

Why the New EDS Classification Matters

This article deals with the major shake-up in the way doctors look at Ehlers Danlos Syndromes, that occurred in 2017. Early that year, an international committee changed the definitions of some kinds of EDS, and invented a new term for many of the conditions that previously were called “hypermobile EDS.” If you don’t understand the new naming system, then other things you read about EDS are likely to be quite confusing, as some will be using the new naming system, and others the old. The new system includes a major shift in how experts think about hypermobility disorders, that has some big effects on diagnosis and treatment, as I describe below.

Loose joints: sometimes a problem, sometimes not

Some people have joints that are unusually flexible, loose, lax, or “hypermobile.” All these terms mean the same thing, namely that joints move through a wider range than normal. If someone has many hypermobile joints, this is usually obvious to other people, and that person is identified as “double-jointed.” They have often learned in childhood that they can do contortions that most people can’t: perhaps twining the fingers of one hand round each other, or pulling up a foot all the way to hook around their neck. Or they may have found that they can easily do splits, dance “on point”, or do other maneuvers in dance or gymnastics that require flexibility, without any practising at all, to the amazement and envy of their peers.

Having hypermobile joints is, obviously, not in itself an illness. It’s actually an advantage in some sports like gymnastics, or in dance moves, or in playing certain musical instruments. However, some people with joint hypermobility do get problems with their loose joints. They may get repeated, painful dislocations of their shoulders, knees or other joints, and they may have joint sprains (painful stretches of the ligaments around the joints), and these may take much longer to heal than other people’s. These problems typically develop in childhood or adolescence. This condition, in which joints are not only loose, but vulnerable to injury, used to be called “Joint Hypermobility Syndrome (JHS).” But in 2017, an international panel of experts advised that this term be abandoned, in favor of a new term, “Hypermobility Spectrum Disorder (HSD),” based on the newly-minted concept of a “hypermobility spectrum.”

Hypermobile Spectrum Disorder and the newly-defined EDS types

This new name was part of a move to redefine, or to abandon, many previously used terms for conditions in which a main feature is joint hypermobility. But it will take time for the new classification, and the nomenclature that goes with it, to become known to most doctors. As of 2019, the new term Hypermobility Spectrum Disorder – *which is now the “officially approved” term for the great majority of people having trouble from their loose joints* – is unknown to almost all doctors except geneticists. If you mention HSD to your doctor, you had better say what it stands for, and be prepared for raised eyebrows.



Ehlers Danlos Syndrome (EDS) is actually a number of conditions, in all of which hypermobile joints are present; their other defining feature is that they are all present from birth, and generally run in families (though not all family members are affected). They are therefore assumed to be inherited. Geneticists have spent decades researching, and debating, how many types there are, and how they can be distinguished. This is a work in progress: there are still big areas of uncertainty, and controversy. However, the two thrusts of experts' recent deliberations are clear. The first is to insist that hypermobility conditions be defined, wherever possible, by the *underlying genetic abnormalities* that cause them, and whose presence can be detected by special (and extremely expensive) tests that geneticists can run. The second is, that conditions for which no such abnormalities are known, should be assigned to a putative "hypermobility spectrum." The word "spectrum" acknowledges that this is a broad category including conditions that may, or may not, turn out to be separable through later research. It also suggests that at least some of the conditions on the spectrum blend into one another, so distinguishing them is a matter of convention, or may even be subjective from one observer to another – just as the distinctions between adjacent colors of the rainbow are. Thus, red and orange are adjacent bands in a rainbow, and there is a real, physical distinction between them: the different wavelengths of the light that composes them. However, these wavelengths decrease *continuously* between red and orange. There is no line, step, or other marker to separate the red wavelengths from the orange. So, not surprisingly, if several people are asked to mark off red from orange, on a colored sheet where red fades to orange from one side to the other, each will make the cut at a different point. No amount of research, or negotiation, will change this: a spectrum is *essentially* a continuous span in which we name distinctions by methods other than objective discovery. We may name a part of the spectrum arbitrarily, and agree on the arbitrary definition, just so we all know what we're talking about. We could, for instance, nominate a particular wavelength as the point of separation between red and orange. But there is no physical basis for this distinction: the light doesn't flip a switch of any sort as it shifts across that wavelength. The wavelength-definition could be established by, for instance, physicists or fashion designers to facilitate communication with other physicists or fashion designers. But those definitions would mainly serve the specific interests of those groups of people: they would be arbitrary, in the sense that they could be changed from time to time without any change in knowledge about the light that composes those colors.

I encourage you to reflect carefully on the analogy with the spectrum of rainbow colors, because it is at odds with the big preference of both doctors and their patients, for clearly-distinguished, black-or-white "diagnoses." Medicine, like other sciences, devotes a lot of time and effort to making such distinctions, and finding special tests for them if necessary. There are many medical conditions that can be so distinguished, and in which the distinctions are crucial for getting the treatment right. For instance, cough and breathlessness can be caused by asthma or pneumonia (amongst others diseases). These are "categorical" diagnoses, meaning they are objectively-based, black-or-white diagnoses which are *not* on a spectrum with one another, in the sense that red and orange are on a spectrum. And if you have one of these conditions, the doctor had better get the diagnosis, and the name, right because the treatments for asthma and pneumonia are totally different.



Spectrum conditions in medicine: often misunderstood

There are many medical ailments that are not like asthma versus pneumonia. They are not “categorical,” black-or-white conditions, but true spectrum conditions, just like colors in the rainbow. Some of the most common, and serious, diseases are spectrum conditions. An example is hypertension, meaning blood pressure that’s high enough to be causing trouble. This can’t be a black-or-white, objectively-based diagnosis for several very good reasons. First, everyone has blood pressure – pressure is what makes blood flow, and without blood pressure, the flow stops and you promptly die. Second, blood pressure varies greatly throughout the day, and from day to day. Third, although pressure can be measured at any one point in time or space, there is no basis for identifying a particular pressure as the main or only one: it’s different in big blood vessels and small ones, and different in the arms, the legs, the brain, the stomach, and so on. Fourth, even you pick one time and place to measure blood pressure, there is no specific number at which blood pressure becomes a problem. Rather, the higher it is and the longer it is high, the more likely it is to be doing damage which, over a period of years or decades, may result in illness. All these are typical features of a *spectrum disorder*. It follows that defining high blood pressure as, say, a reading of 160/90 taken in the left arm on one occasion only, is a definition for human convenience rather than an objective way to identify a disease. The doctor wants to be able to tell the patient whether or not they “have hypertension” and the patient wants to know if they should be doing something about their blood pressure. But there is huge uncertainty, and therefore lots of controversy, over what measurements should lead to a diagnosis of hypertension. The definition of this condition is essentially a matter of convenience so as to provide a simple benchmark for deciding whether or not to recommend antihypertensive treatment to particular patients, and to getting physicians to have (at least some degree of) consistency in the treatment strategies. At levels of blood pressure that are very high, and sustained over time, we know that treatment reduces long-term illness, but in the wide “fuzzy” zone that slides towards normality, we really don’t know what is a disease, what is a risk for disease, and what is just part of the normal span of pressures from one well person to another well person. Moreover, *demanding to know if you “have hypertension” or not, is a mistaken question if your blood pressure is somewhere within that wide zone*. Better questions might be, “Is this something I need to worry about?”, or “Do we know enough about blood pressures in this range, to know if there’s a benefit to trying to reduce it?”

Other common and serious illnesses that are spectrum conditions are diabetes, depression and fibromyalgia. Sadly, doctors are not formally taught about the distinction between categorical diseases and spectrum conditions, and they often get this wrong. They often mistakenly imagine that a spectrum disorder is a categorical one, and that they should be as definite, and as unbending, in making the diagnosis as if they were diagnosing appendicitis. Much of the time, they act as if they believe that there are no spectrum disorders: that all diagnoses really refer to categorical entities that you either have, or haven’t “got”. They then feel obliged to make the diagnosis or reject it, once and for all. But in doing so, if the condition in question is a spectrum disorder, this process neglects the essential “fuzziness” of the condition’s borders, and the possibility that the patient may slide in and out of seeming to have such a disorder, from one time to another. Pushing for clarity by using arbitrary *numbers* (for blood pressure or blood sugar, for instance), can make the patient and doctor feel more

secure, but at the price of sacrificing a realistic view of the medical reality and what should be done about it, if anything. Treating a spectrum disorder as a categorical one is a convenience that compromises good medical care rather than furthering it.

Problems with EDS classification and Hypermobility Spectrum Disorder

Now to return to Ehlers Danlos Syndrome (EDS). For most of the twentieth century, this term was used for a loose cluster of conditions that were clearly inherited (because several people in a family were affected similarly) and in which hypermobile joints were a major feature. As genetic testing developed, the hope was that each of these conditions would prove to be caused by a specific genetic abnormality, and this abnormality would therefore define the condition. This goal was achieved for a number of the conditions called EDS. But the process foundered when applied to what was called the *hypermobile* type of EDS, referred to as H-EDS, and which was also called “Type 3.” Extensive genetic testing in large numbers of patients failed, and continues to fail, to identify its genetic basis. And this is by far the most common of the Ehlers Danlos Syndromes. So we have the awkward situation that the EDS that we most often see – common enough that, for instance, in North Carolina there seem to be one or several students with it in every big high school – is nevertheless out of step with all the other kinds of EDS in the very way that we’d like to be the defining feature of the group as a whole. What ought to be the flagship diagnosis that typifies the whole group, is in fact its major outlier.

On the next page is a summary of the concepts underpinning the new classification that distinguishes hypermobile EDS (hEDS) from hypermobility spectrum disorders (HSD).



The new classification: EDS and HSD in a nutshell

Now, armed with the important concepts of “categorical” versus “spectrum” disorders, we can return to the medical problems that sometimes go with having loose joints.

In a nutshell:

- 1.** Most people with loose joints have little or no trouble from them, ever. In athletics, dance and playing musical instruments, some of the loose joints may be a positive advantage. These people may have one or two dislocations or sprains due to their lax joints, but no long term or serious problems from them. They are now said to be *on the hypermobility spectrum*, but do *not* have a hypermobility spectrum disorder or HSD.
- 2.** Some people have some loose joints that are causing trouble over a longer period, with *recurrent* dislocations or sprains, *joint pain* that goes on for long periods, or evidence of *joint damage* on physical examination or x-rays. Or they may have other evidence of “musculoskeletal involvement,” not just loose joints. They are now said to have a Hypermobility Spectrum Disorder (HSD).
- 3.** A few people not only have loose joints, some of which are causing trouble (as in HSD), but also have evidence of abnormal tissues outside of joints. These people are now said to have hypermobile-type Ehlers Danlos Syndrome, which has been given the confusing acronym hEDS (with a small “h”). There are complex criteria to determine what features count towards the determination of tissue abnormalities outside the joints, and there is plenty of room for disagreement on this. Another area of disagreement is an added criterion, that if you are short of some of the criteria for hEDS, but you have at least one close family member who clearly does meet all those criteria, then you are deemed to have hEDS by virtue of that familial connection.
- 4.** Fewer people still have other conditions in which there are features outside of their loose joints, that merit a different diagnostic label for them. Some of these are conventionally included in the EDS group, such as “vascular-type Ehlers Danlos Syndrome (vEDS)” or “classical-type Ehlers Danlos Syndrome” (cEDS). Other conditions, such as Marfan syndrome, are deemed to be outside the EDS group altogether.

There is much to discuss, and to disagree with, in this new classification system. But for the purposes of this article, and assuming you are rather new to this whole topic, let’s return to the medical problems that can go along with having loose joints, always remembering that only a minority of people with loose joints do have these problems.

Hypermobility conditions: much more than just loose joints

In the last few years, it has become clear that the loose joints, which have always been regarded as the hallmark of the Ehlers Danlos Syndromes, are by no means the only problems for people with an EDS, or with HSD. They also often have *widespread pain* that doesn't seem to come from joints at all. They are often plagued with *exhaustion*, of several kinds: they may need to nap during the day, even if they sleep longer than most people at night; or they may be unable to sustain activities on their feet for as long as their peers. They may have periods when it's *difficult to think or remember things clearly*. They may have various *stomach and bowel problems*. They may sometimes have a *very fast heart rate*, and *low blood pressure*, which can cause them to be dizzy or faint. They may *bruise and bleed* more easily than others. They may notice that their *skin takes longer to heal* than most people's. They may suffer from *anxiety and/or panic attacks*. Women may have *chronic pelvic pain*. And there are various other medical problems that are more common in people with hypermobile joints. We call conditions like this, that have manifestations in several body functions and organs, "multisystem disorders."

Hypermobility conditions as multisystem disorders

A common outcome is that when doctors are presented with such a variety of symptoms, without a common thread to connect them, they are puzzled and they feel overwhelmed. Their response is to send the patient off to see several specialists, none of whom "connects the dots" by realizing that all these problems fit together as part of a hypermobility condition. If this is not realized, there's a strong tendency to infer that some, or all, of the patient's complaints are "in their minds." The fact that *anxiety* is a feature of hypermobility conditions, and may be obvious as an anxiety about one's medical condition, reinforces this inference. The diagnosis now becomes, not just, "Could be a mental problem" but more specifically "This patient is a hypochondriac." This conclusion is further solidified when the patient presents the results of their web searches and their belief that they have an Ehlers Danlos Syndrome – which their doctor was taught is a condition of loose joints and nothing more. That's what we were all taught, years ago. The new information on hypermobility syndromes as *multisystem* conditions is only known to the tiny group of doctors, almost all of whom are geneticists, who pay attention to this evolving field.

Because of this dismal process, most of the patients I see have had psychiatric diagnoses mistakenly ascribed to them for physical symptoms that are part of their hypermobility condition. Some of them eventually get resigned to their problems and stop seeking medical help. But then they stumble across "Ehlers Danlos Syndrome" by internet searching, and suddenly, for the first time, "find themselves" in what they are reading. At this point they quite rightly wonder why their condition was never properly evaluated and treated.



It matters to get a diagnosis

There's a real question as to whether the new diagnostic labels matter to someone with a hypermobility condition. There are four reasons why they do matter. First, if the patient has one of the more rare kinds of hypermobility condition such as vEDS or Marfan Syndrome, these can have dangerous or even fatal complications, that can be checked for and sometimes prevented. Second, the diagnostic label helps when the patient is seeking advice on whether or not to have a child, and wants to know the chance that her child might have the same condition as she does, or a worse one. Third, it's helpful to validate seemingly-vague problems like "fatigue" or "brain fog" that may have been brushed off in the past, rather than allowing patients to believe they are non-problems or signs of a fragile personality. Fourth and most importantly, patients deserve be treated; and they are much more likely to be treated if their condition is known and named, and all their symptoms are recognized.

An important question is, does someone with a hypermobility condition need to be cared for, in the long term, by a doctor with a special interest in this condition? For most patients, the answer is that they don't, which is a good thing since so very few doctors know anything worthwhile about how to treat the manifestations of hypermobility disorders. What most patients need is mainly to learn enough about their condition – which is usually HSD - to collaborate with their primary care practitioner (PCP), who is usually a family doctor, to make sure that each part of their condition is being addressed. Input from a doctor familiar with hypermobility syndromes may be helpful to get this process started, but ongoing follow-up by an EDS-aware doctor is only needed for unusually severe cases. This is because most of the problems of hypermobility syndromes also occur in other medical contexts, and the treatment is similar whether or not the patient has that problem as part of the larger condition of hypermobility syndrome.

The main exceptions - problems that are indeed treated differently when part of an EDS or HSD, are in three groups:

1. Problems specifically coming from the loose joints, whether pain, recurrent joint displacements or damage.
2. Stomach and intestinal problems. I recommend that unless these respond promptly to the usual remedies known to family doctors, they should be managed by a gastroenterologist, who should have taken a little time to read about hypermobility disorders.
3. Spinal problems, since these have implications for potential surgery, that are distinctive if the patient has a hypermobility disorder.

For information on what you need to know if you are to see these, or other, specialists, see my article *"EDS: What Your Specialists Need to Know."*



Life planning with a hypermobility condition

Apart from specifically medical help, people with hypermobility conditions deserve counseling on how their condition should affect their future choices in life. Should the school athlete continue to play contact sports despite recurrent dislocations? Should the college student who's falling asleep in class struggle on, or work towards a degree part-time instead? Should people with these conditions aim to be as active as possible physically, or avoid sports and exercise? Should they have children? What will happen to them as they age? In the long run, making the best decisions about these choices may have more effect on the person's long-term happiness and fulfillment than anything else. I've addressed some of these questions in the article *Common Questions from People with Hypermobility Conditions*, on this website.

Information resources on hypermobility conditions

Finally, everyone with a hypermobility condition should continue to educate themselves about their condition and its treatment. For the foreseeable future, patients will continue to need to do this so as to educate their doctors to help them better. Specific suggestions on how to develop such a collaborative relationship with primary care doctors are in the article, *I Think I May Have EDS*, on the same website as this one.

The main starting point for general information on EDS is the website of the Ehlers Danlos Society at ehlers-danlos.com. This site is evolving, so it's worth checking every few months to see what's new there. It is developing informational materials of all sorts, for doctors as well as patients, and also hosts online support groups and message boards through which people with EDS, and their families, can get in touch with others and share experience and advice. Other useful sites are EDSAwareness.com, and the support groups at Inspire.com. Brad Tinkle's book, *Joint Hypermobility Handbook*, predates the new classification, but otherwise is a good source of information on many aspects of joint hypermobility conditions.

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