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## **MEET ADVOCATE EXTRAORDINAIRE<sup>®</sup>™ CYNTHIA COLABELLA AND LEARN ABOUT EHLERS-DANLOS SYNDROME (EDS)**

**By Marla Schwartz**

What is Ehlers-Danlos syndrome? In the Ehlers-Danlos National Foundation (EDNF) home page ([www.ednf.org](http://www.ednf.org)), EDS is defined as a heterogeneous group of heritable connective tissue disorders, characterized by particular (joint) hypermobility, skin extensibility, and tissue fragility. There are six major types of EDS, which are classified according to their manifestations of signs and symptoms. Each type is a distinct disorder that "runs true" in a family, which means that an individual with the vascular type of EDS will not have a child with classical type EDS. Once thought to be a rare condition of the connective tissue, EDS is now believed to affect 1 in every 5,000 people.

The condition was named after the two doctors who first put the symptoms together and realized that they had discovered a very distinct disease. Edvard Ehlers was a Danish physician specializing in dermatology and Henri-Alexandre Danlos was a doctor at the Hospital Tenon, Paris, France. Although it was not until 1936, that Frederich Parkes-Weber proposed that the name "Ehlers-Danlos syndrome" be applied to the disorder, the condition was already noted by medical professionals.

The six distinct types of EDS are:

Classical Type: Patients have hyperextensible (stretchy) skin that scars and bruises easily, some hypermobility of the joints, and smooth velvety skin. They can suffer from sprains, dislocations/subluxations of the joints, flat feet, etc., along with the risk of hernias and surgical complications.

Hypermobility Type: The main symptom is generalized joint hypermobility, although the skin may be involved as well. Often, there is chronic pain in the joints and recurring dislocations/subluxations. There is often a family history with this disorder.

Vascular Type: Often thought to be the most serious type of EDS, patients have thin transparent skin; the face may be slender, with a pinched nose, prominent eyes and ears; easy bruising and susceptibility to rupture of the arteries, intestines, and uterus. The joints are often hypermobile; there can be rupture of the tendons and muscles, clubfoot, and varicose veins. The danger with this type is that there is a very high likelihood of early death in the third or fourth decade of life, although patients have been known to survive into their 50s. In the family history, there will often be sudden death in a close relative or sibling.

Kyphoscoliosis Type: Symptoms include generalized laxity of the joints, extreme hypertonia of the muscles at birth, scoliosis of the spine, and scleral fragility. The patient bruises easily due to the fragility of the tissues, and there can be arterial rupture and Marfanoid habitus. There will often be a family history.

Arthrochalasia Type: There is extreme general hypermobility of the joints, with recurring subluxations. There can be congenital hip dislocation. The skin is hyperextensible; there is easy scarring because of tissue fragility, hypotonia of the muscles, and Kyphoscoliosis.

Dermatosparaxis Type: Symptoms include extreme skin fragility and sagging skin that has a soft dough-like texture. There are often umbilical and inguinal hernias and premature rupture of the fetal membranes in pregnancy.

Symptoms are skin problems; soft velvet-like skin; fragile skin that bruises or tears easily; stretchy rubber-band-like skin; easy or severe bruising; poor and slow wound healing (usually taking weeks to months to heal); small harmless bumps under the skin, especially along the tops of the shins; joint problems; loose unstable joints, which cause frequent dislocations--usually occurring in the shoulders, knees, hips, collar bone or jaw; double jointedness (hyperextensible joints); extreme in some cases; joint pain from frequent dislocation that can often lead to osteoarthritis; eye problems; nearsightedness; occasionally extreme eye twitches; muscle twitches in various parts of the body; bradycardia; mitral valve prolapse or regurgitation, which can also occur in tricuspid valve; pulmonary hypertension; prolapse of pelvic organs (bladder, rectum, vagina, uterus); fragility of uterine walls; hiatal hernia; degenerative disc disease; Eagle syndrome, which is an elongated, ossified styloid process; and hearing problems due to inner ear cartilage problems.

Less common symptoms that may occur in the more rare forms of EDS include gum disease (EDS VIII); curvature of the spine (EDS VI); problems with blood clotting (EDS X); more serious eye conditions (EDS VI); pulmonary problems (EDS IV); weak blood vessels, intestines, or uterus that may lead to more serious complications (EDS IV).

Now meet a fellow P.A.N.D.O.R.A. member, Cindy Colavita, who has EDS. She is a very active member in P.A.N.D.O.R.A.'S Advocate Extraordinaire program. So how did her involvement in P.A.N.D.O.R.A. begin?

"Two-years ago I attended the P.A.N.D.O.R.A. conference in Ft. Lauderdale via {a recommendation} from my friend Charlotte, who happens to be on the Board of Directors for EDNF," Cindy explained. "Because with EDS the scale can range from day to day and patient to patient, there is no rhyme or reason [to it], even the weather affects us all [in an adverse way]."

"(In fact), the mutated gene [that causes EDS] has not been identified to date," Cindy continued. "[Therefore], there is no cure, and all the people I know with EDS have had more than mild symptoms [at] one time or another. It takes a toll on our daily lives; therefore, most are not able to function as a normal healthy person. The pain can vary from severe to chronic, never knowing whether it will be internal, headaches, or even foot pain. I have been close to death more than once in my life. Most people who have one type have several. I have classical, hypermobile, periodontal, and slight vascular."

So how do people find out that have EDS?

“Testing for EDS is limited, although some types can be tested for by skin biopsy. Others require a clinical diagnosis that is confirmed by a geneticist who specializes in rare disorders. In the case of EDS, if a family medical history accompanies a patient's symptoms, this helps in making a definitive diagnosis. The majority of people who have EDS will experience a mixture of symptoms from among the types rather than fitting neatly into a predetermined category.”

“The severity of the syndrome can vary even within members of the same family, with each person being affected differently. And everyone in the P.A.N.O.R.A. family will be able to relate to this statistic: It wasn't until 1997 that the chronic tiredness and pain accompanying EDS were taken into account in diagnosis of the disorder, and so many patients were told they were seeking attention and that their condition was "all in the mind." Perplexingly enough, this attitude still prevails in the minds of many physicians. What's even more disturbing about this fact is that the excessive bruising seen on youngsters who have EDS has often led to Social Services investigating parents for child abuse. And let your imagination soar on this one—at one time traveling circuses and sideshows featured individuals with EDS as freaks. They were put on display and the paying audience were “entertained” by seeing sufferers' dislocated joints and watching them pull their skin out to great lengths.”

One would think that in this day and age people would have more sense. Perhaps, finally, times are changing in the medical profession. So how does someone like Cindy cope with having such a devastating illness?

“I am 51 years of age and worked 20 years of my life. I climbed the corporate ladder and owned a home, but [because of EDS, I eventually] lost my job, benefits, husband, and home,” she pointed out.

So you see – it's not an easy road to travel.

“My lost benefits were due to me looking the picture of health, so misunderstood [is] this awful sickness,” Cindy continued. “So [with the] loss of a job...you're not eligible for unemployment benefits. I retired on long term disability (LTD) and then social security kicked in. [Although] I'm not allowed to discuss my LTD situation, I can say I took it to the 11th Circuit Court and lost, mainly because I was misunderstood and looked too healthy for a sick woman.”

“... it took years to find a diagnosis [after struggling with symptoms for] 48 years of my life. As a child I struggled in school, failed gym class, and had a broken elbow at age 6 from a fall on the grass. I worked extra hard and graduated with high honors in high school and received the Who's Who Award for students. [Later on] foot surgery in 1989 was a success, so I could walk better. My dental issues started long ago with 15 root canals and still continue to this day. My internal bleeding was misunderstood as a drinking problem (I don't drink).”

Additionally, "Three years ago I was misdiagnosed with a tumor and went for a second opinion at the Moffit Cancer Center. They did not find a tumor, and said, 'We have good news and bad.' The doctors explained there is no sign of a tumor but the bad news is you have EDS .... my reply was, what is that???. They informed me that only two places in the country take patients with this disorder, so off to Cleveland I went. They confirmed my EDS by doing a heart scan and physical exam. Several people in my family have the gene but are in denial; some have died at an early age [from] EDS," Cindy explained. "I'd say the worst thing about EDS is looking good and having smooth good-looking skin. Sounds odd but true...because of the way I look no one believed in me. I was sent to Yale, Lahey Clinic, New York hospitals, Shands, Cleveland Clinic, so on and so forth...tested with EEGs, EKGs, CAT scans, MRIs, and the list goes on...chronic fatigue, FM, ME, Lyme, you name it, I did it. No one knew what I had despite years of testing and no concrete results. I value each day as if it were my first and last. I still get rejection from the medical field, friends, and even family. I ask people to share a smile and be understanding of the challenges in my life. I know hunger and have been to food banks. I give my time to those in need, which helps me have contact with the outside world. My floors are dirty and my laundry not always folded because EDS does not allow my arms, hands, and entire body to complete tasks. At 51 there isn't help. I hear you need to be under 18 or 21 to get medical care. Rejection is a big part of my life. My life has no fringe benefits... no cable TV, No Ipod, No wifi— whatever these things are. My life is growing fruit and vegetables to eat. My nails are not polished, and I cut my own hair. People can say this is odd, but when a budget is in place, it can be done. I am happy and blessed by so many wonderful people who cherish my friendship and ones that believe in me. I say to all who see me SMILE...hold your head high for you are never alone!"

Cindy is right—you are never alone. If you think you may have EDS, get information and help from EDNF, although at this time, the National Office does *not* have a list of physician referrals; this is currently under development and will be made available through the on-line community in the near future.

You can, however, go to Contact Information on the site to find local groups, staff, and other information that will be helpful; the Mayo Clinic website <http://www.mayoclinic.com/health/ehlers-danlos-syndrome/DS00706> is also helpful, in order to get a thorough summation of the disease that covers symptoms, causes, and treatments.

If you are looking for eye doctors who have experience with EDS, contact Therapeutic Optometrist and Optometric Glaucoma Specialists Dr. Rich Driscoll and Dr. Diana Driscoll at: <http://www.TotalEyeCare.net>, email: Press@TotalEyeCare.net; phone, 817-656-5222; 6114 Colleyville Blvd., Colleyville, TX. The Keller office is located at 1834 Keller Parkway, Keller, TX.

The best way to find a good physician to treat any illness is to join a support group and get information from other patients. For information on world-wide support groups, go to: <http://www.edstoday.org/support/index.htm>, to find one near you.

Another highly recommended web-site is <http://www.ehlersdanlosnetwork.org/>, an all volunteer network led by EDS patients and family members who want to make a difference in the life of everyone who is suffering from this disease as well as sharing a passion for EDS research and education.

Finally, you can also use the EDS Support Group as a website, <http://www.ehlers-danlos.org>, because it lists names of physicians who are on its Medical Advisory Panel.

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