So You Think You Might Have EDS?

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with contributions from Liza Sauls

It has been the experience of many of our members that a final diagnosis of EDS is the result of 'connecting the dots' of a person's history and a comprehensive review of the constellation of all symptoms; and includes, for many, thorough review of the medical histories of their families as well. Finally getting the diagnosis can be a relief to know that the symptoms are real and have a name; however limitations occur here as well. There is no cure, no 'fix', simply because the collection of experiences and symptoms now has a name and identity. But it does allow the patient and their families to know what they may be facing and allow them to become educated and proactive about the care they seek and require.

Based on the accumulated experiences of our members, the following essay is an overview of many of the symptoms and conditions that can be associated with EDS. It is not meant as a substitute for thorough medical review and care and supervision, but to help to view some aspects of EDS. Not every patient will have every symptom, as you will see, EDS can manifest in many different ways. The challenge of EDS is not to be able to identify each symptom, but to be able to see a pattern among several. Early diagnosis and intervention are the keys to living the best life possible.
With the recent TV programs on Ehlers-Danlos Syndrome, many people are coming to this website with questions about having EDS. Here is a list to help you determine if you need to see a geneticist who can correctly diagnose you.

**Caution:**

*If after reading the following article, you think you may have EDS, be sure to seek medical advice.*

*Please do not rely solely on this article or end your search with a self-diagnosis.*

EDS is a genetic syndrome and is diagnosed by a specialist, usually a geneticist or rheumatologist with extensive knowledge about EDS and its types:

- Classical,
- Hypermobile,
- Vascular,
- Kyphoscoliosis,
- Arthrochalasia,
- Dermatosparaxis

Ehlers-Danlos Syndrome is a group of disorders that affect connective tissues, which are tissues that support the skin, bones, blood vessels, eyes and other organs. Defects in connective tissues cause the signs and symptoms of Ehlers-Danlos Syndrome, which vary from mildly loose joints to life-threatening complications related to tissue structure and fragility.

A physical examination is required, including taking an extensive family history and using the Beighton scale, which measures Hypermobility. Depending on which type of EDS the physician believes you have, either a blood test or muscle biopsy will be taken.
How You Get It:

EDS is a genetic disorder, a mutation of one or several genes that make different types of collagen in your body, producing a defective tissue. A mutation in a gene on one of the first 22 nonsex chromosomes can lead to an autosomal disorder. Genes come in pairs.

Some types of EDS are autosomal dominant and others are autosomal recessive. Differing types of EDS affect different types of collagen.

- If a disease is autosomal dominant, it means you only need to get the abnormal gene from one parent in order for you to inherit the disease. One of the parents may often have the disease. Each child has a 50/50 chance of inheriting this disease.

- Recessive inheritance means both genes in a pair must be defective to cause disease. People with only one defective gene in the pair are considered carriers. However, they can pass the abnormal gene to their children.

Wrong Diagnoses:

Most people diagnosed with EDS have come the same long road where it seemed that nobody knew what was really wrong with you. Diagnoses of

- osteoarthritis,
- fibromyalgia,
- lupus,
- rheumatoid arthritis,
- rheumatic fever,
- multiple sclerosis,
- “growing pains”,
- and “it’s all in your head” are just some of them.

Often people get several misdiagnoses before finally being correctly diagnosed with EDS.
Symptoms of EDS:

Although all types of Ehlers-Danlos syndrome affect the joints and many also affect the skin, features vary by type and severity. An unusually large range of joint movement, hypermobility, occurs with most forms of Ehlers-Danlos syndrome, particularly the HEDS (hypermobility) and CEDS (Classical) types.

Below is a listing of symptoms that persons with EDS often have. These symptoms are broken down according to body systems. This list is not all-inclusive, but include those most frequently encountered.

Not everyone with EDS has them all and if you have some of them you may still not have Ehlers-Danlos Syndrome.

Joints:

• Different types of EDS have differing degrees of joint problems. Joint dislocation and incomplete dislocation called subluxation is common and recurrent.

• Spontaneous easy reduction or replacement of the finger digits and shoulders occurs.

• Hypermobile joints cause pain, and sometimes the "cracking" or "popping" of them feels like it relieves the pressure.

• ‘Pes planus’ or being flatfooted is common and feet can flatten even more as one ages.

• EDSers can develop osteoarthritis earlier than typical, and they often have difficulty or pain walking. They can appear klutzy.

• Some EDSers’ hands collapse from the pressure of a simple handshake. It is difficult to write, and often finger splints help a great deal.

• Cervical (neck) instability occurs in some types, and some people may have trouble holding up their head.
• Another frequent joint problem is fluid effusions into the knees, ankles and elbows, primarily in Classical or Kyphoscoliosis types. (effusion: The escape of fluid from the blood vessels or lymphatics into the tissues or a cavity)

• In the Kyphoscoliosis type, many infants have severe muscle hypotonia (floppy babies), generalized joint laxity and scoliosis at birth, or develop a progressive scoliosis (a curvature of the spine) within their first year of life.

• With Vascular EDS, joint hypermobility may be limited to the small joints of the feet and hands or may be very lax all over. As with other types, VEDS patients often are first diagnosed as hypermobile, only later learning that they have VEDS!

The range of hypermobility differs greatly among EDSers, even within types. The loose joints throughout life are unstable, prone to subluxation and dislocation, cause chronic pain and early-onset arthritis. Some people are only mildly affected by their EDS; others are completely debilitated.

Orthopedic procedures to stabilize or improve the joint's function sometimes put more than expected strain and stress on adjacent joints, leaving many EDSrs disappointed with the results.

So your back, hips, shoulders, knees, elbows, and other joints go out more often than you do, you might have EDS.
Skin:

General EDS skin traits include:

• Easy bruising, delayed wound healing, differing types of scarring.
• Thinner skin than normal, especially in the Vascular type.
• Those with VEDS have translucent skin where the blood vessels below are clearly visible.
• Soft, velvety skin that is fragile and sometimes highly elastic (stretchy) is found, especially in the Classical type.
• Classical type EDS may experience wounds that split open with little bleeding and leave scars that widen over time to create characteristic shallow "cigarette paper" scars.
• Surgical incisions may present problems with healing, with stitching EDS skin sometimes described as "like sewing butter." often requiring sutures being closer together and left in for a longer time than usual.
• With severe CEDS, even just leaning on the table with your elbows can cause the skin to split or may have molluscoid pseudotumors on elbows and knees.
  (molluscoid pseudotumors are small, spongy tumors consisting of fat surrounded by a fibrous capsule found over scars and pressure points).
• Skin that sags and wrinkles is characteristic of the Dermatosparaxis type of EDS. Extra (redundant) folds of skin may be present as affected children get older.
• Skin hyperextensibility to some degree occurs in all types of EDS except Vascular.

So your skin has so many bruises people think you earn your living as a boxer, you might have EDS.
Cardiovascular:

- People with EDS tend to have low body temperatures, may have trouble controlling their body temperatures when exposed to heat or cold, and many have blood pressure problems.

- Some have dilated aortic roots, incompetent heart valves, and autonomic dystonia or POTS (a syndrome where you have wide and serious blood pressure swings with position changes).

- Many people with EDS bruise very easily and often severely. It can be difficult for a medical professional to "feel" their pulse.

- Mitral valve prolapse is not a sign of EDS, though someone with EDS may have MVP; it is not diagnostic for this syndrome.

In some types, arteries including the aortas are very fragile and can rupture causing a medical emergency.

Note: IV (intravenous) access and even sometimes simply drawing blood for testing may require multiple attempts; using a "butterfly" needle and syringe is much more successful than the use of a vaccutainer which draws the blood rapidly by the use of suction. People with this concern must use extreme care and inform their healthcare providers of these possibilities.

Neurological Symptoms:

- Poor balance, severe headaches including migraines.

- Decreased deep tendon reflexes.

- Intracranial vascular abnormalities.

- Brain "fog", a sense of not being present; absence of focus or a lack of clarity

- Spinal stenosis (narrowing of spinal column) and/or scoliosis.
• Chiari malformation (the brain tonsils protrude down through the foramen at the base of the brain) occurs in some EDS patients.

So you had a school report card that said you were fidgety, uncoordinated, lazy, under-developed, and a complainer, you might have EDS.

Dental:

• Half of all EDS patients have a hypermobile tongue, and are able to touch at least the end of their nose with it easily.

• A high palate and crowded baby and adult teeth are common, even though many EDS patients have smaller than normal teeth. The high palate and smaller teeth can make fitting dentures very difficult even when explained to the dentist prior to the dentures being made.

• Pre-molar and molar teeth often have high cusps and deep fissures with root problems, and enamel hypoplasia can cause decay and possible early extractions. Sometimes teeth actually crumble when losing the enamel.

• Patients with Classical type suffer with juvenile periodontal disease. All EDSers are cavity prone, and have increased bleeding from anywhere in the oral cavity due to the fragility of tissues. Braces can cause problems as they can damage the gums and tongue while moving teeth quickly.

• TMJ (tempomandibular joint) pain and clicking occur in about 30% of the general population, and about the same incidence occurs in EDSers. Often if in a dental chair with your mouth open for an extended period of time, the joint will repeatedly sublux. Taking a pillow so you can prop your hand up to support your jaw during the procedure will prevent it from happening and also reduce your pain level from TMJ.
• Studies have proven that lidocaine (a local anesthetic used during dental procedures) often works poorly or not at all with EDS patients.

• Some people with EDS complain of always feeling like there is a lump in their throat when swallowing, and often have other swallowing and voice problems.

*Please refer to the Dental MRG available at EDNF Shop*

So a dentist ever gave you so much Novocain that his thumb was numb, and you could still feel everything, you might have EDS!

GI system:

Gastrointestinal complications of EDS run literally from one end to the other. Frequently EDSers suffer from reflux and GERD, stemming from an incompetent esophageal sphincter that allows stomach acid to backflow up the esophagus and cause burns in it. Diverticula have been seen throughout the G.I. tract.

• Tissue extensibility and laxity can also cause lack of contraction of the stomach, causing food to not move down into the intestines.

• Megacolon and rectal prolapse may also occur, primarily in childhood but megacolon is also seen in adults. (Megacolon is an abnormal dilatation of the colon (a part of the large intestines) that is not caused by mechanical obstruction. The dilatation is often accompanied by a paralysis of the peristaltic movements of the bowel.)

• Irritable bowel syndrome is a common co-diagnosis. Constipation can result from the flaccidity of the large bowel, more water being pulled from the stool the longer it remains in the colon, and from pain medications.

So your favorite foods are your digestive system’s LEAST favorite foods, you might have EDS.
Eyes:

An EDSer may have many different eye problems depending on the type of Ehlers-Danlos they have including blue sclera, microcornea (very small cornea), epicanthic folds, and wide-spaced eyes. Other common problems are:

• Many EDSers are photophobic, some squint causing an "angry" appearance and angiod streaks.

• Loose tendons and ligaments around the eye create hard working muscles that get tired. Strabismus is the medical terms for eye conditions commonly called by these various names: eye turns, crossed eyes, cross-eyed, wall-eyes, wandering eyes, deviating eye.

• Myopia (near-sightedness), astigmatism, and early presbyopia (a vision condition in which the crystalline lens of your eye loses its flexibility, making it difficult to focus on close objects.) occur often in EDS patients.

• Dry eyes are a common and uncomfortable problem.

• Other EDS related problems are detached retinas and ectopia (displaced) lenses.

• Persons with Ehlers-Danlos syndrome should see an Ophthalmologist annually so the internal eye can be checked for retinal and lens problems among other things. This is not an O.D, a Doctor of Optometry, but an MD with a specialty in eye issues.

Even during an eye exam, the exam itself can cause vertigo, nausea and headache feeling much like carsickness in some people.

*Please refer to the Ophthalmology MRG available at EDNF Shop*

So you change your eyeglass prescription more often than your wardrobe, you might have EDS!
Pain:
Pain with Ehlers-Danlos syndrome can range from none to chronic debilitating pain. It is subjective, individual, and different for each of us. For many patients, this is the worst symptom of all! Causes of this pain can be repeated trauma of constant instability from recent subluxations and dislocations as well as degenerative joint disease. Sometimes poor posture brought on by lax ligaments and weak abdominal muscles cause increased pressure on the spinal joints. Some with EDS do not have pain; others develop it later in life, and others begin to suffer severe pain as children.

- Many things are useful in treating EDS pain such as heated pools (92-94 degrees), gentle stretching, walking (if your joints allow), and emotional support that recognizes the degree of your pain and is non-judgmental.

- Occupational Therapists who make splints and assess what you may need may help to make daily life easier. Heat and cold packs help a lot. Always use cold for the first 24 hours after an injury to decrease swelling and limit bleeding into the area, and then switch to heat.

- Other possibly helpful things are yoga, relaxation therapy, massage, acupuncture or acupressure, diversion, TENS units and chiropractic maneuvers by a knowledgeable chiropractor.

- Common pain management problems are related to medications either in a too low a dose or prescribing the wrong medication, overemphasizing risks, using a "cookbook" approach, patients refusing helpful medications because they worry about addiction, and doctors afraid of prescribing because of their misunderstanding of the DEA laws.

- Often pain is undertreated in children, the elderly, and minorities. Less than 2% of all chronic pain patients (not just EDS patients) using pain medications correctly for pain become addicted. One can become dependent, but can be easily weaned off narcotics in a short amount of time.
• Medications often used with EDS are: muscle relaxants, NSAIDS, steroids, lidocaine patches, antidepressants, narcotic and non-narcotic pain medications. Remember that over 4,000 mg. of Tylenol daily causes liver damage. Different combinations of medications work for each individual.

• Pain can be completely debilitating and keep you from needed sleep. Often family and friends don’t believe you ... the worst part of all.

*Please refer to the Pain Management MRG available at EDNF Shop*

So you have days when you need a nap to rest up from the effort of getting out of bed in the morning, you might have EDS.

Emotional Effects of EDS:

Should either physical disability or chronic debilitating pain make your life feel destroyed, feelings of worthlessness and profound depression may set in; often talking to a counselor or medical professional will help. Regrettably, a tragedy occurs when we not only have to contend with no longer being able to do the things that we have loved doing, but also has to battle for family and friends’ belief, respect and understanding. It appears that everyone with an invisible disability sadly experiences this.

While someone with EDS is mourning their loss of ability and freedom, others often accuse them of just being lazy, malingering, or becoming an addict to the pain medications that allow them to live their life. Because of this, we should not confuse their endeavors to live life and be positive with assuming they are feeling well or doing better.
Knowing you have EDS doesn't suddenly make things worse for you physically, but may allow for better physical management, and ideally allow for the prevention of any real problems, even if none exist currently. So knowing you have it is not necessarily a bad thing. Personal doubt about one's mental and physical abilities can add to the fear that others can't possibly believe or understand what you're going through. Inability to cope with daily tasks or mental confusion can have a demoralizing effect.

But there is hope and help. You can join EDNF and learn how to help yourself, your doctor and your future. You are not alone.

So you are searching for knowledge, learning about EDS and educating others, you might have EDS!