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EHLERS DANLOS SYNDROME:

DIAGNOSIS FOR PRIMARY CARE PRACTITIONERS

- **Ehlers Danlos Syndrome (EDS) is a common, inherited condition[1] causing symptoms in several body systems. Once a diagnosis is made, treatment is straightforward. Much of it is best done in the primary care setting.**
- **EDS used to be diagnosed by geneticists. But most now refuse to see people with possible EDS, because EDS is common, genetics clinics are overloaded, and genetic testing is rarely needed for EDS assessment.[2]**
- **A primary care practitioner can make a firm EDS diagnosis, or rule it out, in most patients. Nearly all cases are hypermobile-type EDS (hEDS).[3] The necessary history, and exam findings, can be established quite quickly.**
- **The following protocol allows patients themselves to collect the diagnostic data, then bring it to their PCP for review. It includes differential diagnosis, ICD coding, and next steps in uncertain cases.**
- **For treatment, guidance is readily available online, via links given on the last page. PCPs can also contact me with questions by phone or email.**

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1. EDS prevalence is thought to be between one in 200, and one in 500, implying one or two children in every high school.

2. Nearly all EDS is the hypermobile type (hEDS), for which no genetic marker is known.

3. The Villefranche diagnostic criteria for hEDS have been in use since 1997. A later classification, in 2017, is of use mainly to researchers. The Villefranche criteria are far more helpful for clinical practice.

THE PATIENT SHOULD COMPLETE THE QUESTIONNAIRE ON PAGES 3 & 4.

THE DOCTOR CAN GO STRAIGHT TO PAGE 5.

- **The next two pages (3 and 4) are a questionnaire for a patient to complete, if they think they may have Ehlers Danlos Syndrome.**
- **They should then take this whole document to their primary care practitioner (PCP).**
- **The PCP should read page one. If they agree to proceed they should check the patient's data, using the instructions on page 5 to decide if a diagnosis of "hypermobile EDS" is appropriate, and if so, what the next steps should be.**

Diagnostic criteria for hypermobile-type EDS (hEDS).

The following criteria are a slightly abbreviated version of the Villefranche criteria, suitable for assessment of possible hEDS – by far the most common type of hEDS - by a patient and their PCP.

CRITERION NUMBER	CRITERION DEFINITION	RELEVANCE TO DIAGNOSIS
1	Widespread hypermobile joints	This criterion is necessary.
2	Joint displacements and pain	} One or more criteria from 2-4 support and confirm criterion 1 above.
3	Other family members with similar features.	
4	Absence of other conditions causing 1-3.	

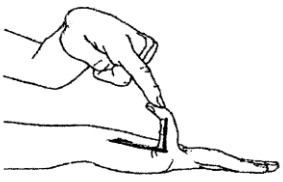
The steps to assess these criteria are given below. **Parts 1, 2 & 3 can be assessed by the patient**, **using the following steps.** There's then a page to help the physician to interpret the patient's responses, assign an ICD code, and check for other conditions that could cause their symptoms (criterion 4). Finally, there are links to handouts on *treatment*.

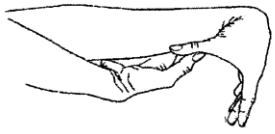
Patient's Name_____ Date of birth _____

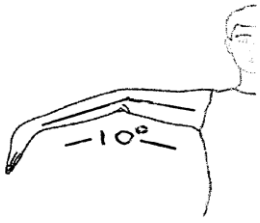
Criterion 1: Are there widespread hypermobile joints?

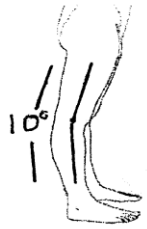
1a: The Beighton scale:

The Beighton scale has been used for several decades as a standard tool for scoring generalized joint hypermobility. Check the following five items. The arm and knee tests require measuring angles with a protractor, which looks like this:  Many schoolchildren have one. You can get one at an office supply store for about $2.









With hand palm-down on a flat surface, pull little finger back. Is the angle between the base of the finger (not the tip) and the back of the hand at least 90 degrees (a right-angle)?	Can the thumb be pulled to touch the inner side of the forearm (the same side as the palm of the hand)?	Arm outstretched, fingers down and palm facing outward, does the elbow bend at least 10 degrees? Measure angle between the bones of the upper arm and the forearm.	Standing, with knees locked, is the knee bent at least 10 degrees? Measure angle between the bones of the thigh and the lower leg.	With knees locked and feet together, can the whole palms of both hands be placed flat on the floor?
One point each side.	**One point each side.**	**One point each side.**	**One point each side**	**One point.**

Write down your score out of a possible total of 9 points: _____

1b The Five Point questionnaire:

This is a substitute for the "Beighton scale," when a patient has a history of widespread joint hypermobility, but has subsequently stiffened up, so some joints are no longer hypermobile.

For each of the five questions below, circle Yes or No in the box beside it.		
 Can you (or could you ever) place your hands flat on the floor, without bending your knees?	Yes	No
 Can you now (or could you ever) bend your thumb to touch your forearm, by pushing it with the other hand as shown?	Yes	No
As a child, did you amuse your friends by doing contortions, or could you do the splits?	Yes	No
As a child or teenager, did your shoulder or kneecap dislocate, on more than one occasion?	Yes	No
Do you consider yourself "double-jointed"?	Yes	No

Write down your score (Yes answers) out of a possible total of 5: _____

Criterion 2: Joint displacements and pain.

2a Do some of your joints get **out of place** at times? **Check one: Yes No .**

2b Do you have **long-term or recurring joint or limb pain**? **Check one: Yes No .**

Criterion 3: Family members

Do any of your family members have **loose joints or pains like yours**?
(Consider parents, grandparents, siblings, aunts, uncles and cousins.) **Check one: Yes No .**

Which relatives have loose joints or pains like yours? _____

_____

SUMMARY

Put your answers to the previous questions here:

1a Beighton score _____

1b 5-Point Questionnaire (if done) **Yes answers:___**

2a Displaced joints **Yes No**

2b Joint or limb pain **Yes No**

3 Similar family members **Yes No**

Physician's interpretation of the criteria

Look at the summary of the patient's diagnostic information on the bottom of page 4.

A diagnosis of hEDS (Villefranche criteria) is appropriate if:

The Beighton score is at least 5,***** and the patient answered **Yes** to at least one question, out of 2a, 2b and 3. (More than one **Yes** strengthens the diagnosis.)

***** Or at least 6 in a pre-pubertal patient, or at least 4 in a patient over 50 years old.

A diagnosis of hEDS (Villefranche criteria) is also appropriate if:

The Beighton score is less than 5,****** but at least 2 on the 5-Point Questionnaire, and the patient answered **Yes** to at least one question out of 2a, 2b and 3. (More than one **Yes** in strengthens the diagnosis.)

****** Or less than 6 in a prepubertal patient, or less than 4 in a patient over 50.

Does the patient have another condition that could mimic the features above?

The hEDS diagnosis becomes questionable if the patient has another condition that can cause joint hypermobility, especially:

- **Vascular-type EDS:** many times more rare than hEDS. But it has a high mortality due to spontaneous arterial ruptures, and perforations of abdominal viscera. It is suspected if the patient has a family history of sudden death before old age, or ruptured aneurysm, bowel or uterine perforations, or severe obstetric hemorrhage. A genetic test can be ordered by a PCP to rule this out. Details on how to order the test are in my article on genetic testing at www.AlanSpanosMD.com.
- **An inflammatory joint condition:** rheumatoid, lupus, psoriatic or other arthritis can lead to hypermobility in affected joints.
- **Marfan Syndrome:** suggested by long fingers, long limbs, visual problems suggesting lens dislocation, personal or family history of aortic arch aneurysm or dissection. A rheumatologist or geneticist can confirm this diagnosis.
- **Osteogenesis imperfecta:** multiple fractures and skeletal deformities, from childhood on.
- **Very rare familial conditions:** these may be suspected if the patient and family have chronic, puzzling symptoms, especially if present since childhood, that are not those of hEDS and not common in the population. If in doubt, seek a genetic consultation but you can still treat the patient for "provisional hEDS."

Coding the diagnosis of hEDS

I suggest you use ICD Q79.60: "Ehlers-Danlos Syndrome, unspecified." This is a reimbursable code. There are more specific codes, including one for the "hypermobile type: Q79.61." But the "unspecificed" code has the advantage that it will still be correct, even if the diagnosis is later changed to another EDS type. It will also survive further reclassifications of the Ehlers-Danlos Syndromes, which we expect to be recommended in future.

Failure to meet the hEDS criteria

Explain to the patient that this basic diagnostic protocol suggests some doubt about what their underlying medical problem is, and offer a referral to a geneticist. Geneticists nowadays usually refuse to see patients for diagnosis of hEDS alone, so you should tell them that you want other diagnoses ruled out. And inform the patient that the wait time for a genetics appointment may be a year or more.

Treatment for hEDS

- Because hEDS is a *variable multisystem patchwork of symptoms,* the first priority is to **listen to the patient**, record their symptoms and believe them. See page 2 of my article for primary care practitioners for a list of the main symptoms (*Medical Care of Patients with a Hypermobility Disorder*, at www:AlanSpanosMD.com).
- The second priority is to have the patient **prioritize the symptoms,** and start by addressing the one that bothers them the most.
- Begin **building a relationship** with the patient in which she researches treatments from sources such as the book *Disjointed, ed. Diana Jovin,* websites like mine at www.AlanSpanosMD.com, and online sites such as www.inspire.com and www.ehlers-danlos.com. Treatments are broadly consistent across resources like these. For most of them, a handout from one of these resources will be straightforward enough to implement in primary care, or will point to a need for specialist referral.
- Over time, most PCPs dealing with people with hEDS find supervising and coordinating their care to be feasible, helpful, and mutually rewarding.

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