

The changing nomenclature of hypermobility

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As GPs, we are probably most familiar with the diagnosis of Joint Hypermobility Syndrome, to describe bendy teenagers with painful joints. This was diagnosed by use of the Brighton Criteria (box 1) which included a Beighton Score (yes, very confusing to have just one single letter difference). The term JHS was introduced by rheumatologists in the 1960s, whereas historically, geneticists had begun to classify and discover the genetic mutations behind the other Ehlers-Danlos Syndromes.

During the years before the new classification in 2017, specialists had begun to recognise that the group of people with JHS could not be distinguished from the group diagnosed with EDS-Hypermobility Type using the previous Villefranche Nosology. They began to use the terms interchangeably, often documenting JHS/EDS-HT. The Brighton Criteria were easy to apply in the primary care setting and were therefore being used by some GPs and even physiotherapists to diagnose patients. It should be remembered that all subtypes other than the hypermobile group are genuinely rare and can be confirmed by genetic testing. The 2017 nosology has given us the very complex hEDS criteria, but also a catch-all of HSD for people with features of hEDS but who do not fulfil all of the criteria. With treatment, prognosis, and associated conditions being similar, this slightly removes the emphasis on meeting particular diagnostic criteria for the common hypermobile patient. There are now so many recognised features and associations it is reasonable to approach the diagnosis by degrees of certainty, depending on how many of these are identified in your patient. It is found to be helpful to patients to receive a 'suspected' diagnosis, rather than remaining 'medically unexplained', as this provides validation for symptoms, a framework for exploring treatment options and access to support groups, both of the traditional and the online variety. As more features are accrued over time, the degree of diagnostic certainty increases. The management of hEDS and HSD remains driven by symptoms and the individual patient's issues.

SNOMED Codes have not caught up with these various changes. It is possible to code 'Ehlers-Danlos Syndrome', but the subtype codes do not correspond with the latest, or even the previous nosology. There are codes for 'musculoskeletal hypermobility (finding)' and for 'Generalized benign joint hypermobility (disorder)', a term which many patients find unacceptable as this condition is anything but benign. There are codes for both Beighton Score and Brighton Criteria and for a finding of hypermobility at individual joints. It is clear that the SNOMED codes will need to be brought up to date to reflect the latest terminology so that we are able to record accurate data going forward.

Brighton Criteria for the (now obsolete) Joint Hypermobility Syndrome

Major Criteria

1. A Brighton score of 4/9 or greater (either currently or historically)
2. Arthralgia for longer than 3 months in 4 or more joints

Minor Criteria

1. A Brighton score of 1, 2 or 3/9 (0, 1, 2 or 3 if aged 50+)
2. Arthralgia (> 3 months) in one to three joints or back pain (> 3 months), spondylosis, spondylolysis/spondylolisthesis.
3. Dislocation/subluxation in more than one joint, or in one joint on more than one occasion.
4. Soft tissue rheumatism. > 3 lesions (e.g. epicondylitis, tenosynovitis, bursitis).
5. Marfanoid habitus - tall, slim, span/height ratio >1.03, upper: lower segment ratio <0.89, arachnodactyly [positive Steinberg/wrist signs].
6. Abnormal skin: striae, hyperextensibility, thin skin, papyraceous scarring.
7. Eye signs: drooping eyelids or myopia or antimongoloid slant.
8. Varicose veins or hernia or uterine/rectal prolapse.

JHS was diagnosed in the presence two major criteria, or one major and two minor criteria, or four minor criteria. Two minor criteria sufficed where there was an unequivocally affected first-degree relative.

JHS was excluded by presence of Marfan or Ehlers-Danlos syndromes.

Major 1 and Minor 1 are mutually exclusive as are Major 2 and Minor 2.

- 400 BC described by Hippocrates as a cause of lax joints and multiple scars
- Edvard Ehlers, in 1901, recognized the condition as a distinct entity, describing a patient with lax joints, hyperextensible skin, a tendency to bruising and frequent subluxations of the knees.
- 1908 Henri-Alexandre Danlos suggested that skin extensibility and fragility were the cardinal features of the syndrome
- 1967 Kirk, Ansell and Bywaters publish 'The hypermobility syndrome' describing generalised joint hypermobility associated with musculoskeletal complaints with autosomal dominant inheritance and variable expression
- 1969 Introduction of the Brighton Score out of 9 for joint hypermobility

- 1988 Berlin, International Nosology of Heritable Disorders of Connective Tissue of which type III corresponds to hEDS/HSD
- 1998 Villefranche Nosology named EDS-Hypermobility Type as one of seven subtypes
- 2000 Introduction of the Brighton Criteria for diagnosing Joint Hypermobility Syndrome
- 2009 Recognition that JHS (the rheumatologists' name) and EDS-Hypermobility Type (the geneticists' name) were the same condition
- 2017 International Consortium on the Ehlers-Danlos Syndromes reclassifies EDS into 13 types of which 12 have known genetic causes. They introduce complex criteria for hEDS (with HSD for anyone with symptomatic hypermobility who does not meet criteria) and JHS is removed as a diagnostic term