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Hypermobility Syndromes Association

Hypermobility Syndromes: an introduction.

by Dr Philip Bull

Dr Philip Bull FRCP

- Consultant Rheumatologist - the One Hospital, Ashford.
- Clinical teacher - East Kent Hospitals
- Honorary Senior Lecturer – GKT Medical School.
- Medical adviser to the HMSA.

Objectives of this talk:

- Helping you understand the Hypermobility Syndromes
- Examine the causation of some "medically unexplained symptoms"
- Relationship between Hypermobility, Pain, fibromyalgia, IBS and PoTS
- How to approach management.
- Education: to look at resources provided on the HMSA website!

(www.hypermobility.org)

Hypermobility: Epidemiology

- Adult population prevalence 10 -15%
- Probably 1% symptomatic (may be higher)
- Female : Male ratio 3 : 1
- Diminishes with age
- Asian and African racial groups more affected
- Autosomal dominant, variable expression

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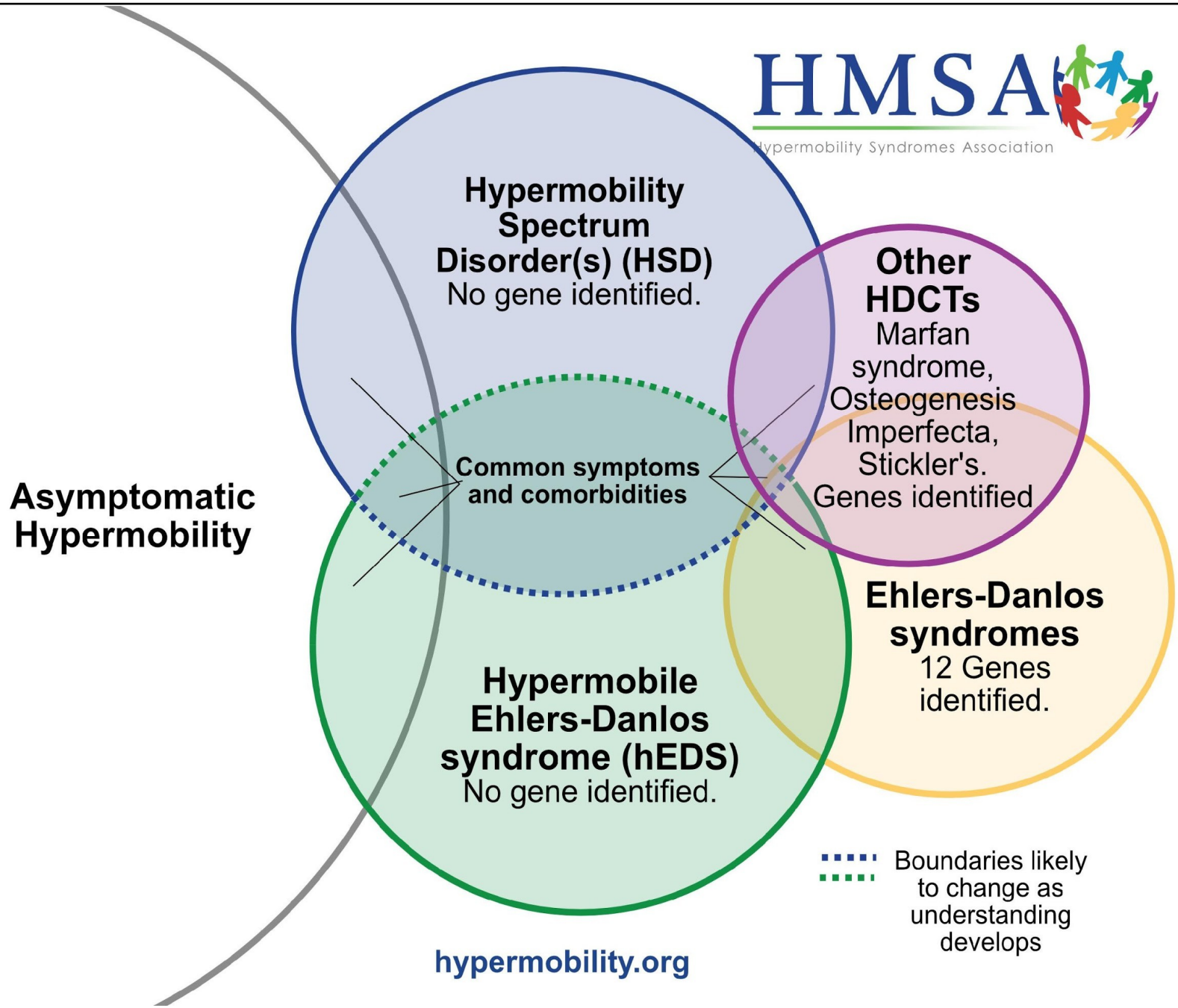

Further resources to access:2017 wellbeing conference videos:

Videos:

Click here to see all the videos from the joint HMSA and EDS UK Wellness Conference 2017

These cover a wide range of topics including: pain, fatigue, pacing, physiotherapy, swallowing and voice, mast cells, the role of primary care and more.

Below is one of these videos - An Overview of the 2017 EDS Classifications by Dr Hana Kazkaz

An here is a presentation by Hannah Ensor on living well with a hypermobility syndrome, given as part of an HMSA masterclass.

Hypermobility can be an advantage

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Daily Mail, Tuesday, May 14, 2019

GOOD HEALTH

When being flexible may not be such a good thing after all

By ANNA BEHRMANN

AS AN accomplished performance artist, Roxani Eleni Garefalaki was able to contort her body into incredible positions — including balancing on stilts for the opening ceremony of the Athens Olympics in 2004.

But while the Greek acrobat may have looked strong and flexible, she would collapse with joint pain and extreme tiredness after shows finished.

She also had unexplained tinnitus, a ringing in her ears, for days at a time.

'I went into performance arts from a very young age,' says Roxani, 35, who lives in London.

'When I was 12 or 13, I started having problems with tinnitus. It was very bad but if I rested it would be fine after two or three days. Doctors didn't understand what was wrong.

'At 18, I started doing aerial acrobatics. I was very successful but shows would be followed by a complete collapse. I'd be in great pain and have to rest. In the end, I was spending everything I earned on osteopaths and physiotherapists to help the pain in my joints.'

After moving to the UK aged 29, her symptoms were repeatedly dismissed by her GP and a specialist.

However, three years ago, a rheumatologist — a doctor specialising in musculoskeletal medicine — finally diagnosed hypermobile Ehlers-Danlos syndrome.

People with the condition have faulty collagen, a protein that normally strengthens connective tissue. The condition means that connective tissue all over the body that supports the skin, tendons, ligaments, blood vessels, internal organs and bones is affected, causing a range of problems including unstable joints, fatigue, digestive problems and bruising.

Some patients, like Roxani, also experience tinnitus.

Could YOU be hypermobile?

THE Hypermobility Syndromes Association has devised this questionnaire for hypermobility. If you answer yes to two or more questions then you may have joint hypermobility and may want to see a doctor about getting help.

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

HYPERMOBILITY comes in a range of different forms, of which hypermobile Ehlers-Danlos syndrome is a severe form. At the other end of the spectrum is joint hypermobility — commonly known as being double-jointed — which affects 10 to 15 per cent of people.

Hypermobility itself is not a disorder. It's a trait, like height, which gives people a greater range of movement than normal in their joints so they are able, for example, to touch their wrists with their thumbs.

It can even be advantageous, particularly in certain sports and performing arts — such as Roxani's acrobatics — as it gives an incredible range of movement.

'A small percentage develop musculoskeletal symptoms as a result of their stretchy collagen and they typically have "clicky" joints which can sometimes dislocate,' says Dr Philip Bull, a consultant rheumatologist and advisor to the Hypermobility Syndromes Association charity.

'These patients usually manage well with specialist physiotherapy.

'A smaller group of patients have hypermobility spectrum disorders such as hypermobile Ehlers-Danlos syndrome, which can cause other problems such as chronic pain, irritable bowel syndrome (IBS) and dizziness on standing.

'Other symptoms include stretchy skin, poor wound healing, easy bruising, hernias, varicose veins, pelvic problems, bladder irritability and clumsiness.

'Many patients with hypermobility experience their first symptoms as a child or in their teenage years; others discover they have a hypermobility syndrome later in life and realise, looking back, that this has been their problem all along, sometimes having been labelled as having "medically unexplained symptoms",' adds Dr Bull.

Roxani's experience of struggling to get a diagnosis is all too familiar, according to the charity, as hypermobility spectrum disorders continue to be missed or not even considered by specialists.

According to a poll in 2012, most people wait ten or more years for a diagnosis — and some don't get diagnosed for decades.

'There are still so many people who are not diagnosed for 30 or 40 years,' says Donna Wick, chief executive of the Hypermobility Syndromes Association. 'I get 80-year-olds ringing me up. It's so sad because they spent their whole lives being disbelieved.'

Part of the problem is that patients can have a wide range of symptoms and so will go to different specialists — for example, a gastroenterologist if they have IBS, or an orthopaedic surgeon if they keep dislocating their shoulders. However, rheumatologists, orthopaedic surgeons and physiotherapists should be in a good position to identify hypermobility.

Early diagnosis is important so that patients can be given the right advice on how to manage their condition.

There is no definitive test for hypermobile Ehlers-Danlos syndrome, so specialists should look out for joint hypermobility, signs of faulty connective tissue throughout the body (for example, hernia or prolapse), a family history of the condition, and dislocations, according to the charity Ehlers-Danlos Support UK.

To help identify if there could be a problem, the Hypermobility Syndromes Association has devised a five-point questionnaire (see box). If you answer two or more of the questions with yes, then you may have hypermobility and may want to consider seeing a specialist about getting help.

There is no specific treatment, but rather different ways of managing the symptoms.

'For many patients, symptoms can be made manageable and patients are able to live active and fulfilling lives,' says Dr Bull.

'This starts with a correct diagnosis and validation of the patient's symptoms — in particular, understanding that this is not just "in their head".

'Most people benefit from expert advice from a physiotherapist skilled in hypermobility management, focusing on core stability and general fitness,' he says. Dr Bull recommends the Alexander technique — which teaches improved posture and movement — for musculoskeletal symptoms.

'If you consider the body as a machine, such as a car, the physiotherapist is like a mechanic ensuring that all your tissues are moving normally, and the Alexander technique teacher is like a driving instructor, teaching you to move more effectively,' he says.

'There is also evidence that tai chi is effective in patients with fatigue, and mindfulness is well established as a management tool for anxiety and depression as well as improving pain management.'

For Roxani, the Alexander technique has helped her manage her condition. But she no longer does gymnastics as she believes this aggravates her symptoms.

'I know how careful I must be with myself,' she says. 'I still get symptoms, but I know not to go to extremes in terms of exercise.'

Painful: acrobat Roxani Eleni Garefalaki, below, suffered from an extreme type of hypermobility

Pictures: GETTY; RANN CHANDRIC

Should our food be fortified

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ROXANI’s story

Why look for Hypermobility?

1. **Increased Injury Risk:**

   - Soft tissue injury, Joint subluxation, dislocation
   - Poor Proprioception, Muscle Imbalance, Physical Deconditioning

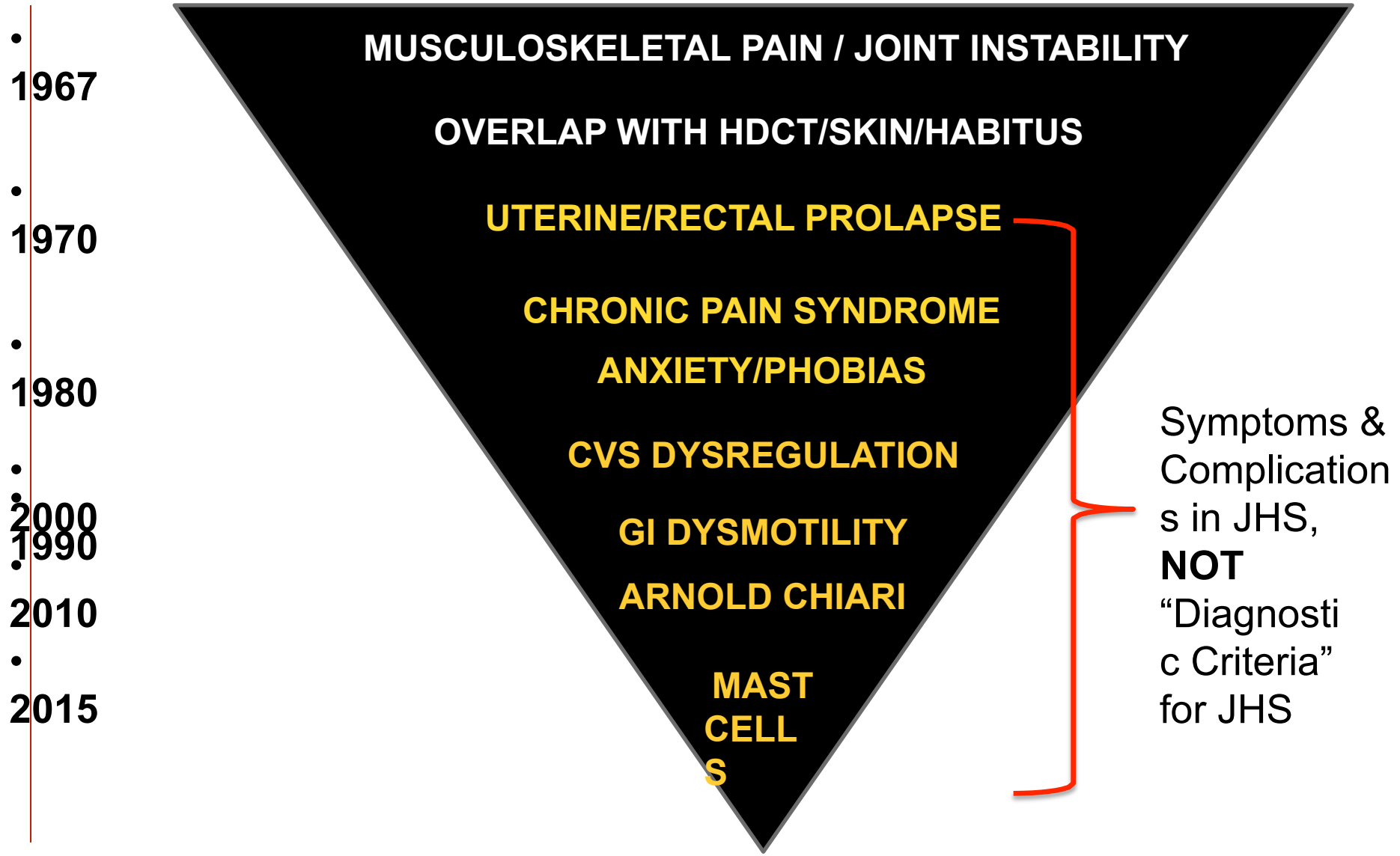
2. **Frequently requires different approaches to physical treatments**

   - Often missed opportunities in cases labelled as Fibromyalgia or Chronic Fatigue Syndrome.
   - Many patients will reflect on the therapy being too much and too painful when the hypermobility has been missed.

3. **May be a feature of Hereditary Disorders of Connective Tissue (HDCT)**

eg: Ehlers-Danlos Syndromes, Marfan Syndrome, Osteogenesis imperfecta.

The ‘Joint Hypermobility Syndrome’ 1960s to the Present Day



R Grahame, 2014

Symptoms and clinical signs

- Joint hypermobility & hyperextension
- Stretchy skin

- Chronic Pain
- Fatigue
- PoTS
- IBS / constipation / reflux / nausea
- Bladder and pelvic problems
- Easy bruising / poor wound healing
- Clumsy/poor proprioception
- Anxiety, phobic states, depression
- Autism/ADHD

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Hypermobility (HSD/hEDS)

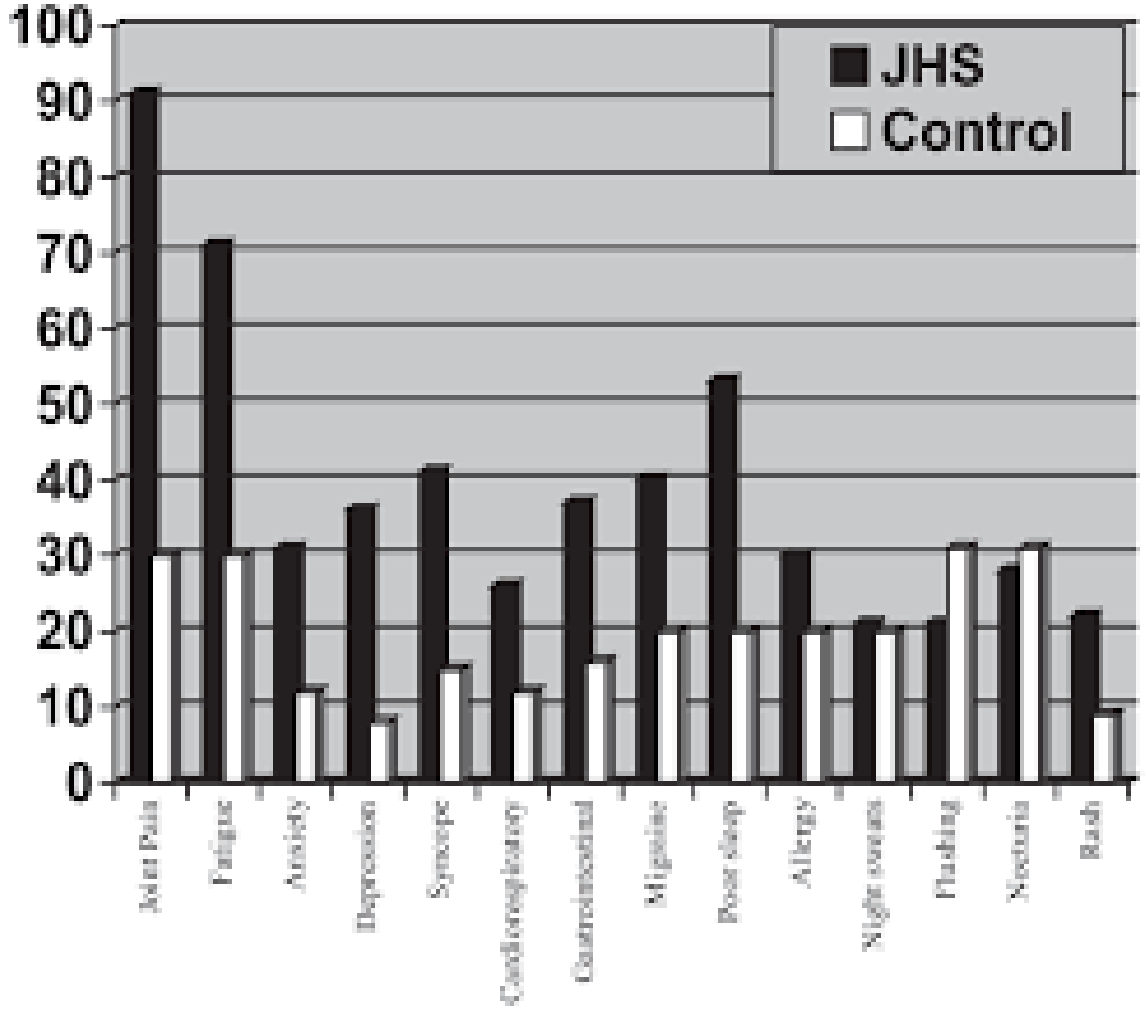
Frequently:

- Overlooked
- Disregarded
- Discounted

- Easy to spot if you look for it
- Easy to miss if you do not
- So: Ask the right questions !

- Significant impact on wellbeing whilst undiagnosed
- AN INVISIBLE ILLNESS

Symptoms associated with hypermobility



Hakim & Grahame (2003)


STOP
LOOK
LISTEN
THINK

The New York meeting May 2016

- International Consortium meeting looking at the evidence base
- The proceedings published in the American Journal of Genetics

The UK contingent

American Journal of Medical Genetics March 2017

Summary of existing evidence:

- Classification
- Pain
- Physio
- Fatigue
- Dysautonomia
- Gastrointestinal
- Anxiety
- Orthopaedics
- Neurological

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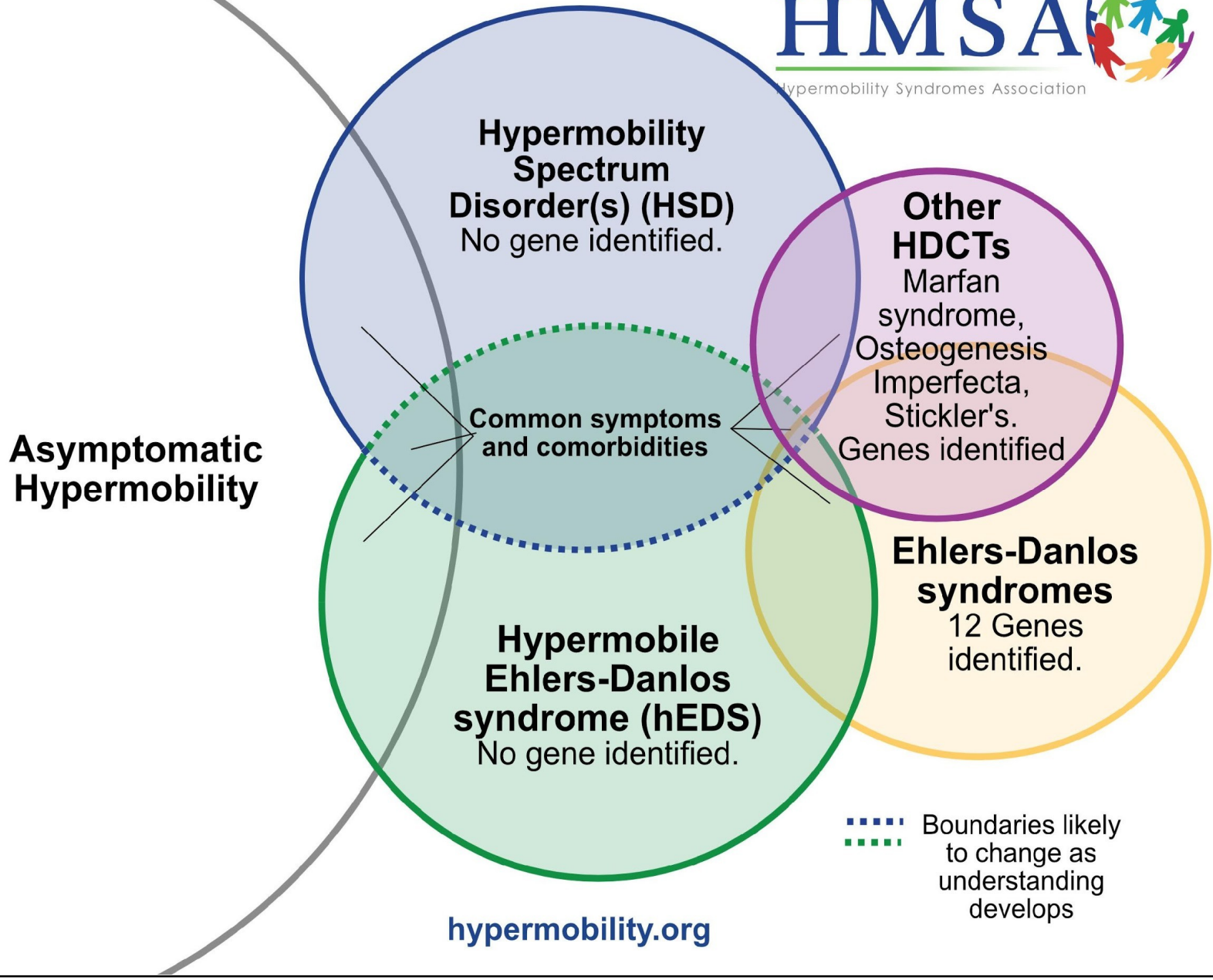
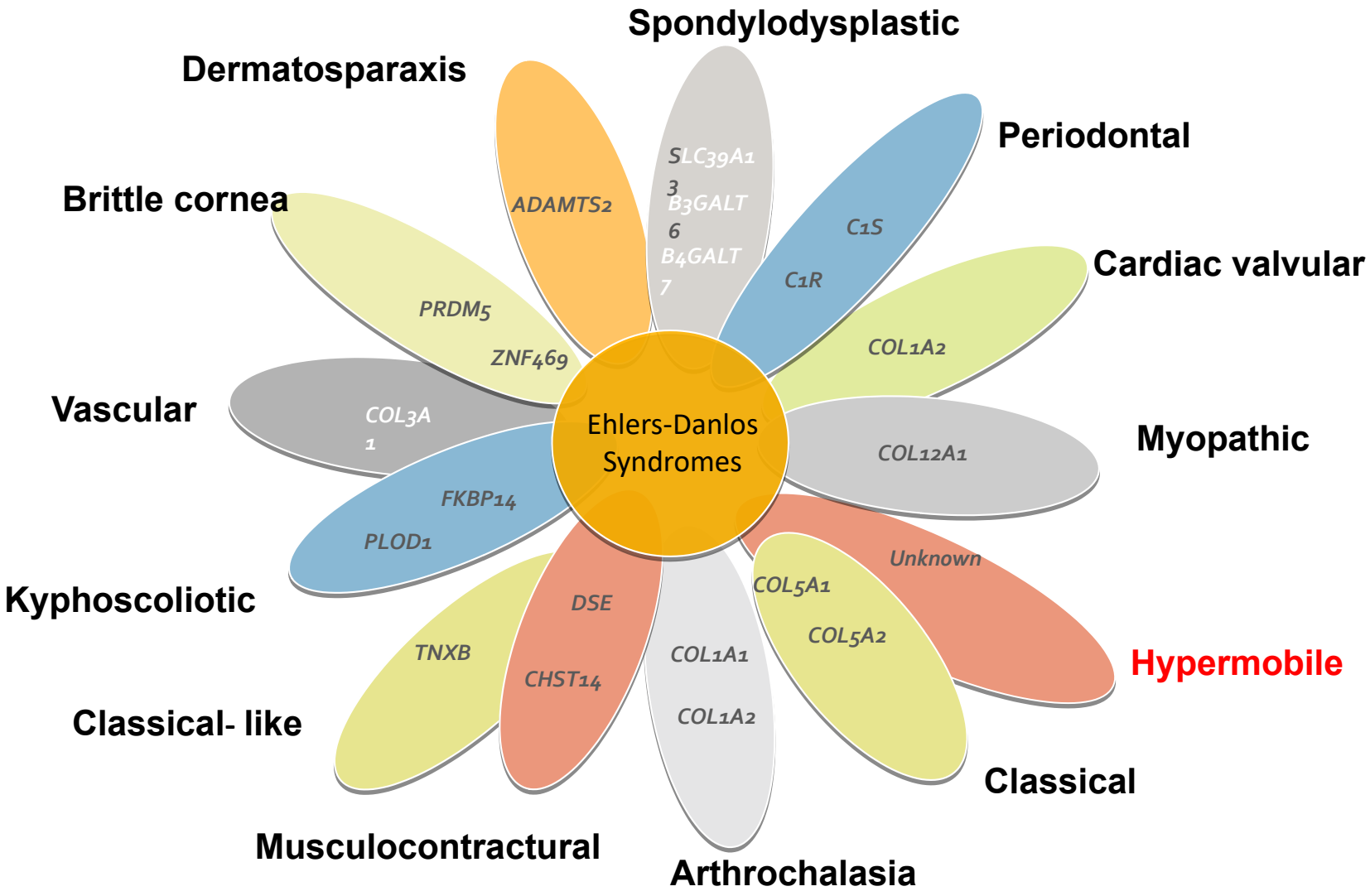


hypermobility.org

EDS Types



+ 14th subtype: *AEBP1 gene*

J Amberger, C Bochinni, A Hamosh, 2018

Why is there confusion?

- 2017 criteria not evidence based…eminence based
- Lack of Education about Hypermobility syndromes
- No coding for HSD
- Paediatrics..2017 criteria not adapted for use in children

- Unconscious bias in the clinical decision making process

Getting the Diagnosis

10 key questions:

- Joint instability or pain
- Bruising, stretchmarks
- Fatigue
- Reflux, nausea, constipation, hernias
- Palpitations, tachycardia or dizziness
- Bladder symptoms
- Worse around menstruation
- Anxiety and or depression
- Family affected
- For adults, remember to look back

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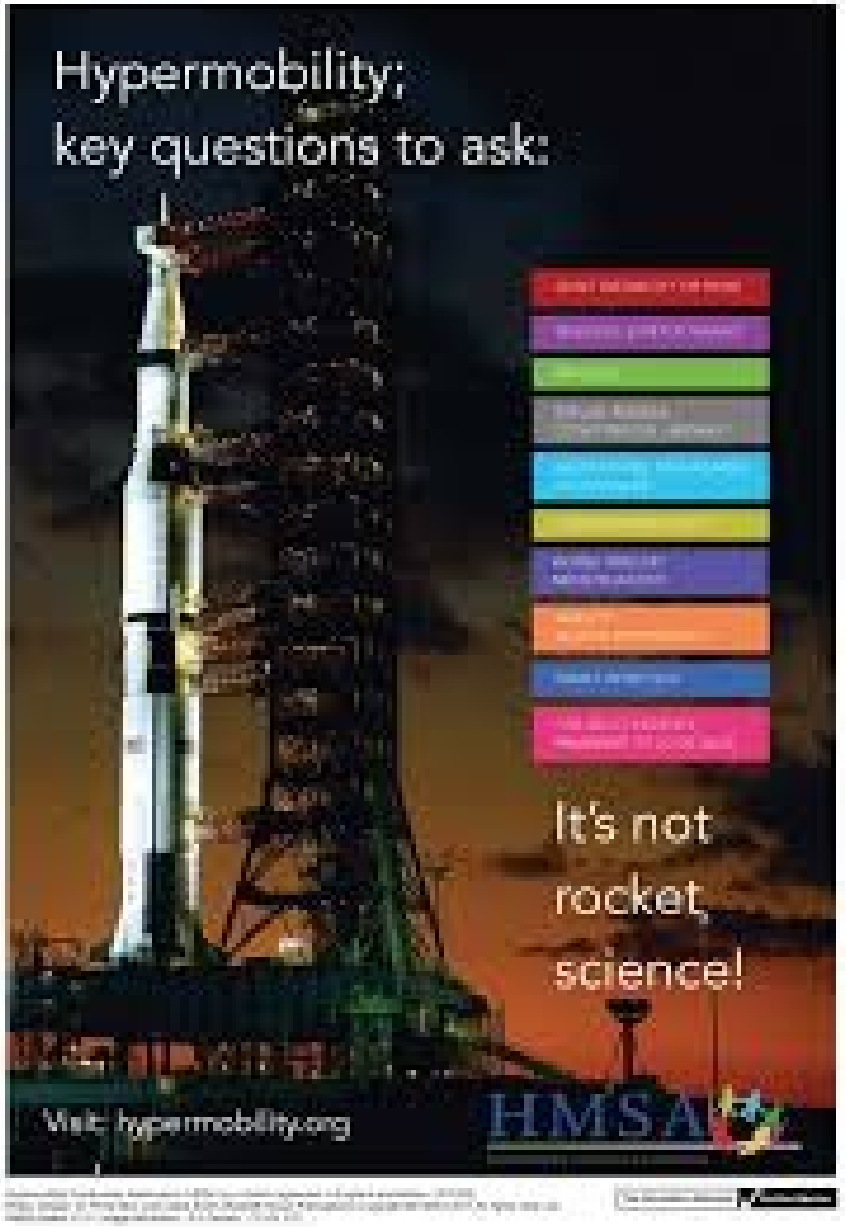
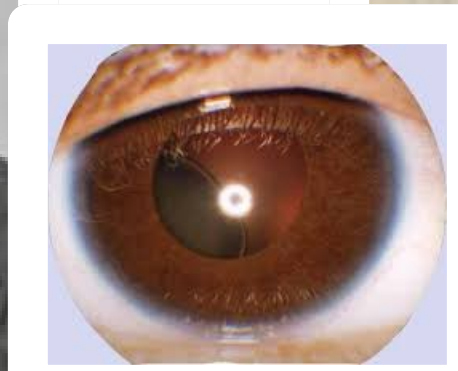


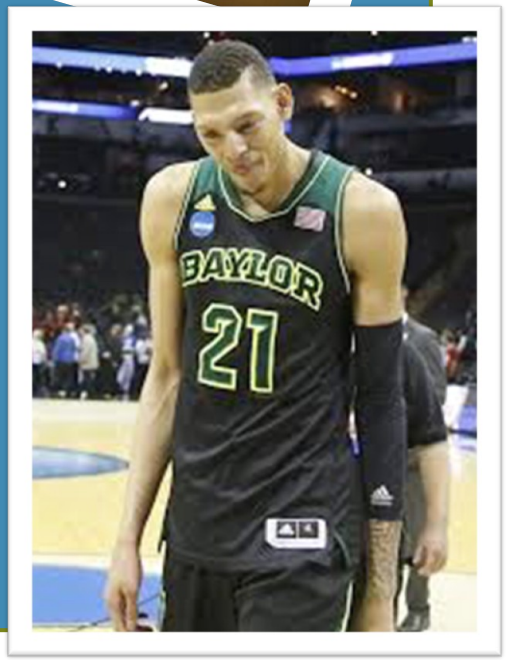
Ehlers-Danlos Syndrome clinical features

Symptoms:

- Skin hyper-elasticity
- Atrophic scarring
- Easy bruising
- Congenital dislocation of hips
- Vascular type

Marfan syndrome: Clinical features





Aortic aneurysm… important

Osteogenesis Imperfecta

https://www.hypermobility.org/Pages/Category/osteogenesis-imperfecta/Tag/oipage

Genetic testing: When is it indicated?

- Marfanoid body habitus
- Aortic root dilatation
- Ocular signs
- Very stretchy skin or unusually widened atrophic scars
- Large unusual bruising/haematomas
- Organ rupture
- Personal or family history of young onset unexplained arterial dissection, aneurysms or significant haemorrhage
- Significant kyphoscoliosis
- Recurrent large hernias

What is happening during the 10 year diagnostic delay?

- Patients go round in circles
- Wasted GP consultations and hospital appointments
- Unnecessary medication and side effects
- No diagnosis
- Patient in limbo
- Patient becomes unwell as a result of lack of diagnosis and appropriate treatment
- TIME TO DO SOMETHING DIFFERENT!

The 10 Year Maze!

The Beighton 9-Point Hypermobility Score

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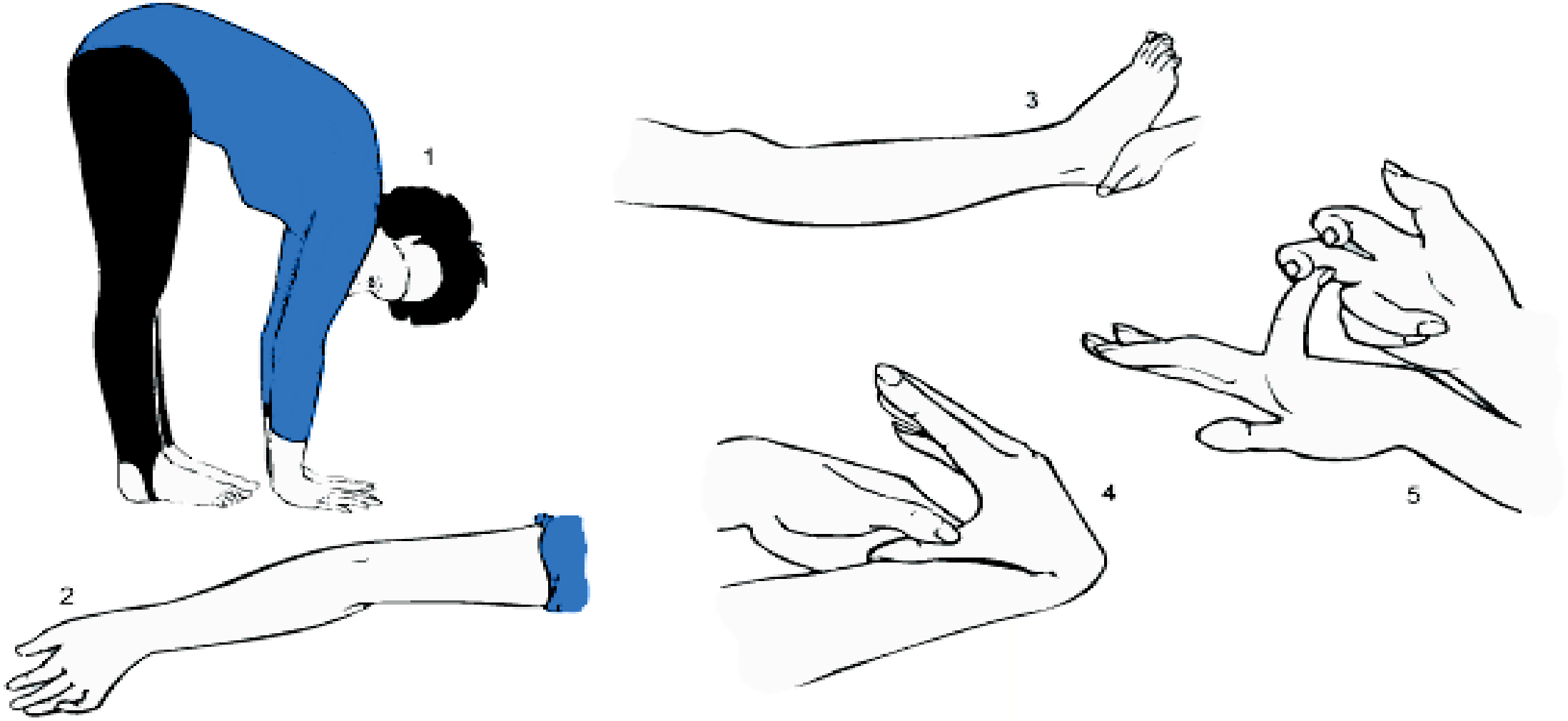
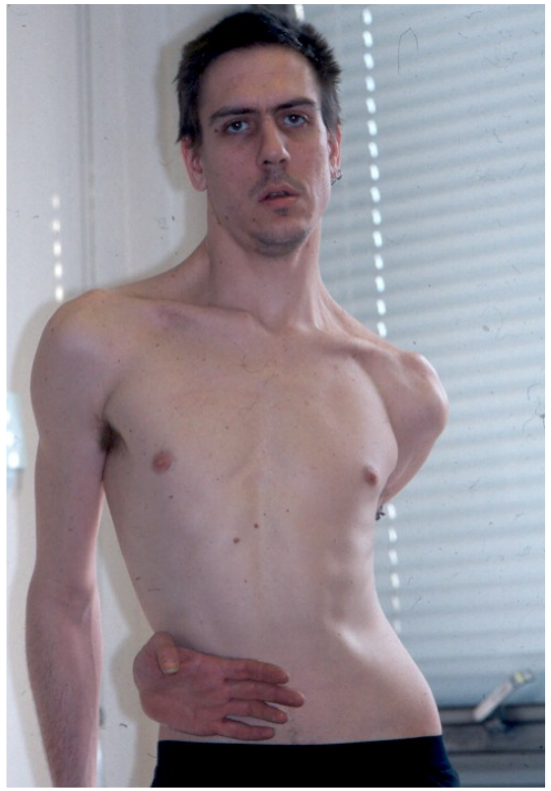


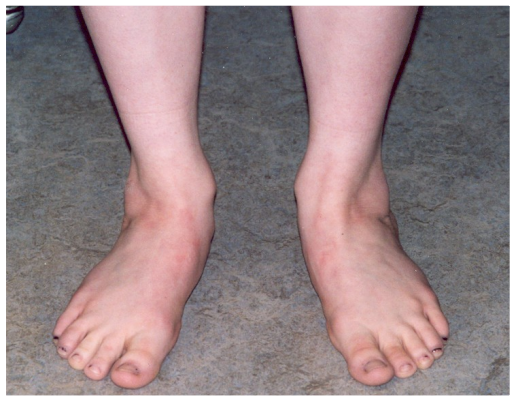
Understanding the Beighton Score

It is very important to understand the Beighton score limitations..only 9 joints.

Look elsewhere, particularly ankles and feet, also TMJ.

A low Beighton score does not exclude hypermobility





5 Point Questionnaire
(Hakim and Grahame 2003)

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

Review Of Associated Features

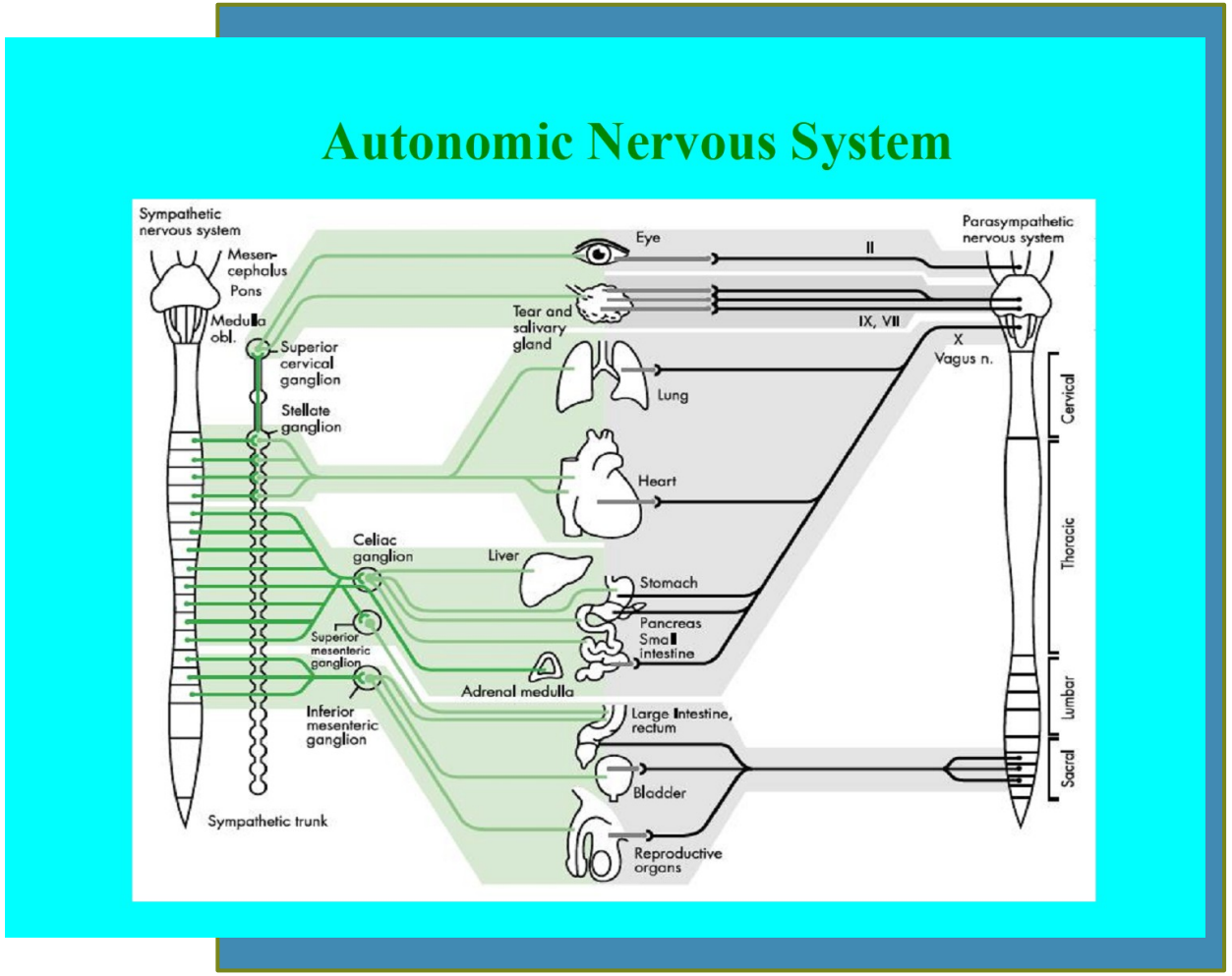
1. Autonomic Dysfunction
2. Gastrointestinal eg.: IBS
3. Mast cell activation
4. Neurological
5. Skin manifestations
6. Urological
7. Respiratory
8. Chronic pain/Fibromyalgia
9. Psychological

NB: This doesn't mean you will get ALL of the above!

Remember: Everyone is different

1. Autonomic Dysfunction Symptoms

- Palpitations
- Dizziness
- Fainting
- Postural hypotension(POTS)
- Temperature control
- Sweating
- Bowels/bladder
- Shortness of breath

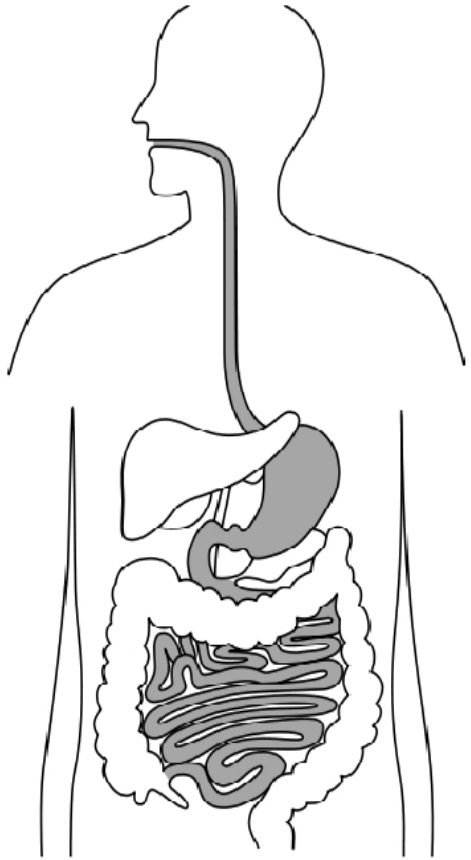


Cardiovascular Dysregulation

• Orthostatic Intolerance	Young women Delayed hypotension
• Postural Orthostatic Ta chycardia (POTs)	> 30 bpm rise in pulse Associated with OI
	Patchy dysautonomia, pooling of blood in peripheral circulation
• Orthostatic Hypotension	Rapid drop in blood pressure > 20/10 mmHg Standing intolerance
	Activation / hypersensitivity of cardiac sympathetic system

Vaeroy et al 1998, Petzke & Clauw 2000, Raj et al 2000, Rosner et al 2000, Cohen et al 2001, Giske et al 2008, Mathias CJ et al. Nat Rev Neurol. 2011; 8(1):22-34, De Wandele I, et al 2014

2. Gut Symptoms in EDS



	Symptoms
Oesophagus	Problems swallowing
Gastro-oesophageal junction	Reflux
Stomach	Fullness, nausea, vomiting, bloating, reflux
Large bowel	Constipation
Rectum	Constipation

Slide courtesy of Prof. Qasim Aziz

2. Bowel Dysfunction in JHS

Mechanical

- Hiatus hernia / weak sphincter
- Gastroparesis: Nausea and Vomiting
- Slow transit: Constipation, Colic, Diarrhoea
- Pseudo Obstruction

(Swallow studies, Manometry, (capsular)endoscopy, Transit studies, colonoscopy)

Autonomic

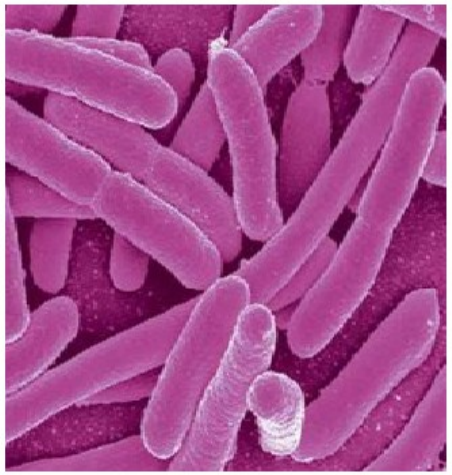
- Neuropathic – dysfunction and pain

Allergies / Intolerances / Chronic Inflammation

(Breath tests, infection studies, stimulation and exclusion trials, colonoscopy / histology)

Zarate N et al. 2010. Fikree et al 2014, Domínguez-Ortega G et al 2014, Fikree et al 2015

Effects Of A Sluggish Gut



Slide courtesy of Prof. Quasim Aziz

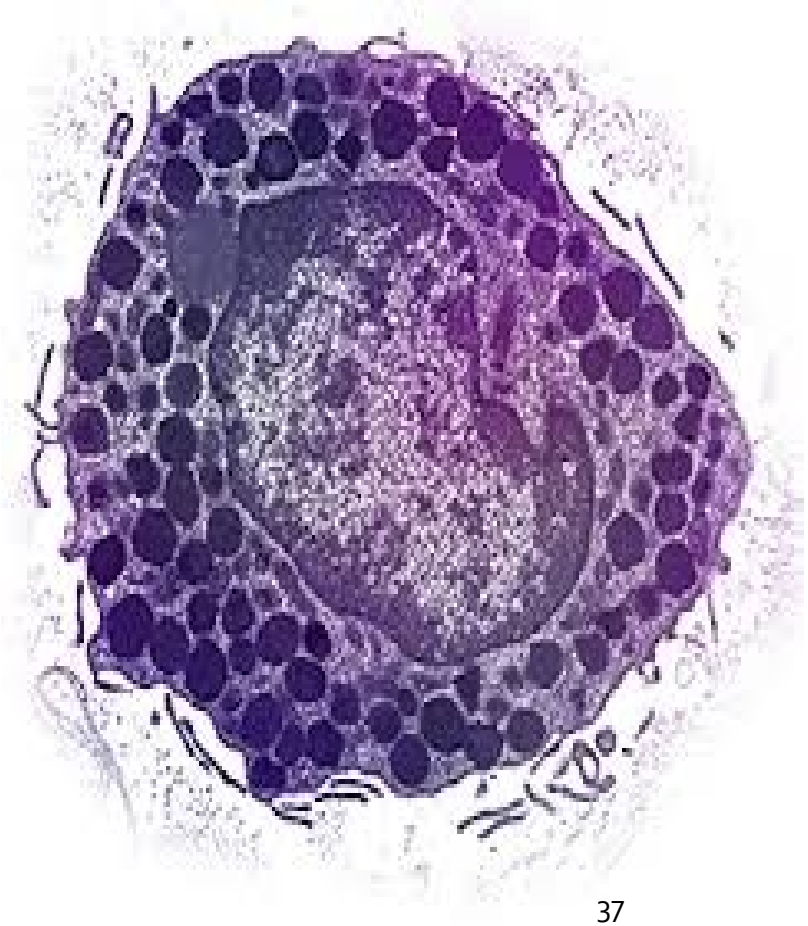
The Microbiome

- Residential gut flora (trillions)
- The hidden metabolic organ
- Association with inflammatory bowel disease, obesity, diabetes, atopy
- Dysbiosis…c. difficile…
- Non diversity of flora…bad
- Fatigue syndrome
- Role in hypermobility not yet fully understood

- Kurzegesagt microbiome….YOUTUBE

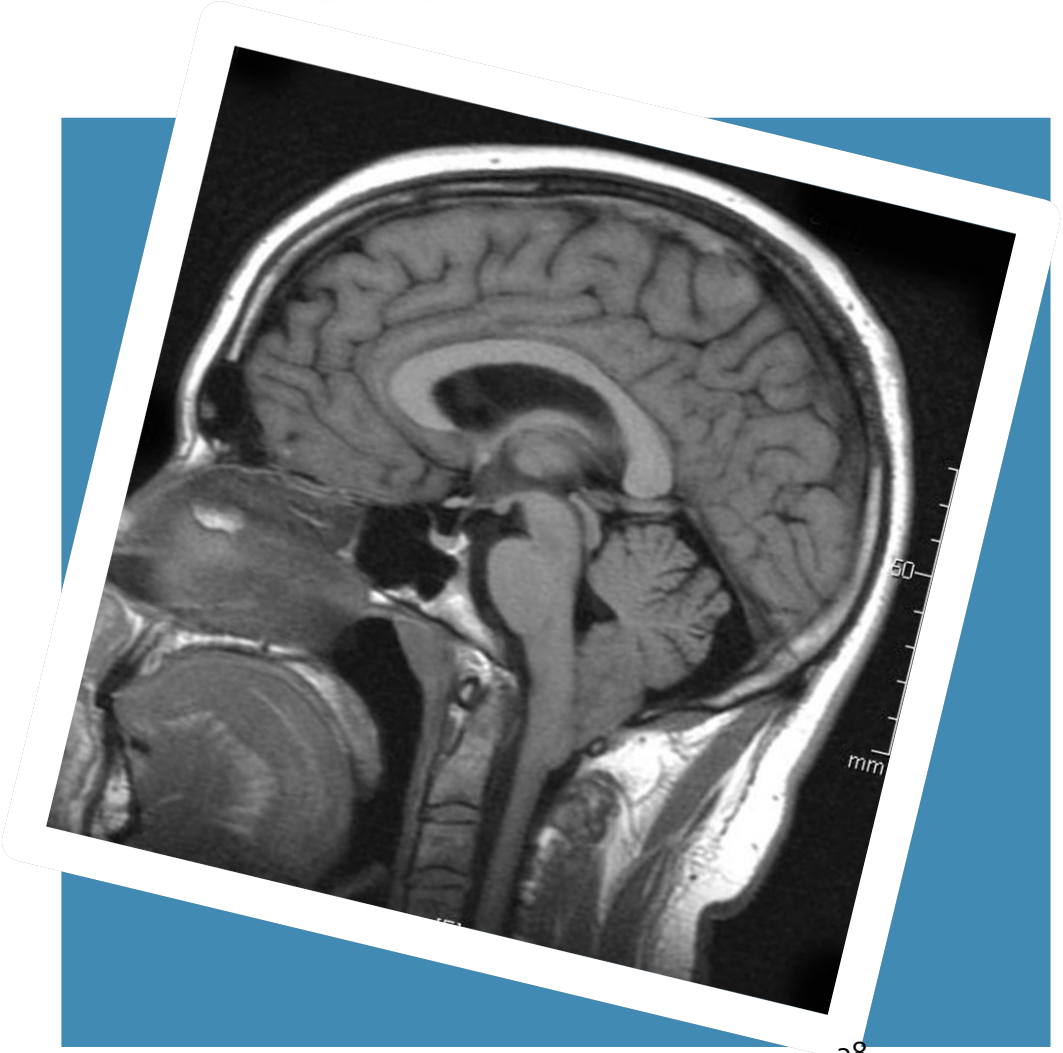
3. Mast cell activation syndrome (MCAD)

- Mast cells inappropriately release mediators
- Dermatological (flushing, itching)
- Cardiovascular (dizziness,syncope)
- GI (diarrhoea, nausea, vomiting)
- Neuro (brain fog, headaches, migraine)
- Respiratory (congestion, cough, wheeze)
- Eye (conjuctivitis)
- General (fatigue, food and drug intolerances)
- Feeling cold all the time
- Anaphylaxis



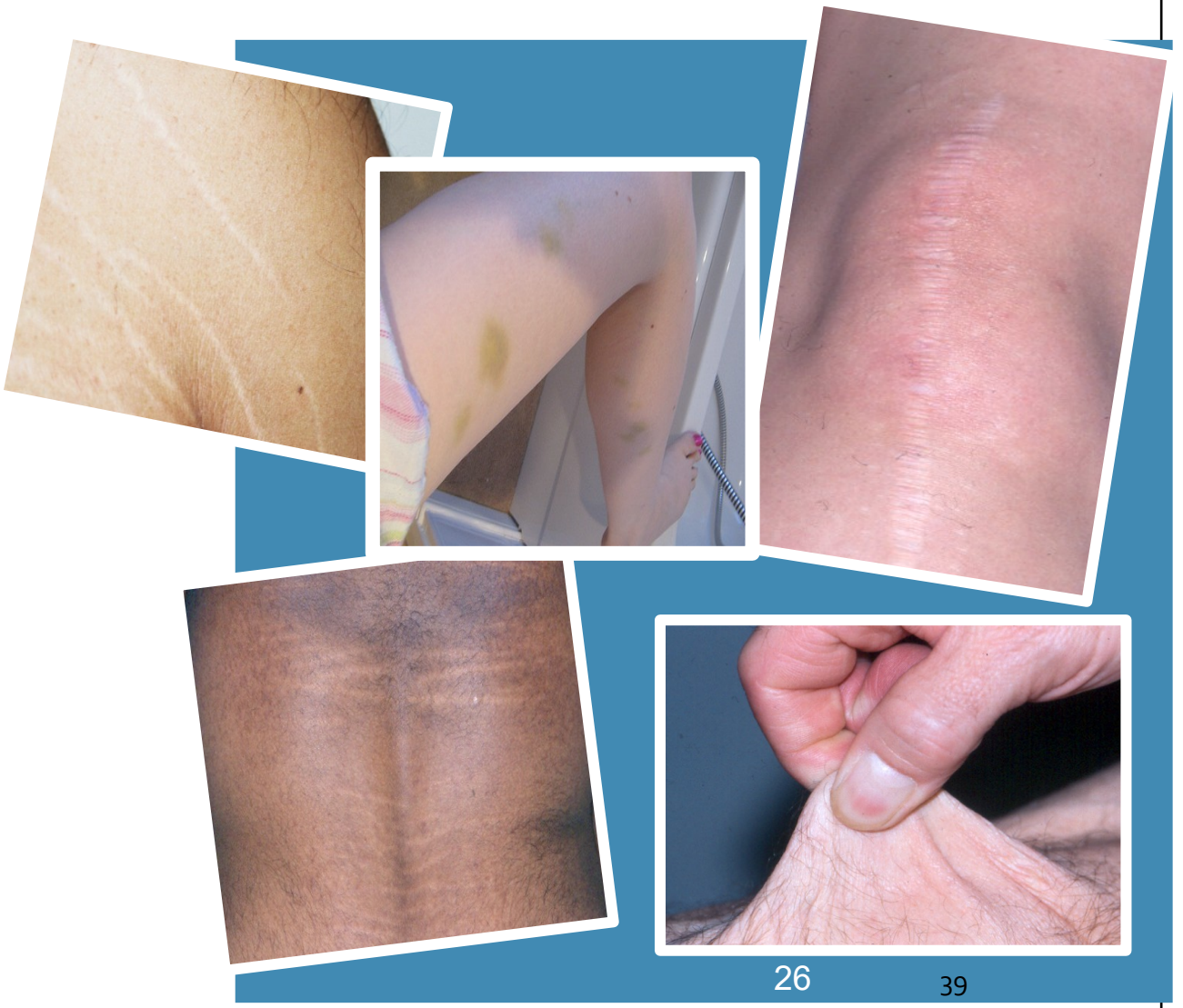
4. Neurological symptoms

- Clumsiness/coordination difficulties
- Small fibre neuropathy on skin Biopsy with EDS (Neurology, 2016: Cazzato et al)
- Arnold-Chiari…Cranio-Cervical instability
- Neurodevelopmental disorders:
  - Dyspraxia, learning disorder, attention deficit/ hyperactivity disorder, autism



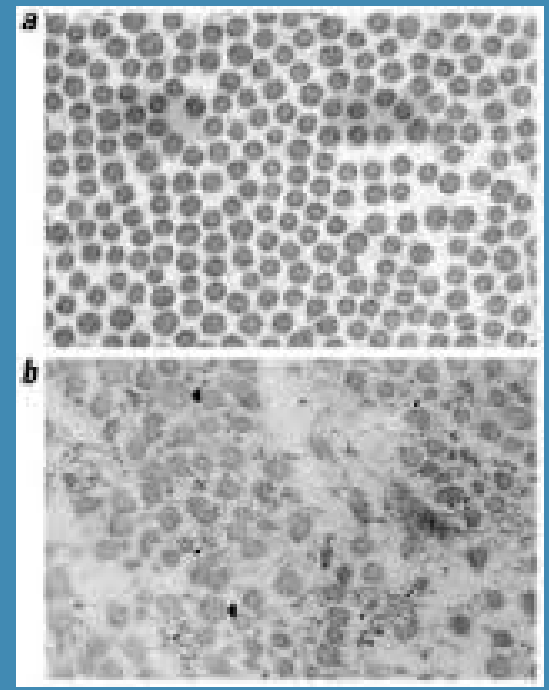
5. Manifestations Affecting the Skin

- Soft, silky, velvety, stretchy
- Slower healing
- Abnormal Scars
- Unexpected stretch marks
- Easy bruising
- Thin, semi transparent
- Nodules



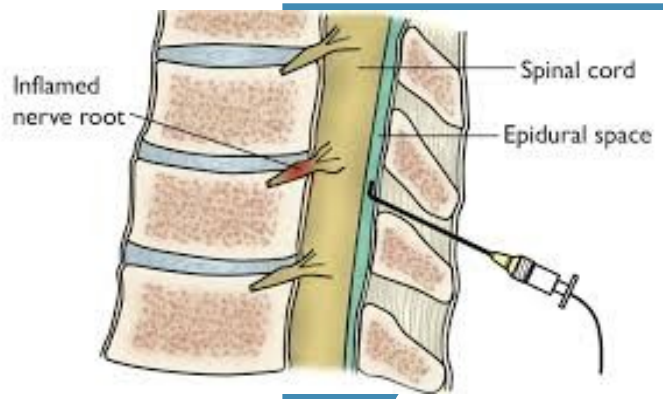
Histology

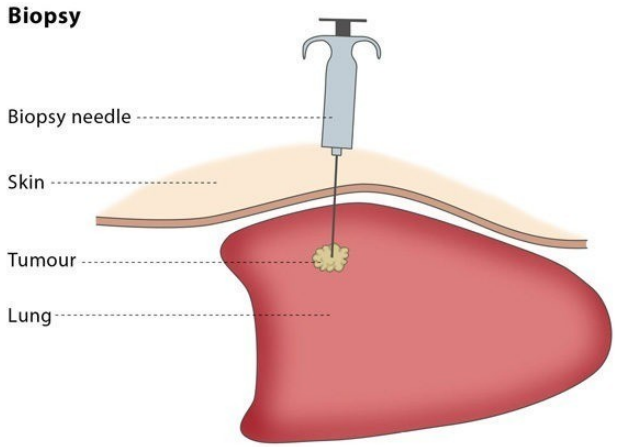
Collagen and elastic fibres in the dermis are abnormal and weaker leading to poor healing.



Insensitivity to Local Anaesthesia

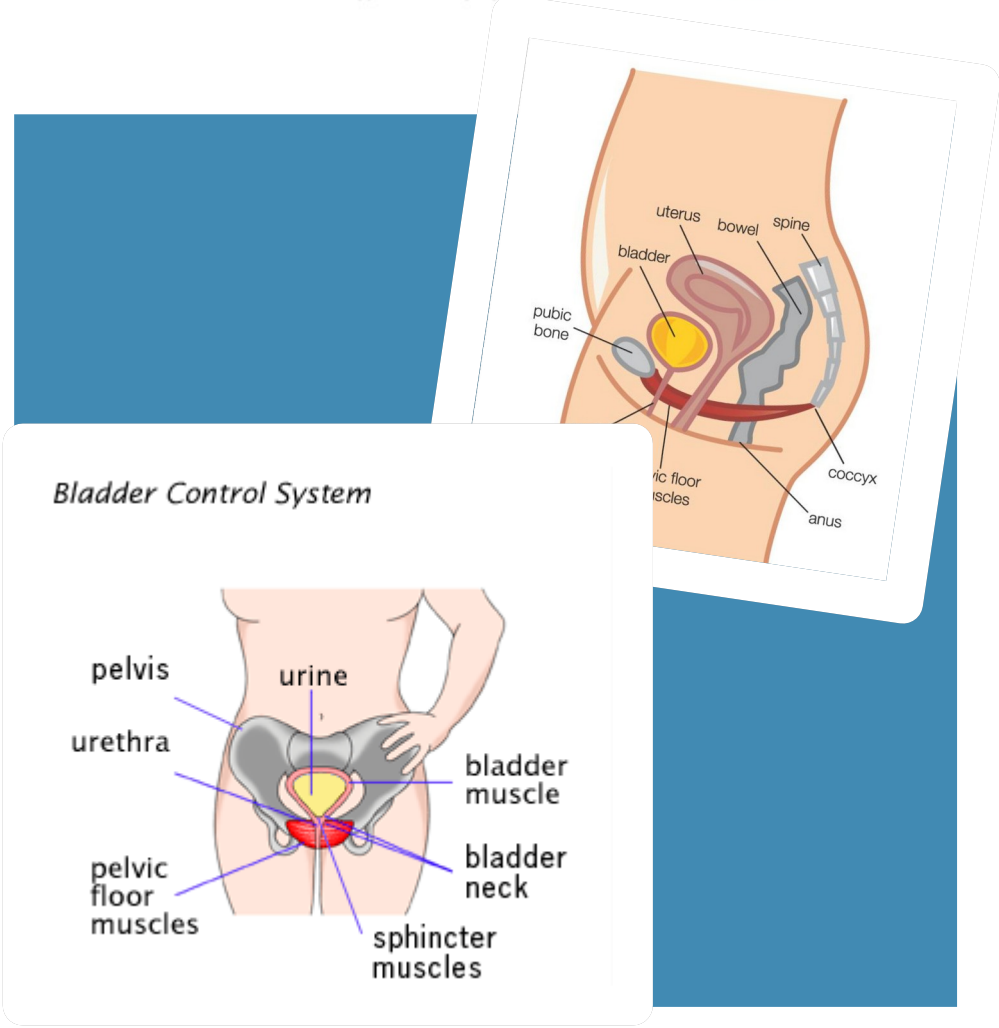
- Dental work
- Regional blocks
- Failed epidural
- Biopsies





6. Bladder & Pelvic Floor

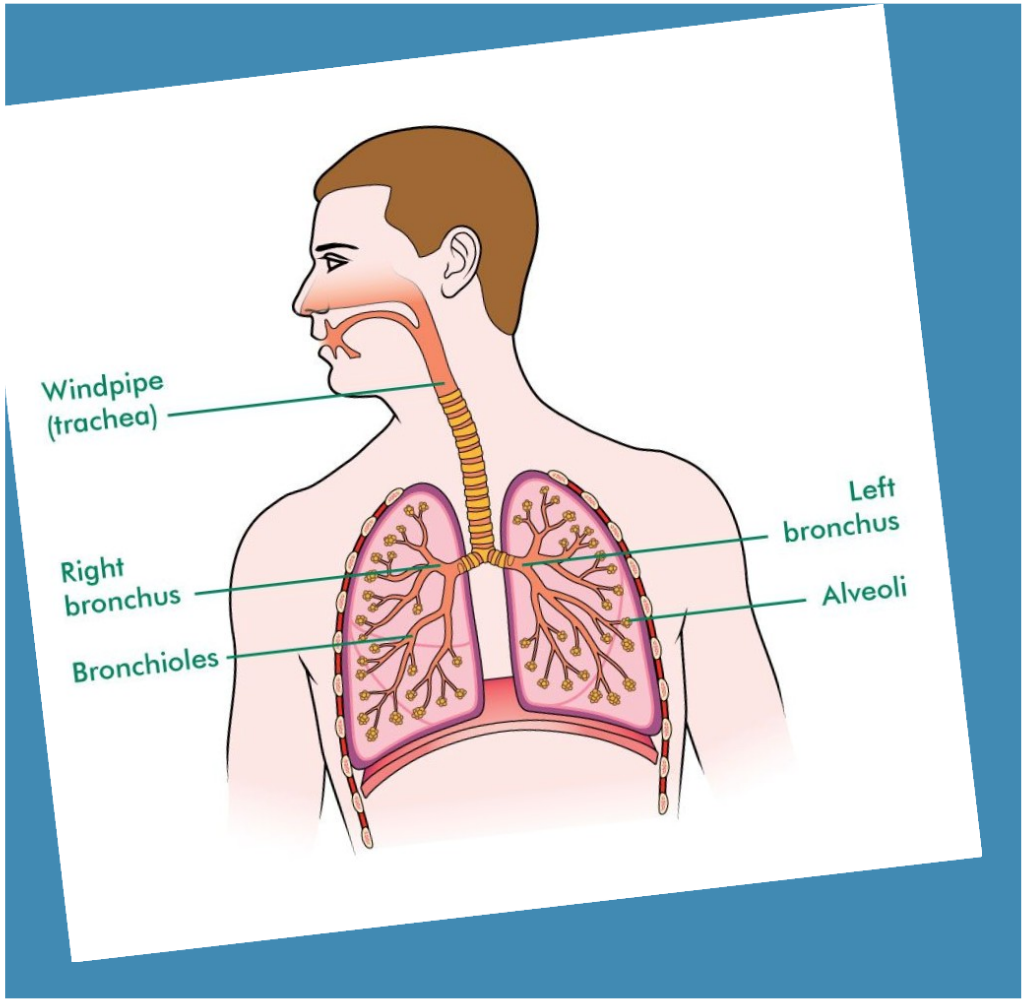
- **Mechanical**
  - Rectal prolapse
  - Rectocele, Cystocele
    - Vaginal / Bladder prolapse
  - Stress incontinence, Urgency
  - Sexual dysfunction
- **Neuropathic and Inflammatory**
  - Bladder instability / irritability
  - Urgency, Frequency
- **Symphysis Pubis Dysfunction**



Al-Rawi ZS, Al-Rawi ZT. 1982, Norton PA et al. 1995, McIntosh LJ, et al 1996, Bai SW, et al. 2002, Karan A, et al. 2004, Mastoroudes H et al. 2013, Mastoroudes H et al. 2013.

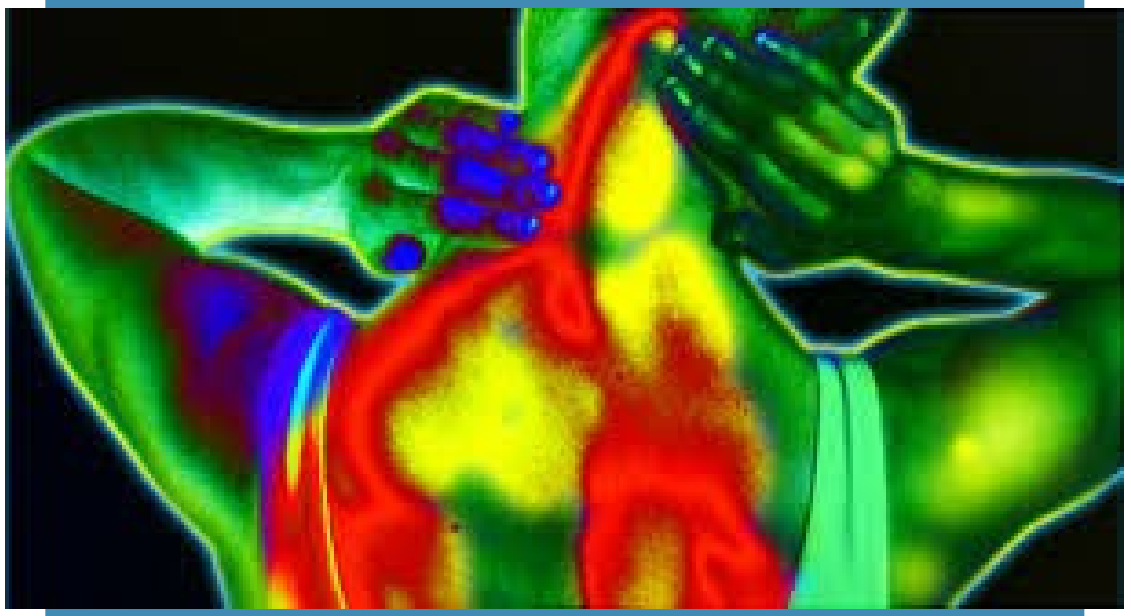
7. Respiratory System

- May present with wheeze which is not bronchospasm
- Association with sleep apnoea
- Pneumothorax



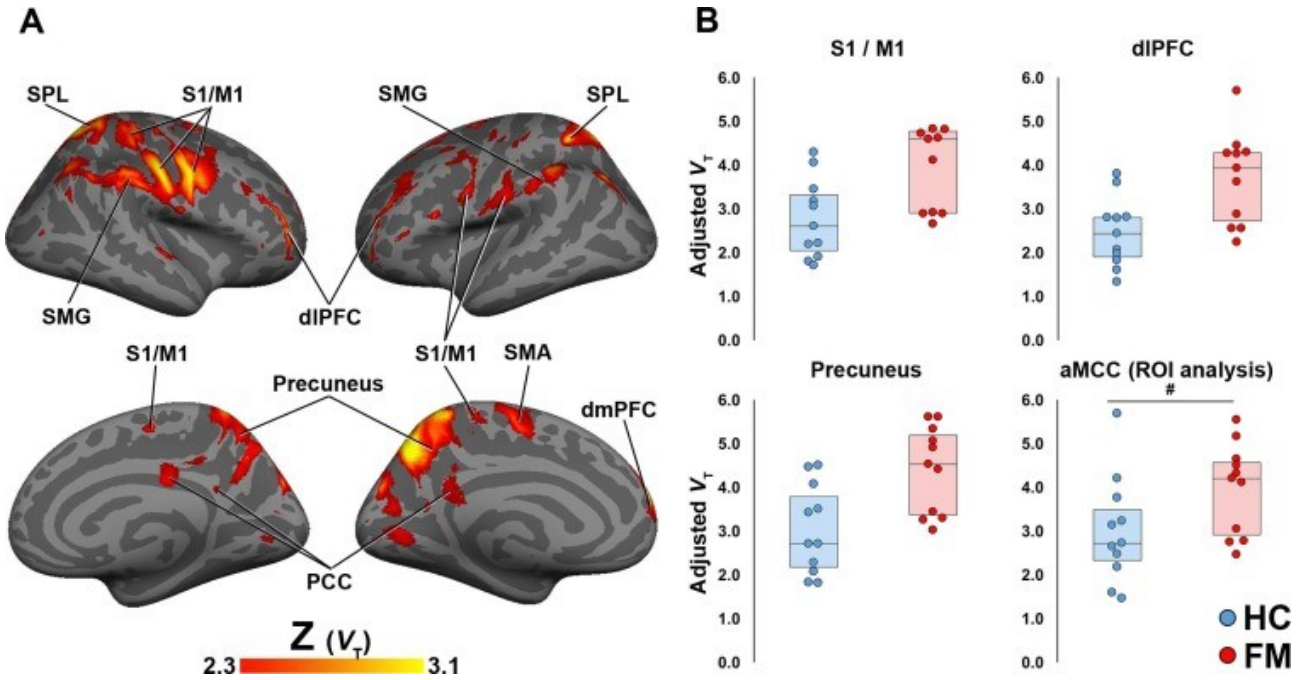
8. Fibromyalgia
(highly significant overlap with hypermobility)

- A long term condition that causes pain all over the body
- Increased sensitivity to pain
- Fatigue (extreme tiredness)
- Muscle stiffness
- Sleep disturbance
- Brain fog (problems with memory and concentration)
- Headaches
- Irritable bowel
- Cause unknown



Glial activation in fibromyalgia

Albrecht 2018



Recap:

10 to 15 percent of the population have evidence of hypermobility. Most are asymptomatic.

For some it is an advantage but, for others:

- Musculoskeletal pain
- Multiple symptoms (hypermobility syndrome…now HSD/hEDS
- May be a Hereditary Disease of Connective Tissue(HCTD) eg Ehlers-Danlos syndrome, Marfans, Osteogenesis Imperfecta

My Approach to Hypermobility

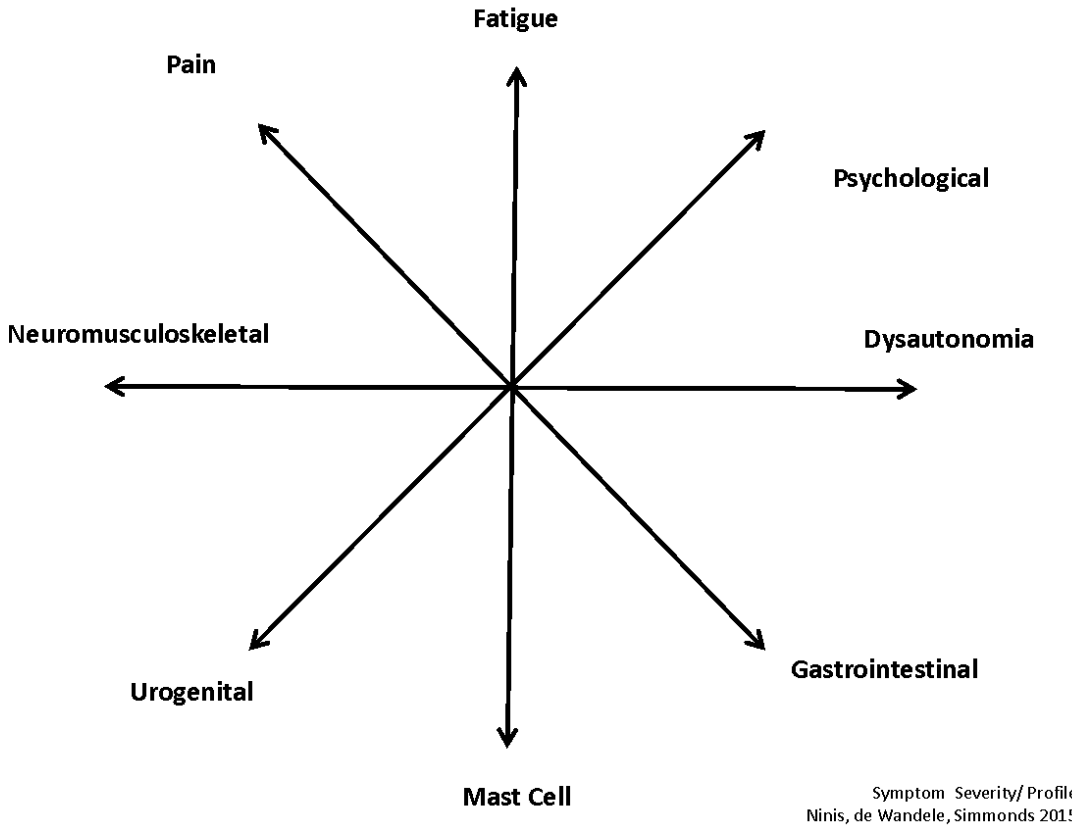
- Listen carefully, give time
- Examine and Investigate appropriately
- Check that hypermobility is relevant
- VALIDATE symptoms and give workable explanation.
- Medication review…medications usually don't work!
- Make a management plan:

- Patient understanding: getting on the same page… resources, signposting.

- Detailed physiotherapy assessment to identify mechanical factors
- Mindful self compassion and solution focused psychotherapy for psychological wellbeing
- The right type of exercise is important eg: yoga, aqua, tai chi, Modified Pilates
- Alexander technique
- Empathic follow up, with guidance
- Holistic approach i.e. pacing, sleep, dietary advice

Overall assessment

Helping the patient to organise their thinking.



Symptom Severity/ Profile
Ninis, de Wandele, Simmonds 2015

My Approach to Hypermobility

- Listen carefully, give time
- Examine and Investigate appropriately
- Check that hypermobility is relevant
- VALIDATE symptoms and give workable explanation.
- Medication review…medications usually don't work!
- Make a management plan:

- Patient understanding: getting on the same page… resources, signposting.

- Detailed physiotherapy assessment to identify mechanical factors
- Mindful self compassion and solution focused psychotherapy for psychological wellbeing
- The right type of exercise is important eg: yoga, aqua, tai chi, Modified Pilates
- Alexander technique
- Empathic follow up, with guidance
- Holistic approach i.e. pacing, sleep, dietary advice

Dr. Philip Bull

https://www.hypermobility.org

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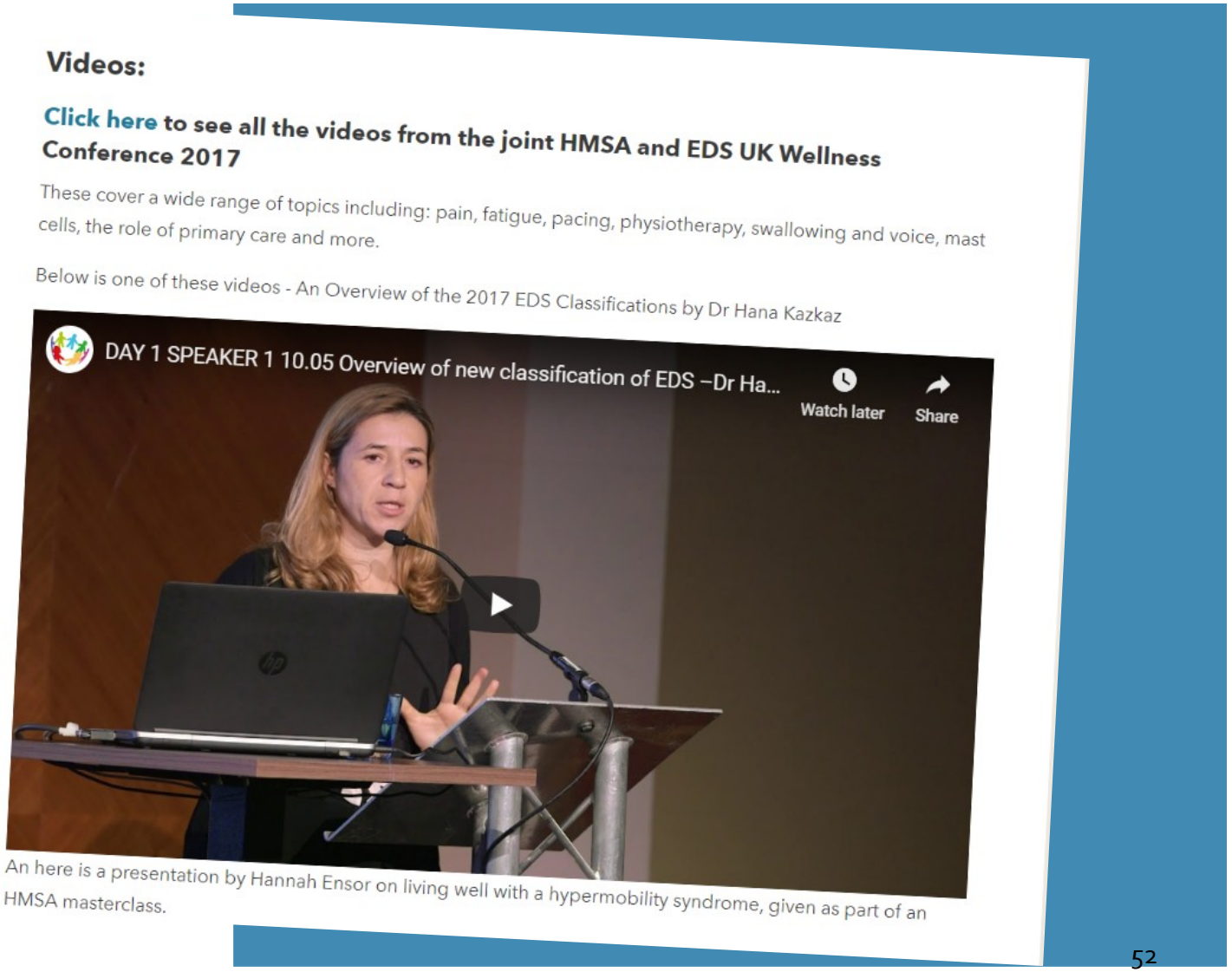
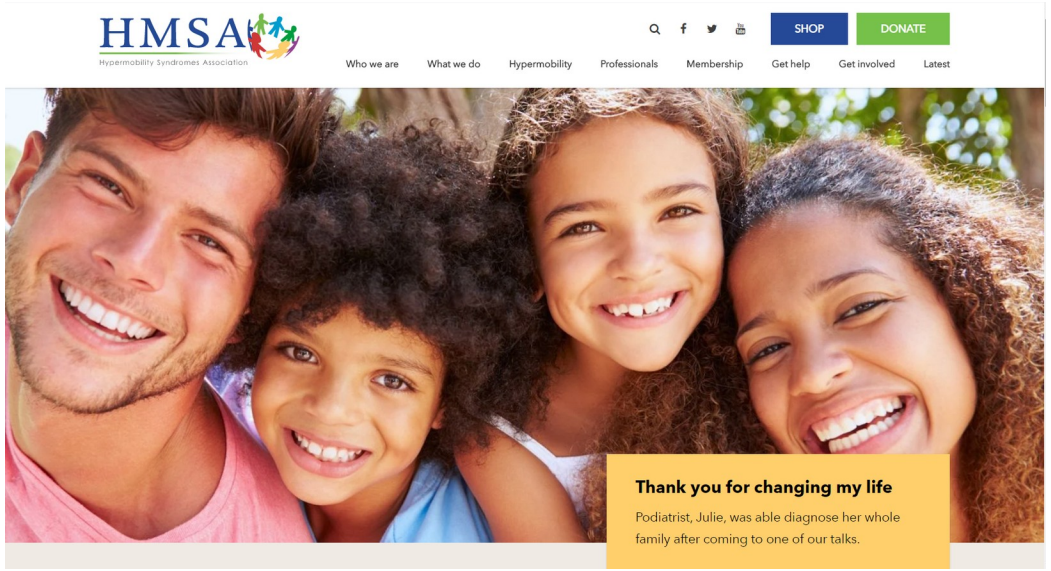
Further Resources To Access

- Video
- Leaflets
- Bi-Annual Journal
- Workshops
- Webinars
- Booklets
- Schools HUB



Great Resources for professionals and patients



https://www.hypermobility.org

- Professional membership
- The HMSA Education Model
- Resources

https://www.hypermobility.org/kent-model-resources

The HMSA Education model:
(the Kent model)

- Increasing understanding through education
- Utilising existing resources
- Creating a network

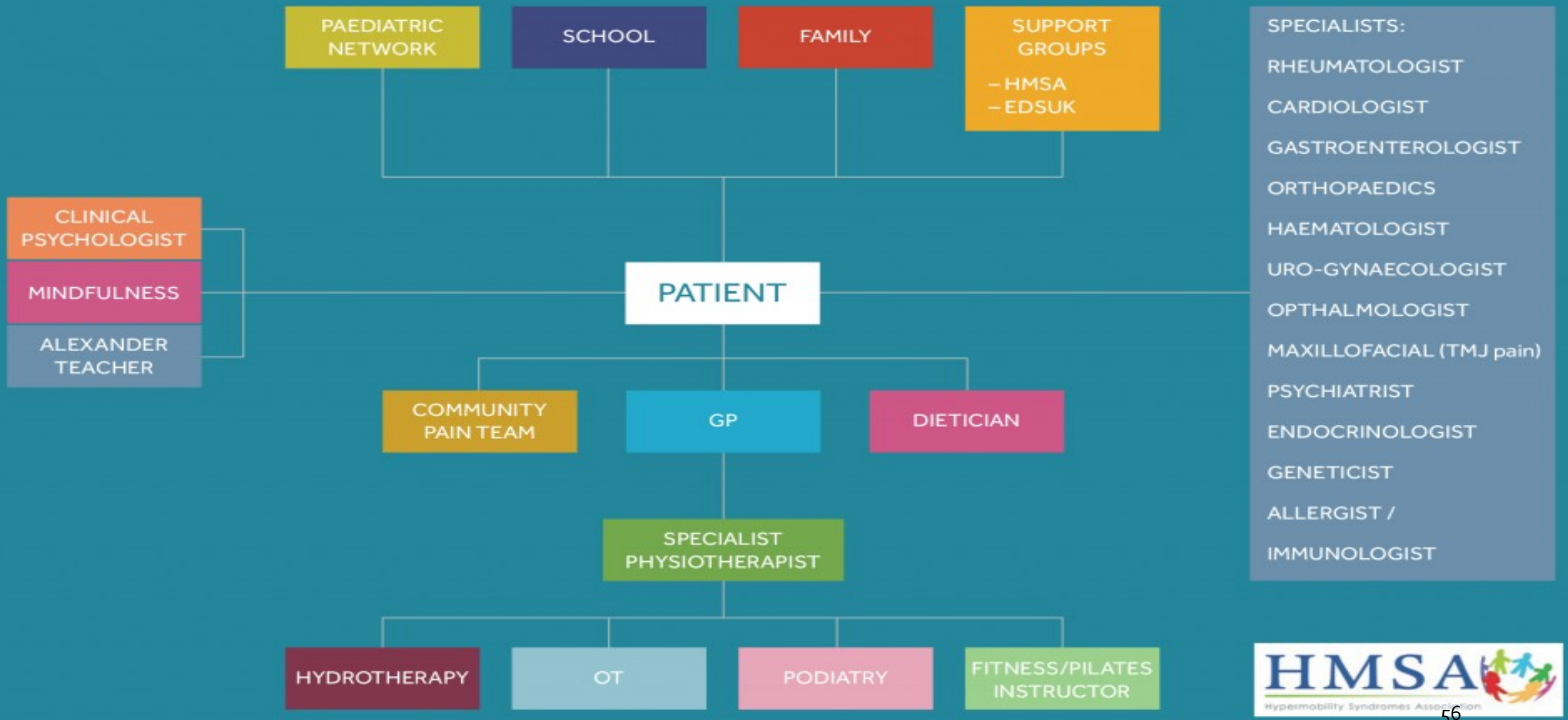
OPPORTUNITIES:

- Self management
- Reduced consultations.
- Reduced referrals
- Potential reduced drug costs
- Well managed patients
- Opportunities for early diagnosis and better outcomes.

Masterclasses:

THE HYPERMOBILITY NETWORK

(multidisciplinary care web)



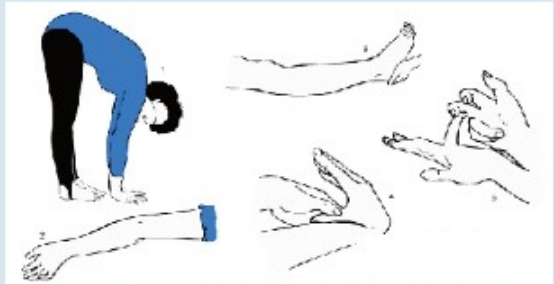
HMSA
Hypermobility Syndromes Association

Hypermobility Syndromes: Key messages for health professionals

1. Be aware of the 5 point questionnaire

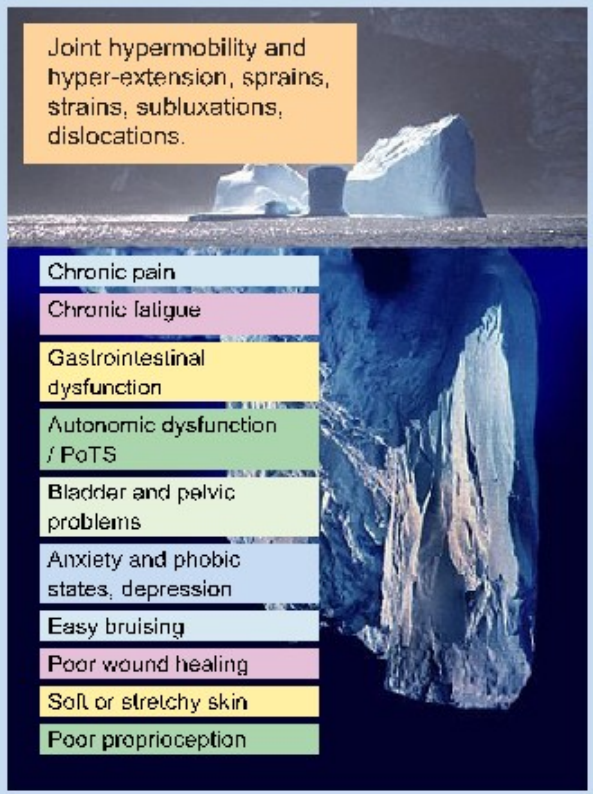
Can you now (or could you ever)
Place your hands flat on the floor without bending your knees?
Bend your thumb to touch your forearm?
Do party tricks?
Dislocate on more than one occasion?
Consider yourself double-jointed?

2. The Beighton Score (not a diagnostic test)

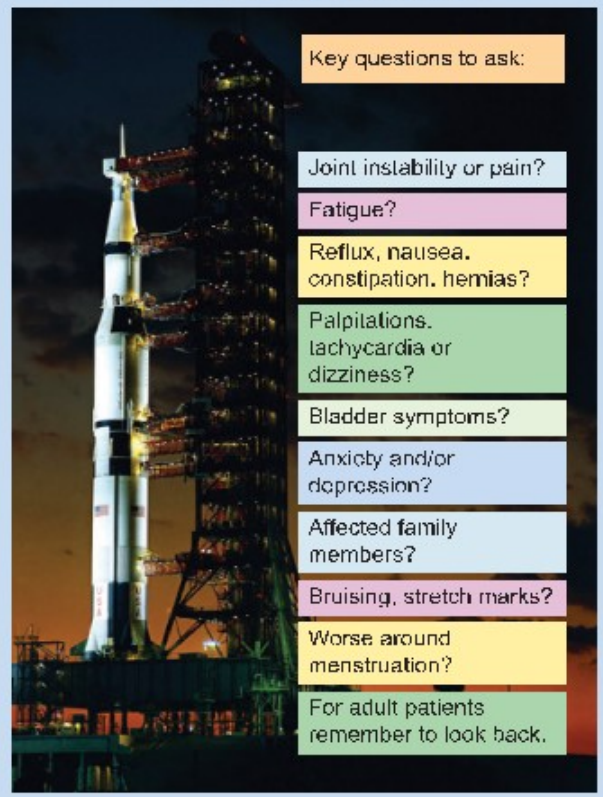


Low Beighton score does not exclude hypermobility, which can affect other joints or other planes of movement.

3. Know the common associations



4. It's not rocket science.



5. When to refer for Genetic Testing

- Marfanoid habitus + aortic root or ocular signs
- Very stretchy skin or unusually widened atrophic scars
- Large unusual bruising/haematomas
- Organ rupture
- Personal or family history of young onset unexplained arterial dissection, aneurysms or significant haemorrhage
- Significant kyphoscoliosis
- Recurrent large hernias

6. What can you do?

- Listen carefully
- Make a diagnosis (asymptomatic hypermobility, HSD, hEDS, query rare HDCT)
- Ensure you have access to a hypermobility orientated physiotherapist with a holistic approach.
- Refer for podiatry assessment.
- Use HMSA website as a resource for the patient and yourself (see 'professionals' section).
- Be aware that medications are often ineffective.
- Try to avoid codeine, opioids, and morphine.
- PoTS? Increase fluid and salt in moderation
- IBS-like symptoms? See website for more info.
- Address anxiety and depression consider evidence based intervention such as mindfulness and mindful self compassion.
- Consider the Alexander Technique and Tai Chi.
- Review and support

For more information visit

Hypermobility.org/Kent-Model

HMSA
Hypermobility Syndromes Association

The Information Standard Certified Member