- **Definitions of joint hypermobility:**
  - Joint hypermobility (JH):
    - Capability for a joint to move beyond normal physiological limits.
    - Descriptor, not diagnosis
    - Also known as joint laxity, double-jointedness, or joint hyperlaxity.
  - Localized joint hypermobility (LJH):
    - Hypermobility in fewer than 5 types of joints (i.e. Beighton Score/Scale)
    - May be inherited or acquired
  - Generalized joint hypermobility:
    - Hypermobility in 5 or more joints.
    - Not straightforward to diagnose
    - More likely to be congenital than LJH, can also be caused by inflammatory or degenerative diseases or endocrine disorders,
  - Additional speculative classifications:
    - Peripheral joint hypermobility: hypermobility in hands and/or feet only
    - Historical joint hypermobility: older adults who have lost GJH due to age.
- **Joint instability (JI):**
  - Implies a detrimental effect on the joint, whereas JH has neutral implication.
  - Hypermobile joints need not be unstable, and vice versa.
- S**econdary manifestations of joint hypermobility:**
  - Trauma
    - Dislocations, subluxations, and other soft tissue injuries likely result of trauma due to excessive joint movement, typically leads to acute pain and loss of function
    - Microtrauma typically not noticed as it occurs, but can predispose to pain or early joint degeneration over time.
  - Chronic pain
    - Can be a long-term complication of JH
    - Hyperalgesia (increased pain sensitivity) suggested in patients with EDS and chronic pain
      - Could be a direct result of connective tissue defects

        - Could be that a common biological mechanism gives rise to various forms of chronic pain, independent of the cause of joint disease
    - Disturbed proprioception (sense of the relative position of parts of the body)
        - Mechanism of relationship to JH poorly understood.
    - Other musculoskeletal traits
        - May be the result of interactions of "soft" musculoskeletal tissues with mechanical forces during growth.
        - Include flat feet, elbows, feet, and toes angled outward (valgus deformity), spinal curvature (scoliosis, kyphosis, lordosis), and flat head syndrome
- **Pathogenesis and pleiotropy:**
    - JH-associated musculoskeletal traits are secondary effects, mediated by JH as well as independent factors, not a direct result of the underlying cause of JH.
    - Pleiotropy: mutations in a single gene giving rise to multiple, seemingly unrelated effects.
    - Combination of JH and musculoskeletal symptoms does not indicate a genetic syndrome, since musculoskeletal symptoms do not share the same underlying cause as JH
- **Genetic syndromes with joint hypermobility:**
    - Hereditary disorders of the soft connective tissue
        - E.g. EDS, Marfan syndrome, Loeys-Dietz syndrome, Beals syndrome
    - Other genetic syndromes
        - Skeletal dysplasias (disorders of bone & cartilage development)
        - Hereditary myopathies (muscular diseases)
        - Chromosomal disorders (e.g. Down syndrome)
        - Disorders with multiple congenital anomalies/intellectual disabilities (e.g. Fragile X)
    - An annotation on hEDS
        - "Hypermobile Ehlers-Danlos syndrome" (hEDS) refers to type of Ehlers-Danlos with no known genetic marker
        - New set of criteria stricter than those for JHS or EDS-HT, excludes many patients with who suffer from secondary JH manifestations.
- **Classifying joint hypermobility:**
    - Asymptomatic or isolated LJH, PJH, or GJH
    - Well-defined syndrome with JH, including hEDS
    - Hypermobility spectrum disorders (HSDs): symptomatic JH that does not satisfying the criteria for a specific syndrome
- **Hypermobility spectrum disorders:**
    - Usually limited to musculoskeletal symptoms

    - Refers to patients with symptomatic JH, but do not meet criteria for hEDS under new nosology
    - Includes patients meeting old criteria for JHS and EDS-HT
    - Family history of hEDS not sufficient to distinguish HSD from hEDS.
    - Four types of HSDs, corresponding to GJH, LJH, PJH, or HJH, plus additional musculoskeletal symptoms.
    - GJH often associated with anxiety and other extra-articular disorders, but does not automatically qualify as genetic syndrome.
- **Important Points:**
    - Need clarification on if Ehlers-Danlos syndromes and hEDS fall under the umbrella term of Hypermobility Spectrum Disorders (HSD) (I think they do), or if separate because this The Ehlers-Danlos Society webinar suggested something different than how paper reads – In my opinion.
    - Acquired = possible heritable component, but "switch" is "turned on" at some point in our lifetime (ie. RA, Type 1 diabetes, etc.). Are present only in certain cells, not in every cell in the body.
    - Inherited = mutations passed down from parents that are present throughout a person's life in virtually every cell in the body – ie. Mutations distinctly found in one or more of our 23 pairs of chromosomes that come from the egg and sperm that were conceived from. Example – Down's syndrome, Vascular EDS, etc.

"A gene mutation is a permanent alteration in the DNA sequence that makes up a gene, such that the sequence differs from what is found in most people. Mutations range in size; they can affect anywhere from a single DNA building block (base pair) to a large segment of a chromosome that includes multiple genes.

Gene mutations can be classified in two major ways:

"Hereditary mutations are inherited from a parent and are present throughout a person's life in virtually every cell in the body. These mutations are also called germline mutations because they are present in the parent's egg or sperm cells, which are also called germ cells. When an egg and a sperm cell unite, the resulting fertilized egg cell receives DNA from both parents. If this DNA has a mutation, the child that grows from the fertilized egg will have the mutation in each of his or her cells.

Acquired (or somatic) mutations occur at some time during a person's life and are present only in certain cells, not in every cell in the body. These changes can be caused by environmental factors such as ultraviolet radiation from the sun, or can occur if a mistake is made as DNA copies itself during cell division. Acquired mutations in somatic cells (cells other than sperm and egg cells) cannot be passed on to the next generation."

Source - https://ghr.nlm.nih.gov/primer/mutationsanddisorders/genemutation"

- We do not know all ways hypermobility is either inherited or acquired yet. Hundreds of connective tissue disorders cause hypermobility and other conditions are also associated with hypermobility. We need research to determine the exact links – ie. Causes and Correlations.
- Epigenetics and Genetics both play roles, but only research will help us understand how much and which factors help and hurt us. Early detection and integrative preventative methods is key. Lifestyle plays a big role, as does access to adequate healthcare.
- Yes, the new classification states that essentially HSD (previously referred to as JHS) is not one in the same as hEDS. And yes, all of us have to go back and correct this information on our sites, even me (EDS Wellness).
- **JHS = HSD, and HSD does not = hEDS**. BUT not equaling or not being the same DOES NOT MEAN LESS THAN or one is MORE SEVERE vs. the other. They can be equal in severity and symptoms, issues and needs should be treated with the same seriousness and urgency.
- The distinction and changes to the diagnostic criteria, most specifically for hEDS and cEDS, are not downgrades as Lara Bloom stated. It's critical to hone the new terminology and understand the bigger picture.
- ***Why the change again?*** Because our understanding of connective tissue disorders as a whole, Ehlers-Danlos syndromes, and related comorbidities have increased, but we have so much more to understand. We need more research and clear diagnostic guidelines to continue the momentum that we've made and those the medical community has made over the last 5-10 years.
- I was diagnosed clinically 3x over the last 12 years by the best EDS specialists with Classical EDS, but I'm negative according to some genetic tests, and possibly according to the new criteria. ***Let's walk through it, factor in other new types of EDS as well, and see where I may now fall according to the new criteria.***
- ***My own diagnosis has most likely changed, but that doesn't change the name of my organization, what I do to help myself, or how I view my condition.*** It just is what it is. The new criteria will help all of us understand the how's and why's we are built the way we are, what makes us different and the factors that help and hurt us.
- ***We know so much that we know so little.*** Research has truly just begun – in my opinion. It's an exiting time for the broader HSD and EDS communities, and for the medical community as a whole.
- A separate document with Q&A from The EDS Society webinar will also be provided.

- **Resources:**
  - Q&A on 2017 EDS Classification by The EDS Society - http://ehlers-danlos.com/wp-content/uploads/QandA-2.pdf
  - Update on the 2017 Ehlers-Danlos syndromes (EDS) International Classification – Updated EDS Nosology to be Released March 15th,

2017 - http://edswellness.org/2017/02/19/update-2017-ehlers-danlos-syndromes-eds-international-classification-updated-eds-nosology-released-march-15th-2017/
- *'A Framework for the Classification of Joint Hypermobility and Related Conditions'* (2017) - http://hdl.handle.net/1854/LU-8510229
- *'Ehlers–Danlos Syndrome, Classical Type'* (2017) - http://hdl.handle.net/1854/LU-8510231