Summary of diagnostic criteria for Hypermobile EDS (hEDS) (Malfait et al, 2017)

Autosomal dominant; molecular basis unknown; Diagnosis remains clinical and requires presence of criteria 1, and 2 and 3

Criteria 1 Generalised joint hypermobility ; Beighton score 5/9, may need adjustment for age, as score decreases with age eg 6 in pre-pubertal and adolescents, 5 from puberty to age 50, 4 in over 50.

Criteria 2 Two of the following, A, B and C:

A; systemic manifestations of a more generalized connective tissue disorder (a total of five must be present)

* 1. Unusually soft or velvety skin
* 2. Mild skin hyperextensibility (>1.5cm)
* 3. Unexplained striae such as striae distensae or rubrae atthe back, groins, thighs, breasts and/or abdomen in adolescents, men or prepubertal women without a history of significant gain or loss of body fat or weight
* 4. Bilateral piezogenic papules of the heel
* 5. Recurrent or multiple abdominal hernia(s) (e.g., umbilical, inguinal, crural)
* 6. Atrophic scarring involving at least two sites and without the formation of truly papyraceous and/or hemosideric scars as seen in classical EDS
* 7. Pelvic floor, rectal, and/or uterine prolapse in children, men or nulliparous women without a history of morbid obesity or other known predisposing medical condition
* 8. Dental crowding and high or narrow palate
* 9. Arachnodactyly, as defined in one or more of the following: (i) positive wrist sign (Steinberg sign) on both sides; (ii) positive thumb sign (Walker sign) on both sides
* 10. Arm span-to-height 1.05
* 11. Mitral valve prolapse (MVP) mild or greater based on strict echocardiographic criteria
* 12. Aortic root dilatation with Z-score > þ2

*NB;, if Marfanoid features, present, must exclude conditions such as: Marfan syndrome, Loeys–Dietz syndrome, congenital contractural arachnodactyly, Shprintzen–Goldberg syndrome, Stickler syndrome, Homocystinuria, multiple endocrine neoplasia type 2B, and the familial thoracic aortic aneurysmal disorders*

B; positive family history, with one or more affected relatives

C; musculoskeletal complications (must have at least one):

* 1.Musculoskeletal pain in two or more limbs, recurring daily for at least 3 months
* 2. Chronic, widespread pain for 3 months
* 3. Recurrent joint dislocations or frank joint instability, in the absence of trauma

a. Three or more atraumatic dislocations in the same joint or two or more atraumatic dislocations in two different joints occurring at different times

b. Medical confirmation of joint instability at two or more sites not related to trauma

Criteria 3; All the Following Prerequisites MUST Be Met

* 1.Absence of unusual skin fragility, which should prompt consideration of other types of EDS
* 2. Exclusion of other heritable and acquired connective tissue disorders, including autoimmune rheumatologic conditions.