

# EXTERNALLY LED PATIENT-FOCUSED DRUG DEVELOPMENT MEETING

31 OCTOBER 2023

## PRESENTATION

Introduction to the Ehlers-Danlos  
Syndromes and Hypermobility  
Spectrum Disorders

## SPEAKER

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# The Ehlers-Danlos Syndromes



- These are a group of hereditary disorders, all involving the connective tissue
- There are 14 different types, and these have a wide range of differing presentations
- The underlying gene(s) causing 13 of these types are known
- These genes affect the structural proteins of the connective tissue and the enzymes involved in the processing of these proteins.
- The genes underlying the hypermobile type of EDS are not yet known

# Classifying Joint Hypermobility: Hypermobility Spectrum Disorders



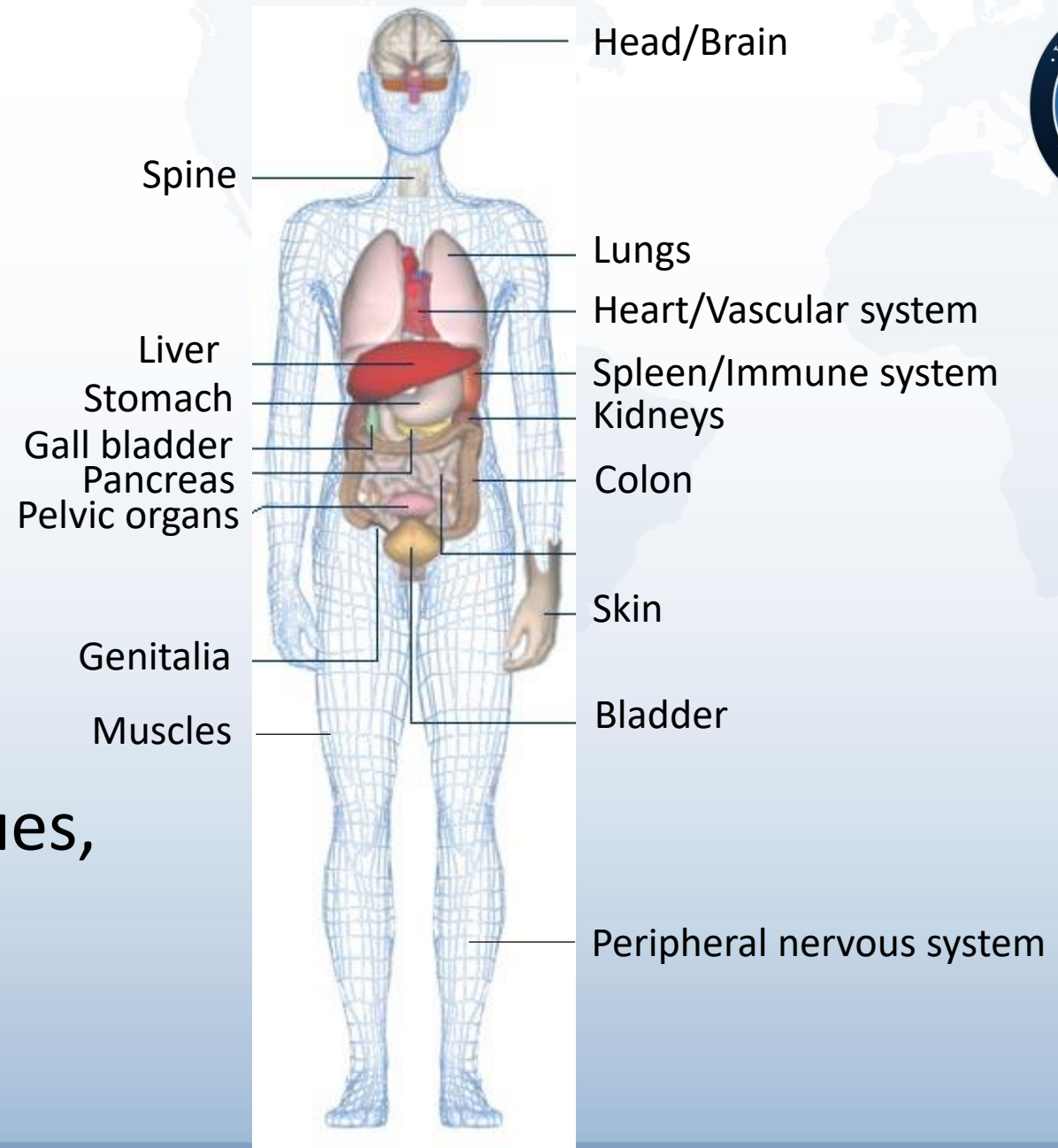
- Persons with asymptomatic joint hypermobility
  - Localized
  - Generalized
  - Peripheral
- Individuals with symptomatic joint hypermobility, not meeting diagnostic criteria for a syndrome
- Persons with a well-defined syndrome with joint hypermobility

EDS and HSD are complex,  
multi-disciplinary disorders



“If you can’t connect the issues,  
Think connective tissues”

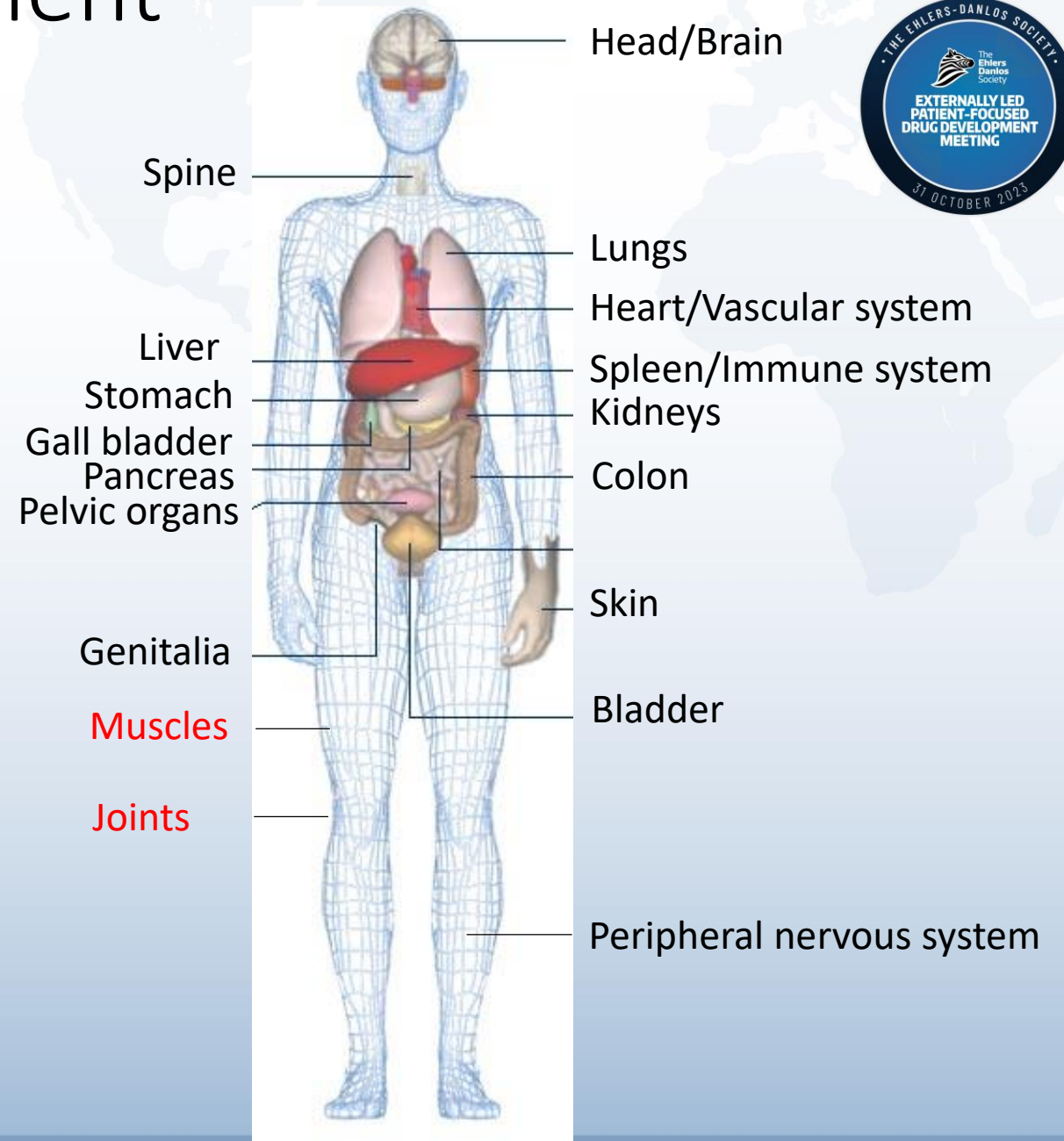
Dr. Heidi Collins





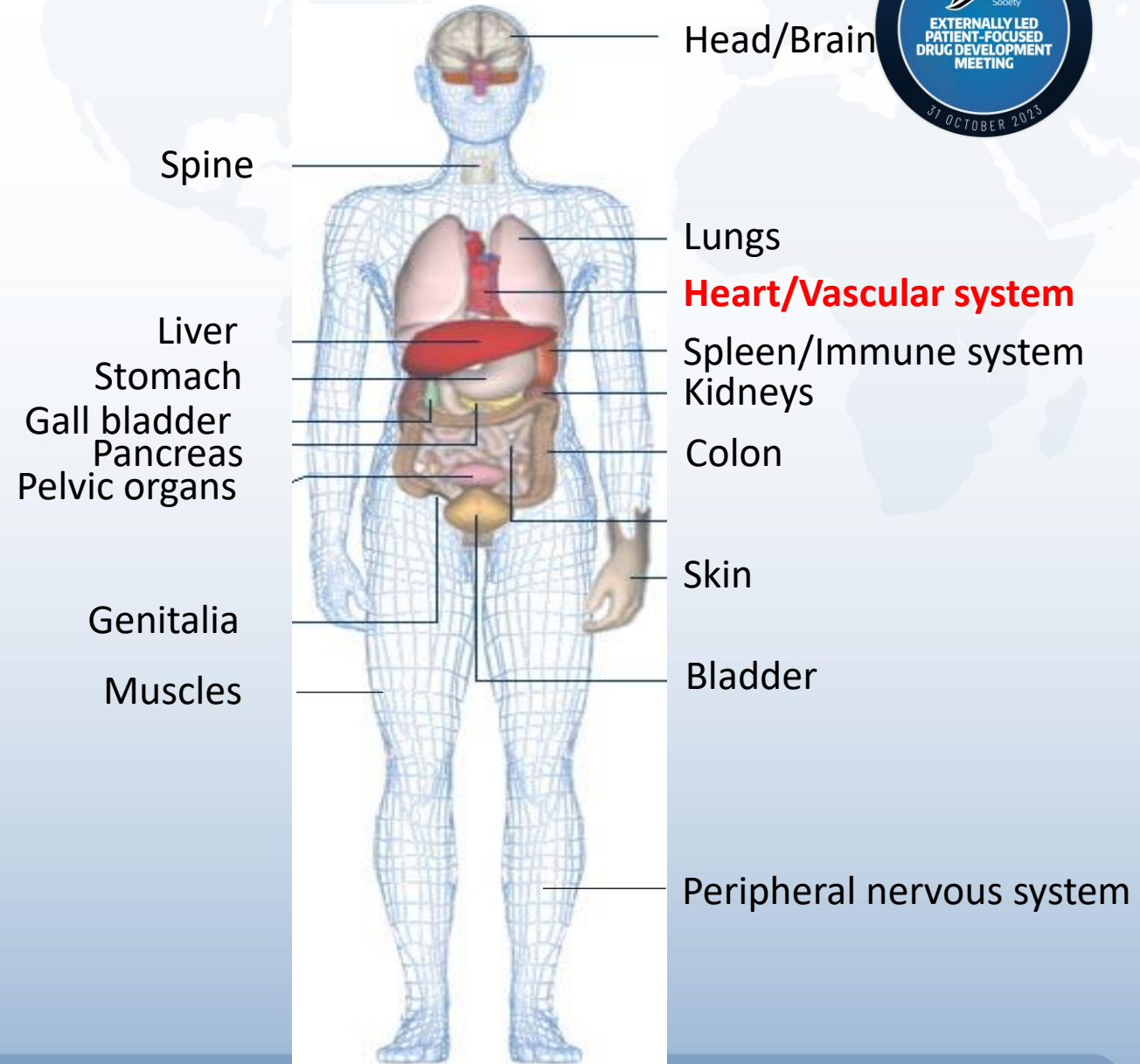
# Musculoskeletal involvement

- Joint laxity/instability
- Subluxations/dislocations
- Tendency for tendon rupture/tears
- Chronic musculoskeletal pain
- Frequent sprains



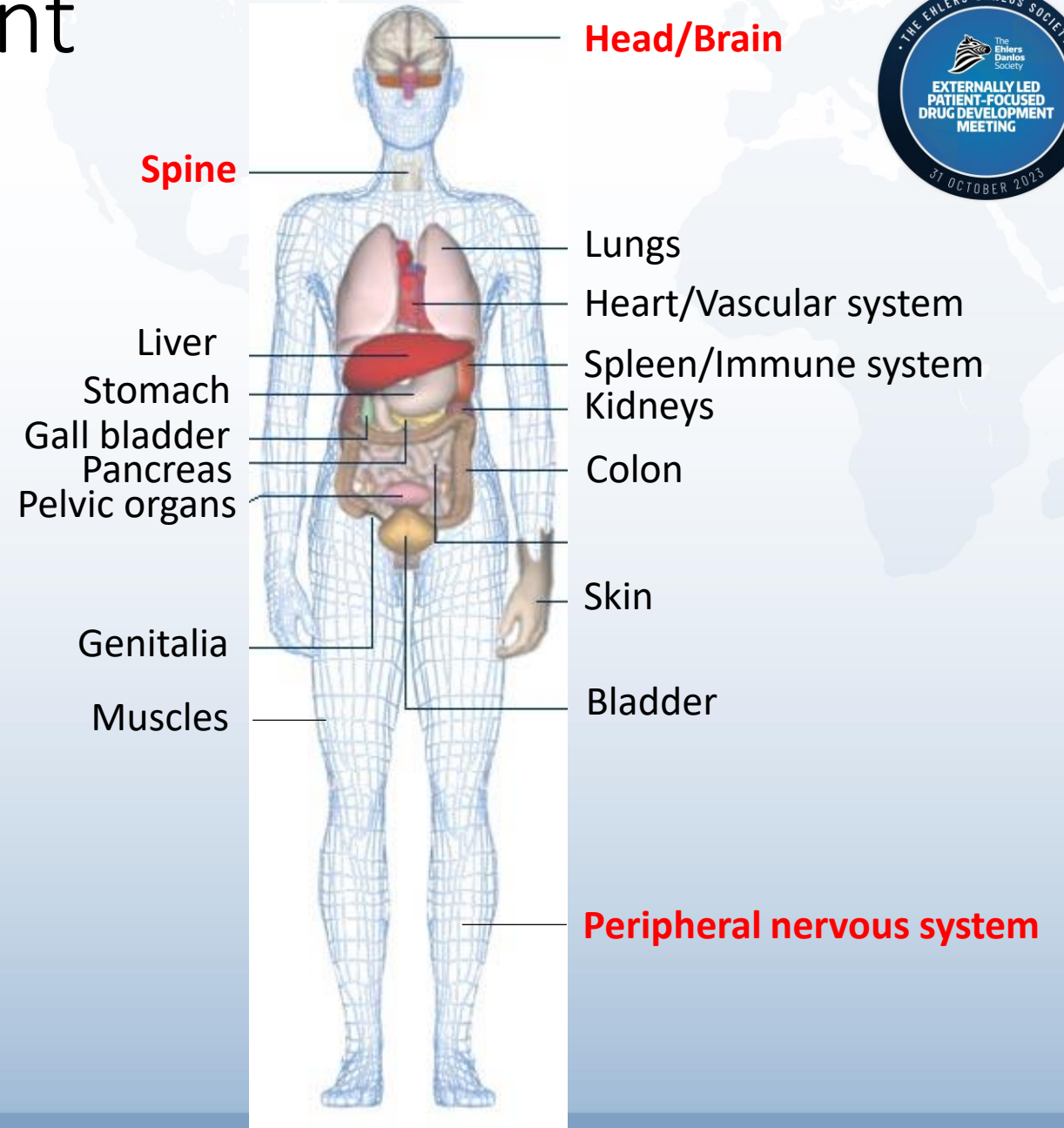
# Cardiovascular Manifestations

- Orthostatic intolerance –
  - Postural orthostatic tachycardia syndrome
  - Neurally mediated hypotension
- Mitral valve prolapse
- Aortic root dilation
- Chronic fatigue



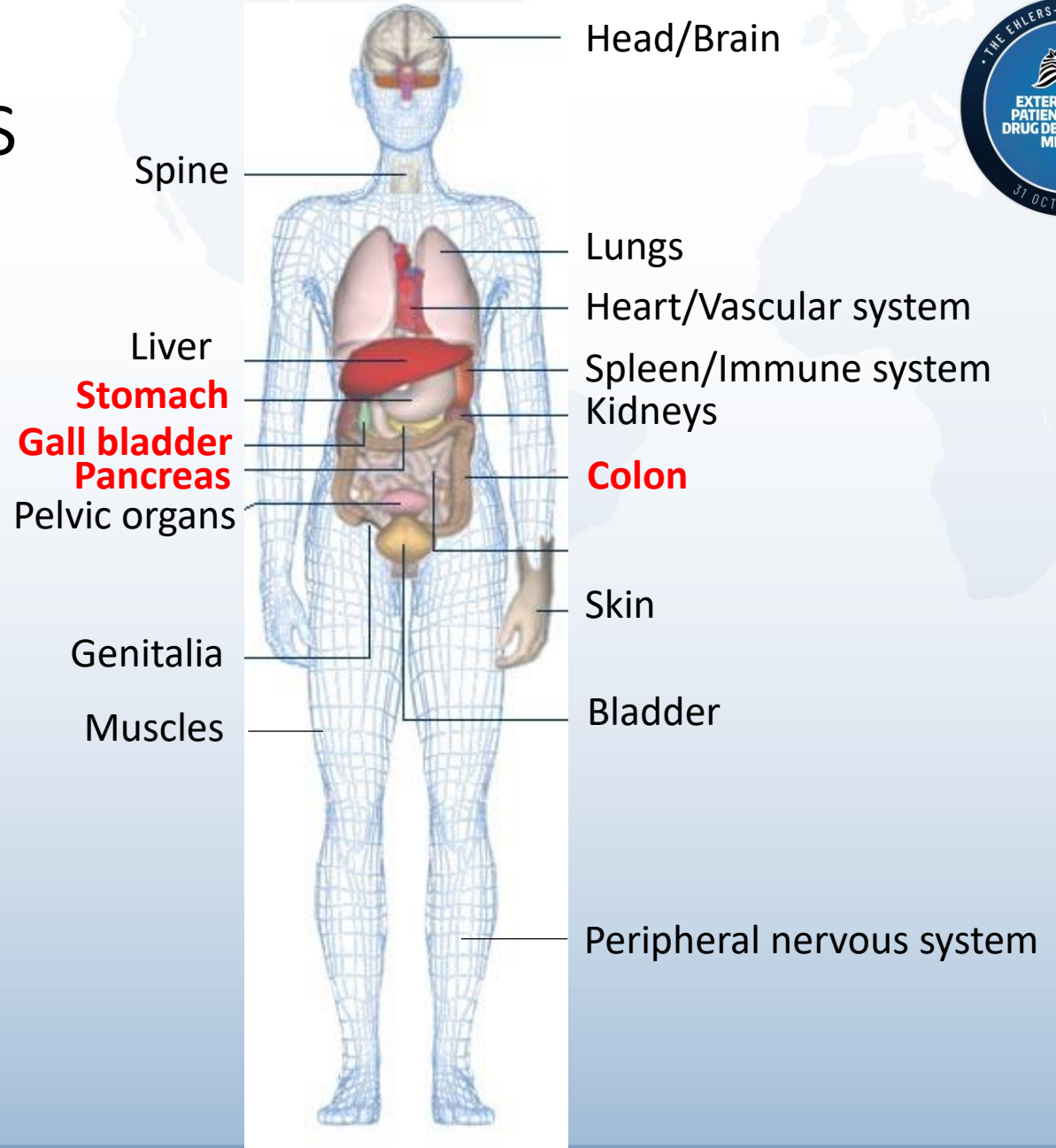
# Neurologic Involvement

- Headaches
- Cerebral venous insufficiency
- Cervico-medullary syndrome
- Degenerative disc disease
- Spinal instability
- Occult tethered cord
- Neuropathic pain
- Small fiber neuropathy
- Brain fog



# Gastrointestinal manifestations

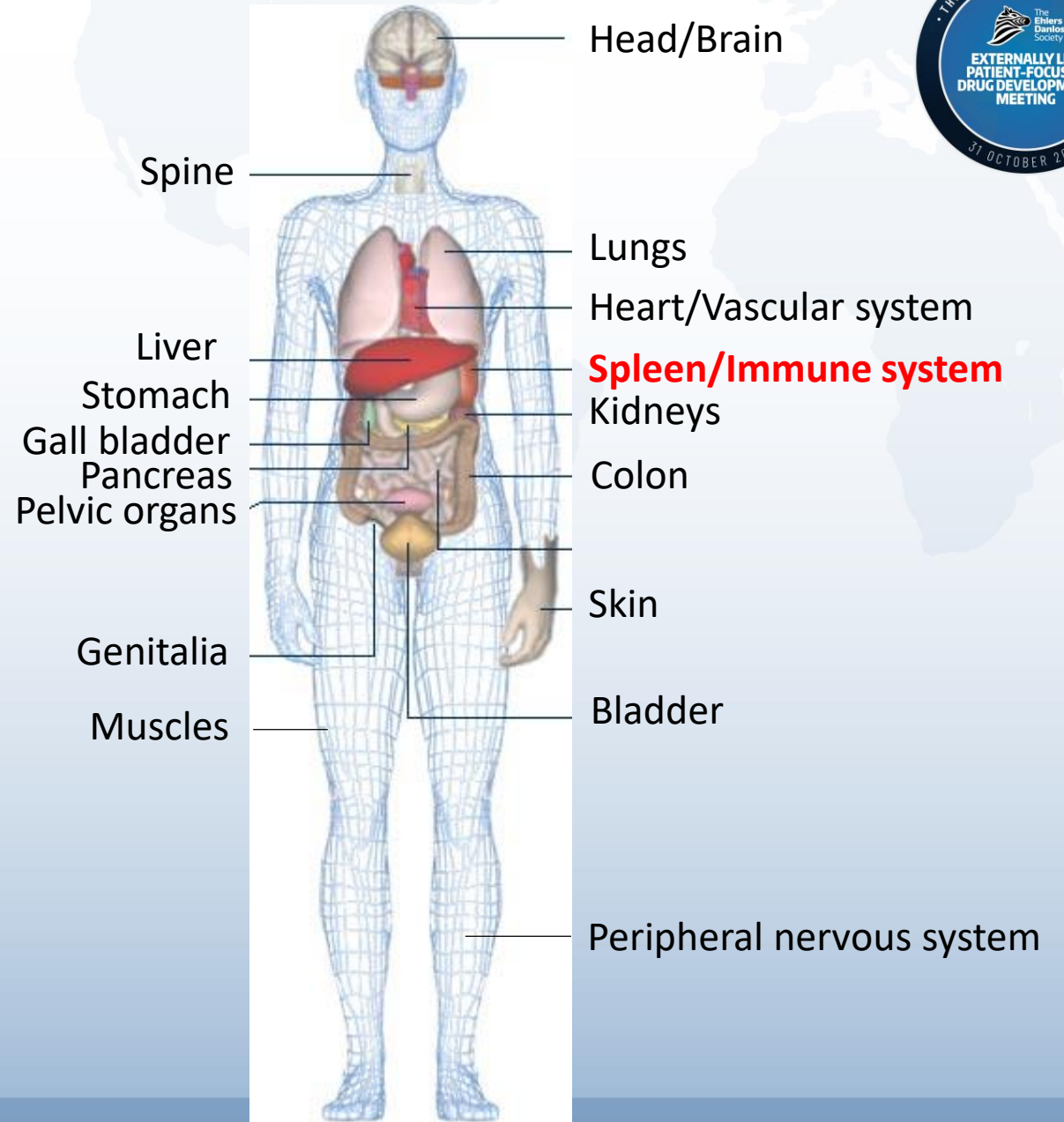
- Food intolerance
- Esophageal reflux
- Abdominal pain
- Bloating
- Irritable Bowel Syndrome
- Diarrhea
- Constipation
- Fecal incontinence
- Spincter of Odi dysfunction
- Hernias





# Hematology/Allergy

- History of hives
- Rashes
- Flushing
- Itching
- Frequent infections
- Severe allergies
- Mast Cell Activation Disorder
- Auto-immune disorders
- Blood clots
- Easy bruising





Our ability to diagnose the Ehlers-Danlos Syndromes and Hypermobility Spectrum Disorders has far out-paced our ability to treat them.

# Why are we here?



- The multiple symptoms of EDS and HSD often stop highly productive individuals in their tracks. Lives are interrupted. Some people never regain their former level of functioning.
- Symptoms are managed individually, often by different specialists
- As many patients have multiple symptoms, polypharmacy is a huge issue
- Research on therapeutics is desperately needed to help people living with EDS and HSD lead energetic, healthy, productive lives without pain.



**Thank you for  
your attention**