
HMSA
Hypermobility Syndromes Association

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Latest News

Hypermobility Disorders — An Update for Clinicians

Posted By Alan Hakim, March 16, 2017

Hypermobility Disorders – An Update for Clinicians

This article is a summary of the more common concerns that can present in hypermobility disorders and an update on terminology arising as a consequence of the 2017 International Criteria on Ehlers-Danlos syndrome. The article is written primarily for non-specialist clinicians but is also suitable for a layperson with background knowledge. We have deliberately presented the information in a bullet format to highlight the key messages and advice, and cited recommended articles should the reader wish to seek more detail.

Why look for Hypermobility?

1. It may explain musculoskeletal symptoms and loss of physical function:

- Isolated or widespread, and recurrent injury to joints, ligaments, tendons and other soft tissues around joints may occur
- Acute and chronic joint pain, and neuropathic symptoms can arise
- There may be associated instability leading to joint subluxation or dislocation, or vertebral listhesis; and /or poor proprioception increasing the risk of injury
- The ability to undertake daily activities of living, or exercise, schooling, or work may be significantly compromised.

2. It may be associated with a chronic pain syndrome and chronic fatigue, requiring adaptation to treatments to account for hypermobility / joint instability.

3. There is a growing recognition of an association with other concerns such as:

- Cardiovascular symptoms and dysautonomia (tachycardia, hypotension, syncope)
- Mechanical and neuropathic bowel dysfunction (hernia, reflux, sluggish bowel and constipation, and chronic inflammation (including mast cell activation))
- Myopia, astigmatism
- Poor response to local anaesthetic
- Pelvic floor weakness, rectal and/or uterine prolapse, chronic bladder inflammation (including mast cell activation)
- Influence of progesterone – worsening musculoskeletal symptoms; also heavy and painful menstrual cycle
- Musculoskeletal and pelvic complications of pregnancy, and
- Anxiety disorders, such as panic disorder and agoraphobia.

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'Coping in school'! Be #HMSAware!
February 22, 2017 by - Donna Wicks

4. There may be an underlying heritable disorder of connective tissue that explains concerns such as:

Multiple fractures

Poor wound healing, bruising, thin or atrophic scarring, excess stretch marks

Eye problems – cataracts, retinal detachment

Heart valve disease and arterial vascular pathologies such as dissection / aneurysm

Spontaneous rupture of viscera.

Identifying Hypermobility

It is important to note that hypermobility may be local (one or two joints), peripheral (fingers and toes), or generalised. Clinicians are advised to use a goniometer to measure the range of movement at a joint.
It is also important to note that while there are screening tools for generalised joint hypermobility such as the Beighton Score, joints not included in these scores should also be assessed, particularly if they are sites of pain / injury. For example, the shoulder, hip and ankle are common sites of pain and instability but are not included in the Beighton Score. In such a situation only using the Beighton Score to decide whether hypermobility might explain a presentation is inappropriate.
Also one should be aware that an injured hypermobile joint might appear to have a 'normal' range of movement i.e. beware the stiff hypermobile joint.

Two screening tools for generalised joint hypermobility are commonly used; the Beighton Score and the 5-Point Questionnaire. Specialists and researchers will also use other tools including the Contompasis.

The Beighton Score (**Figure 1.** Maneuvers in the Beighton Score)

http://hypermobility.org/wp-content/uploads/2017/03/Beighton_Score.png

A total of 9 points are collated from 5 maneuvers comprising:

1. Passive dorsiflexion of the little fingers beyond 90° – 1 point for each hand
2. Passive apposition of the thumbs to the flexor aspects of the forearm – 1 point for each thumb
3. Hyperextension of the elbows beyond 10° – 1 point for each elbow
4. Hyperextension of the knee beyond 10° – 1 point for each knee
5. Forward flexion of the trunk with knees fully extended so that the palms of the hands rest flat on the floor – 1 point

The 5-point Questionnaire – an answer in the affirmative to a 2 or more of the questions has 85% sensitivity and specificity (tested internationally and in different languages

1. Can you now (or could you ever) place your hands flat on the floor without bending your knees?
2. Can you now (or 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 your shoulder or kneecap dislocate on more than one occasion?
5. Do you consider yourself double-jointed?

10 things to explore in clinic that might begin to identify a hypermobility-related disorder

These are a recommendation – the start of an enquiry that should lead to more detailed assessment. There may be a number of reasons why certain symptoms are present. It would not be appropriate to assume that they are always associated with a hypermobility disorder. They should in their own right be assessed and managed accordingly, but equally multiple symptoms of this nature may arise from an over-arching condition that might not be realized if each issue is considered in isolation.

Do your joints feel like they twist easily or injury easily? Does it feel like certain joints may be slipping in and out of place? Which ones?

Do you bruise very easily, or have you noticed widened scars or lots of stretchmarks on different parts of the body? Has it been noted that (or do you think that) your skin is more stretchy than other peoples?

Do you constantly feel tired (physical or mental) – perhaps not refreshed after sleep?

Is there a lot of stomach acidity / reflux, nausea, or constipation – perhaps multiple food intolerances? Any hernias?

Do you regularly notice a fast heart rate or feel dizzy as if you might pass out? When does this happen?

Have you had any bladder concerns? Perhaps difficulties in passing or controlling urine, or repeated burning / painful urine?

Have you noticed your symptoms are worse around the time of your menstrual period?

Do you consider yourself to be anxious or depressed? What do you think is driving that?

Is there anything like this in your family history? Possibly even eye, vascular, or bowel problems?

Are there any other symptoms or concerns that are worrying you?

The Most Common Diagnoses – The 2017 terminology for HSD and hEDS

The most common diagnosis of a hypermobility-related disorder was previously called Joint Hypermobility Syndrome (JHS). The JHS diagnostic criteria covered a wide group of patients, some of whom had signs and symptoms that might equally be described as the Hypermobile variant of Ehlers-Danlos syndrome (EDS). As such, some confusion arose over JHS/EDS co-terminology.

The 2017 International Classification on EDS aims to address this by giving clarity to the criteria for hEDS. The criteria are presented later in this article. The term JHS has been dropped. Those individuals with hypermobility-

related problems that do not have hEDS or any other Heritable Disorder of Connective Tissue (HDCT) are now given the diagnosis of Hypermobility Spectrum Disorder (HSD), and their individual concerns described and managed accordingly (Figure 2).

Figure 2 – schematic to describe the relationship between HSD and hEDS

Using the 2017 criteria for the diagnosis of Hypermobile Ehlers-Danlos syndrome (hEDS) an individual must fulfill each of the 3 domains (Table 1). In the second domain they must fulfill at least 2 of the 3 descriptors (A, B and C) by achieving sufficient scores where relevant. The domains are summarized below.

Explanation of all aspects of the classification of Ehlers-Danlos syndrome are presented on a webinar viewed by clicking HERE (an explanation of the criteria for hEDS beginning 12 minutes in to the webinar).

Table 1: The 2017 criteria for the diagnosis of Hypermobile Ehlers-Danlos syndrome (hEDS)

Domain 1 The presence of generalized joint hypermobility (based on the Beighton Score or 5-part questionnaire) **Domain 2** (A) – skin or fascia signs and/or pelvic floor concerns and/or Marfanoid features (having at least 5 of all the features mentioned (B) a family history (C) At least 1 of the following 3 presentations: Musculoskeletal pain in 2 or more limbs recurring daily for at least 3 months, or Widespread pain for ≥3 months, or Recurrent joint dislocations in the absence of trauma – 3 or more atraumatic dislocations in the same joint, or atraumatic dislocations in 2 different joints occurring at different times, or medical confirmation of joint instability at 2 or more sites not related to trauma **Domain 3** The absence of any other underlying Heritable Disorder of Connective Tissue including other variants of EDS

The features that constitute Domain 2 A are: **Skin/fascia**

Unusually soft or velvety skin

Mild skin hyperextensibility

Unexplained striae such as striae distensae or rubrae

Bilateral piezogenic papules of the heel

Atrophic scarring involving at least two sites and without the formation of truly papyraceous and/or hemosiderotic scars as seen in classical EDS

Recurrent or multiple abdominal hernia

Pelvic floor, rectal, and/or uterine prolapse in children, men or nulliparous women without predisposing medical condition

Marfanoid features:

Dental crowding and high or narrow palate

Mitral valve prolapse (MVP) mild or greater based on strict echocardiographic criteria

Arm span-to-height ≥1.05 AND/OR upper segment/lower segment ratio <0.89

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; (iii) hand/height ratio > 11% on both sides; (iv) foot/height ratio > 15% on both sides

Aortic root dilatation with Z-score >+2 on echocardiography

One must also bear in mind that another heritable disorder of connective tissue (Figure 3) may be present. If there is any concern a patient should be referred for expert opinion, and further assessment might include for example vascular imaging, ophthalmic review, and genetic testing.

Figure 3 – the relationships between the more common HDCT

Investigations and Management of HSD and hEDS

A patients' concerns may be protean. A long list of investigations and treatments is inappropriate for a summary of this nature. Detail regarding specific concerns is presented on other pages on the HMSA

website and in the Reviews cited in the reference literature. Many aspects of care should employ guidance over self-management, and likely include physical treatments, medicines, and therapies, often running in parallel and managed in a multidisciplinary way.

The more common areas of investigation include:

Musculoskeletal and Fatigue blood tests:

If there is any concern that joint and/or muscle pain may be due to an inflammatory or autoimmune disorder then the relevant blood tests should be undertaken

Blood tests may be required to exclude haematologic, endocrine, and metabolic causes for fatigue.

Neuro-Muscular Imaging:

Radiographs, Ultrasound, MRI : imaging of joints / soft tissue may help to determine whether mechanical or inflammatory damage is present, impingement at the joint or of a nerve has arisen, or whether subluxation/listhesis etc. is occurring.

Neuropathic concerns might require central nervous system imaging; peripheral tests including NCS/EMG

Echocardiography if there is any concern on examination, or as part of the diagnostic work up for hEDS and other HDCT

Bowel and Urogynaecologic Investigations:

Tests for helicobacter, coeliac, bacterial over-growth

Upper or lower GI endoscopy and functional bowel tests

Urodynamics and cystoscopy might be required to delineate a problem, as might hysteroscopy

The more common areas of management include (recent reviews cited in brackets):

Physical therapies (Engelbert et al, 2017)

Pain Management (Chopra et al. 2017)

Anxiety and Mood management (Bulbena et al. 2017)

Fatigue (Hakim et al. 2017a)

Reflux, nausea, and sluggish bowel (Fikree et al. 2017)

Cardiovascular autonomic dysfunction (Hakim et al. 2017b)

Management of gynaecological concerns

Very recent literature reviews in this field of medicine detail the current understanding of the associations with hypermobility-related disorders (in particular HSD and hEDS) and the treatment options available. These are cited in the references below, and marked in green with an *. We encourage all to read these. Also the pages of the HMSA website ‘hypermobility.org’ detail a number of the concerns and provide further

references to the general literature.

We recognize that the problems a person may have (medical and social) often require a team of doctors, therapists, and social support with complementing skills. The HMSA is here to help patients and professionals in any way it can with the support from our own medical and therapy advisors and our trained volunteers, sign posting you to information and services as required.

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