Welcome!

Capital Area

Ehlers-Danlos Syndrome Support Group

Ehlers-Danlos Syndrome

AN OVERVIEW

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What is Ehlers-Danlos Syndrome (EDS)?

- EDS is a group of inherited disorders of connective tissue.
- First described by Hippocrates in 400 B.C.
- Additional cases were documented in the late 1800s.

What is Ehlers-Danlos Syndrome (EDS)?

- The Danish Dr. Ehlers, and French Dr. Danlos, both dermatologists, had patients with the syndrome in the early 1900s. Together, they defined its characteristics. The name, Ehlers-Danlos Syndrome, was coined in 1936.
- The symptoms of EDS are caused by defects in the structure, production, or processing of collagen or proteins that interact with collagen. Each subtype of EDS has its characteristic defect.
- Collagen is the most abundant protein in the body, and is found throughout the body.

What is Ehlers-Danlos Syndrome (EDS)?

- Affects men and women of every race and ethnicity.
- Men and women are equally as likely to have EDS.
- However, men may experience less of a problem with hypermobile joints as they get older (possibly because men have more muscle mass, which may develop stronger connective tissue).

Prevalence

- More than 1.5 million with EDS world-wide
- All types combined: 1 in 5,000-10,000
- Most common: Hypermobilty type, 1 in 5,000-20,000; but may be under-diagnosed (only 5% of EDS cases recognized per Prof. Rodney Grahame). Likely *at least* 5-10% of the general population, may be as much as 15%. (Horse rather than zebra?)
- Classical type: 1 in 20,000-40,000.

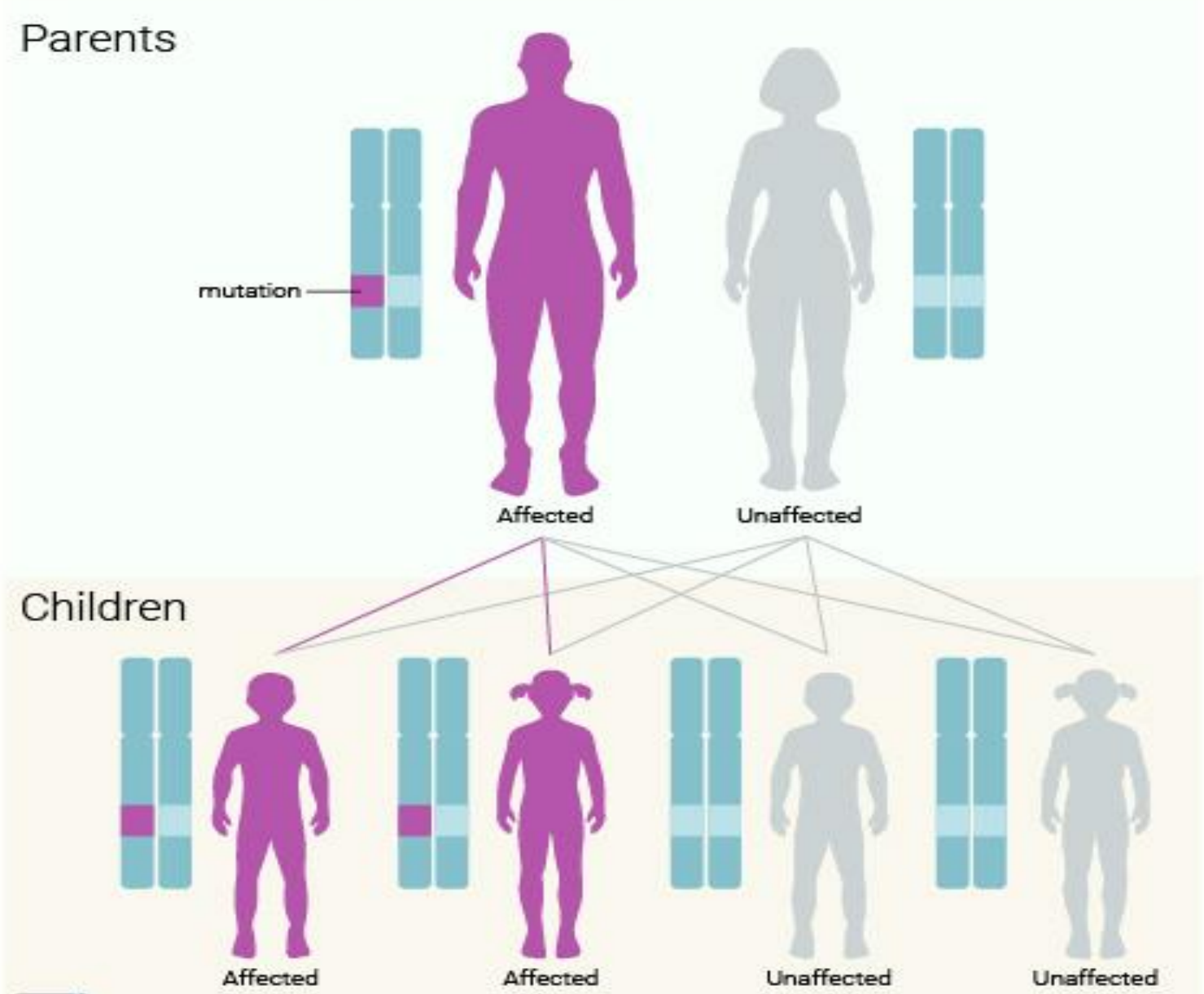
Prevalence

- Other forms of Ehlers-Danlos Syndrome are more rare.
- EDS may frequently be misdiagnosed as fibromyalgia, chronic fatigue syndrome, or as psychosomatic or hypochondriac (“It’s all in your head!”).
- Patients may go years, even decades, (or a lifetime) without a diagnosis.

Inheritance patterns

- Some types of EDS are transmitted in an *autosomal dominant* manner. Only one copy of the gene mutation(s) is needed for an individual to have the disorder. If one parent has EDS, each child has a 50% chance of inheriting the disorder.
- Likely more than a single gene mutation is involved in causing EDS.

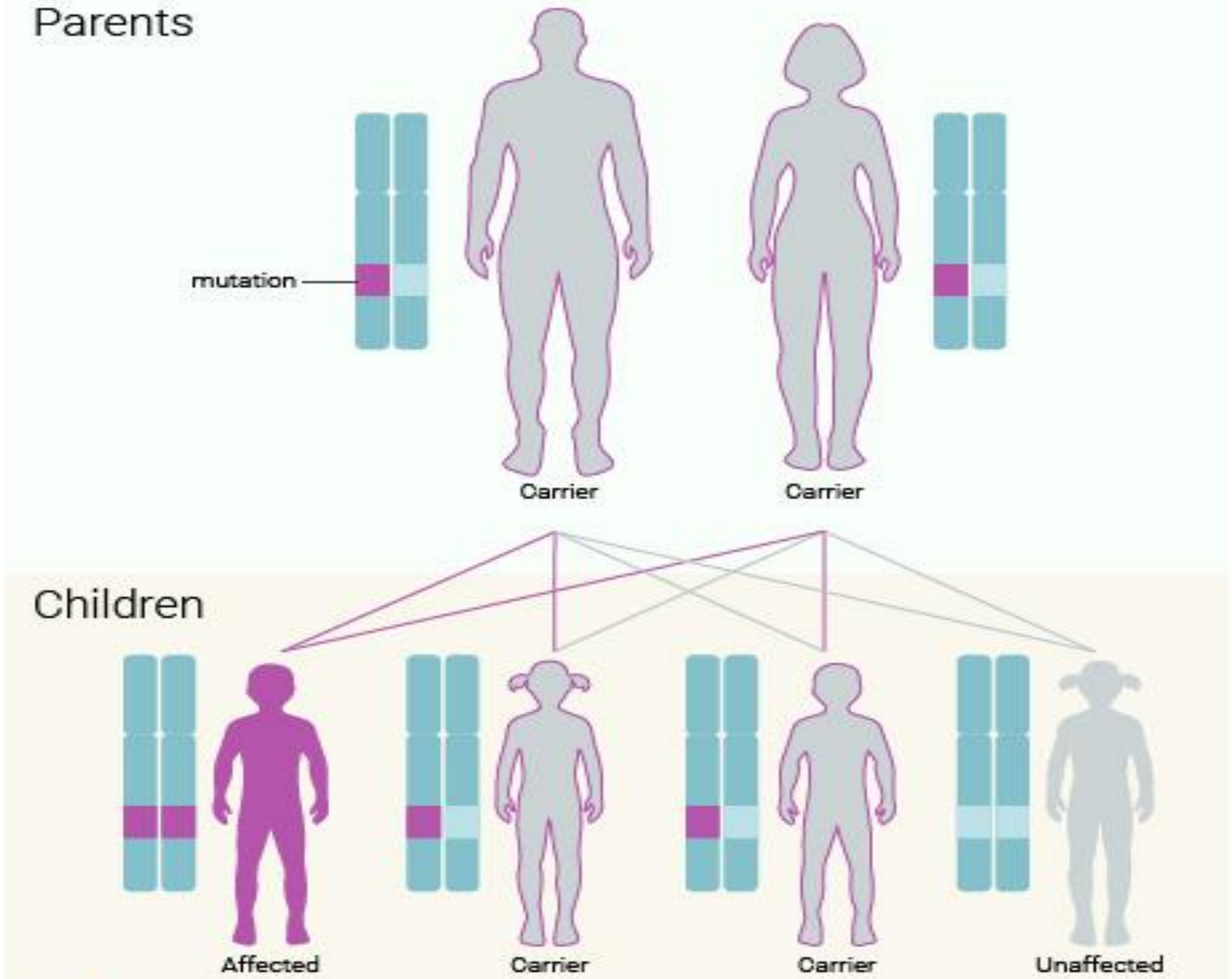
Autosomal Dominant



Inheritance patterns

- Some types of EDS are transmitted in an *autosomal recessive* manner. Both parents must have the EDS gene mutation(s) and pass the gene to their child.
- The parent may just “carry” the trait and not have EDS.
- Each child has a 25% chance of inheriting the disorder by receiving an EDS gene from both parents.
- There is a 50% chance of a child becoming a carrier of the trait without symptoms by receiving an EDS gene from just one parent.

Autosomal Recessive



Inheritance patterns

- EDS may also occur as a new mutation in an individual with no family history of connective tissue disease ("*de novo*").

What Is Connective Tissue?

- Connective tissues are found between other tissues throughout the body. Collagen is the main protein in connective tissue.
- Connective tissues are present around the brain and spinal cord (meninges), in skin, bone, cartilage, tendons, ligaments, fatty tissues, blood vessels, eyes, blood, lymph, and in the walls of hollow organs (such as stomach, esophagus, intestines, urinary bladder).

What Does Connective Tissue Do?

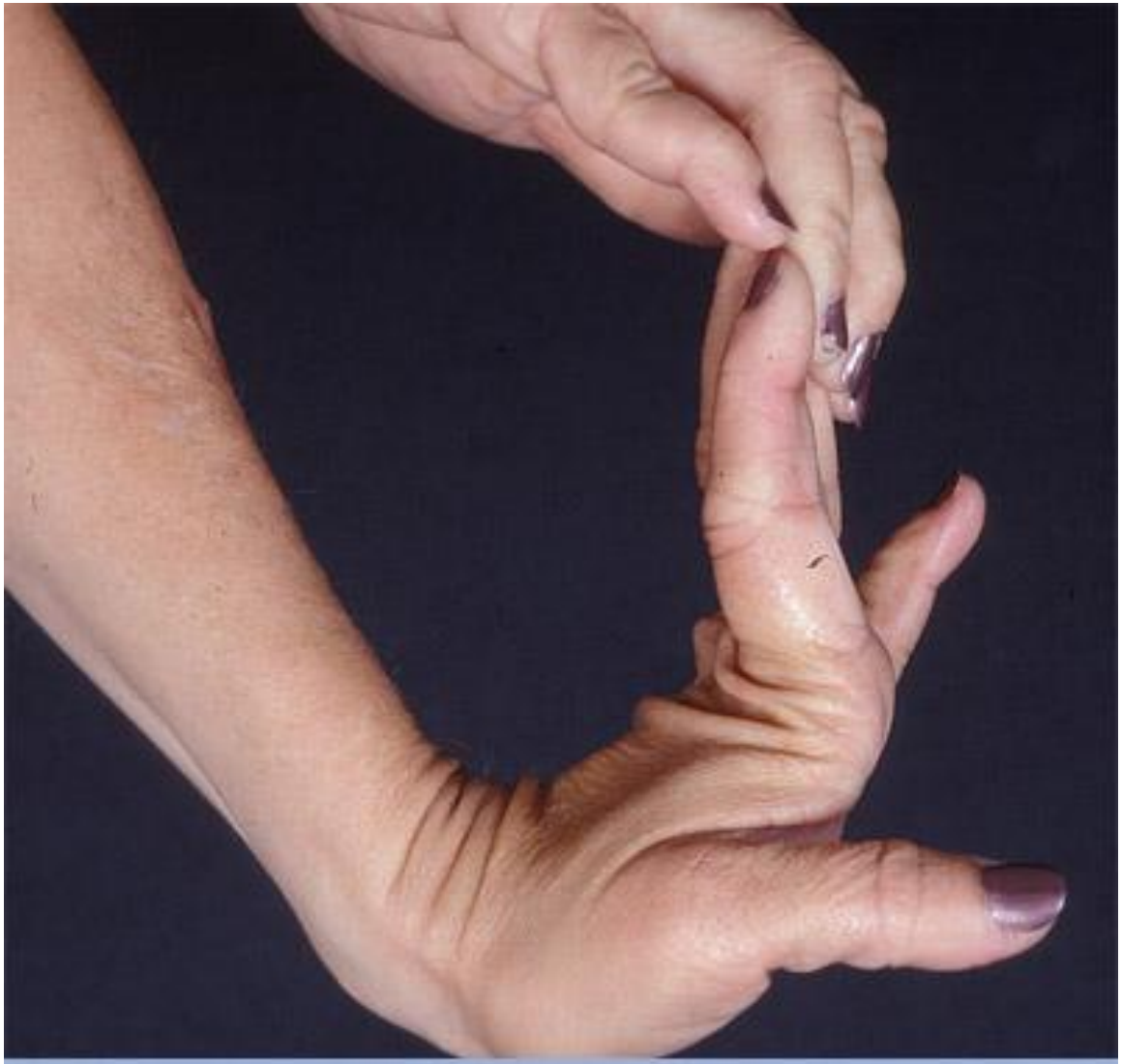
- Gives strength to structures like joints, organs, muscle attachments (e.g., tendons, ligaments).
- Provides flexibility and elasticity to blood vessels, skin and other structures.
- Enables functions such as transport of oxygen and nutrients from capillaries to cells.
- Is the matrix for immune system function.
- Makes up specialized tissue such as cornea.
- The wide-spread location of connective tissue explains the many diverse symptoms of EDS.

Abnormal Connective Tissue

- Individuals with Ehlers-Danlos syndrome (EDS) have abnormal connective tissue because of genetic defects in its strength, elasticity, integrity, or healing properties.
- Each type of EDS has a genetically different abnormality of the connective tissue proteins.

Symptoms of EDS - Joints

- Joint laxity/hypermobility; “double-jointed”
- Spontaneous (without injury) dislocations or subluxations (partial dislocations) of joints
- Frequent sprains
- Chronic joint pain
- Early degenerative arthritis
- Temporomandibular joint (TMJ) dysfunction
- Spine problems, neck and back pain, instability



Beighton score

Give yourself 1 point for each of the manoeuvres you can do, up to a maximum of 9 points

Ehlers-Danlos Support UK

Registered Charity 1157027

Can you bend your thumb back onto the front of your forearm?

left thumb 1 point

right thumb 1 point

Can you bend your knee backwards?

left knee 1 point

right knee 1 point

Can you put your hands flat on the floor with your knees straight?

1 point

Can you bend your elbow backwards?

right arm 1 point

left arm 1 point

Can you bend your little finger up at 90º (right angles) to the back of your hand?

left hand 1 point

right hand 1 point

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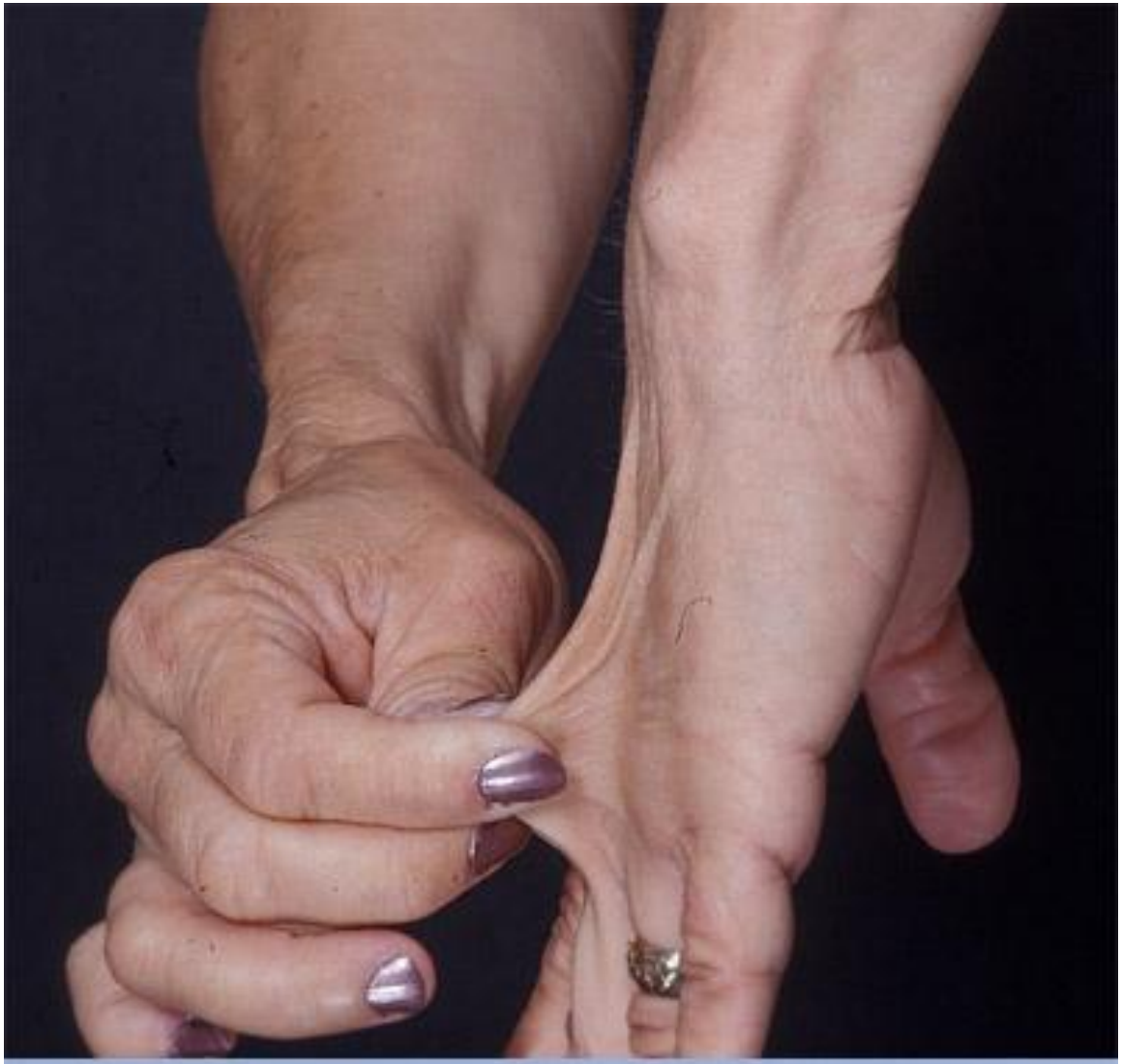
www.ehlers-danlos.org
T: 020 8736 5604

Symptoms of EDS – Skin/Vessels

- Soft, velvety, loose, stretchy, thin, fragile skin
- Easy bruising/fragility of blood vessels
- Poor wound healing
- Tissue fragility
- Prolonged bleeding time
- Parchment-like, thin scars in some types
- Periodontal (gum) disease



Symptoms of EDS – Hernias, Genitourinary

- Organs in abnormal places , e.g., stomach or bowel in pelvis when person is upright, organs not attached properly to intra-abdominal wall
- Hernias, including hiatal hernia
- Rectal prolapse
- Rectocele, cystocele and uterine prolapse in women
- Possible complications of pregnancy
- Bladder problems

Symptoms of EDS - Gastrointestinal

- Slow GI motility, distension of bowel
- Gastroparesis (slow stomach emptying)
- Gastric reflux (GERD), gastritis
- Chronic constipation
- Possible intermittent bowel obstruction from twisting of bowel loops
- Small Intestine Bacterial Overgrowth (SIBO)

Symptoms of EDS – Cardiovascular

- Prone to abnormalities of arteries, such as aneurysms, or unusually “twisty” (tortuous) arteries
- Dilated aortic root (where aorta, largest artery, leaves the heart)
- Mitral valve prolapse (valve between left heart chambers)
- Heart arrhythmias (skipped beats, fast heart rate)

Symptoms of EDS – Neurovascular

- Postural Orthostatic Tachycardia Syndrome (POTS)
- Neurally Mediated Hypotension (NMH) – low blood pressure with prolonged standing
- Small fiber neuropathy
- Raynaud's Syndrome
- Poor balance and proprioception (sense of where body parts are in relation to self and environment)

Symptoms of EDS – Immune System

- Mast Cell Activation Syndrome (MCAS) – inappropriate activation in the mast cells of the immune system, causing reactions like rash/hives, swelling, nausea/vomiting, and diarrhea
- Allergies and sensitivities
- Gut-related immune dysfunction?

The Ehlers-Danlos syndromes

NAME OF EDS SUBTYPE	INHERITANCE	GENETIC BASIS
Classical EDS (cEDS)	AR	*COL5A1, COL5A2* (rarely *COL1A1*)
Classical-like EDS (clEDS)	AR	*TNXB*
Cardiac-valvular EDS (cvEDS)	AR	*COL1A2*
Vascular EDS (vEDS)	AD	*COL3A1* (rarely *COL1A1*)
Hypermobile EDS (hEDS)	AD	Unknown
Arthrochalasia EDS (aEDS)	AD	*COL1A1, COL1A2*
Dermatosparaxis EDS (dEDS)	AR	*ADAMTS2*
Kyphoscolitic EDS (kEDS)	AR	*PLOD1* *FKBP14*

AD = autosomal dominant; AR = autosomal recessive

www.ehlers-danlos.com/eds-types/

The Ehlers-Danlos syndromes

NAME OF EDS SUBTYPE	INHERITANCE	GENETIC BASIS
Brittle cornea syndrome (BCS)	AR	*ZNF469* *PRDM6*
Spondylodysplastic EDS (spEDS)	AR	*B4GALT7* *B3GALT6* *SLC39A13*
Musculocontractual EDS (mcEDS)	AR	*CHST14* *DSE*
Myopathic EDS (mEDS)	AD or AR	*COL12A1*
Periodontal EDS (pEDS)	AD	*C1R*

AD = autosomal dominant; AR = autosomal recessive

www.ehlers-danlos.com/eds-types/

Vascular EDS (vEDS)

Rare disorder known to cause:

- Arterial aneurysms, dissections, and ruptures;
- Rupture of bowels;
- Rupture of uterus;
- Reduced lifespan (median 49 for males and 53 for females, but a very large range from 10 to 80 years).

It is important those with vEDS have a team for care and for emergencies (primary care, geneticist, vascular and general surgeons).

Treatment includes:

- Blood pressure maintenance in normal or low range;
- Surgical interventions.

For more information:

"Diagnosis, natural history, and management in vascular Ehlers-Danlos syndrome"

http://bit.ly/EDS2017papers

Hypermobile EDS (hEDS)

- Believed to be the most common genetic connective tissue disorder.
- Can experience:
  - Joint hypermobility with subluxations and dislocations;
  - Skin issues;
  - Other symptoms.
- Some of the possible associated features include:
  - Chronic pain and fatigue;
  - Dysautonomia;
  - Gastrointestinal issues;
  - TMJ and dental problems
  - Spine problems;
  - Mast cell activation disorder.

For more information:

"Hypermobile Ehlers-Danlos Syndrome"

http://bit.ly/EDS2017papers

How is EDS managed?

- Early diagnosis is critical for person's health and safety.
- Care is largely preventative to prevent physical damage.
- Routine screening for heart and blood vessel disease, especially in Vascular EDS.

How is EDS managed?

- Exercise is *essential* to strengthen muscles to support joints, improve posture and balance, and strengthen core muscles to decrease pain and risk of injury.
- It is important to avoid physical activities which may result in injury, like contact sports, heavy weight-lifting.
- Also, should try to limit activities which result in hyperextension of joints, so activities like yoga and gymnastics should be avoided or done with extreme care. Teaching on joint limits needed.

How is EDS managed?

- Myofascial release (trigger point therapy), massage, manipulation by experienced DO, PT.
- Bracing or orthotics may be helpful.
- Surgery needs careful consideration and should be done by a surgeon with experience in treating those with EDS. Special suturing techniques may be needed, etc.

How is EDS managed?

- Pain management should be tailored for each individual.
- Balance of rest and activity is crucial for management of pain and fatigue.
- Team approach helpful: medical, physical therapy, geneticist, psychologist.
- Each person with EDS has a unique set of symptoms!

Resources

- The Ehlers-Danlos Society (formerly ednf.org) https://www.ehlers-danlos.com/
- EDS Awareness https://www.chronicpainpartners.com/
- Ehlers Danlos International Registry http://www.edsregistry.org/
- Ehlers-Danlos Syndrome Network C.A.R.E.S. Inc. http://www.ehlersdanlosnetwork.org/

Support

- Inspire https://www.inspire.com/
- EDS Today~Advocates https://themighty.com/partner/edstoday/
- Rare Connect https://www.rareconnect.org/en/community/ehlers-danlos-syndrome
- Strength/flexibility/health/EDS http://strengthflexibilityhealtheds.com/

Topics for Future Meetings?

- How is EDS diagnosed?
- Pain Management
- Exercise, Physical Therapy
- Self Care
- Gastrointestinal Issues
- Psychological Issues (depression and anxiety, ADD, Autism Spectrum Disorders)
- What's Your Story?

Thank You!

Time for us to talk!

Any questions?