EXTERNALLY LED PATIENT-FOCUSED DRUG DEVELOPMENT MEETING

31 OCTOBER 2023

PRESENTATION

Living with hEDS & POTS

SPEAKER

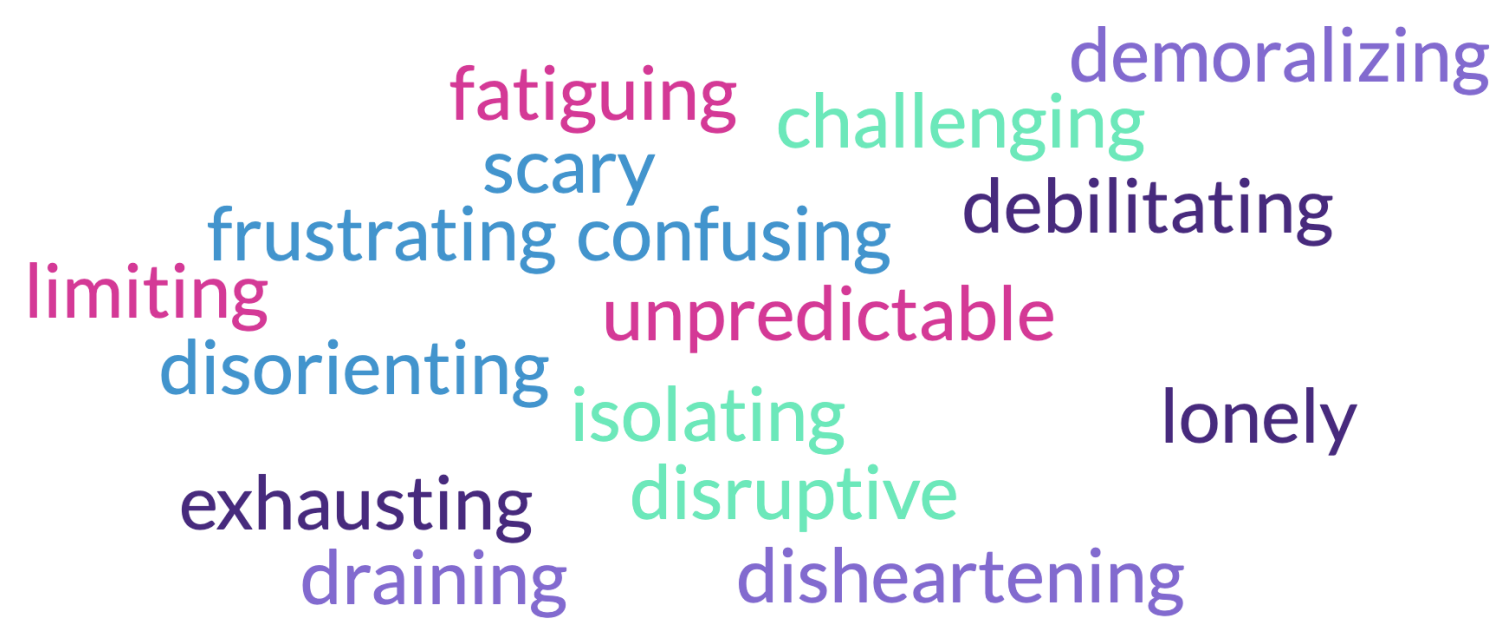
Francie Fitzgerald, LSW
Ebright Collaborative

Let’s start in 2010…

NCAA FIELD HOCKEY

“Glandular Fever”

What it's like to live with the condition(s)



As a result of this experience I have sought help from **>100** physicians in the past 13 years

April 10, 2013

The Children's Hospital of Philadelphia®

Metabolic Disease

Phone 215-590-3376 Fax 215-590-4297

ASSESSMENT/PLAN:
Frances is a 19 year old female with POTS, Joint laxity and bruising. While she does have laxity, her skin findings do not support a formal diagnosis of Ehlers Danlos syndrome to account for the bruising.
I have broadened the work up a bit during this evaluation and will seek a hematology consult if labs are abnormal. The comprehensive palatelet function test must be scheduled through HUP labs. Requisitions were provided.

Case in point…
The pains of getting diagnosed.

January 6, 2014

MAYO CLINIC

200 First Street SW
Rochester, Minnesota 55905
507-284-2511
mayoclinic.org

January 6, 2014

Salman Kirmani, M.B.B.S.
Department of Medical Genetics

It was a pleasure to participate in your care during your recent visit to Mayo Clinic.

Our final diagnoses were:

- POTS
- Question gastroparesis
- Chronic fatigue
- Does not meet criteria for any of the subtypes of the Ehlers-Danlos syndrome
- Less than ideal body weight

April 21, 2014

Penn Medicine

Division of Translational Medicine and Human Genetics

Hospital *of the* University *of* Pennsylvania

Staci Kallish, D.O.
Clinical Genetics

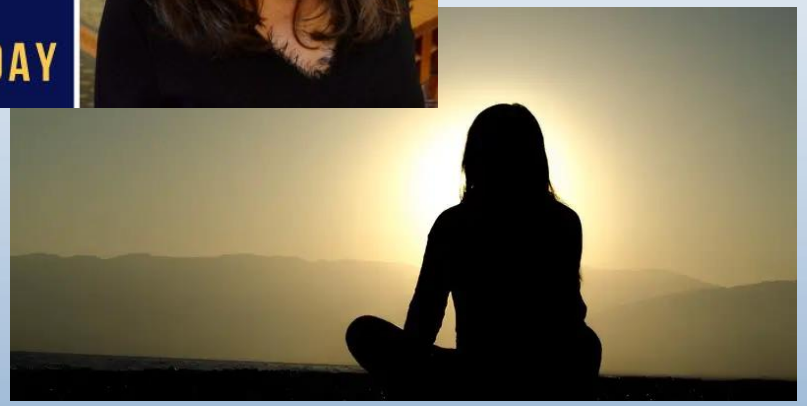
SUMMARY AND RECOMMENDATIONS:
Frances Fitzgerald is a 20 y.o. female with a medical history significant for chronic headache, fatigue and dysautonomia symptoms beginning in the setting of EBV infection. Her examination today is notable for normal mobility in most joints with subtle hypermobility in her left shoulder, right patella, and hips. Together these findings not consistent with a diagnosis of Ehlers Danlos syndrome or other connective tissue disorder.

We discussed that there is a spectrum of joint mobility within the normal range. Joint hypermobility can be a feature of a genetic syndromes that involve the connective tissue, such as EDS. In addition to joint involvement, individuals with EDS can have differences in the skin and other organ systems. We reviewed that Ms. Fitzgerald, while at the more flexible end of the normal spectrum, does not have abnormally flexible joints. There is no indication that she has an inherited connective tissue disorder. We reviewed that an underlying hereditary connective tissue disorder would not likely explain Ms. Fitzgerald's autonomic dysfunction or other complicated symtpoms.

What has helped

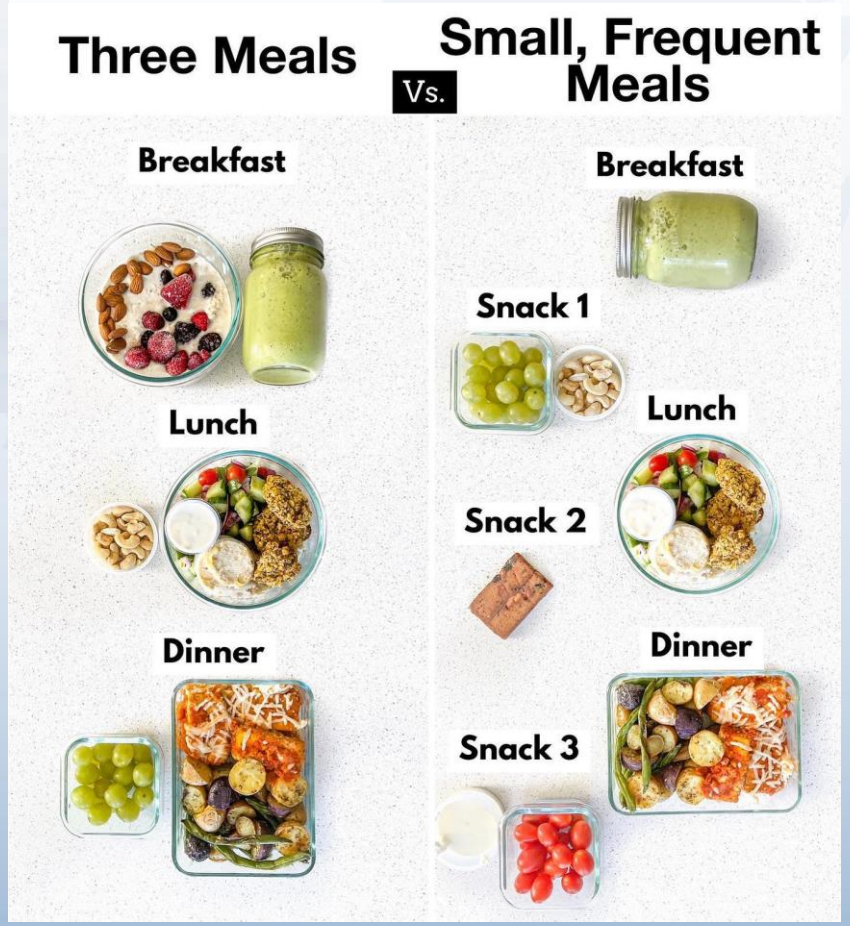
Acceptance / Cognitive Modification

Redefining what a good life looks like



School & workplace accommodations

What has helped



The key to everything is pacing & adapting

It has taken a TREMENDOUS amount of **trial and error** to find a regimen that is currently enabling stability that *minimizes interruptions* in my work & personal life…

"Quarterback"

Treatment polarization
(of Low prestige diseases)

Dismissal & Neglect

Polypharmacy & Iatrogenic harm

Treatment polarization
(of Low prestige diseases)

Dismissal & Neglect

Evidence based guidelines & standards of care

Polypharmacy & Iatrogenic harm

Moral of the story:

We need better ways to diagnose and treat these comorbidities or people will continue to lose their livelihood

Thank you for your attention

Low prestige disease

Leads to:
Invalidation
Getting lost in the system