The EDS Predicament and How To Get Through It

If you think you may have Ehlers Danlos Syndrome (EDS), your predicament is that hardly any doctors know how to diagnose it. If you have been diagnosed with it, your predicament is that hardly any doctors know how to treat it. This article aims to help you deal with this dual predicament, and get better medical care.

The EDS predicament

Many people think they have EDS – or rather, one of them, since there are several Ehlers Danlos Syndromes. Some of these people have medical problems that resemble those of a relative who has been diagnosed with an EDS, so they wonder if they have one too. Or, they've been surfing the web to learn more about some ailment, and they find it can be part of an EDS, and then they read more, and it all seems to fit them. Or, they see a doctor who notices they have some loose joints and wonders aloud about EDS.

If you ask, say, an orthopedist or a rheumatologist whether you have an EDS, and if so what to do about it, you are likely to get a version of one of the following responses:

"It's just a name, don't worry about it."

"It's inherited, so there's nothing you can do about it."

"See a geneticist."

"It's probably fibromyalgia."

"Have you thought of getting counseling?"

None of these is helpful, if you are hurting and tired all the time, and getting worse, with an assortment of other symptoms that your doctors discount, or perhaps hint may be all in your mind.

The next step ought to be straightforward: see a doctor who does know about EDS, can tell you if you have it or not, and will get you started on the right treatment. This is certainly what you'd do if you suspected you might have pneumonia, or a brain tumor, or almost any other medical problem. But it doesn't work for EDS, for three reasons:

1. The only doctors whose business it is to diagnose EDS are geneticists (the specialists in inherited conditions). But there are very few of them, and they typically have waiting times of one to two years, or even longer.
2. Geneticists can *diagnose* the Ehlers Danlos Syndromes, but almost none of them *treat* it: they see their job as diagnosis only. These are, after all, lifelong conditions requiring lifelong treatment: geneticists could not possibly deal with the multitudes of new patients seeking diagnoses, if they also had to follow their patients and treat them for the rest of their lives. So, if you've been diagnosed with an EDS by a geneticist, you will have quickly learned that this doesn't mean you now have a doctor who can treat you for it.
3. Since the only doctors knowledgeable about EDS are the geneticists, it follows that *treatment of these conditions falls to doctors who know little or nothing about it*. Let me explain these worrisome statements a bit further:

Rheumatologists and EDS

The Ehlers Danlos Syndromes are categorized as "connective tissue disorders," and it is *rheumatologists* who are supposed to specialize in such conditions. So they should know about EDS, and in other countries than the USA, they often do. However, American rheumatologists have robustly refused to learn about *inherited* connective tissue disorders, preferring to focus instead on the interpretation of an ever-increasing number of laboratory tests they can order, and an ever-increasing number of medications they can prescribe. Only a few rheumatologists know enough to identify which patients should be referred to a geneticist for a diagnosis. I have yet to see a clinic note by a rheumatologist that displayed awareness of the range of medical problems that come with the Ehlers Danlos Syndromes, or how to treat them. And I have yet to meet an American rheumatologist at a national conference of EDS specialists (though rheumatologists *from other countries* are routinely among the speakers at such meetings). In the few cases where a rheumatologist has made an EDS diagnosis, they have failed to apply the required diagnostic criteria so their diagnosis is questionable; and then they have either given no treatment advice, or made recommendations that are at odds with what EDS specialists advise. (Having written this, I should add that it gives me no pleasure to point out the deficiencies of an entire medical specialty, but my responsibility is to inform my patients, not to stand by my colleagues.)

Orthopedists and EDS

Can orthopedists help people with an EDS? Things are brighter here – many orthopedists do know something about EDS and other "loose-ligament disorders," at least about those aspects that affect their surgical practice. However, if you have such a condition, it's important for you to be well-informed, because the orthopedist's knowledge may need some upgrading in order for them to help you, and avoid treatments that could make you worse. This is because in the course of their daily work, orthopedists see plenty of people with loose ligaments *who do not have EDS or any other medical problem.* A large part of orthopedists' work is with young, fit people with injuries sustained in athletics or dance. Some of these have loose ligaments but they are not causing trouble. In fact, loose ligaments can be a distinct benefit for a gymnast, or a dancer. For these fit young people with "hypermobile joints," injuries heal normally, and orthopedic surgery works as well as in other patients. Because most of the people with loose ligaments are like this, orthopedists easily overlook the occasional patient whose joints are causing far more trouble than the average athlete's, and whose tissues don't heal well, and who may not get success from surgery. But once alerted to the fact that someone with EDS has fragile tissues, they can adjust their surgical techniques to mitigate the increased risk of failure this could bring to some surgical procedures. They may also decide *not* to advise surgery for a condition for which that would usually be a good option. For more, see the article *EDS and Orthopedic Surgery* at *AlanSpanosMD.com*.

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The wide range of EDS symptoms

If an American doctor knows anything about the Ehlers Danlos Syndromes, it's usually only that people with EDS have loose joints, and maybe loose skin too. However, over the last 20 years it has become very clear that these are *multi-system conditions,* causing a wide array of symptoms, in different organs and body systems. Here is a partial list:

- Widespread pains, with various causes, and not just coming from joints.
- Headaches, often seeming to come from the back of the neck.
- Need for more sleep than most people, often going back to early childhood.
- Poor stamina compared to other people, often starting in childhood or adolescence.
- Feelings of faintness and "brain fog" on staying upright for periods that most people would easily tolerate. (This is called "orthostatic intolerance" or OI.)
- Chronic anxiety, dating from childhood or adolescence.
- Easy bruising.
- Slow healing of cuts and other injuries.
- Gastro-intestinal disorders, from nausea and bloating, to chronic constipation, often not responding well to standard treatments.
- Clumsiness, noticed from childhood.
- Poor response to local anesthetics, so it's hard to prevent pain during dental procedures.
- Pelvic pain with periods.

It's implausible that all these problems are due to "loose ligaments" or even to the broader category of "defective connective tissue." It's much more likely that the Ehlers Danlos Syndromes are truly multisystem conditions, in which different organs and tissues misbehave in different ways, for reasons we don't know. If this sounds strange, consider a condition like diabetes. This can cause weight loss or weight gain, excessive thirst, tiredness, burning pains in hands and feet, heart and eye trouble amongst other problems. However, the difference is that we understand why diabetes does all these things, but we don't know what it is about the Ehlers Danlos Syndromes that causes all the problems listed above. Nor do we know why each individual patient gets a unique mix of problems, with a different mix in each affected family member.

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The triple predicament for people who may have an EDS

To regroup: if you think you may have an EDS, then you have a triple predicament:

1. It will likely take a long, long time to see a geneticist to establish the *diagnosis.*
2. The diagnostician won't be providing you with *treatment*.
3. If you do have an EDS, this is a complex condition affecting different body systems, so you could spend a lot of time and money seeing a number of medical specialists to help with it, who know little or nothing about EDS. And they probably will not be communicating with each other about your care, which further reduces your chance that your care will be good care.

The diagnostic predicament changed in 2017: mainly for the worse

The diagnostic predicament – having to wait for ages to see someone who can make a diagnosis of EDS –changed in a big way in 2017. Early in that year, an international committee of experts agreed to rename the Ehlers Danlos Syndromes, and to recommend different *criteria* for their diagnosis. The committee was part of the "International Consortium on the Ehlers Danlos Syndromes," and had spent several years deliberating before they reported their recommendations. The results have mainly been bad for patients, as described below, but there is also a silver lining, as I explain later.

By far the most common type of EDS is the "hypermobile type", which had been defined in 1997 by what came to be known as the Villefranche criteria, after the town in France where an expert committee developed them. This was commonly abbreviated to HEDS or H-EDS.

What changed in 2017, was that the Consortium changed the criteria that had to be met, in order for a diagnosis of "hypermobile EDS" to be made; and they advised we abbreviate this to "hEDS" with a small "h." The criteria for this new diagnosis (hEDS) became much more restrictive than those for the previous diagnosis of HEDS. *This meant that many people who had previously been diagnosed with HEDS did not have hEDS by the new criteria.* For these patients, the experts invented a brand-new label for their condition, namely "Hypermobility Spectrum Disorder" or HSD. They specified criteria for this new "diagnosis". They were quite tentative both about the name (HSD), and its diagnostic criteria. It's not clear whether there is actually a biological basis for the name or the diagnosis, so HSD is really not a *diagnosis*, but rather a *label* for *a range of conditions,* that *may or may not turn out to be "real"*. (For more on this, see my article, "Why the New EDS Classification Matters.)

Patients naturally protested that being told their diagnosis had changed, looked like the doctors who previously made that diagnosis had been wrong, and that the patients had something else. Those who were applying for disability benefits were worried – rightly – that this apparent change could prejudice their application. The experts apparently hadn't thought of this, and after some hasty discussions, they reassured patients that if they already had a diagnosis of HEDS, then that wouldn't change. But *new* diagnoses would

have to follow the new criteria. So now we have a lot of people who get to keep a diagnosis HEDS (but with the first letter made lower-case), but only because it was diagnosed before 2017, and a lot of other people who have *exactly the same medical problems as them* but are being told they have something else because they were diagnosed in 2017!

It gets worse. Applying the new diagnostic criteria, a person might "fail" to meet criteria for hEDS. However, the criteria allow that in some such situations, if *another family member* is found, any time later, to meet criteria for hEDS, then that means the first person's diagnosis gets switched to hEDS too. This sounds like telling Mary she failed her SAT, then a year later, coming back and saying her younger sister Rachel just passed the test, so now Mary gets her SAT changed from "fail" to "pass." We don't normally think of a medical diagnosis as something that can be tipped one way or the other by diagnosis of another person, whether a family member or not. Patients are quite right to be disconcerted by this. They rightly feel that they had better corral other family members and get them to the geneticist in case their relatives' diagnoses change their own. But given the extraordinary wait-times to see geneticists, this tactic will take years to follow, even if the geneticist agrees to see the other family members.

And, it gets worse still. Some geneticists have become sufficiently discontented with the shifting diagnostic criteria for hEDS and HSD, that *they refuse to make these diagnoses at all*. This is because efforts to find laboratory tests that confirm hEDS or HSD have consistently failed (although *other* kinds of EDS can be confirmed by such tests). These geneticists take the view that if they can't back up their diagnoses with lab tests, they should not be in the business of making these diagnoses at all. So it is now possible to wait a year or two for a consultation with a medical specialist for the purpose of finding out what's the matter with you and other family members, only to be told that the specialist doesn't see that as his or her business. (A cynic might wonder if the reason some geneticists are abandoning this group of patients is just because their income depends on ordering expensive genetics tests, so why see patients with a condition that doesn't lead to any?)

And finally: none of these confusing new developments have been publicized outside the tiny world of card-carrying geneticists: about 1,500 in the entire USA. (For comparison, there are 32,000 cardiologists.) Nearly all other doctors,who only know a sentence or two (if that) about the Ehlers Danlos Syndromes, have no idea that the top experts just changed the meaning of the commonest EDS diagnosis, and invented a new name for many patients' conditions.

All of this makes effective medical care even less likely, for people with what-we-used-to-call Ehlers Danlos Syndromes. And it further reinforces the belief of many doctors that EDS, and anything like it, is "only a name" and "nothing that can be helped."

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Hypermobile EDS (hEDS) or Hypermobility Spectrum Disorder (HSD)?

The new diagnostic criteria that define hEDS are complex, confusing and arcane. They include words and concepts whose meaning most doctors (including me) had to look up when we first saw them. It requires some study and practice – as well as learning what all the words mean! - to implement them correctly. The important point is that the 2017 criteria are much more restrictive than the previous, so-called Villefranche criteria. The result is that most people diagnosed as having hypermobile EDS (HEDS) by the Villefranche criteria do not meet the 2017 criteria for hEDS with a small "h."

Instead, most of them meet criteria for the new diagnosis of "Hypermobility Spectrum Disorder." *The criteria for HSD are much simpler than for hEDS*. Essentially, the criteria are that the subject has a number of loose joints, plus also "musculoskeletal involvement" which could include:

- Chronic pain,
- Any evidence of joint damage,
- Abnormal curvature of the spine,
- Misaligned bones in the elbows or big toes,
- Clumsiness due to poor body-awareness ("disturbed proprioception"),
- Flat feet,
- Scoliosis,
- Kyphosis (forward curvature) of the upper spine
- "Swayback" in the lower spine.

There's no specification of how many of these features should be present to justify a diagnosis of HSD. Moreover, most of these features don't have clear-cut criteria themselves. - How much spinal curvature is "abnormal?" What defines a "flat foot" or a "swaybacked spine?" The Consortium was quite cautious and tentative in proposing this new term, and mentioned that it would be reassessed in future, at two-year intervals.

A very important point is that hEDS is thought of as being at one end of the Hypermobility Spectrum, but this does <u>not</u> mean that people with hEDS are more severely affected than people elsewhere on the spectrum: the spectrum is not a spectrum of severity of pain, or of joint looseness, or of anything else. It's more like a washing line, with various medical conditions hanging on it, rather randomly at different points. At one end is hEDS. Several other conditions hang on it elsewhere, and several of these are referred to as Hypermobility Spectrum Disorders in the new scheme. For our purposes we don't need to go into the different HSDs here, we only need to distinguish them from hEDS, which is at the end of the "washing line."

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The silver lining: something very helpful in the new diagnostic scheme

Now for the "silver lining" in the new EDS classification. So far as we know, people who meet criteria for an HSD (but not hEDS), can have *all the symptoms listed on page 3, at any degree of severity, and meriting exactly the same treatment as if the patient had hEDS*. Moreover, as is clear from the criteria listed on the previous page, *HSD is easy to identify* and this can be done by any doctor, and indeed by patients themselves.

So the striking change is that hEDS has now been redefined so that the diagnosis is quite hard to make, and far fewer people will now get this diagnosis than in the past; but the much larger group of people, with a Hypermobility Spectrum Disorder (but not hEDS), can be diagnosed very, very much more easily than in the past.

The straightforwardness with which HSD can now be assigned has the enormous advantage that *nonspecialist doctors can now confidently diagnose HSD, an inherited connective tissue disorder, closely related to hypermobile EDS, which can and should be treated in the same way as hEDS.* A geneticist isn't needed to make the diagnosis of HSD or to confirm it (especially since some of them actually refuse to do so, as mentioned above!). If the patient or their primary care physician wants a geneticist to confirm their diagnosis, that can be arranged (with a wait of up to two years), but it need not delay the patient's *treatment*. That treatment will be broadly appropriate, whatever more refined diagnosis the geneticist may make. (Often the geneticist's diagnosis will make no difference at all to the generalist's presumptive treatment.) The geneticist may perhaps suggest some *additions* to that treatment, but is most unlikely to suggest *changes* to it.

Your primary care doctor can make the diagnosis

To make a diagnosis of HSD, your doctor should use the protocol in the article accompanying this one, called *Joint Hypermobility Diagnosis for Non-Specialists*. I suggest that you make a copy and take it to your doctor, after reading it carefully yourself. It has nine pages. The first six explain why it is generalists, not specialists, who should be doing this, and summarize how hypermobility conditions are multisystem disorders. The diagnostic procedure itself is on pages 5 to 7 and will take about half an hour, so if your doctor wants to do it, he or she will need to set aside a longer-than-usual appointment. A nurse practitioner or physician assistant can also do the diagnostic procedure, though their supervising doctor should review the diagnosis and confirm it.

Medical treatment for Hypermobile Spectrum Disorder (and hEDS)

This is dealt with in the article, at *AlanSpanosMD.com*, entitled *Medical Care of Patients with a Hypermobility Disorder.* That article is aimed mainly at primary care practitioners and is appropriate whether the patient has HSD or hEDS. Family doctors can and should manage most of the problems that come with these diagnoses.

Helping your doctors to provide better medical care

Almost all doctors, in almost all specialties, are quite unaware of the information on the preceding seven pages, and I do not see this dismal situation changing any time soon. So if your doctors are to help you, *you will need to learn about your condition, and interest them in learning about it too*. This is an unfamiliar task, and one that takes care, patience and tact. *Your medical care is very likely to depend on your success at this, for a long time to come.* It is what the rest of this article deals with.

If you have an EDS, or if you think you might have one, then your best medical ally should be your Primary Care Practitioner (PCP), namely a family physician, pediatrician or general internist. Most of the medical problems listed on page 3 can be managed by your PCP. And they *should* be managed by your PCP, because this makes for much better medical care than farming out each symptom to a different specialist. There is simply no substitute for a good primary care doctor. A good doctor is one who *pays attention to you* and makes serious attempts to help you, rather than hustling you quickly out the door with a prescription or a referral to another doctor. Some PCPs are too busy and stressed to do this. But many can, and want to, so long as they don't feel overwhelmed by the complexities of your "case." I suggest the following tips for encouraging your doctor to get interested enough in your *condition*, and concerned enough about *you*, that they can overcome their unease at the difficulties of your case, and provide you with the medical care you need and deserve.

Your job is to learn enough about your condition that you can help your doctor to help you. This requires that, as you learn more yourself, you practise three general tactics in working with your doctor:

1. ***Let your doctor know about each of your medical problems.*** Several of the items on the list on page 3 are things people often don't seek help for – for instance, poor stamina, or a need for more sleep than most people, or feelings of faintness or "brain fog." Each of these merits medical help, and a good primary care doctor can provide that help. *Write a list and give it to the doctor*: much faster and less disconcerting than delivering it verbally, which is likely to sound just like the "moaning and whining" that makes doctors want to back out of the room.
2. ***However, propose only one, or at most two, problems to be dealt with at each visit.*** So you may need to see your doctor rather frequently at first, to deal with all your symptoms. Be really clear at the start of each appointment, which issues (no more than two!) you want to deal with each time. The more you add to the agenda, the more likely your doctor is to feel out of her depth, and hassled as the minutes tick by and she's getting more behind. You can help your doctor feel better about the consultation if you acknowledge this, by saying that there are other things you want help with "but we can deal with them another time so I don't put you behind."

3. ***Be tactful in dealing with your doctor.*** It's not easy to work with a professional when you know more than they do about the problem in hand. Most of us feel awkward in that situation, whichever side we're on. To help you, with a condition they know they are under-informed about, the doctor needs to feel you are supporting her and seeking her help, rather than berating her for her ignorance and twisting her arm to do what *you* want. Developing a "therapeutic alliance" with your doctor is crucial.

In the box below, I've suggested specific steps you can take to interest your doctor in getting involved in helping you figure out if you have a hypermobility disorder, and if so, what to do about it.

Six steps to help your doctor take better care of you

1. **Do some reading on EDS**, in addition to what is at my website at *AlanSpanosMD.com*. The best starting point is the website of the Ehlers Danlos Society, at *ehlers-danlos.com*. Others are *EDSAwareness.com,* and the support group organization *Inspire.com.* The standard book on hypermobility conditions is Brad Tinkle's *Joint Hypermobility Handbook*. This has a lot of good material in it, but it hasn't been updated with the changes in diagnostic criteria explained above. Make notes on what you learn from any of these sources: and remember, you now know more than the great majority of doctors do, about your condition. You will need to tactfully educate your doctor, to enable him or her to help you.
2. **Make two copies** of the articles *Joint Hypermobility Diagnosis for Nonspecialists,* and *Medical Care of Patients with a Hypermobility Disorder,* from my website*;* one set is for you and one for your doctor.
3. **Make a list** of the symptoms you have from the list on page 3 of this article.
4. **See your doctor.** Take along copies of the two articles, and the symptom list. Explain that you've been reading about EDS, that you've listed the medical problems you have that could be connected with HSD or EDS. Hand over your list. Explain that you've learned that most such cases are now called "Hypermobility Spectrum Disorder" and that this can be diagnosed by a primary care doctor, using a straightforward protocol in one of the articles.
5. **Ask your doctor** to take a look at the articles and see if he/she can help you, at least with some of your problems if not all of them. Request an appointment in a week or two to discuss that.
6. **Study the two articles** before you return for your next appointment. This is the appointment at which you may have to tactfully press your doctor to get involved in your diagnosis and treatment. You are much more likely to succeed if you can both refer to those two articles as you discuss this.

Seeing a geneticist

A good PCP can help with most of the problems that come with HSD or hEDS. But if they suspect you have one of the rarer forms of EDS, or something that is not in the EDS group at all (like Marfan syndrome), then they can refer you to a geneticist to assess those possibilities. In my article, *Joint Hypermobility Diagnosis for Non-Specialists,* I mention the features that should raise suspicions about the two main diagnoses that can resemble HSD and hEDS and which merit such an evaluation. *If your PCP calls a geneticist and explains their concern, the geneticist may agree to see you without the usual delay of many months for a new appointment.* But remember: geneticists do *diagnosis*, but they almost never get involved in long term *treatment.* You will still need your PCP for that.

Another option, which some PCPs prefer, is to refer <u>all</u> patients to a geneticist for confirmation of the diagnosis, not just the ones with "red flags" for rarer and more serious disorders, while the PCP treats them based on the presumptive diagnosis of HSD or hEDS. This is fine in principle, but if your doctor suggests this, remind her that the waiting time to see the geneticist is likely to be over a year, and that *some geneticists decline to assess patients with hypermobility conditions altogether*. The important thing is, to get on with treating all aspects of the patient's condition right now, whether or not you plan to see a geneticist.

Seeing other specialists

Sometimes, people do need to see specialists other than their PCP about their hEDS or HSD. Before seeing any specialist, I suggest you read about the problem you're seeking help with, in articles on my website, and at sources such as *ehlers-danlos.com*, *EDSAwareness.com*, and support groups such *Inspire.com*. And before you see a specialist, read the relevant parts of my article, *EDS: What Your Specialists Need to Know*, which summarizes what specialists may need to be reminded of, about your condition. It also contains handouts both for you, and for the specialist, to help you both focus on practical questions arising out of your condition. Also on my website are two articles on specific specialties, namely orthopedics, and gastro-enterology.

But does this work?

It would be rash to claim that if you follow the suggestions above, then you are bound to get prompt and expert medical help. The reality is that only a few primary care doctors are interested in learning how to help people with hEDS/HSD; and those with the interest may be too overloaded to have the time to do so. But some family doctors have risen to this challenge when approached tactfully by their patients, in the ways I suggest above. Those that have, have found that collaborating with their patients has been a rewarding experience both for the patients and the doctors too.

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