Ehlers-Danlos Syndrome Hypermobility Type III

I. Do I have Ehlers-Danlos Syndrome or EDS, or am I just simply hypermobile and have diffuse pain all over? I hurt all over. These are questions that I am often asked from patients that present to our office practice.

There are several types of Ehlers-Danlos Syndrome. We will discuss the various types subsequently. However, the most common type that we see in clinical practice is the type known as Type III. This is also known as the hypermobile type of Ehlers-Danlos Syndrome (hEDS). It has also been referred as benign joint hypermobility, or joint hypermobility syndrome (JHS).

The typical patient presents to us with diffuse pain, especially in the shoulders, upper back, interscapular area and elsewhere. The patients typically do not stand for long periods of time and get dizzy and may have a history of fainting or near-fainting. Oftentimes, the patients are extremely tired, cannot get going in the morning, and oftentimes have to lie down when standing for periods of time. Bright light or sound may disturb them. The patients oftentimes complain of their joints being quite flexible and oftentimes subluxing, or popping out. It is not unusual for the patients to have their knees, elbows, ankles, wrists and even jaw pop out, or their cervical spine hypermobile. Even hyperflexibility in the lumbar spine may make the patient able to touch the ground with their palms to hands quite easily.

Many patients state that they were quite athletic and were good dancers, or good gymnasts during high school and grade school. They even comment that they believe they are double-jointed. A family history of this is often important in determining whether a person has hypermobile Ehlers-Danlos Syndrome. There is a way to type for hypermobility in a scoring system known as Beighton score. We will subsequently discuss that also. While physicians can do this simple scoring system in the office setting, the patient can also perform it at home. Oftentimes, the physician uses a measuring instrument known as a goniometer to get more precise information on the flexibility of the various joints tested, which include the joints of the hand, lower back, elbow, and knee for this assessment.

The skin is oftentimes soft and very hyperextensible in patients who present to our office with suspected hypermobile Ehlers-Danlos Syndrome. Specifically, the skin and the nondominant forearm should be tested to see if it stretches more than 2 cm. The skin may also be velvety and mildly hyperextensible. Patients may have striae on their back, thighs, breast areas and abdomen, and they may also have a history of recurrent hernias of the abdomen or inguinal area. Interestingly, some patients give a history of pelvic floor abnormalities with rectal or uterine prolapse as children. Some patients may present with arm spans greater than their height spans. However, other entities such as Marfan syndrome, which we will discuss below, should be considered first with this presentation.

There are definite criteria to diagnosis hypermobile Ehlers-Danlos Syndrome and we will discuss that below. These criteria should be met for a precise definition. Unfortunately, there is no blood test, lab test, or imaging modality than confirms a diagnosis and the diagnosis completely clinical. There is no genetic testing that has been identified for hypermobile Ehlers-Danlos Syndrome. What may complicate the issue is there are some patients who present with this syndrome complex de-novo and do not have a family history despite a careful search from the physician and clinician. These are the minority of cases, however, but the exact percentage is not known.

Many patients present to our office with prior diagnoses of depression and anxiety or psychiatric illness attributed to them, but they know they have something real and abnormal that is not purely psychiatric. The patients hurt all over and have diffuse pain, which keeps them from functioning properly. Many cannot perform any gainful employment. Certainly, they become anxious and depressed because of their non-functional status. They have poor exercise tolerance. Autonomic nervous system features, such as exercise intolerance, orthostatic intolerance, where one cannot stand up without getting brain fog or dizzy, and chronic fatigue are almost universally present in these patients. There is a high percentage of females with this problem, but we do also see males in addition, since it is believed that if a person has this disorder they can transmit it genetically to one or two of their children (autosomal dominant transmission).

Therefore, when a patient presents with many of these symptoms that we have described above, we will do a clinical test for joint hypermobility, which can be performed quickly in the office setting and give a scoring system up to nine points. We look for systemic features that reflect a syndrome, such as family history or many musculoskeletal complications such as pain in several limbs that has been occurring for more than three months, or pain that is widespread and recurrent joint dislocations or joint instability without any trauma having occurred; that is they occur spontaneously. We also look to exclude other possibilities that may mimic hypermobile Ehlers-Danlos Syndrome, which will be discussed below. These syndromes have abnormal collagen tissue and abnormal connective tissue as does Ehlers-Danlos and can give joint laxity and hypermobility, but have other features, some of which can be life-threatening and need to be identified. Many rheumatologists will send us patients whom they have evaluated for connective tissue disease and have excluded these (example, excluded lupus, rheumatoid arthritis, scleroderma, and so forth) and have us evaluate the patients who have hypermobility and pain for autonomic dysfunction, which we will detail below also.

It is not difficult to diagnosis hypermobile Ehlers-Danlos Syndrome (hEDS) but a very detailed history and physical examination is required. A very thorough family history is also required, and this may even involve examining some of the family members. Excluding other entities that present with abnormal collagen composition or other types of hereditary tissue diseases is extremely important and consultation with a skilled rheumatologist and an autonomic nervous system expert is often needed.

We believe that the fatigue and exercise intolerance and oftentimes the shortness of breath, palpitations, and chest pains that many patients with hypermobile Ehlers-Danlos Syndrome present with are all the result of the dysfunction of the autonomic nervous system, which almost always goes hand in hand with the hypermobile Ehlers-Danlos Syndrome. This we can test for objectively with even some diagnostic test modalities that give us quantitative information.

However, as mentioned before, there is no genetic testing or lab testing that is diagnostic of Ehlers-Danlos Syndrome. For example, diabetes mellitus can be diagnosed with a certain blood sugar above a known level, or a certain hemoglobin A1c level. High cholesterol and other lipid disorders can be diagnosed with abnormal elevated lipid values above a certain level. Lyme disease can be diagnosed with various serology for Lyme blood testing. No such lab test, blood test or genetic test is possible with hypermobile Ehlers-Danlos Syndrome. That is not to say that we will not in the future hone down on a specific gene loci or other biomarkers that may be supportive of hypermobile Ehlers-Danlos Syndrome. However, to date, there are none.