

Hypermobility and Ehlers-Danlos Syndromes

An Overview of the Classification

Anna Higo MCSP



Overview

- The 2017 International Classification of the Ehlers Danlos Syndromes
- Where does Hypermobility fit?
- Current concepts and understanding of treatment of such conditions



Introduction

- Physical Therapists are playing a central role in the management of individuals with hypermobility related disorders (Simmonds and Keer, 2007; Grahame and Hakim, 2008; Scheper et al., 2013, 2016a)
- Prevalence of JHS/hEDS in adult physical therapy outpatient MSK settings is reported to be between 30% (Connelly et al., 2015) and 55% (Clarke and Simmonds, 2011)
- Despite high incidence many clinicians are not familiar with the diagnostic criteria, prevalence, or common clinical presentation of affected individuals (Billings et al., 2015; Lyell et al., 2016; Russek et al., 2016)

2017 International Classification OF EDS

- EDS are a group of heritable connective tissue disorders (HCTDs) characterized by joint hypermobility, skin hyperextensibility, and tissue fragility.
- 13 subtypes which are classified according to their genetic basis and molecular confirmation
- For us as treating clinicians we need to be concerned with Hypermobility Ehlers Danlos (hEDS) and classical Ehlers Danlos (cEDS)

Terminology

- **1967 - Hypermobility Syndrome**

Musculoskeletal symptoms in the presence of generalized joint laxity in otherwise healthy individuals

Kirk JA, Ansell BM, Bywaters EGL. Am rheum, 1967 419-25

- **2012 - Joint Hypermobility (JH)**

A range of movement that exceeds what is considered to be normal for that joint taking into consideration the individual's age, gender and ethnic background

Rodney Grahame (2012)

The 2017 Classification

- HSD - Hypermobility Spectrum Disorder
- Ehlers Danlos Syndrome
 - 1) hEDS
 - 2) cEDS
- HDCT - Heritable Disorders of Connective Tissue

The 2017 Criteria

The Hypermobility
Spectrum Disorder

To the left are those
who do not meet the
criteria for EDS - people
who have their own sets
of problems due to
hypermobility but who
do not fit into hEDS

Hypermobile EDS

Hypermobility Spectrum Disorder

- Signs

1. Beighton Score
2. Isolated or widespread, and recurrent injury to joints, ligaments and tendons
3. Acute and chronic joint pain
4. Associated instability leading to joint subluxation or dislocation, or vertebral listhesis and /or poor proprioception
5. The ability to undertake daily activities of living or exercise or schooling or work may be significantly compromised

Clinical Conditions Associated with HSD

- Clumsy children
- Congenital hip dysplasia/ dislocation
- Chondromalacia Patella
- Down syndrome
- Dyspraxia
- Fibromyalgia
- Growing pains
- Infantile hypotonia
- Larsen syndrome
- Marfan syndrome
- Motor developmental delay
- Spondylolysis /
Spondylolysis
- Stickler syndrome

How to Diagnose hEDS - 3 Criterion

- **Criterion 1**

Generalized Joint
Hypermobility (GJH)

- a. Beighton Score
- b. 5 Point Hypermobility
Questionnaire

- **Criterion 2** - has 3 features

1. Feature A
2. Feature B
3. Feature C

- **Criterion 3**

- ✓ The absence of any other
underlying HDCT including
other variants of EDS

Criterion 1

- **Beighton Score**

Score 1 point each side and there is a maximum score of 9

- Important to note that caucasian is a score of 4+ and black/asian is a score of 5+
- Beighton does NOT allow for age - joint range of motion decreases with age (Soucie et al., 2011; McKay et al., 2016). The cut off points may prompt over diagnosis in children and under diagnosis among adults and elders
- Also Beighton does not consider other joints such as TMJ, Shoulders and FEET

Beighton score



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Give yourself 1 point for each of the manoeuvres you can do, up to a maximum of 9 points

Can you bend your thumb back into the palm of your forearm? **1 point**

Can you bend your knee backwards? **1 point**

Can you do your hair tie on the floor with your knees straight? **1 point**

Can you bend your elbow backwards? **1 point**

Can you bend your little finger up at 90° (right angles) to the back of your hand? **1 point**

Can you bend your foot backwards? **1 point**

Can you bend your wrist backwards? **1 point**

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5 Point Hypermobility Questionnaire (Grahame & Hakim, 2003)

1. Can you/could you ever place your hands on the floor without bending your knees?
2. Can you/could you ever bend your thumb to touch your forearm?
3. As a child did you amuse your friends by contorting your body into strange shapes or could you do the splits?
4. As a child or teenager did you dislocate your shoulder or kneecap on more than one occasion?
5. Do you consider yourself double jointed?

MUST HAVE AT LEAST 2 POSITIVE ITEMS

Criterion 2

- **Feature a** - at least 5 of the following to proceed from GJH into the diagnostic area of hEDS

- Velvety skin
- Mild skin hyperextensibility
- Unexplained striae
- Bilateral prezogenic papules of heels
- Recurrent hernias
- Atrophic scarring at at least 2 sites
- Pelvic floor, uterine, rectal prolapse
- Dental crowding
- Arm span to height
- Mitral valve prolapse
- Aortic root dilation
- Wrist sign or Thumb sign positive on both sides



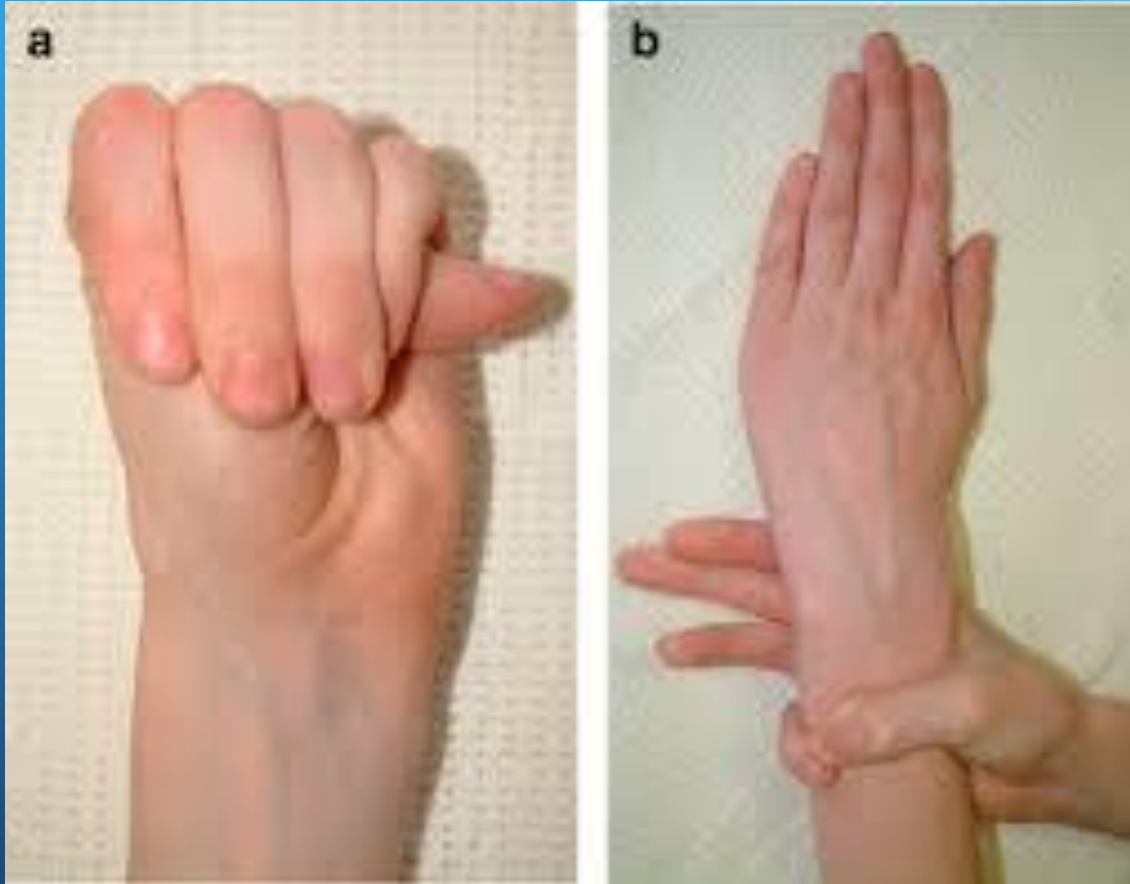






Steinberg Sign

Walker-Murdoch Sign



Feature b

- A family history with one or more first degree relatives



Feature c

- Musculoskeletal Complications **(must have at least 1)**
 - Musculoskeletal pain in 2 or more limbs, recurring daily for at least 3 months
 - Chronic widespread pain for 3 or more months
 - Recurrent joint dislocations in the absence of trauma

Classical EDS (cEDS)





cEDS

- Final diagnosis would be on molecular testing as more than 90 percent of cEDS harbor a heterozygous mutation in the gene encoding type 5 collagen



Comparing hEDS and cEDS



**KEEP
CALM
AND
RECAP**

- Major Skin Hyperextensibility
- Atrophic scarring - most cEDS have extensive atrophic scars at a number of sites.
- Genetic testing with cEDS
- No reliable genetic etiology in the vast majority of hEDS patients.
- hEDS **MUST** have the presence of criteria 1 **AND** 2 **AND** 3 to be able to diagnose.

Skin Hyperextensibility

hEDS



cEDS



Atrophic Scarring

hEDS

cEDS



Physiotherapy & Pilates - Where do we fit?



- Symptoms we will be assessing, treating and need to be aware of are:
 - PAIN - correlating with hypermobility, frequency of subluxations/dislocations, soft tissue injury, Hx previous injury, myalgias (Sacheti et al., 1997; Mulvey et al., 2013). Pain can be MSK or widespread and can be acute or chronic
 - FATIGUE - heterogenous in nature and can vary from mild to severe. HOWEVER most report fatigue as the most disabling complaint
 - PROPRIOCEPTION - Decreased in hEDS. 2 hypotheses 1) Increased jt mvt may damage proprioceptive receptors in jts (Fatoye et al., 2009) or 2) sensation of pain in jt decreases proprioception (Felson et al., 2009). EXERCISES TO INCREASE PROPRIOCEPTION DEMONSTRATE AN IMPROVEMENT IN PAIN (Ferrell et al., 2004)
 - MUSCLE STRENGTH/ BALANCE
 - JOINT INSTABILITY
 - EXTRA-ARTICULAR FEATURES (any organ consisting of collagen; heart, gut, bladder and bowel)
 - PSYCHOLOGICAL SYMPTOMS - increased risk of anxiety, depression and panic disorders (Smith et al., 2014). Low self esteem in children and adolescents (Pacey et al., 2013)

Principles of Management

- Strength, core stability and endurance training in addition to education in pain management (Bathen et al., 2013)
- With adolescents the best adherence to exercise is parental motivation adapting family routines, making exercise a family activity (Birt et al., 2014)
- Education, reassurance, manual therapy, tape, hydrotherapy and relaxation (Lyell et al., 2015; Palmer et al., 2015; Rombaut et al., 2011; Billings et al., 2015)
- Treatment individualised and undertaken with care to avoid exacerbation of pain
- Cardiovascular, MSK and physical fitness training parameters encouraged in both children and adults (NSCA and ACSM)
- Carefully graduated exercise programme, with main emphasis on motor learning (Smith et al., 2014)
- Make sure avoidance of injury and overtraining as it can lead to loss of confidence in the physio
- PAIN, FATIGUE and FEAR of injury are common barriers to exercise (Simmonds et al., 2016)
- Graduated return to sport with emphasis on training loads and adequate recovery

The Future

HSD and EDS are conditions we all need to be more aware of when treating our clients in the MSK setting.

The management of the often severe, debilitating pain in patients with these conditions is currently insufficient.

Much more studies are required into not only pain management itself but also the management of fatigue, dystonia, proprioception and energy consumption.



THE FUTURE IS OURS TO CREATE.

In life, you will realize there is a role for everyone you meet. Some will **test** you. Some will **use** you. Some will **love** you and some will **teach** you. But the ones who are truly important are the ones who bring out the **best** in you. They are the rare and amazing people who remind you why it's **worth** it.

**Always
stay
humble and
kind.**

- Tim McGraw

THANK YOU



DAILY INSPIRATIONAL QUOTES

[FACEBOOK.COM/D.I.QUOTES](https://www.facebook.com/D.I.QUOTES)

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