

# **Ehlers Danlos Syndrome (EDS) and the Joint Hypermobility Syndrome (JHS)**

Some people have joints that are unusually flexible, loose, or “hypermobile.” Having hypermobile joints is not in itself an illness. Indeed, it can be a real advantage in some sports like gymnastics, or in playing 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 that take much longer to heal than other people’s. These problems typically develop in childhood or adolescence. “Joint Hypermobility Syndrome (JHS)” refers to the condition of having such problems with a number of loose joints. Defined this way, JHS is quite common, often showing up as the teenager who wins awards at gymnastics, but is now having dislocations in one joint after another. This condition used to be called “Benign Joint Hypermobility Syndrome (BJHS)” to distinguish it from more serious conditions like Ehlers Danlos Syndrome, in which there were more things going on than just loose joints.

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 are therefore assumed to be inherited. Over eight types have been distinguished, and they used to be identified by numbers, so you will still sometimes see “EDS 1” or “EDS 4” referred to in doctors’ notes – sometimes, confusingly, using latin numerals.

Until recently, it was thought that JHS was different from EDS. This is now believed, by most experts, to be a mistake. It is now thought that one particular type of EDS – formerly called “Type 3” and now called “Hypermobility Type”, is the same as Joint Hypermobility Syndrome. EDS Hypermobility Type is abbreviated to EDS-H, EDS-HT or H-EDS. I’ll refer to it as H-EDS. It is now thought to be simply a more severe kind of the same condition as JHS, with the addition of features in addition to the loose joints that are the hallmark features of both. This view, however, is unproven at present. It may yet turn out that there’s an underlying difference between people with JHS who “just” have loose joints , and those who have the other features of H-EDS. If there is, then this may important in helping to counsel people about their likely future health, and whether the condition is relevant for their children. For now, though, the consensus is that H-EDS and JHS should be regarded as the same condition, and that the term JHS should probably be retired.

In practice, people still tend to get diagnosed as having EDS when their symptoms are severe, or when they have many of the features of H-EDS, and JHS when their symptoms are less severe, and they have less features of H-EDS. This is because very, very few doctors stay abreast with the evolving expert

opinions in this field, so the term JHS is far from defunct. But in this article, from now on I will use the term H-EDS to apply to people who were formally classified with either of these titles. H-EDS is by far the most common form of EDS – I estimate its prevalence in North Carolina as around one in five hundred, which means there are around 19,000 people with this condition in our State. But the great majority of these people will never see a doctor who is familiar with H-EDS. So people with H-EDS must get educated about their condition, so they can help their doctors to help them better. Hence this website.

In the last few years, it has become clear that the loose joints, which have always been regarded as the hallmark of H-EDS, are by no means the only problems for patients with this condition. They also have widespread pain that doesn't 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 properly. 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 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 H-EDS.

A common outcome is that people with symptoms like these find that doctors are puzzled by them and have little or no treatment to offer. They eventually get resigned to their problems and stop seeking medical help. Then they stumble across "Ehlers Danlos Syndrome" by internet searching, and suddenly seem, for the first time, to find themselves in what they are reading. At this point they quite rightly wonder why their condition hasn't been properly evaluated and treated.

There is at present no lab test or x-ray that confirms H-EDS. The diagnosis is made by identifying the pattern in the patient's medical history, their current symptoms, and findings from the doctor's physical examination. A standard formula, or set of criteria, has been proposed to standardize recognition of this pattern, but its value hasn't been proven. A recent International Symposium (in May 2016) was charged with developing new criteria for diagnosis, but its work is still in progress, and its participants don't expect to report on their recommendations till about March of 2017.

Sometimes the diagnosis is very obvious to anyone familiar with the condition; sometimes it's quite difficult and uncertain. But it's important to make the right diagnosis if possible, for several reasons. First, the patient should learn whether they have one of the other forms of EDS, because some of these carry major medical risks such as aneurysms and internal ruptures that can be life-threatening, and should be searched for. Second, the patient should know what the implications are for their children: are they likely to have the condition, and if so how severe might it be? Third, it's helpful to validate seemingly-vague problems like "fatigue" or "brain fog" that may have been brushed off in the past. Finally and most importantly, patients deserve be treated! This begins with assessing their various medical problems and

prioritizing them: for many people the first priority is pain, but for just as many, it's fatigue, or one of the other problems mentioned above. Most of these problems can be helped, even though they can't be permanently cured.

When EDS is suspected, a patient may be referred to a geneticist to make the diagnosis. Seeing a geneticist is no small thing: the waiting list is typically several months to a year long – and sometimes even longer! - and the geneticist may feel obliged to run lab tests that are not covered by insurance, and may cost many hundreds of dollars. But H-EDS is by far the commonest type of EDS, and as yet there are no known tests that confirm it, though there are tests to confirm the other, much rarer forms. For this reason, it's questionable whether a person with clearcut H-EDS, and no features of the other types, should see a geneticist. This is an area of uncertainty, and therefore of controversy. It may be that when the members of the International Symposium publish their conclusions, they will present a consensus on this.

That said, there are cases of H-EDS for which a geneticist's assessment should be obtained. These are when the pattern of features, or "presentation", is vague, or a mixture of features of more than one type of EDS; or the patient seems to clearly have H-EDS but a family member has features of one of the other types, or of another condition altogether, such as Marfan Syndrome. It should be added, though, that the geneticist's role is diagnosis only, not treatment – just as a radiologist may detect the fracture in an x-ray picture, but it's someone else's job to apply the plaster cast or straighten the bone.

An important question is, does someone with H-EDS 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 much about this condition! What most patients need is mainly to learn enough about H-EDS to collaborate with their main physician (usually a family doctor), to make sure that each part of their condition is being addressed. Input from a doctor specializing in H-EDS may be helpful to get this process started, but ongoing specialist follow-up is only needed for unusually severe cases. This works for most of the problems because they mainly 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 H-EDS. But that entails first acknowledging the problem and its level of severity. As mentioned above, many patients with H-EDS have problems other than loose joints, which have been ignored or passed over as trivial for years or decades by their physicians. Their local doctors will need reminding that symptoms such as excessive sleepiness, exhaustion and so on are not normal and can be treated.

That said, a few of the problems are indeed treated differently when part of EDS/JHS. This is especially true of problems usually dealt with by an orthopedist, a physical therapist (PT) or a chiropractor. Patients themselves can learn how the treatment they are being offered should be altered to take account of their H-EDS. Rarely, such alterations may be large enough that it may be worth travelling to see an orthopedist or PT with a special interest in H-EDS. Other articles on this website deal specifically with this, and other H-EDS-specific aspects of treatment.

Also, people with H-EDS deserve counseling on how their condition is likely to 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 H-EDS 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. The person asking such questions deserves to have them addressed by someone who has seen many H-EDS patients as well as studied the scientific research on this condition.

Finally, everyone with H-EDS should educate themselves about their condition, and there are two main ways to do that. First, get Brad Tinkle's splendid book, *Joint Hypermobility Handbook*. If you read it and "find yourself" in its pages, then you almost certainly have made the correct diagnosis, and you now know more than most doctors about your condition. Second, go to the website of the Ehlers Danlos Society at [ehlers-danlos.com](http://ehlers-danlos.com), and start getting familiar with its various departments. This site is rapidly changing, as the Society is a new creation, as of late 2015, which has taken over from its predecessor, the Ehlers Danlos National Foundation. So it's worth checking on the site every few months to see what's new there. It is developing learning materials of all sorts, 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.

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