Supplementary material

Table S1: The Villefranche criteria for major EDS subtypes 1

Subtype	Major criteria	Minor criteria
Classical	Skin hyperextensibility Widened atrophic scars Joint hypermobility	Smooth, velvety skin Molluscoid pseudotumors Subcutaneous spheroids Complications of joint hypermobility Muscle hypotonia, motor delay Easy bruising Manifestations of tissue extensibility and fragility Surgical complications Positive family history
Hypermobility	Hyperextensible and/or smooth, velvety skin Generalized joint hypermobility	Recurring joint dislocations Chronic joint/limb pain Positive family history
Vascular	Thin, translucent skin Arterial/intestinal/uterine fragility or rupture Extensive bruising Characteristic facial appearance	Acrogeria Hypermobility of small joints Tendon and muscle rupture Talipes equinovarus Early-onset varicose veins Arteriovenous, carotid-cavernous sinus fistula Pneumothorax/pneumohemothorax Gingival recessions Positive family history, sudden death in a close relative
Kyphoscoliosis	Generalized joint hypermobility Congenital hypotonia Congenital and progressive scoliosis Scleral fragility and rupture of the ocular globe	Tissue fragility, including atrophic scars Easy bruising Arterial rupture Marfanoid habitus Microcornea Osteopenia/porosis Positive family history
Arthrochalasia	Generalized joint hypermobility with recurrent subluxations Congenital bilateral hip dislocation	Skin hyperextensibility Tissue fragility, including atrophic scars Easy bruising Hypotonia Kyphoscoliosis Osteopenia/porosis
Dermatosparaxis type	Severe skin fragility Sagging, redundant skin	Soft, doughy skin texture Easy bruising Premature rupture of fetal membranes Large hernias (umbilical, inguinal)

The presence of one or more major criteria is either necessary for clinical diagnosis or highly indicative and warrants laboratory confirmation whenever possible. A minor criterion is a sign of lesser diagnostic specificity. The presence of one or more minor criteria contributes to the diagnosis of a specific type of EDS. However, in the absence of major criteria they are not sufficient to establish the diagnosis.

Table S2: The revised (Brighton 1998) criteria for the diagnosis of benign joint hypermobility syndrome [2]

Major criteria	
	Beighton score ≥ 4
	Arthralgia for longer than 3 months in 4 or more joints
Minor criteria	
	Beighton scores 1-3
	Arthralgia in 1 to 3 joints or back pain or spondylosis, spondylolysis and/or spondylolisthesis
	Dislocation in more than one joint or in one joint on more than one occasion
	Three or more soft tissue lesions (eg, epicondylitis, tenosynovitis, bursitis)
	Marfanoid habitus
	Abnormal skin: striae, hyperextensibility, thin skin, papyraceous scarring
	Eye signs: drooping eyelids, myopia, or antimongoloid slant
	Varicose veins, hernia, or uterine or rectal prolapse

Joint hypermobility syndrome is diagnosed in the presence of two major criteria; one major criterion plus two minor criteria; or four minor criteria. Two minor criteria will suffice where there is an unequivocally affected first degree relative.

[1] Adapted from Beighton P, De Paepe A, Steinmann B, Tsipouras P, Wenstrup RJ. Ehlers-Danlos syndromes: revised nosology, Villefranche, 1997. *American Journal of Medical Genetics*. 1998;77:31–37.

[2] Adapted from Grahame R, Bird HA, Child A. The revised (Brighton 1998) criteria for the diagnosis of benign joint hypermobility syndrome (BJHS). J Rheumatol, 2000; 27:1777–1779