Ehlers-Danlos Syndrome (EDS) refers to a set of inherited disorders affecting connective tissue, such as skin, joints, and the walls of blood vessels. Hypermobility is one of the most common presentations of EDS.
PHOTOGRAPH BY U.A., ALAMY STOCK PHOTO

SCIENCE

Is Ehlers-Danlos Syndrome really so rare—or is it misdiagnosed?

Chronic pain. Extra-flexible joints. Stretchy skin. These are just some of the symptoms of the debilitating genetic disorder that's incredibly hard to diagnose.

BY ERIN BLAKEMORE

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For 41-year-old Baltimore physician Alissa Zingman, the first confusing symptoms—dislocated kneecaps and chronic pain—began when she was a child; by age 19 she had already undergone two orthopedic surgeries. Without a firm diagnosis for what was causing her various symptoms, Zingman doubted her own experiences and suffered through medical school, worrying she might be a hypochondriac. "For many years, I felt like the fact I was in pain all the time was something to be ashamed of," she says.

But after suffering for years with joint problems, chronic pain, and repeated illnesses, Zingman finally received a diagnosis in 2017: Ehlers-Danlos Syndrome, a genetic disorder that affects her body's ability to produce the collagen necessary to support her body's connective tissues.

Zingman isn't alone: Ehlers-Danlos is a growing watchword among a community of patients with a variety of health issues that range from unusually bendy joints to strange scars and chronic fatigue. Here's what's behind the wide range of syndromes—and why the genetic disorder can be so difficult to diagnose and treat.

What is Ehlers-Danlos Syndrome?

Ehlers-Danlos was first described by Danish physician Eduard Ehlers and French physician Henri-Alexandre Danlos in the early 20th century. Since then, the name has been extended to a host of inheritable connective tissue disorders with a variety of symptoms and severities. Though often referred to simply as "Ehlers-Danlos" or "EDS," the term officially covers 13 genetic connective tissue disorders.

Most forms of EDS are characterized by hypermobile joints and stretchy, velvety skin, but symptoms of the disorders vary. The most common subtype, hypermobile EDS (hEDS), involves joint instability, dislocations, joint pain, and fatigue.

Other subtypes affect different bodily systems: In brittle cornea syndrome (BCS), for example, patients' corneas become thin and fragile; in periodontal EDS (pEDS), the tissues that support patients' teeth break down. EDS syndromes cover everything from the skin to the skeleton and the internal organs, and the conditions can range from annoying to life-threatening.

An elusive diagnosis

Zingman's experience isn't rare: Despite the importance of early diagnosis, patients typically wait years and even decades after symptom onset to learn they have EDS—an average of 14 years, concluded one 2019 study; the same study noted a quarter of the patients waited more than 28 years for an EDS diagnosis.

Misdiagnosis is also common, and sex disparities plague EDS patients; the same 2019 study, which looked at a cohort of patients in Wales, found that men were diagnosed 8.5 years earlier on average than women.

Often, the joint issues, pain, and fatigue associated with many cases of EDS are only the beginning. The disorders are commonly associated with other conditions including POTS, digestive disorders, and sleep and anxiety orders. For Zingman, an immune system disorder made her EDS diagnosis that much more complicated. When she finally received a diagnosis of hEDS, she says, it was a relief—and an explanation for all of the orthopedic surgeries, disc herniations, and pelvic instability that affected her ability to walk.

"Getting a diagnosis changed everything," she says. "I now knew that my symptoms were real and that there had to be a way to get myself better."

A rare disease?

Self-advocacy and integrated health care made it possible for Zingman to finish her medical residency, and after watching her slowly rehabilitate herself, Zingman's EDS physician suggested she start her own practice devoted to treating the conditions. Today, Zingman is a physician specializing in orthopedics and preventive medicine, and her private practice in Silver Spring, MD treats many patients with EDS.

For Zingman, injury prevention, provider education, and coordinated health care are all key to managing EDS. But she's among a tiny subset of providers who specialize in the suite of genetic syndromes. Even among the small, but committed EDS community, the condition can present more questions than answers.

For example, the prevalence of the conditions are hotly contested; though some estimate that 1 in 5,000 people have an EDS subtype, it's still considered a rare disease. One 2019 estimate of the hypermobile subtype found that 1 in every 500 people in Wales has that syndrome. But "since misdiagnosis is common," a spokesperson for the Ehlers-Danlos Syndrome Research Foundation says, "we believe the actual number is much higher."

Nor is there a single treatment or cure for the condition, and awareness can be lacking even among medical professionals. But the tide is slowly turning in the world of EDS research and awareness. Today, the Ehlers-Danlos Society recognizes 18 "Centers of Excellence" nationwide—facilities that meet strict criteria for patient-centered care, proficiency in the condition, and specialized EDS services. And as research on the conditions slowly mounts, more and more providers are becoming proficient at caring for patients with EDS.

That wouldn't be possible without grassroots advocacy from EDS patients and caretakers committed to pushing research and treatment forward. Peer support ranges from a 50,000-user-strong subreddit to in-person groups at hospitals and clinics. Patients who went without diagnoses or effective treatments for years can direct one another to providers and provide empathy and anecdotal evidence from their own experiences. But Zingman notes that without systemic changes in diagnosis, treatment, and patient-centered care, many patients with Ehlers-Danlos are likely to remain in the shadows.

Nevertheless, Zingman, her colleagues, and many with EDS diagnoses note that even though EDS has no cure, it's both survivable and treatable. "Patients can live really productive, healthy lives," she says. One day, she hopes, medicine will catch up to patients' everyday experiences—and help them thrive, one small step at a time.

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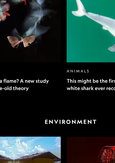

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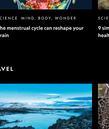

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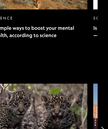

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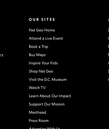

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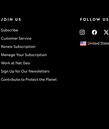

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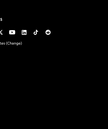

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