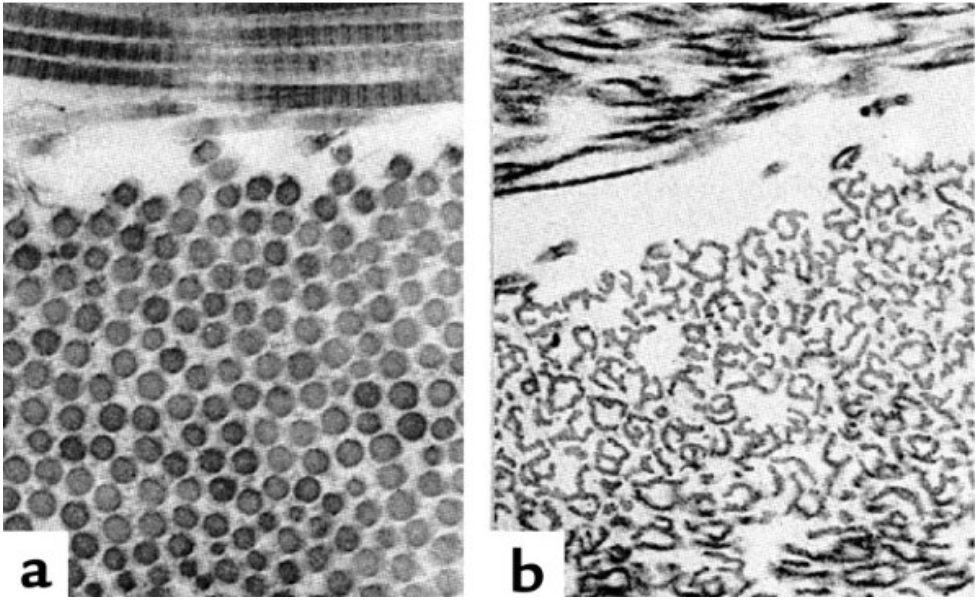
# EHLERS-DANLOS SYNDROME (EDS) & HYPERMOBILITY EDS (hEDS)

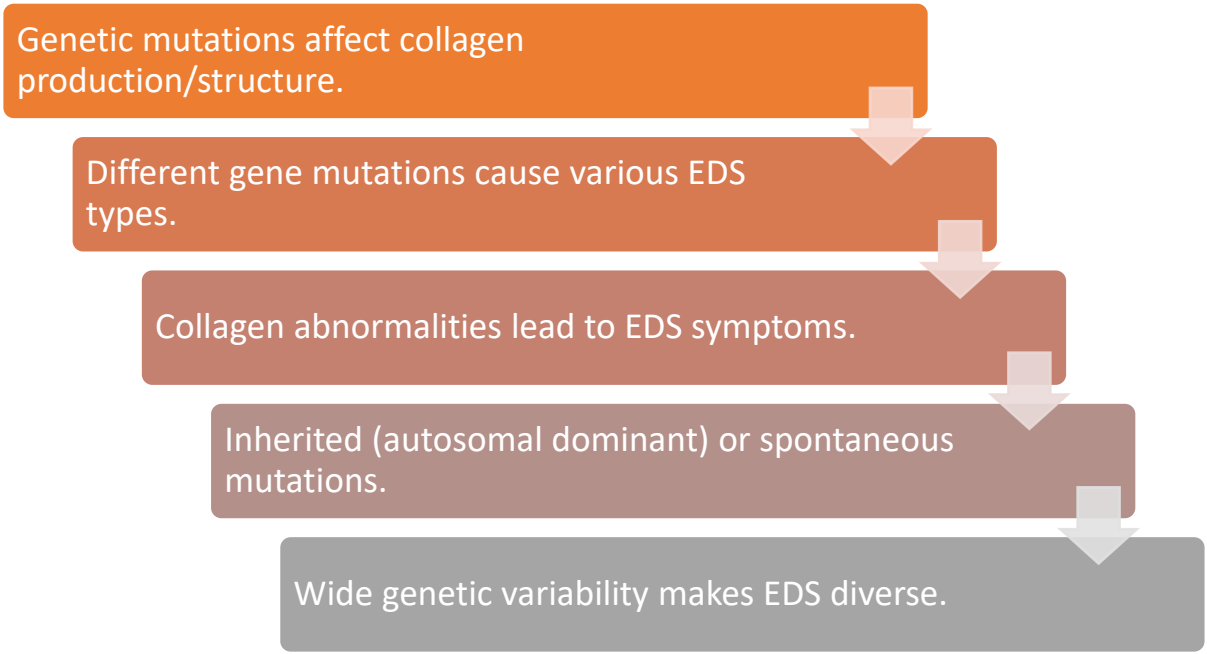
## What is EDS?

It is a group of connective tissue disorders caused by genetic defects in collagen.

*Collagen = the body's "glue." When faulty, tissues stretch too easily or fail to support joints/skin.*




The collagen fibril and EDS. (a) Normal collagen fibrils are of uniform size and spacing. Fibrils from a patient with dermatosparaxis (b) show dramatic alterations in fibril morphology with severe effects on tensile strength of connective tissues.


Genetic mutations affect collagen production/structure.
Different gene mutations cause various EDS types.
Collagen abnormalities lead to EDS symptoms.
Inherited (autosomal dominant) or spontaneous mutations.
Wide genetic variability makes EDS diverse.

# MOST COMMON

**Classical EDS (cEDS):** Stretchy, fragile skin, hypermobility, scarring

**Hypermobile EDS (hEDS):** Most common, joint hypermobility, chronic pain

**Vascular EDS (vEDS):** Fragile blood vessels, high risk of rupture/aneurysms

- **CLASSICAL EDS (cEDS):**
    - Hypermobile joints and stretchy, smooth skin that is prone to bruising.
        - identified genetic mutations
        - Caused by mutations in **genes COL5A1 and COL5A2**, which are involved in the production of **type V** collagen.
- **HYPERMOBILE EDS (hEDS):**
    - Joint hypermobility and frequent joint dislocations or subluxations.
    - Often associated with chronic joint pain and soft, velvety skin.
        - Genetic cause **is not well-defined** for hEDS. → many patients fall through the cracks
- **VASCULAR EDS (vEDS) or EDS Type IV**:
    - The most severe type, characterized by a risk of arterial and organ rupture due to weakened blood vessels.
    - Can cause life-threatening complications.
        - Caused by mutations in **the COL3A1** gene, which affects the production of type **III collagen**.

**Table 3**: Types of HSD

| Type of HSD | Beighton Score [1] | Musculoskeletal Involvement |
|---|---|---|
| **Asymptomatic GJH** | Positive | Absent |
| **Asymptomatic PJH** | Usually Negative | Absent |
| **Asymptomatic LJH** | Negative | Absent |
| **Generalised HSD** | Positive | Present |
| **Peripheral HSD** | Usually Negative | Present |
| **Localised HSD** | Negative | Present |
| **Historical HSD** | Negative [2] | Present |
| **hEDS** | Positive | Possible |

- LJH localized joint hypermobility 1 to 4 joints involved
- GJH generalized joint hypermobility 5 or more joints involved
- PJH Peripheral joint hypermobility – affects Hands and feet only
- HJH Historical joint Hypermobility – found in older adults who have progressively lost joint hypermobility

# COMMON PRESENTATION

# Early Symptoms

Simple tasks (e.g., typing, writing) can be painful or tiring with hands cramping

In sports with recurrent dislocations of the shoulder or ankles, knee cap.

# Late Symptoms

Chronic pain

Fatigue

Skin allergies / sensitivities

May present with GI issues

Migraines –due to OA in the neck if not fused

Multiple subluxations/ dislocations
Multiple surgeries –different joints
Surgeries on the same joint multiple times due to failed procedures
Arthritis in multiple joints
Multiple trigger fingers
Multiple nerve releases bilaterally

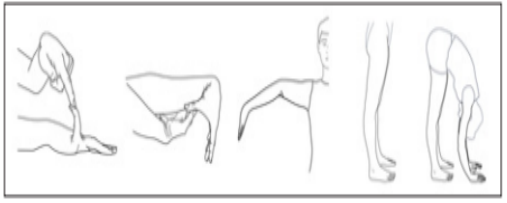
# DIAGNOSIS (EARLY)

The Beighton Score, a 9-point scale assessing joint hypermobility

One of the following selected:

- ☐ ≥6 pre-pubertal children and adolescents
- ☐ ≥5 pubertal men and woman to age 50
- ☐ ≥4 men and women over the age of 50

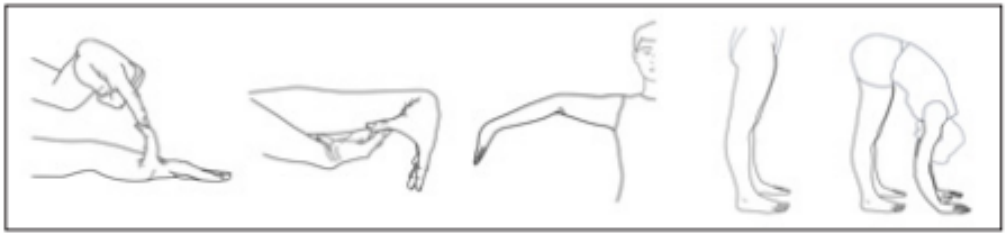
Beighton Score: ____/9



*Take obesity into account if they can't touch the floor*

# DIAGNOSIS (LATE)

The Beighton Score, a 9-point scale assessing joint hypermobility



*Take arthritis, multiple surgeries, obesity into account*

*If Beighton Score is one point below age- and sex-specific cut off, two or more of the following must also be selected to meet criterion:*

- ☐ 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"?

# DIAGNOSIS IS DERIVED BASED ON

*-History*

*-Symptoms*

*-Clinical judgment*

*-And Genetic testing*

# How does EDS affect you?

*<u>-Ligament Support</u>*

*EDS being a connective tissue disorder the ligaments are insufficient*

*They do not hold the joint in place*

*So, when ligaments fail to hold the joint in place the muscles need kick in for patients to stabilize the joints*

*However, the patients rely on the joints to stabilize themselves, thus not engaging the muscles to help support them to maintain normal mechanics*

*Even the muscles have the same connective tissue and thus cannot stabilize properly*

*This may result in subluxations and even dislocations of the joints with minimal strain.*

# How does EDS affect you?

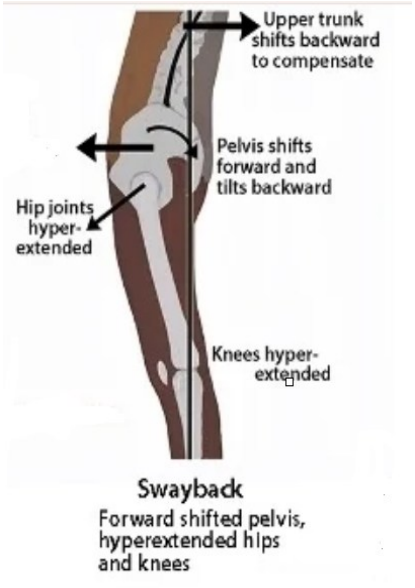
*-Weak stabilizing muscles*

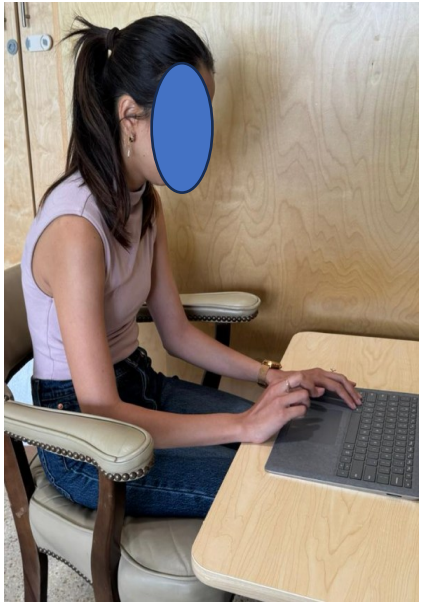
*Since the EDS patient locks their joint to stabilize themselves to support an upright posture*

*They fail to engage the muscles*

*This results in lack of deep tone in the muscles*

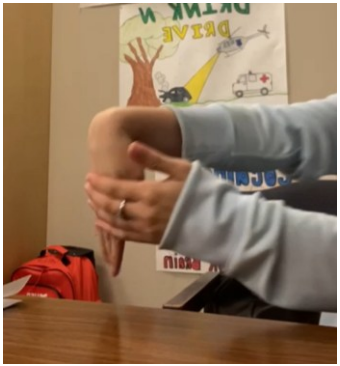
# EDS POSTURE


Upper trunk shifts backward to compensate
Pelvis shifts forward and tilts backward
Hip joints hyper-extended
Knees hyper-extended
Swayback
Forward shifted pelvis, hyperextended hips and knees



# How does EDS affect you?

*-Compensatory tightness*



*Since muscles are not engaged, they tend to tighten*

*So, to find relief from the tight muscles these patients constantly keep stretching their joints*

*Unfortunately, the joints give before a stretch can be felt*

*Thus, engaging in the vicious cycle by causing the already stretched ligaments to overstretch*

# How does EDS affect you?

*-Neurological Involvement*

*The joint gives before the patient could experience relief from flexibility*

*The muscular tunnels tend to tighten and synch down on the nerves*

*This may cause entrapment syndromes in the muscular tunnels*

*This nerve involvement in muscular tunnels results in traffic jam effect causing multiple hierarchical nerve entrapment sites*

*These multiple nerve entrapment sites result in tenderness at these muscular tunnels along the path of the nerve*

# How does EDS affect you?

*Overstretched nerves – Eg:* ***Snapping Ulnar nerve***

*Overstretched ligaments allow excessive motion in the joints*

*The osseous tunnels (ligaments supporting them are over stretched)*

*Thus, allowing increase movement*

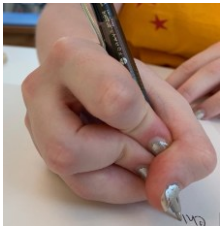
*Nerve snaps in and out resulting in sensory symptoms/ pain*

*Adding to that poor posture due to lack of deep tone adds to the already burdened nervous system*

*Resulting in pain and tenderness all over the body*

*Thus, the fibromyalgia diagnosis*

# How does EDS affect you?



EDS Normal

*-Poor Proprioception*

*Poor proprioception (joint sense)*

*These patients cannot experience end feel, this is lack of Joint proprioception*

*Since the patients cannot tell where the end range is as they cannot feel the end point of the stretch*

*They tend to lock their joints or put excessive pressure to stabilize the object*

*Eg: holding a pen*

*This overpressure and locking their joints results in the development of early arthritis*

# How does EDS affect you

*Mast cell activation syndrome (MCAS) is a condition characterized by the abnormal activation of mast cells, which are immune cells that release various chemicals, including histamine, tryptase, and leukotrienes.* ***Eg: Tape allergy***

*They are diagnosed with functional gastrointestinal disorders (FGIDs), which are conditions characterized by persistent and recurring GI symptoms without an identifiable physical cause.* ***Eg: Cannot handle a lot of foods***

*Just like RA patients these patients tire easily, since the entire body is affected by this connective tissue disorder.* ***Eg: Excessive fatigue***

# 3 PHASES OF EDS LIFE

**Hypermobile Phase**

*1st several years of life*

**Pain phase**

*2nd -4th decade*

**Stiffness phase**

*Adults and older adults*

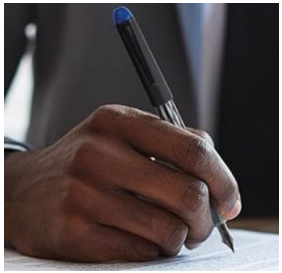
# MULTIDISCIPLINARY APPROACH

*Personalized therapy changes the course of disease*

*Symptoms managed through a multidisciplinary approach.*

- *Therapy*
- *Pain management,*
- *and lifestyle adjustments.*

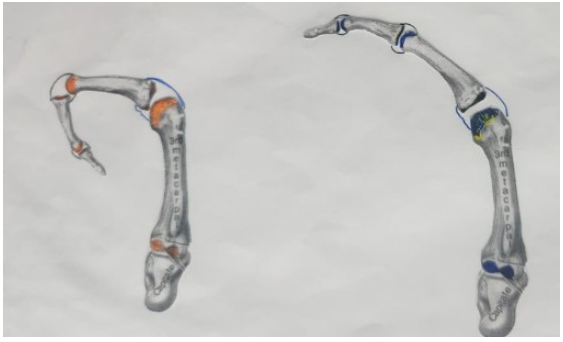
# GADGETS AND FUNCTIONAL BRACES



# Living with EDS – Exercises and Pain Management

- *Exercising in mid-range*
- *Avoiding end-range movements*
- *Proprioception exercises*
- *Myofascial Manipulation with nerve glides for pain management and sensory complaints*

# Arthritis in EDS



*Since the ligaments are insufficient*

*They do not hold the joint in place*

*They allow for excessive ROM in the joint (acute and obtuse angles)*

*This impinges on the cartilage*

*If mild subluxation is present, add loading with functional activities*

*Overtime the cartilage erodes*

*Results in Arthritis of the joints*

# Living with EDS – late stages

*Heat in the morning or hot showers to warm up the body to allow for movement*

*Followed by gentle passive ROM*

*Biking/ swimming*

*ROM exercises*

*Ice at the end of the day especially if overdone to calm the inflammation down*

*Certain Yoga poses may not be ideal for EDS due to excessive joint stretching risks in the early stages*

*However, when arthritis settles in Yoga may help maintain ROM as long as its pain-free*

# SURGERIES FOR THE EDS POPULATION – LATE STAGES

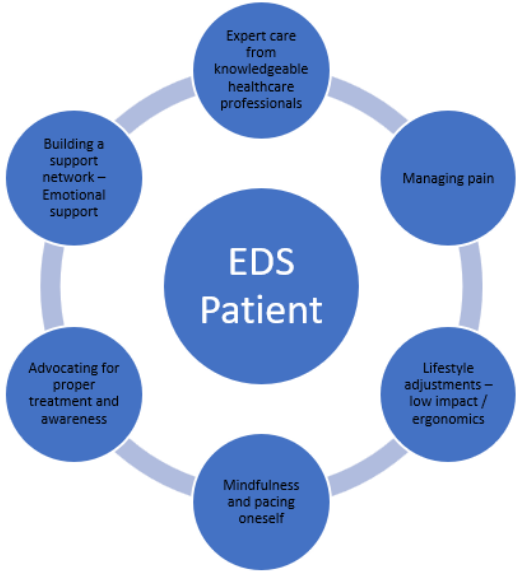
- For severe arthritis/dislocations
- **Neck:** Mobile fusion- Mobi-C device
- **Shoulder recurrent dislocation:** Latarjet procedure
- If OA has set in and RTC is redundant then reverse shoulder
- **Wrist:** PRC or fusion
- **Thumb CMC:** Tightrope or FiberLock™ instead of LRTI
- **MCP / PIP joints:** Silicone or hemi hamate arthroplasty
- Multiple trigger fingers

# EDS CONTRAINDICATIONS

*Cortisone shots –*

*Locking their joints / Yoga*

*Stretching vs Myofascial Manipulation to relieve painful symptoms*



# EDS SUPPORT GROUPS

The Ehlers-Danlos Society

EHLERS-DANLOS SUPPORT GROUP
INTERNATIONAL ZEBRAS TRYING TO DAZZLE TOGETHER

*Facebook group*

**Watch Billie Eilish documentary**

EDS.clinic

*Virtual support group*