

# Hypermobility

Primary care assessment &  
management

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# Objective

Provide an brief overview of the diagnosis and management of Hypermobility Spectrum Disorder (HSD)

- Classification (2017 criteria- 13 types)
- Mechanisms of pain
- The use of diagnostic tools
- When to refer on
- Practical signposting

Can be functionally Advantageous (Baeza-Velasco et al. 2013) when pain not present

- Asymptomatic

- Positive Beighton score



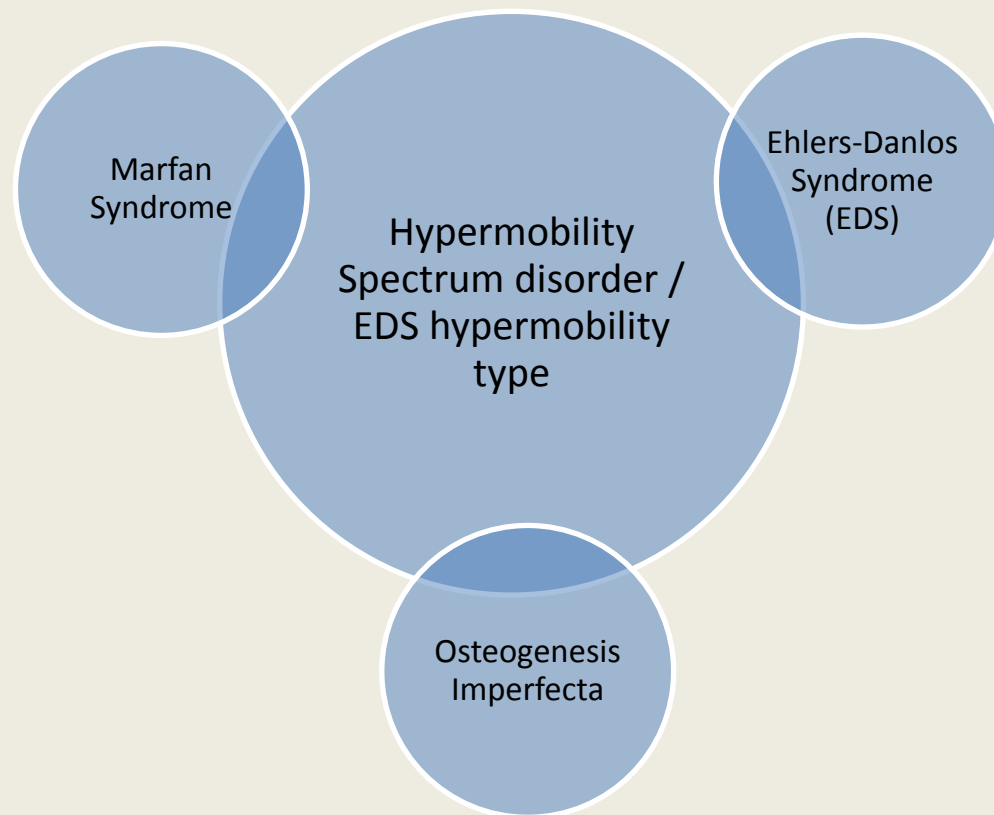
*Royal Ballet School and Royal Ballet company  
95% Women 82% Men (McCormack et al. 2004)*

*66% Dance students (Scheper et al. 2013)*

# Classification

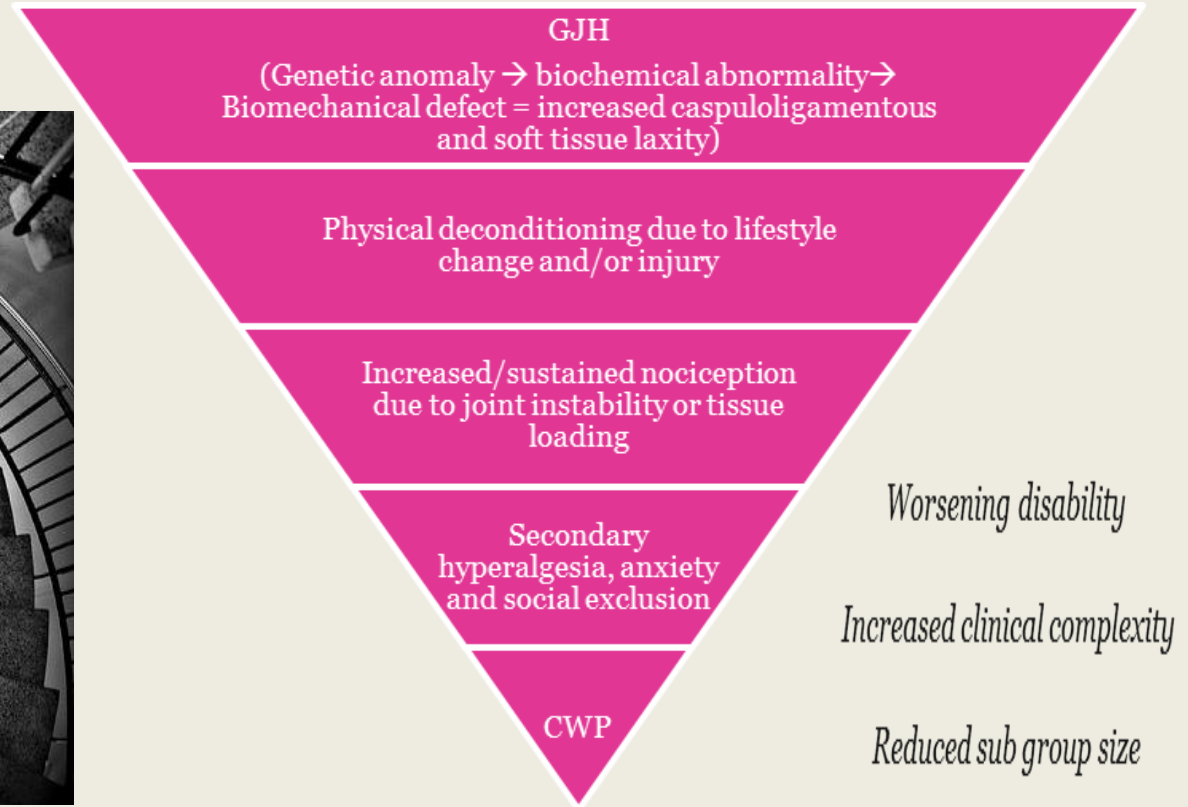
- 1997 to 2017 6 types of EDS- simpler /descriptive
- 2017- 13 types, includes rare, severe types
- Hypermobile EDS (hEDS) most common, same or similar to HSD
- Rarer: Classical (atrophic scarring/ impaired wound healing), Vascular(most catastrophic/ life threatening), Kyphoscoliotic

# HSD can be indicative of Hereditary Disorder of the Connective tissue (HDCT)



*Hakim et al. (2010); Hakim & Sahota (2006); Tinkle et al. (2011)*

# Progression of HSD



# Diagnosis

- A diagnosis of HSD is given after other possible causes are excluded, such as any of the EDS syndromes including hEDS.
- A diagnosis of HSD is made when a patient has hypermobility but does not fulfil the criteria for hEDS found at <https://www.ehlers-danlos.com/eds-diagnostics>
- There is no genetic test for HSD

# Features of hEDS

- Joint hypermobility
- Loose unstable joints, dislocations
- Joint pain and clicking
- Fatigue
- Easy bruising
- GIT- heartburn / constipation
- PoTS- Postural ortostatic tachycardia syndrome (dizziness / syncope/ palpitations, sweating etc.)
- Mitral valve / organ prolapse
- Stress incontinence

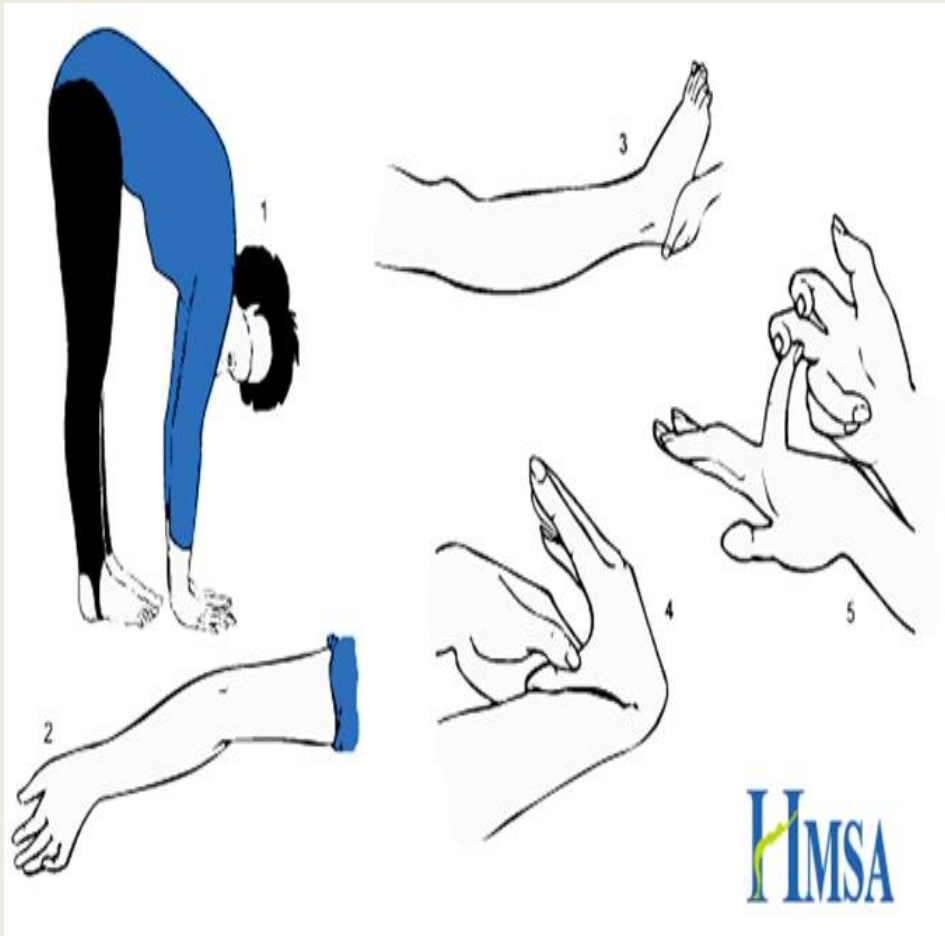


# The Beighton Score (9 points) & Quick questionnaire

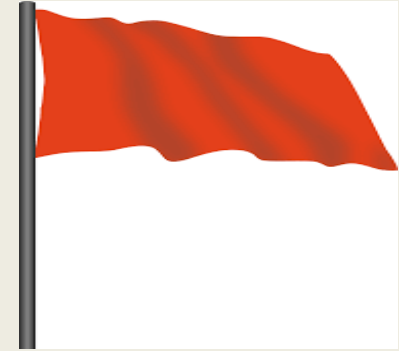
- *Can you now (or could you ever) place your hands flat on the floor without bending your knees?*
- *Can you now (or could you ever) bend your thumb to touch your forearm?*
- *As a child did you amuse your friends by contorting your body into strange shapes OR could you do the splits?*
- *As a child or teenager did your shoulder or kneecap dislocate on more than one occasion?*
- *Do you consider yourself double-jointed?*

*Answer yes for 2 or more questions suggests hypermobility, sensitivity 80-855, specificity 80-90%*

*Beighton et al 1973, Hakim & Grahame 2013*



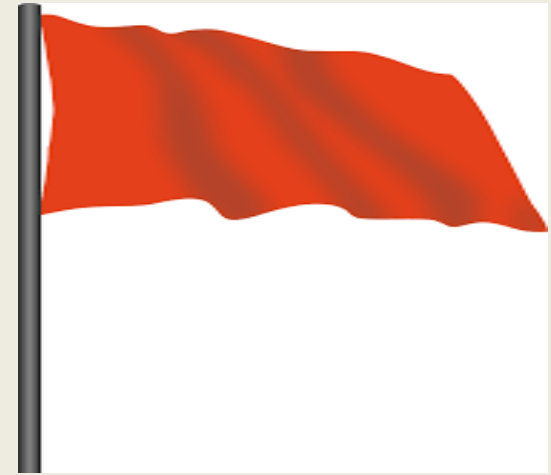
# Red flags



## Personal Hx red Flags

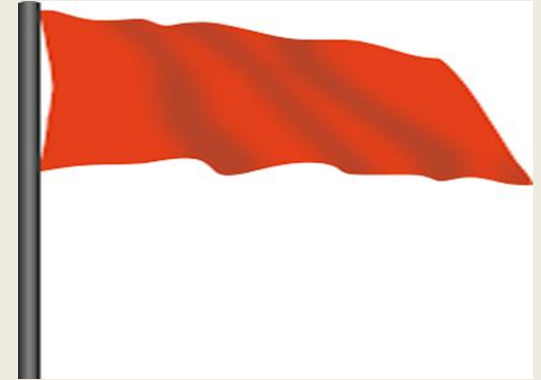
- Past medical history of Aneurysm, Arterial rupture at a young age, or Spontaneous pneumothorax
- > 7 Long bone fractures
- Obstetric History of Uterine rupture during the third trimester in the absence of previous C-section +/- or severe peripartum perineum tears
- Carotid-cavernous sinus fistula formation in the absence of trauma  
Spontaneous perforation of sigmoid colon in the absence of diverticular/bowel disease
- Marfanoid Features (a constellation of symptoms including long limbs, with an arm span that exceeds the height of the individual, a crowded oral maxilla (sometimes with a high arch in the palate), arachnodactyly, and hyperlaxity)

# Family history



- Marfan's syndrome
- Osteogenesis Imperfecta
- Vasculitis EDS
- Sudden unexplained death <40 years of age
- 7 long bone fractures in a close relative
- Thoracic aneurysm or dissection
- At least 2 relatives (one first degree) with any aneurysm or dissection at any age

# Physical examination



- Beighton score of score of  $\geq 5$  up to 50 years,  $\geq 4$  if  $> 50$  years old,  $\geq 6$  prepubertal children and adolescents
- Cardiac Murmur
- True Cutis Laxa\* (not just soft skin) or other excessive skin fragility
- \* Potential life threatening associations with the rarer forms of EDS are the vascular features that can have cutis laxa related with it. Cutis laxa is characterized by skin that is loose (lax), wrinkled, sagging, redundant, and lacking elasticity (inelastic). When stretched, inelastic skin returns to place abnormally slowly. The skin around the face, arms, legs and trunk is most commonly affected

# Cutis laxa



(a)



# Striae and atrophic scar



# Treatment strategies

- Treatments follow the same lines:
- Reassure / de-medicalise / encourage activity  
<http://bit.ly/startactive>
- Aim to reinforce non-medical model of care / promote effective self-management
- Physio, OT- if pain & functional difficulties
- Treatment plans may include; tailored exercise advice (low impact) , pain & fatigue management (pacing), clinical psychology (often CBT based)

# When to refer to Rheumatology

- Hypermobility with pain
- Failed to respond to first line treatment strategies
- If concerned regarding concurrent connective tissue disease and inflammatory arthritis ie to exclude



# Tertiary / Specialist services

## UCLH / RNOH Hypermobility Services

- Tertiary referral centre
- Referrals accepted via local Rheumatology consultant following clinic review
- For suspected or confirmed JHS, EDS, Marfan's, Osteogenesis Imperfecta
- Main treatment strategies, pain management, exercises and activity guidance
- May recommend Genetic testing after Ax

# RNOH Hypermobility service

- Severe and complex end of spectrum
- Out of area referrals accepted from local NHS Rheumatology consultant
- 3 week inpatient programme
- Collaborates with UCLH service
- Combined pain management / rehab approach

# SGH Genetics Unit

Hypermobility, mild skin involvement, joint dislocations, chronic joint pain, autonomic dysfunction, positive family history

Associated features of classic, vascular, kyphoscoliotic, arthrochalasia, dermatospraxis EDS

Associated features of Marfan syndrome spectrum

# London NW NHS Trust (Northwick park)

- National EDS diagnostic service
- Consultant referral only, initial local genetics testing prior to referral recommended
- Service is funded by NHS England for people with suspected complex EDS in England and Scotland.
- Accepts referrals for patients where:
- Diagnostic testing does not confirm diagnosis of a suspected rare type of EDS
- Diagnostic criteria of more than one type of EDS identified
- there are significant additional findings aside from diagnostic criteria
- The patient has a confirmed diagnosis of a rare type of EDS and you are requesting expert advice

# Signposting

- <http://hypermobility.org/>
- <http://www.ehlers-danlos.org/>
- <http://bit.ly/startactive>
- <https://www.nhs.uk/conditions/ehlers-danlos-syndromes>

Thank you!

