2017 INTERNATIONAL EDS CLASSIFICATION

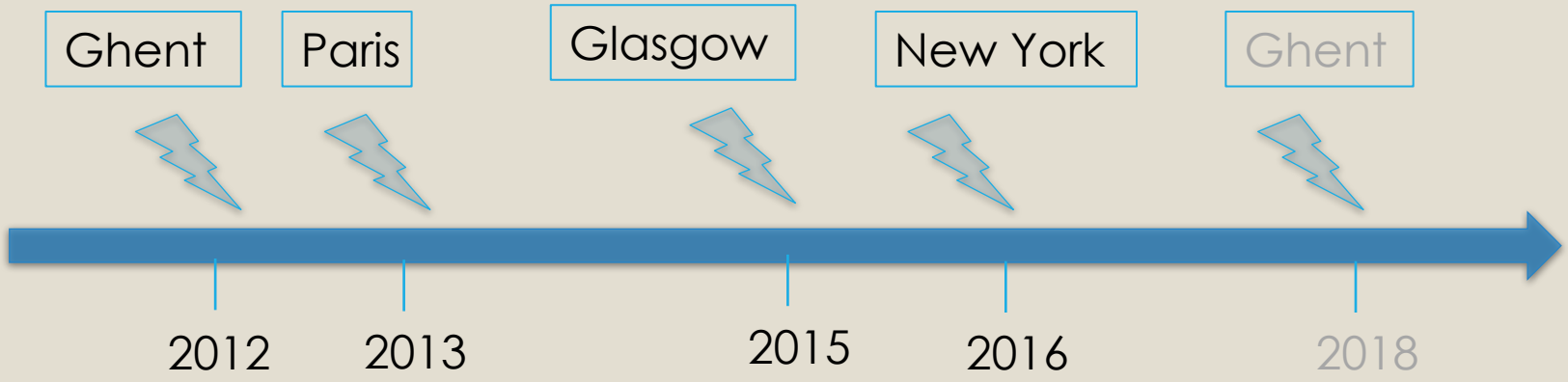
Clair A. Francomano, MD
on behalf of the
The International Consortium on EDS and Related Disorders

Classification of the Ehlers-Danlos syndrome based on the Villefranche nosology

New	Gene	Protein	Transmission
Classical	*COL5A1* *COL5A2*	Type V procollagen	AD
Hypermobility	*?*	?	AD
Vascular	*COL3A1*	Type III procollagen	AD
Kyphoscoliosis	*PLOD1*	Lysyl hydroxylase	AR
Arthrochalasis	*COL1A1* *COL1A2*	Type I collagen (N-propeptide-processing)	AD
Dermatosparaxis	*ADAMTS2*	Procollagen N proteinase	AR

Beighton et al, AJMG, 1998

Time for a revision of the EDS classification!



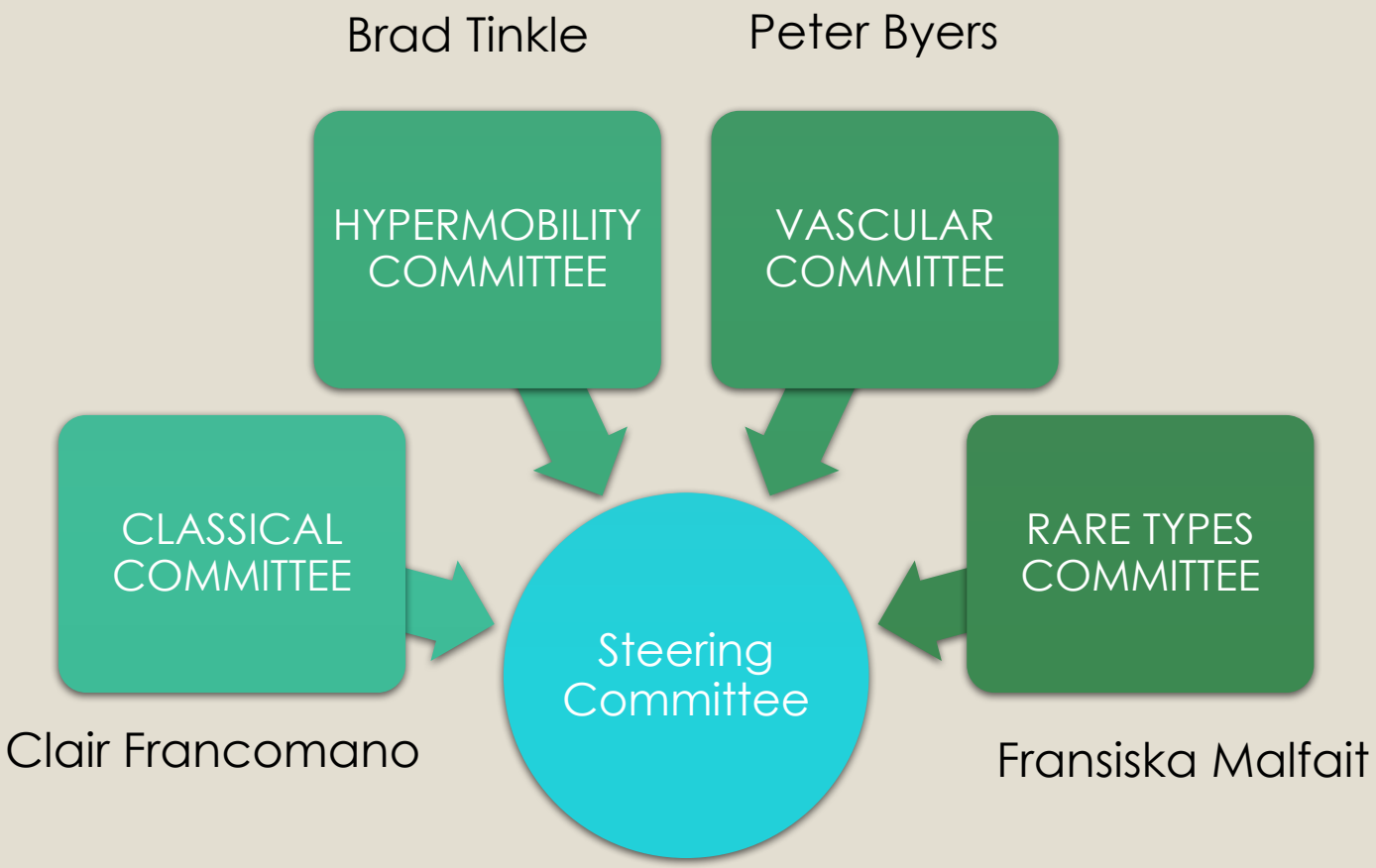
EDS UK and EDNF engaged to fund this enterprise

EDS Symposium May, 2016
New York

Goals

- Build a revised nosology that defines diagnostic criteria
- Define new EDS types where necessary
- Begin the process by which management and care guidelines are being developed for each EDS subtype

Committees


Brad Tinkle
Peter Byers
HYPERMOBILITY COMMITTEE
VASCULAR COMMITTEE
CLASSICAL COMMITTEE
RARE TYPES COMMITTEE
Steering Committee
Clair Francomano
Fransiska Malfait

Working Groups

Beighton score: Birgit Juul-Kristensen

Physiotherapy: Jane Simmonds

Orthopaedic surgery: William Ericson

Autonomic Dysfunction and fatigue: Alan Hakim

Pain : Pradeep Chopra

Psychological aspects: Antonio Bulbena

Gastro-intestinal involvement: Qasim Aziz

Neurology: Fraser Henderson

Allergy/Immunology in EDS: Surinjith Seneviratne

Oral manifestations: John Mitikades

Classification of EDS Types: 2017

Classical type	AD
Classical-like EDS (clEDS)	AR
Cardiac-valvular EDS (cvEDS)	AR
Vascular EDS (vEDS)	AD
Hypermobile EDS (hEDS)	AD
Arthrochalasia EDS (aEDS)	AD
Dermatosparaxis EDS (dEDS)	AR

Kyphoscoliotic EDS (kEDS)	AR
Brittle cornea synsrome (BCS)	AR
Spondylodysplastic EDS (spEDS)	AR
Musculocontractural EDS (mcEDS)	AR
Myopathic EDS (mEDS)	AD or AR
Periodontal EDS (pEDS)	AD

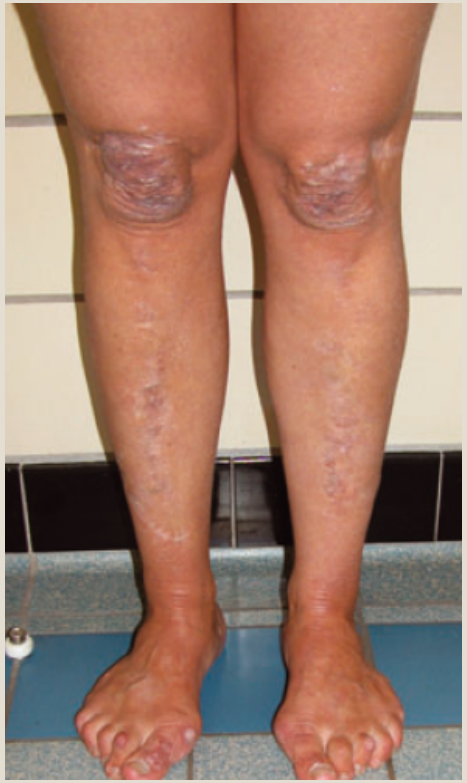
The Spectrum of Joint Hypermobility

Type	Beighton score	Musculoskeletal involvement	Notes
Asymptomatic GJH	Positive	Absent	
Asymptomatic PJH	Usually negative	Absent	JH typically limited to hands and/or feet
Asymptomatic LJH	Negative	Absent	JH limited to single joints or body parts
G-HSD	Positive	Present	
P-HSD	Usually negative	Present	JH typically limited to hands and/or feet
L-HSD	Negative	Present	JH limited to single joints or body parts
H-HSD	Negative	Present	Historical presence of JH
hEDS	Positive	Possible	

Classical EDS (cEDS): 2017 Criteria

Major criteria

1. Skin hyperextensibility and atrophic scarring
2. Joint hypermobility



DePaepe and Malfait, 2012

http://www.epharmapedia.com/diseases/profile/167/Ehlers-Danlos-syndrome.html?lang=en

Beighton score

Ehlers-Danlos Support UK

Registered Charity 1157027

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

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

left thumb 1 point

right thumb 1 point

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

1 point

left hand 1 point

right hand 1 point

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

Can you bend your knee backwards?

left knee 1 point

right knee 1 point

Can you bend your elbow backwards?

right arm 1 point

left arm 1 point

MAKING OUR INVISIBLE **VISIBLE**

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

Classical EDS: Minor Diagnostic Criteria

- Easy bruising
- Soft, doughy skin
- Skin fragility (or traumatic splitting)
- Molluscoid pseudotumours
- Subcutaneous spheroids
- Hernia (or history thereof)
- Epicanthal folds
- Complications of joint hypermobility (e.g. sprains, dislocation/subluxation, pain, flexible flatfoot)
- Family history of a first degree relative who meets clinical criteria

Clinical Diagnosis of Classical EDS: 2017 Criteria

Major Criterion (1): Skin hyperextensibility and atrophic scarring

Plus

- Either: Major criteria (2) – joint hypermobility
- Or: three of the eight minor criteria

cEDS: Verification of Clinical Diagnosis

- Confirmatory analysis is recommended for any patient meeting the recommended clinical criteria
- Molecular analysis of *COL5A1* and *COL5A2* genes identifies a causal mutation in more than 90% of the patients and should be used as the standard confirmatory test

Vascular EDS (vEDS): 2017 Criteria

Major criteria

- Family history of vEDS with documented causative variant in *COL3A1*
- Arterial rupture at a young age
- Spontaneous sigmoid colon perforation in the absence of known diverticular disease or other bowel pathology
- Uterine rupture during the third trimester in the absence of previous C-section and/or severe peripartum perineum tears
- Carotid-cavernous sinus fistula (CCSF) formation in the absence of trauma

Vascular EDS: Minor Criteria

- Bruising unrelated to identified trauma and/or in unusual sites such as cheeks and back.
- Thin, translucent skin with increased venous visibility
- Characteristic facial appearance
- Spontaneous pneumothorax
- Acrogeria
- Talipes equinovarus
- Congenital hip dislocation
- Hypermobility of small joints
- Tendon and muscle rupture
- Keratoconus
- Gingival recession and gingival fragility
- Early-onset varicose veins (under age 30 and nulliparous if female)

Minimal criteria suggestive for vEDS

- Family history of the disorder
- Arterial rupture or dissection in individuals <40 years of age
- Unexplained sigmoid colon rupture
- Spontaneous pneumothorax
- in the presence of other features consistent with vEDS, any of these criteria should lead to diagnostic studies to determine if the individual has vEDS.
- Testing for vEDS should also be considered in the presence of a combination of the other ‘minor’ clinical features listed above

vEDS: Diagnostic Confirmation

The diagnosis of vEDS rests on the identification of a causative variant in one allele of *COL3A1*

Hypermobile EDS (hEDS)

- New criteria designed to emphasize syndromic nature of the condition, reduce clinical heterogeneity and facilitate research into underlying cause(s)
- It is expected that further clinical experience and research will lead to revision of these criteria with time

1997 Criteria for Hypermobility Type EDS

Major Criteria:

- Skin involvement (hyperextensibility and/or smooth velvety skin)
- Generalized joint hypermobility

Minor Criteria:

- Recurring joint dislocations
- Chronic joint/limb pain
- Positive family history

The presence of one or both of the major criteria is necessary for clinical diagnosis

Beighton et al., 1998

Hypermobile EDS: 2017 Diagnostic Criteria

Clinical diagnosis of hEDS requires the presence of

Criteria 1, 2, AND 3

Hypermobile EDS: Criterion 1

Generalized Joint Hypermobility (GJH)

Beighton Score

Prepubertal children and adolescents	≥ 6
Men and women, post-puberty up to age 50	≥ 5
Men and women older than 50	≥ 4

If the Beighton score is 1 point below the cutoff and the 5PQ is "positive" (at least 2 positive items), a diagnosis of GJH may be made.

Generalized Joint Hypermobility

5-Point Questionnaire

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

Hypermobile EDS: Criterion 2

2 or more of the following features (A and B, B and C, or A and C)

A: Systemic manifestations of a more generalized connective tissue disorder

B: Positive family history

C: Musculoskeletal complications

Feature A: Systemic manifestations of a more generalized connective tissue disorder

At least 5 of the following must be present:

- Unusually soft or velvety skin
- Mild skin hyperextensibility
- Unexplained striae without a history of significant weight change
- Bilateral piezogenic papules of the heel
- Recurrent or multiple abdominal hernia(s) (e.g. umbilical, inguinal, crural)
- Atrophic scarring involving at least two sites and without the formation of truly papyraceous and/or hemosideric scars as seen in classical EDS

(continued on next slide)

Feature A: Systemic manifestations of a more generalized connective tissue disorder (cont.)

- Pelvic floor, rectal, and/or uterine prolapse in children, men or nulliparous women without a history of morbid obesity or other known predisposing medical condition
- Dental crowding and high or narrow palate
- Arachnodactyly, as defined in one or more of the following: (i) positive wrist sign (Steinberg sign) on both sides; (ii) positive thumb sign (Walker sign) on both sides
- Arm span-to-height ratio ≥1.05
- Mitral valve prolapse (MVP) mild or greater based on strict echocardiographic criteria
- Aortic root dilatation with Z-score >+2

Feature B: Positive Family History

- One or more first degrees relatives independently meeting the diagnostic criteria for hEDS

Feature C: Musculoskeletal Complications

One of the following:

- Musculoskeletal pain in two or more limbs, recurring daily for at least 3 months
- Chronic, widespread pain for ≥3 months
- Recurrent joint dislocations or frank joint instability, in the absence of trauma

  a. Three or more atraumatic dislocations in the same joint or two or more atraumatic dislocations in two different joints occurring at different times

  b. Medical confirmation of joint instability at 2 or more sites, unrelated to trauma

Criterion 3: All prerequisites required

- Absence of unusual skin fragility, which should prompt consideration of other types of EDS
- Exclusion of other heritable and acquired connective tissue disorders, including autoimmune rheumatologic conditions.
  - In patients with an acquired/autoimmune connective tissue disorder additional diagnosis of hEDS requires meeting both Features A and B of Criterion 2. Feature C of Criterion 2 (chronic pain and/or instability) cannot be counted towards a diagnosis of hEDS
- Exclusion of alternative diagnoses that may also include joint hypermobility by means of hypotonia and/or connective tissue laxity.

Syndrome: Definition

- A pattern of anomalies, at least one of which is morphologic, known or thought to be causally related (Hannekam et al, 2013)
- The presence of joint hypermobility in combination with secondary musculoskeletal anomalies does not suffice for delineation of a genetic syndrome
- Joint hypermobility may occur independently or in the context of multiple genetic disorders

Classifying joint hypermobility

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

Hypermobility Spectrum Disorders

APPROACHES TO INTEGRATIVE HEALTH FOR PERSONS WITH EHLERS-DANLOS SYNDROME

With thanks to Kendra Neilsen-Myles

Common Symptoms

- Musculoskeletal pain
- Fatigue
- Headache/ migraine
- GI issues
- Brain fog/cognitive issues
- Sensitivities (food, chemicals, or other)
- ADD/ADHD
- Anxiety/Depression
- Joint issues
- Pelvic pain
- Bruising

Co-Morbid Conditions

- Mast Cell Disease
- POTS/Dysautonomia
- Fibromyalgia
- Chronic Fatigue Syndrome
- Auto-Immune Conditions
- Functional GI Disorders
- Allergies (seasonal, food, bug & other)
- Endocrine Disorders
- Pelvic Dysfunction
- Arthritis

YOU ARE NOT ALONE

Many people live an active and full life with Ehlers-Danlos Syndrome.

This brochure is meant to provide some guidance in managing the symptoms of EDS. Many patients manage their health by traversing the winding path of care themselves. Our hope is to provide some actionable steps for living well with EDS.

KEY POINTS TO REMEMBER

- Work to improve one area of health at a time. (e.g., nutrition or sleep)
- **TAKE ONE STEP AT A TIME. *BABY STEPS.***
- ***BE PATIENT.***
- Focus on what you can do each day, not what you can't.
- Little things count! (e.g, moving more throughout the day, even if it means taking 10 more steps than yesterday.)
- **Something is more than nothing.**
- Work with your doctor for a fully integrative plan. Targeted therapies should improve overall success.
- Try focusing on "Just 5 Minutes" a day

"Start low, go slow."
-Dr. Chopra

MOVEMENT

Movement gets the blood flowing and makes you feel better. Learn your limits.

- Track daily steps (e.g., Fitbit)
- Physical & Aquatic Therapy (Caution: heated pools may compound POTS.)
- Tai Chi
- Yoga
- Cycling
- **Supplements** -- *Epsomsalts, Arnica & Rhodiola*

Try focusing on just 5 minutes a day.

RESOURCES

Books as well as online resources are helpful.

BOOKS

- The Last Best Cure
- Move Your DNA
- Full Catastrophe Living
- The Trigger Point Therapy Workbook
- The Whole Life Nutrition Cookbook & Nourishing Meals
- The Anti-inflammation Diet & Recipe Book
- The Whole 30
- The Whal's Protocol

FINDING A PRACTITIONER

- Physicians: www.naturopathic.org
- Institute for Functional Medicine: www.functionalmedicine.org
- American Board of Integrative Holistic Medicine: www.abihm.org

Questions?

EDS WELLNESS
info@edswellness.org

DISCLAIMER -- Tell all of your health care providers about any complementary health approaches you use. Give them a full picture of what you do to manage your health. Doing so will help ensure coordinated and safe care. Use all supplements with discretion and at recommended dosage. EDS Wellness and the authors of this brochure, assume no liability for injuries as a result of using any of the therapies and supplements listed, or their lack of effectiveness. You are responsible for your own healthcare.

integrative healing

Ehlers-Danlos Syndrome

NATURAL APPROACHES TO LIVING WELL

A holistic approach for supporting individuals living with Ehlers-Danlos syndrome (EDS) — this pamphlet provides resources for incorporating mind-body therapies as part of an integrative treatment plan. Viewing the body as a complex connection of various systems, removing barriers to health, and supporting the body's own ability to heal, is critical for chronic illness, health, and overall well-being.

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Building the foundation of health is as important as managing symptoms. **NUTRITION, MOVEMENT, SLEEP AND STRESS REDUCTION** are the cornerstones of a healthy, vibrant lifestyle.
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“Start low, go slow.” -Dr. Chopra

NUTRITION:

Studies show proper nutrition is vital to staying healthy

- Eat real food; avoid processed foods, nothing artificial
- Anti-inflammatory diet (www.histaminintoleranz.ch)
- Significantly limit inflammatory foods:
- gluten, dairy, sugar, soy & alcohol
- Probiotics
- Salt & proper hydration
- Supplements

CORE NUTRIENTS

- Vitamin C
- Vitamin D3
- Vitamin B6 & B12
- Magnesium
- Calcium
- Zinc
- Iron

STRESS REDUCTION

Stress causes physical pain. It is vital to keep it under control

- Goal is REST AND DIGEST vs. FIGHT OR FLIGHT
- Acupuncture
- Mindfulness techniques
- Biofeedback
- Cranio-sacral Therapy
- Exercise (movement!)

SLEEP
A lack of sleep can wreak havoc on your body

- Relaxation apps (e.g., Relax Melodies)
- Mindfulness techniques
- Use white noise machine
- Adjust room temperature
- No blue light (tv/phone) one hour before (blue light reduces melatonin production)
- Warm shower before bed
- Supplements -- Melatonin & Ashwaganda

Pain Management

- TENS Unit
- Ultrasound
- Massage
- Infrared & Cold Laser
- Castor oil packs
- Massage pillow
- Trigger Point Therapy
- Myofascial Release
- Supplements – Turmeric, Arnica

Tumeric

A potent anti-inflammatory super-food
Inhibits NF-kappa B, the master inflammatory cytokine

Arnica

- Tincture of arnica is an external remedy for bruises, sprains, and sore muscles and joints. It is made by crushing whole plants and soaking them in alcohol.

- Arnica is toxic if it gets inside the body. Never apply arnica in any form on broken skin or on an open wound.

Movement

- Track daily steps (e.g., Fitbit)
- Physical & Aquatic Therapy (Caution: heated pools may compound POTS.)
- Tai Chi
- Yoga
- Cycling
- Supplements – Epsom salts, Arnica & Rhodiola


A mental state achieved by focusing one's awareness on the present moment, while calmly acknowledging and accepting one's feelings, thoughts, and bodily sensations, used as a therapeutic technique
Mindfulness

Books/Resources

- The Last Best Cure – Donna Nakazawa
- Move Your DNA - Katy Bowman
- Full Catastrophe Living – Jon Kabat-Zinn
- The Trigger Point Therapy Workbook – Claire Davies
- The Whole Life Nutrition Cookbook & Nourishing Meals
- Is Food Making You Sick? Josh Gibbs
- The Whals Protocol – Terry Whals

TIPS FOR SUCCESS
Look for support groups online and try the following

- When trying new therapies, keep a critical eye on the results of your trials
- Throw out what DOES NOT work and hold on to what DOES!
- For mast cell issues - focus on general stress reduction and anti-inflammatory approaches including stress reduction, movement, and eliminating inflammatory foods
- Anti-inflammatory strategies such as diet, proper nutrition, sleep, movement, and stress management are key for all

THANK YOU!

Kendra Neilsen-Myles
The Ehlers-Danlos Society
EDS-UK
Shane Robinson
Lara Bloom
Fransiska Malfait
Peter Byers
Brad Tinkle

My patients and their families

The Art of Self Care: Creating a Supportive Framework
A Retreat of Healing and Friendship

Cliff Springs Ranch, New Mexico
September 27-October 1, 2017
Margo Landon: landonmassage@gmail.com