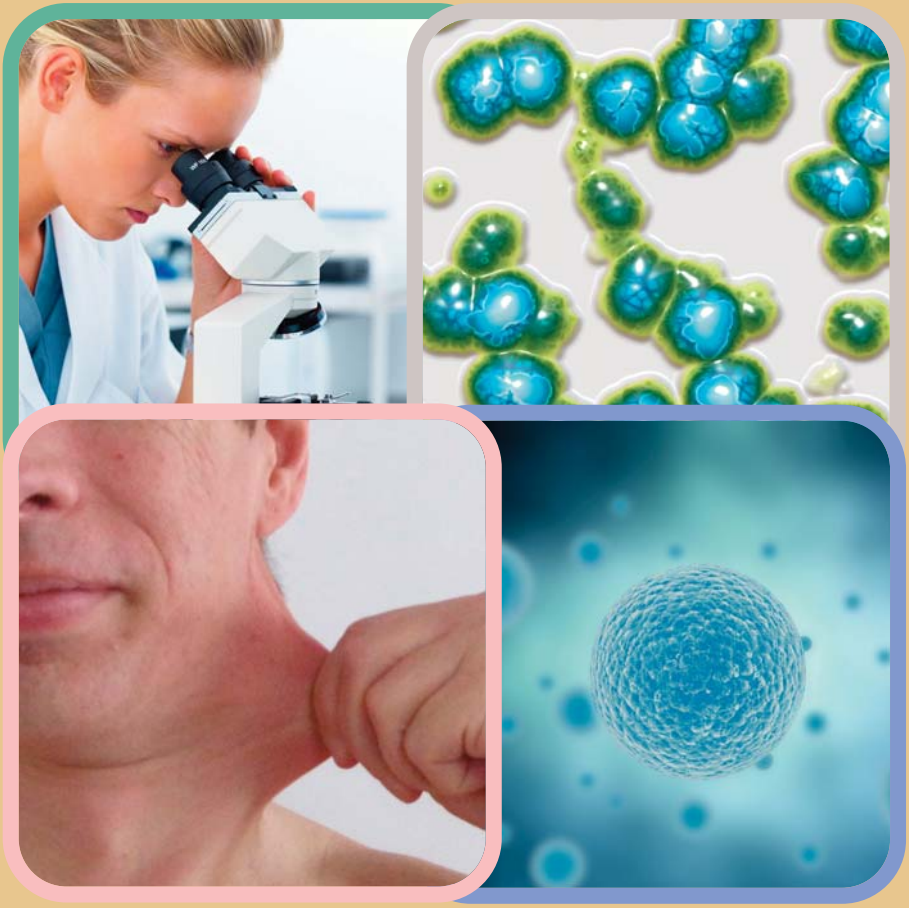

Information to go!

Health Library brochures on a num-ber of health-related topics are available free of charge at various patient contact points, self help groups, doctors' surgeries and clinics, rehab centres, pharmacies, medical stores and drugstores and also on Health Library online.

These brochures are not to be used as a substitute for a medical consultation, but rather offer additional information.

www.Health-Library.org
www.Health-Library.eu

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