Hypermobility Syndromes

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- They work at a specialist HM Unit for the diagnosis & management of all HM-related disorders. It is based at The Hospital of St John & St Elizabeth, London.
- There is a large MDT comprising of many of the world's leading experts on HM. The unit is unique in providing treatment & services for HM patients with complex problems under one roof. The unit receives referrals from all over the UK & across the world & is actively involved in research & education.
- The team comprises of:
  - 3 consultant rheumatologists
  - 2 consultants specialising in GI & visceral pain
  - 1 neurologist specialising in neurovascular disorders
  - 1 paediatrician specialising in HM, paediatric infectious diseases & immunology
  - 1 geneticist specialising in HM
  - 2 pain psychologists
  - 11 physiotherapists including women's health & paediatrics
  - 1 podiatrist
  - 2 OTs
Epidemiology

- HM is a normal phenomenon within the population
- Population Prevalence in adults 10-30%. HM is a normal phenomenon within the population.
- Female : Male ratio 3:1
- Diminishes with age
- Asian/African > Caucasian racial groups. More Caucasians complain of symptoms.
- Strongly genetically determined
- Moderate association with widespread pain. Within the chronic pain population 20% are HM – equivalent to the incidence in the general population.
HM Signs & Symptoms

HM may be of no medical consequence & even confer advantages for gymnasts, dancers. However it may be associated with;

- Ligament / tendon injuries
- Acute pain as a consequence of acute injury
- Sustained chronic pain due to sensitization & amplification of pain in peripheral nerves & spinal cord
- Poor proprioception & spatial awareness
- Joint subluxation / dislocation
- Weakness of abdominal & pelvic wall with herniation & prolapse
- Is a common feature of Hereditary Disorders of connective Tissue (HDCT) e.g Ehlers Danlos Syndrome, Marfan’s syndrome
Hypermobility & Medical Conditions

- **Hypermobile**
  - Asymptomatic/ Mild non-impairing symptoms
  - Even advantageous (e.g. gymnastics, dance, sports)

- **Focal Joint Injury/dislocations**

- **Multiple injuries & other structural pathologies**

- **Hereditary Disorders of Connective Tissue**
Why Look for Hypermobility

• Injury Risk- soft tissue injury. Poor proprioception, physical deconditioning, ROM, muscle imbalance
• Often requires different approaches to physical treatments- often missed in cases of chronic pain and/or chronic fatigue
• Feature of hereditary disorders of connective Tissue (HDCT)
• Consequences of missing HM may lead to poor treatment outcomes
HM Misconceptions

- Presence of HM ≠ Presence of a HM syndrome
- A low Beighton Score ≠ Absence of HM
- Absence of HM ≠ Absence of HDCT
Beighton Scale

Beighton's modification of the Carter and Wilkinson scoring system. Give yourself 1 point for each of the manoeuvres you can do, up to a maximum of 9 points.

1. Can you put your hands flat on the floor with your knees straight? .......................... 1 1
2. Can you bend your elbow backwards? ........................................................................... 1 1
3. Can you bend your knee backwards? ............................................................................... 1 1
4. Can you bend your thumb back on to the front of your forearm? ............................... 1 1
5. Can you bend your little finger up at 90° (right angles) to the back of your hand? .... 1 1

SCORE

Left  Right

1  1
Brighton Criteria

1998 Brighton criteria for classification of joint hypermobility syndrome

- Joint hypermobility syndrome is diagnosed in the presence of two major criteria; one major criterion plus two minor criteria; or four minor criteria. Two minor criteria will suffice where there is an unequivocally affected first degree relative.
- The syndrome is excluded by the presence of Marfan’s or Ehlers-Danlos syndromes (other than the hypermobility type of Ehlers-Danlos syndrome) as defined by the Ghent 1996\cite{ghent} and Villefranche 1998\cite{villefranche} criteria respectively.
- **Major criteria**
  - Beighton score of ≥4 (either currently or previously)
  - Arthralgia for longer than three months in four or more joints
- **Minor criteria**
  - Beighton score of 1, 2, or 3 (0, 1, 2, or 3 if aged >50 years)
  - Arthralgia in one to three joints or back pain or spondylosis, spondylolysis and/or spondylolisthesis
  - Dislocation in more than one joint or in one joint on more than one occasion
  - Three or more soft tissue lesions (eg, epicondylitis, tenosynovitis, bursitis)
  - Marfanoid habitus (tall, slim, ratio of span to height greater than 1.03 and/or ratio of upper segment to lower segment less than 0.89, arachnodactyly)
  - Abnormal skin: striae, hyperextensibility, thin skin, papyraceous scarring
  - Eye signs: drooping eyelids, myopia, or antimongoloid slant
  - Varicose veins, hernia, or uterine or rectal prolapse
- *Although originally designed for use as a research tool in defining a cohort of patients for recruitment into clinical studies, in practice the criteria have proved to be a useful diagnostic aid in the clinical setting.*
Five-part questionnaire for identifying hypermobility

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?

(Hakim, Grahame 2003)
Affirmative answer to 2 or more questions suggests HM with 80-85% sensitivity & 80-90% specificity
Joint Hypermobility Syndrome JHS

Definition- “Musculoskeletal symptoms in the presence of generalised joint laxity in otherwise normal subjects”
(Kirk JA et al Ann Rheum Dis1967)

- A complex spectrum of S&S.
- Diagnosis based on S&S including historical pathologies & use of Brighton Criteria
- Considered synonymous with HM variant of EDS
- Patients typically report to their GP 2 years after symptoms begin
- On average diagnosis may take up to 10 years to be made. (Hakim 2012) partly by not piecing seemingly unrelated symptoms together & poor clinician awareness.
JHS signs & symptoms

- HM of axial as well as peripheral joints
- Joint dislocation/subluxation
- Degenerative changes of joints
- Recurrent soft tissue injuries typically taking longer to heal, may become chronic, persistent injury.
- Skin fragility with easy bruising, scarring, stretch marks
- Weakness of the abdominal / pelvic walls with herniation / prolapse
- CV & GI autonomic dysfunction that manifest as hypotension, blackouts, tachycardia/palpitations, gastroparesis & IBS
- Cardiac mitral valve prolapse
- Varicose veins
- Resistance to local anaesthetics- often have to give more in HM patients
- Chronic regional widespread pain
- Chronic fatigue
- Anxiety, kinesophobia
- Impaired proprioception, coordination & clumsiness
- Reduced peak flow/impaired pulmonary function, pneumothorax
- Allergic rhinitis/interstitial cystitis in absence of pathogen linked with HM (fairly new)
Other commonly associated physical signs

The Marfanoid Habitus- presence
Arm span to height ratio > 1.03,
Upper to lower body ratio < 0.89,
Hand to height ratio > 11%
Foot to height ratio > 15%
Presence of cardiac and/or ocular symptoms- consider Marfan’s syndrome
Skin signs

Skin signs –
- easy bruising
- scarring & delayed healing
- striae
- doughy skin
Abdominal Wall & Pelvic floor

- Gastro-oesophageal reflux/hiatus hernia, gastroparesis, nausea, bloating, constipation, diarrhoea, anorexia. Gastroscopy/endoscopy often not enough- may need to swallow camera to monitor symptoms over a time period. GI symptoms can be so severe peg feeding required. Referral to GI specialist with good understanding of HM is key.
- Other hernia
- Rectal Prolapse
- Vaginal/bladder prolapse
- Bladder instability/ irritability, urgency, frequency, stress incontinence
- Symphysis Pubis Dysfunction
May need referral to urogynaecology
Autonomic Nervous System Dysfunction

- Light headedness/dizziness/faint
- Vascular sympathetic Dysautonomia
- Orthostatic hypotension- uncommon, rapid drop in blood pressure >20/10mm Hg, standing intolerance
- Orthostatic Intolerance- common, young women, delayed hypotension
- Postural Orthostatic Tachycardia >30bpm rise in pulse rate, associated with orthostatic intolerance. Patchy dysautonomia with pooling of blood in peripheral circulation. Activation/hypersensitivity of cardiac sympathetic nervous system. Related to deconditioning rather than HM, also seen in CFS, chronic pain
- May need tilt table test, hot/cold suppressor tests, epinephrine/Norepinephrine levels/stimulation
- Gustatory tests
- Endocrinology/cardiology- a simple echocardiogram not enough, 24 hour monitoring of pulse/bp
POTS

• **POTS- Postural tachycardia syndrome**

• An abnormal response by the ANS to becoming upright. There is an abnormally high increase in heart rate & altered blood supply to the brain. This results in symptoms of dizziness, fainting, tiredness & palpitations.

• When lying down 25% of our blood lies in our chest cavity. Under normal circumstances when we stand up, up to 800 millilitres of blood will be pulled down by gravity from our chest to the abdomen & legs. To maintain blood supply to our brain, the sympathetic nervous system will react by immediately narrowing blood vessels. heart rate increases by 10-15 beats per minute & there is a very slight increase in blood pressure.

• In some people these mechanisms fail, altering the return of blood to the heart & brain. Extra noradrenaline can be produced altering the return of the blood to the heart and brain. Within 10 minutes of standing up, patient experience an increase in heart rate of 30 beats per minute & this is associated with symptoms of POTS.

• Causes of POTS in JHMS patients is thought to be due to deconditioning. The heart does not pump as efficiently as before triggering symptoms of orthostatic intolerance so patients avoid exercise leading to physical inactivity & worsening of POTS. Also blood vessels may be more elastic causing pooling of blood in the lower body.
Potential Treatments for CVS Autonomic Dysfunction in JHS

• **Non-pharmacological** - hydration, salt intake, isotonic fluids, avoid caffeine/alcohol, control meal size & frequency, head up bed tilt, elastic stocking support, exercise & muscle conditioning

• **Pharmacological** - alpha stimulants (midodrine, clonidine)
  B blockade, Fludrocortisones
Potential Treatments for Bowel Dysfunction in HM

• **Non-Pharmacological** - hydration, salt intake, isotonic fluids, avoid caffeine/alcohol, control meal size & frequency, fibre
  
  • FODMAD diet
  
  Involvement of dietician

• **Pharmacological** - bowel stimulants, antispasmodics, antacids, anti-emetics, neuropathic analgesics
HM & Children

• Growing pains
• Clumsy Child
• Impaired hand writing & sports skills
• Association with dyspraxia/ ADHD
• Impact of ill health on education and social development
Hereditary Disorders of Connective Tissue

- Ehler’s Danlos Syndrome
- Marfan’s Syndrome
- Osteogenesis Imperfecta
- Stickler Syndrome
- Pseudo Xanthoma
- Congenital contractural arachnodactyly
- Alport Syndrome
The Hereditary Disorders of Connective Tissue

- Hypermobility/ Dislocations
- Skin pathology
- Morphometry (e.g. Marfanoid Habitus)
- Osteoporosis
- Osteoarthritis
- Vascular Pathology
- Ocular disease/ lens dislocation /Dental disease ? Deafness
- Visceral pathologies (lungs, bowel)
- Neuropathies/autonomic disturbance
- Morbidity & Mortality
Management

MDT is frequently required

• Medical- rheumatology, paediatrician, endocrinologist, gastroenterologist, cardiologist, psychiatrist, pain management, orthopaedics.
• Therapies- physiotherapy, OT, podiatry, psychology, dietician/nutritionist, social worker, hypnotherapy
• Educational- teachers, school nurse
• Family
• Charitable organisations- HMSA, EDSSG
Management cont.

• Haematological, biochemical & immunological tests may be undertaken to exclude other causes of rheumatic disease or fatigue
• Biochemical blood sampling & skin biopsy may be undertaken to identify collagen, fibrillin or elastin disorders if there is concern regarding the presence of HDCT
Rehabilitation

Patient has to be at a point where they are willing to actively engage in rehabilitation. List all clinical features of concern rather than make a diagnosis during assessment. Adapt to individual patient. Engage holistically with the individual to develop shared realistic goals:

- LISTEN
- Reassurance, education, advice.
- Core Stability/ Pilates/Tai Chi
- Enhance proprioception, body awareness, posture, balance
- Restore normal (hyper) mobility with control
- Reverse Deconditioning
- Behavioural strategies to manage e.g. anger, depression
- Self-management & function
- Pacing strategies
- Avoidance of unhelpful posture/activities e.g. W sitting
- Splints/orthoses provision - podiatry/OT
- Facilitate participation
- Joint protection, activity modification
Rehabilitation cont.

Exercise - goal orientated/ patient specific & pain free

Hydrotherapy - pain relief, facilitates proprioception, buoyancy

Fatigue is one of the greatest enemies causes need to be addressed
Outcomes

- 80% of patients with complex pain & mechanical problems from JHS would be expected to remain stable or improve unless a new event arose
- 60% would report good to excellent results & 30% very good to excellent
- Up to 20% may report ineffectiveness

Average period of recovery is 12-18 months from the onset of corrective interventions - some will take longer.
Taking things forward locally

- HM Group Meetings with Dr Clarke/ therapists
- Group looking at existing services - care fragmented/ compartmentalised, poorly co-ordinated, lack of understanding
- Education of healthcare team / raise awareness
- Mapping exercises of patient journeys
- Business plan
- Development of a care web/ specialist unit

Any Questions?