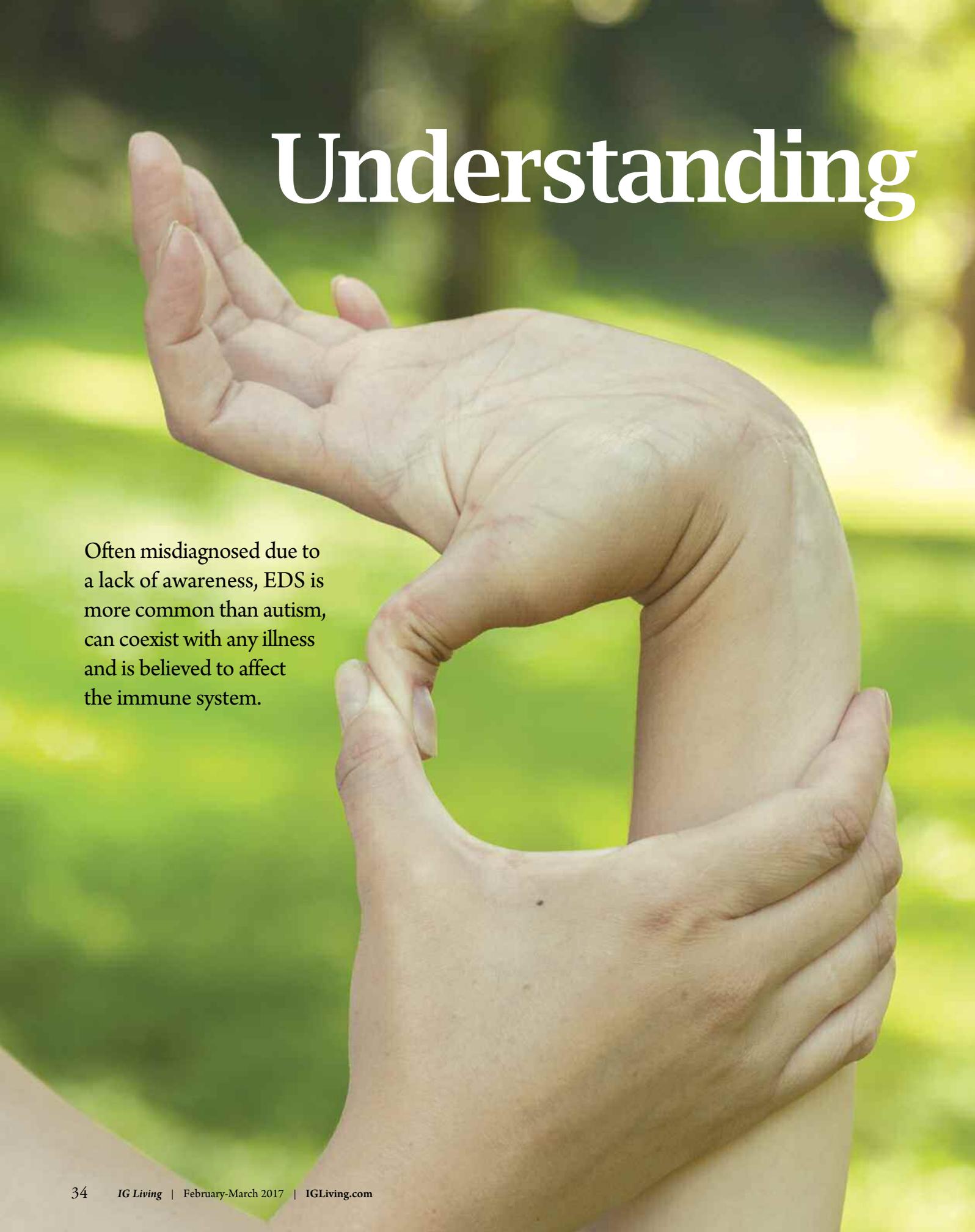


Understanding

A close-up photograph of a person's hands, one holding the other, against a blurred green background. The hands are positioned in a way that suggests a gesture of support or understanding. The lighting is soft and natural, highlighting the texture of the skin.

Often misdiagnosed due to a lack of awareness, EDS is more common than autism, can coexist with any illness and is believed to affect the immune system.

Ehlers-Danlos Syndrome

By Meredith Whitmore

SOME PEOPLE HAVE a list of health-related “justs” that they have grown up with. “My skin just bruises easily,” or “I’m just clumsy,” they might say. Maybe they’re “just tired a lot,” or they “just have migraines” or “just get dizzy” when they stand. Sometimes their heart “just races,” too. People who have struggled since childhood with these symptoms, and many more, often believe everyone else has them, too.

A healthy person would certainly say no, they do not have such a list. But for individuals with Ehlers-Danlos syndrome, a condition that few doctors understand, let alone are able to identify, this list is par for the course and rarely questioned — until symptoms worsen to the point of pain or even debilitation.

What Is Ehlers-Danlos Syndrome?

Commonly called EDS, this syndrome is a group of six genetic connective tissue disorders that often seem to masquerade as a variety of other illnesses. As a result, EDS patients spend an average of 20 years looking for a proper diagnosis.¹ During that time, they are often misdiagnosed with conditions such as irritable bowel syndrome, fibromyalgia, multiple sclerosis, scleroderma, depression, anxiety, seronegative rheumatoid arthritis, osteoarthritis, chronic fatigue syndrome, asthma, hypochondria, lupus, Sjögren’s syndrome and other autoimmune disorders. Like its cousin osteogenesis imperfecta, EDS is sometimes even misdiagnosed as child abuse since joint dislocations and bruising during normal activities are not uncommon.²

EDS can coexist with virtually any illness. Although it is not an immune disorder, EDS is thought to affect the immune system. The theory is — and at this stage, it is only a theory — EDS can predispose a patient to acquiring an immune-mediated disorder. And, immune-mediated disorders can definitely be exacerbated by preexisting, and usually undiagnosed, EDS. Symptoms such as infections, diarrhea, malabsorption, fatigue and headaches can all be worsened by EDS since it can share many immune system disorders’ problems and further weaken the body.²

EDS is caused by heritable defects in how the body produces collagen. Despite its relative obscurity, it is not a new condition. Around 400 B.C., Hippocrates was the first to describe the symptoms. It wasn’t until centuries later, however, that Edvard Ehlers, in 1901, and Henri-Alexandre Danlos, in 1908, conducted further research and discovered that hallmark features of the disorder are skin extensibility and fragility.³

Since collagen is found in bones, skin, tendons, ligaments, muscles, teeth, organs, corneas and blood vessels, all of which can be affected by the disorder, symptoms are diverse. An undiagnosed or diagnosed EDS patient’s doctors are typically inexperienced in diagnosing or treating a connective tissue disorder that manifests in virtually every bodily system. A significant problem, too, is that only 5 percent of those who have EDS know about the condition. Patients suspect that something is physically wrong with them at times — or perhaps much of the time — but the symptoms they suffer from often seem to be entirely unrelated to one another.²

EDS CAN COEXIST WITH VIRTUALLY ANY ILLNESS.

Heidi Collins, MD, who specializes in physical medicine and rehabilitation at Beacon Healthcare Medical Group, South Bend, Ind., is an EDS patient. She often quips: “If you can’t connect the issues, think connective tissues.” She explains that EDS is misunderstood and underdiagnosed because doctors are not unlike the parable of the four blind men touching an elephant. Each man feels something unique and real about the area they touch, but none is able to put all of the parts together as a whole. Likewise, doctors see individual symptoms and, unless they speak to each other about a common patient, are usually unable to diagnosis a unifying disorder.²



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President and Founder, Immune Deficiency Foundation

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Unexpected But Not Common

Some statistics indicate that between one in 2,500 and one in 5,000 people have some form of EDS.⁴ Clinical observation, however, shows that 3 percent of the population has hypermobility EDS, by far the most common type.⁵ This is more people than those diagnosed on the autism spectrum. While virtually everyone knows about autism, EDS is only now beginning to be better known in the medical community. Beyond the difficulty of recognizing the disorder by the untrained eye, it was thought for decades to be rare enough to require little to no study in medical schools. In fact, EDS is one of the few disorders to have a mascot, one it shares with the primary immunodeficiency disease community. The zebra represents the disease because of a somewhat folksy saying among doctors: “When you hear hoofbeats, think horses, not zebras.” In other words, doctors are trained to look for the most common conditions, not the exotic. Even so, more and more physicians and patients are learning that EDS is not that exotic at all.²

CLINICAL OBSERVATION,
HOWEVER, SHOWS THAT
3 PERCENT OF THE POPULATION
HAS HYPERMOBILITY EDS, BY
FAR THE MOST COMMON TYPE.

Other EDS Types

Beyond hypermobility, other varieties of EDS include the vascular and classic types, as well as the extremely rare kyphoscoliosis, arthrochalasia and dermatosparaxis types. Each has in common joint laxity, velvety skin, easy bruising and various systemic manifestations. Still, each type is a distinct condition that runs in families, affecting both men and women of all racial and ethnic backgrounds.⁶ Vascular type, seen in 5 percent of the EDS population, often results in a shortened lifespan; aortic dissections, ruptured organs and aneurysms are among its symptoms.⁷ Classic and hypermobility types, though disabling, do not shorten a patient’s life but often result in joint pain, poor healing, dysautonomia (an impairment of the autonomic nervous system) and POTS (postural orthostatic tachycardia syndrome).²

Overall, symptoms vary widely depending on the EDS type because the illness is a spectrum disorder, with some patients

mildly affected and others completely incapacitated. The most common signs, however, include hypermobility of joints, fatigue, chronic joint and musculoskeletal pain, migraines, dizziness when standing, velvety or hyperextensible skin, unexplained stretch marks, various gastrointestinal problems, easy bruising, seemingly spontaneous joint dislocations (known as subluxations), scarring, tachycardia, hernias and, more seriously, aortic dissection and aneurysms.⁸

Only a few of the genes responsible for causing EDS have been discovered. Biopsies and blood tests are used to identify some rarer types. There is no genetic test available for hypermobility type, so physicians rely on family history and a clinical diagnosis.⁶

The Road to Diagnosis — And Why It’s Important

Shani Weber, 48, of Mount Airy, Md., has experienced doctors’ mishandling of her symptoms. At age 16, after a doctor told her she did not have Marfan syndrome (a related condition), he deemed her “fine.” She attributed her widespread pain to gymnastics training and went on with life, although her “just” list was significant. She says that over the years, she had “lots of diagnoses for each symptom separately. Each knee had its own diagnosis of instability and tendonitis. My blood pressure drops were diagnosed as orthostatic hypotension. My shoulder was diagnosed with bursitis. And on and on.”

Doctors could not connect her problems until after an accident, when her shoulder would not heal. “I spent countless hours Googling to try to learn why I was not improving. I stumbled upon EDS, and it was a perfect match for my lifetime of joint issues, extreme hypermobility and more.” After educating herself and her physician about EDS and waiting a year to see a geneticist (doctors who can diagnose are in short supply), she was finally properly diagnosed and treated.¹⁰

Since EDS mimics other illnesses, it’s crucial in some cases to determine whether a diagnosis such as rheumatoid arthritis, lupus, multiple sclerosis or another condition could instead be EDS — or perhaps even be compounded by EDS. Dr. Collins recalls a colleague who had informally diagnosed herself with ALS, or Lou Gehrig’s disease. The woman was in tears of relief after Dr. Collins properly diagnosed her with EDS. “Everybody needs to keep this disorder on their radar,” says Dr. Collins. “She went through a box of tissues because I was telling her Ehlers-Danlos syndrome was causing her weakness rather than Lou Gehrig’s disease. She, a physician herself, was completely unaware that her symptoms were a poster child manifestation of Ehlers-Danlos syndrome.”²

Diagnosing EDS is much simpler than many doctors realize with a physical examination and two diagnostic scales, the Beighton and

Brighton. “My then 11-year-old son accurately applied the diagnostic criteria to some of his elementary school fellow students,” Dr. Collins explains, “And, funny enough, one of the students separately appeared in my clinic. So, my 11-year-old can diagnose it. If somebody is obviously hypermobile and very obviously hypermobile on the Beighton or Brighton, then a physician or patient can look at those lists of questions and apply them pretty simply.”²

The Beighton scale, most commonly used, is a series of flexibility tests. Patients are asked to bend their little fingers back beyond 90 degrees, push each thumb to their forearms, put their palms on the floor without bending their knees and hyperextend their knees and elbows. For each extension an individual is able to do, one point is given, for a total of nine points. A Beighton score of 4 or more, along with other symptoms such as chronic joint pain, is required for a diagnosis.¹⁰

Despite the relative ease of identifying EDS, Dr. Collins understands the frustration many patients experience while trying to find answers for their various symptoms. Undiagnosed “EDSers” are often accused of doctor shopping because they consult so many specialists — usually with no definitive answers. Tests such as MRIs, CT scans, blood work and X-rays usually come back negative or normal, so EDSers are frequently labeled hypochondriacs, even though nothing could be further from the truth.²

Treating EDS

Since EDS is a genetic disorder, it has no cure. Treatments for each type are palliative, designed to help patients manage the most debilitating symptoms. Physical therapy can help to strengthen muscles and joints, adding to their stability, but patients must be sure to see a physical therapist who is familiar with the condition or willing to learn about it. The wrong exercise or articular manipulation can damage a patient’s already hypermobile joints and muscles. Ring splints for fingers and other braces for major joints are commonly used to prevent hyperextension and, thus, further strain or damage them.

In extreme cases, surgery may be necessary to repair damaged ligaments and tendons. The risk for some patients is poor healing, especially with the classic type, because the skin is prone to scarring and is slow to recover.

Chronic pain is another struggle, but a doctor can help patients determine which pain medications and diets work best to manage it. Opioids are used only for acute pain, as long-term use can cause dependence. Migraines can be managed or even controlled with a variety of medications. Blood pressure, which is often low, can be improved with proper hydration, exercise, diet and various medications.

Though treatment is somewhat limited, many patients still find hope as they learn how to modify their lifestyles to remain healthy.¹¹

A Person’s List of “Justs” Could Be Significant

The “just” list that most EDSers have, whether or not they realize they suffer from EDS, usually means something despite a person’s occasional ability to rationalize it. “I haven’t met an EDS patient yet who doesn’t say, ‘This is how I’ve been all my life, so I didn’t think it was a problem,’” says Dr. Collins.²

DIAGNOSING EDS IS MUCH SIMPLER THAN MANY DOCTORS REALIZE WITH A PHYSICAL EXAMINATION AND TWO DIAGNOSTIC SCALES, THE BEIGHTON AND BRIGHTON.

Symptoms could be a path to something significant, however. If an individual suspects that they or someone they know has a form of EDS, the next step is to learn as much as possible about the disorder. They should educate themselves, and then educate their physicians, or find physicians who are experienced in dealing with EDS. Comprehensive information on EDS types and symptoms can be found at ehlers-danlos.com and other EDS organizations. Being proactive in the search for answers just might spare people years of pain and many, many “justs” that could be treated rather than accepted or deemed mysterious. ■

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