TABLE 1

Overlap between Joint Hypermobility, Hypermobility Spectrum Disorders, and Ehlers-Danlos Syndromes

Type	Beighton score	Musculoskeletal involvement*	Notes
Asymptomatic joint hypermobility			
Asymptomatic generalized joint hypermobility	Positive	Absent	—
Asymptomatic peripheral joint hypermobility	Usually negative	Absent	Joint hypermobility typically limited to hands and/or feet
Asymptomatic localized joint hypermobility	Negative	Absent	Joint hypermobility limited to single joint or body parts
Hypermobility spectrum disorders			
Generalized hypermobility spectrum disorders	Positive	Present	Does NOT meet criteria for hypermobile EDS based on limited findings in skin and musculoskeletal systems and lack of family history No genes identified Screening with echocardiography unnecessary
Peripheral hypermobility spectrum disorders	Usually negative	Present	Joint hypermobility typically limited to hands and/or feet
Localized hypermobility spectrum disorders	Negative	Present	Joint hypermobility limited to single joints or body parts
Historical hypermobility spectrum disorders	Negative	Present	Historical presence of joint hypermobility
EDS – Joint hypermobility with more pronounced skin and musculoskeletal findings and/or positive family history			
1. Hypermobile EDS	Positive	Possible	Meet criteria based on supportive findings in skin and body systems and/or positive family history (see Figure 2) No genes identified AD inheritance pattern Obtain screening echocardiography

Source:
132. Yew, K.S., Kamps-Schmitt, K.A. and Borge, R., 2021. Hypermobile Ehlers-Danlos syndrome and hypermobility spectrum disorders. American Family Physician, 103(8), pp.481-492.

Type	Beighton score	Major features	Gene affected
EDS			
2. Classical	Positive	Skin hyperextensibility Abnormal scarring	*COL5A1*, *COL5A2* genes Rare *COL1A1* gene AD inheritance
3. Classical-like	Positive	Skin hyperextensibility Easy bruising	*TNXB* gene AR inheritance
4. Cardiac-valvular	Positive or negative, general hypermobility or restricted to small joints	Cardiac valvular problems Skin involvement	*COL1A2* gene AR inheritance
5. Vascular	Positive or negative	Family history of vascular EDS History of early arterial rupture or uterine rupture, sigmoid colon perforation, or atraumatic carotid-cavernous sinus fistula formation	*COL3A1* gene Rare *COL1A1* gene AD inheritance
EDS *(continued)*			
6. Arthrochalasia	Positive	Congenital bilateral hip dislocation Skin hyperextensibility	*COL1A1*, *COL1A2* genes AD inheritance
7. Dermatosparaxis	Positive or negative	Extreme skin fragility Characteristic craniofacial features	*ADAMTS2* gene AR inheritance
8. Kyphoscoliotic	Positive with history of dislocation and subluxation	Congenital hypotonia Kyphoscoliosis	*PLOD1*, *FKBP14* genes AR inheritance
9. Brittle cornea syndrome	Positive or negative	Thin cornea with or without rupture Keratoconus Keratoglobus Blue sclerae	*ZNF469*, *PRDM5* genes AR inheritance
10. Spondylodysplastic	Positive or negative	Short stature Muscle hypotonia Bowing of limbs	*B4GALT7*, *B3GALT6*, *SLC39A13* genes AR inheritance
11. Musculocontractural	Positive or negative	Congenital multiple contractures Characteristic craniofacial features Skin involvement	*CHST14*, *DSE* genes AR inheritance
12. Myopathic	Distal joints affected	Congenital muscle hypotonia and/or atrophy that improves with age Proximal muscle contractures	*COL12A1* gene AD or AR inheritance
13. Periodontal	Positive or negative	Periodontitis Lack of attached gingiva Pretibial plaques Family history of periodontal EDS	*C1R*, *C1S* genes AD inheritance

AD = autosomal dominant; AR = autosomal recessive; EDS = Ehlers-Danlos syndrome.

*—Musculoskeletal involvement includes the following: (1) pain; (2) musculoskeletal/soft tissue trauma, including dislocations, subluxations, soft tissue damage, and microtraumas (microtraumas include small tears of muscles, sprained ligaments, strained muscles, and overstretched tendons); (3) disturbed proprioception; and (4) other musculoskeletal conditions (e.g., flexible flat feet; valgus abnormality of the elbow, hindfoot, and hallux; kyphosis; scoliosis; deformational plagiocephaly).

Information from references 1, 4, and 5, and personal communication from Karyn Laursen, MD.