Why doctors won't diagnose or treat Ehlers Danlos Syndrome

Nearly everyone with EDS is now aware that finding a doctor who can diagnose it, or treat it, is almost impossible in America. This fact is actually the reason why I started writing articles about EDS. Until now, I've resisted writing an explanation of how and why this shortage happened, mainly because that would entail saying some grumpy things about my own profession and my colleagues. But I've changed my mind on this, because so very many people are harmed by doctors' neglect of their condition and its treatment, and they deserve to know why. Over years, this neglect grinds them down and causes many of them to wonder if the problem is somehow not real, or is only in their minds – which is what some of their doctors have implied. Indeed, I am now seeing people who need long term psychotherapy, to deal with "complex post-traumatic stress disorder (CPTSD)" caused by the harmful interactions they've had with doctors. This is terrible, and a scandalous indictment of our medical system.

I reassure my patients that their problem is indeed real, not "in their minds", and that the fault for their abandonment by the medical system lies in that system, not in themselves. I hope that this little article will convey that reassurance to some of the many, very many, people with EDS who will never get a diagnosis or treatment for their condition.

There is a notion abroad in the EDS world that doctors are being educated around the country on this subject and soon everyone can look forward to access to doctors who know about EDS, and what to do about it. This is a fantasy. The prevalence of the Ehlers Danlos Syndromes is thought to be at least one in five hundred, and quite likely twice that. At one in five hundred, we can expect about 670,000 Americans to have EDS: a number equal to the entire population of Nashville, Tennessee, or of Oklahoma City. Even on an optimistic estimate of the EDS clinics that could be created around the country, they could maybe diagnose this number of patients *in about thirty years.* And that's without allowing any time to *take care of* all those patients as well.

So how did we get here? I'll try to explain this in the smallest nutshell I can. The failure of the American medical system to deal with people with EDS is due to *institutional failure*. All such failures are explained by the history of the relevant institutions. Here's what happened to the institutions pertinent for people with EDS.

The Ehlers Danlos Syndromes have been written about and discussed for over a hundred years, as a loosely-defined group of inherited conditions in which the common thread was laxity of joints, so they move around more than normal, causing pain and injuries. Rheumatologists were the specialists dealing with joints, so they "owned" this topic. However, they had little to do with it, because they believed EDS was very rare, and anyway that nothing could be done for it. But in the late 20th century, *geneticists* started making important discoveries about EDS. They found that many EDS patients had specific mutations in their DNA, that could explain their joint laxity. The idea was floated that EDS could be defined as a group of conditions in which collagen, the protein that ligaments are made of, is defective because of a mutation in the DNA that designs or "encodes" it. This made geneticists the natural masters of this topic, and the rheumatologists' lack of interest was matched by the geneticists'new enthusiasm for diagnosing EDS and defining which type the patient had.

Genetics is almost entirely unintelligible to most medical doctors. It requires a knowledge of areas of biochemistry and cell biology that are so arcane and challenging that other specialists cannot hope to keep up with it, or even know how to interpret the tests that geneticists order. So it became accepted that as geneticists were busy *defining* the Ehlers Danlos Syndromes, they were also the only doctors who could *diagnose* them.

But there was a problem. The commonest type of EDS, previously known as Type 3, didn't actually seem to have a mutation to account for it. So it wasn't clear how to define it. After several earlier attempts to clean up and standardize the definition, in 2017 an "International Consortium" of researchers published new EDS definitions that they thought would help researchers to know they were all talking about the same things. However, they promoted their new definitions as items that *practicing doctors,* as well as researchers, should accept. The main novelty was that they recognized that Type 3 EDS graded imperceptibly into more minor conditions, in a range that included people who had very loose joints *that were not causing any trouble*, and

indeed might be an advantage in such activities as ballet dancing or gymnastics. Clearly, these people should not be identified as having a "medical condition."

The Consortium's solution was to create a new name to cover the people who were having less trouble from loose joints than the most severe EDS Type 3 patients, but not so little that they should be ignored and not offered help. For this large population, the term "Hypermobility Spectrum Disorder (HSD)" was coined. Only the most severe condition was now to be called hypermobile Ehlers Danlos Syndrome (hEDS).

Unfortunately, this coinage has been a disaster for the patients it was made to denote. The reasons are these:

1. The criteria proposed for HSD and hEDS are so complex that only a tiny number of enthusiasts is prepared to employ them. It takes a whole page even to display a summary chart of the criteria. In the last five years I have written summaries showing how to operate the new diagnostic framework, and I've recommended it to many doctors in many specialties. But not one doctor has chosen to learn how to use it (and I can't blame them for that).
2. The term "spectrum disorder" is usually used for psychiatric or behavioral conditions. Most people have heard of "autism spectrum disorder" but there is also "obsessive-compulsive spectrum disorder," "schizophrenia spectrum disorder" and on and on. The "spectrum" title carries two connotations. One is that it's a mental problem: but EDS is not. The second is that it's not very bad: autism spectrum disorder is not as bad as autism, schizophrenia spectrum disorder isn't as bad as schizophrenia, and so on. But hypermobility spectrum disorder (HSD) can be every bit as bad as the most severe cases of hEDS. So, the term HSD carries two false and demeaning implications: that HSD is in the mind, and that it's not very bad. These implications don't seem to have occurred to the writers of the 2017 EDS classification, but they are real and harmful.
3. It dawned on the geneticists that HSD and hEDS together were quite common, and that thanks to the internet, far more people than previously were seeking these diagnoses for themselves or family members. Genetics practices were getting overwhelmed by these patients, with the added irritation that the geneticists' calling-card – their ability to interpret arcane genetic tests – couldn't be applied to this population, since neither HSD nor hEDS come with a known mutation or mutations. So one after another, geneticists simply stopped seeing patients who wanted these conditions assessed. At present, the great majority of geneticists in the USA either refuse to see these patients altogether, or tell them they will be waiting for one to three years for their first appointment.
4. The obvious solution would be for the Consortium who invented the new categories, to educate other specialties with more manpower to take over EDS patients' diagnosis and care. But the Consortium was a self-selected club of international researchers. It had no status or even identity with bodies such as the World Health Organization, the Centers for Disease Control (CDC), the National Institutes of Health (NIH) or the American Medical Association (AMA). To my knowledge, the Consortium has made no attempts to promote its recommendations to these or other bodies. The result is that one searches in vain for review articles on EDS in the journals of most medical specialties. Even worse, the very few articles that do exist regurgitate the Consortium's dauntingly complex classification system, in a way that can only convince busy doctors that they should have nothing to do with this topic.
5. This lack of outreach included a lack of collaboration with American insurance companies. This is important because these companies specify which diagnoses they cover. Each covered diagnosis has a specific numerical code. But there is no code for HSD. And the code for hEDS doesn't specify what criteria are used to define it. Codes that do exist include ones for "hypermobility syndrome" and other terms that the Consortium said should be discarded. Because of this confusion, whether you will get reimbursed for treatment of an EDS-like condition is anyone's guess.

6. So much for the institutional failures circling around Ehlers Danlos Syndrome. The last and perhaps biggest failure can't be blamed on an institution but rather on the *absence* of one. The USA, unlike other developed countries, doesn't have a *national health system.* If it did, that system would nominate which specialties were responsible for which medical problems, how they should be paid for, and would ensure that doctors competent to manage them were available in all areas of the country. But in the USA there is no such oversight. Rheumatologists still announce on their national website that they are the people to see if you have an "inherited connective tissue disorder," a group which includes EDS and anything like it. But no one is charged with ensuring that their members do actually know about these conditions, or even see people who have them.

I hope what I've written above makes sense, and helps you understand your predicament and appreciate that it isn't your fault. As an afterword, I should comment that all the factors depriving people with EDS of good medical care operate just as much in big hospitals and prestigious universities, as in your local doctor's office. In fact, those institutions have *additional* handicaps that can further restrict their scope of practice: so much so that "We don't do that" could almost be their company motto.

Tips on how to work around these institutional defects are in several of the other articles on this website. A patient who read them told me they reminded her of a saying from her native Poland: "If you can't get in by the door, try the window." I wish you well, and I hope you find your window.

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