

Quantifying the POTS Patient Experience

Presented by Lauren Stiles, JD
President & Co-Founder

DYSAUTONOMIA INTERNATIONAL



AWARENESS



ADVOCACY



ADVANCEMENT

Beth Israel Deaconess Medical Center
May 6, 2016

Disclosures

- Consulting fees: Vaelant Pharmaceuticals
- I might discuss off-label use of medications.
- I am not a physician.

Common Challenges for POTS Patients

- Physical symptoms that may result in significant disability
- Many comorbidities
- Frequent diagnostic delay & misdiagnoses
- Difficulty finding physicians with POTS expertise
- Misconceptions about POTS being “a teenage syndrome that everyone grows out of”
- “Normal” physical appearance may lead to disbelief amongst classmates, co-workers, family, friends & doctors

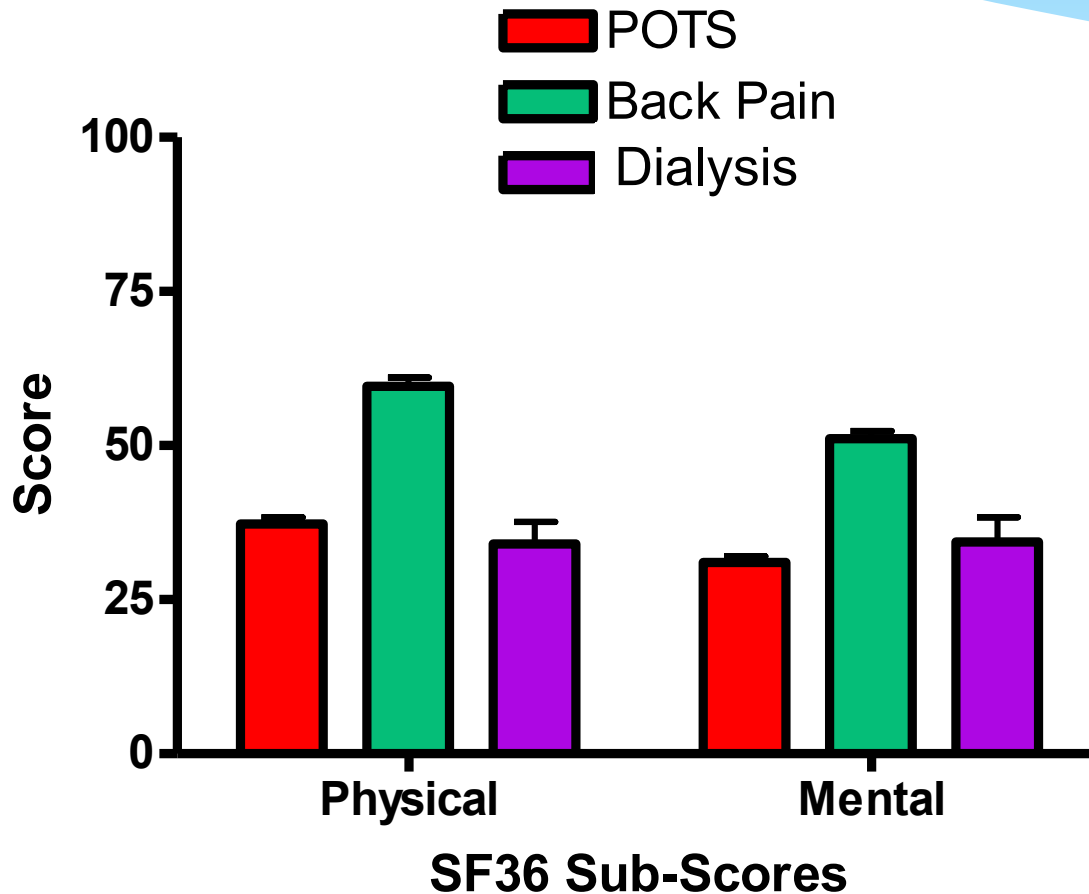
Disability

- SF-36 physical functioning, role functioning, bodily pain, general health, vitality and social functioning significantly impaired in POTS vs. healthy controls.¹
- **POTS disability similar to disability seen in congestive heart failure and COPD.**¹
- Psychological domains similar amongst healthy controls, CHF, COPD and POTS.¹

¹ Benrud-Larson LM, Dewar MS, Sandroni P, Rummans TA, et al. Quality of Life in Patients with Postural Orthostatic Tachycardia Syndrome. *Mayo Clin Proc.* 2002;77:531-537.

Disability

Health Related Quality of Life (SF-36) – Chronic Illnesses



² Modified from K Bagai et al., J Clin Sleep Med 2011. Slide courtesy of Dr. Satish Raj.

Comorbidities

“The Big POTS Survey”

- IRB approved structured online survey
- Collaboration between Dysautonomia International, Vanderbilt University & University of Calgary
- Patient input into study design
- 3300+ POTS patients, 400+ variables
- Patients from more than 15 countries
- Largest POTS study to date – follow up surveys in progress, plans to link to clinical data to develop larger POTS research database



UNIVERSITY OF
CALGARY

DYSAUTONOMIA INTERNATIONAL



AWARENESS



ADVOCACY



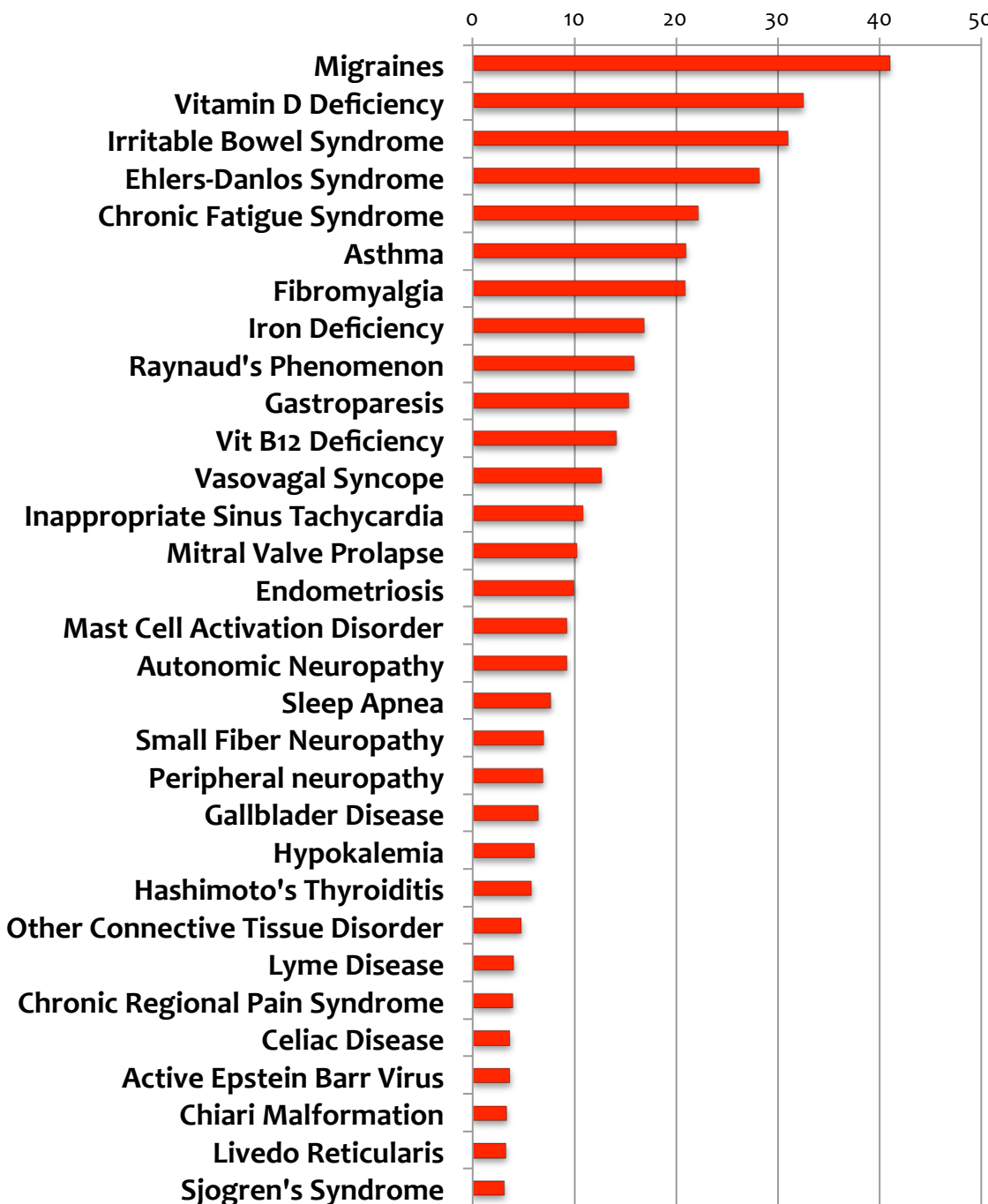
ADVANCEMENT



VANDERBILT

³ Satish R. Raj MD MSCI, Lauren E. Stiles JD, Brett H Shaw MSc, Elizabeth A. Green MD, Cindy A. Dorminy MEd, Cyndya A. Shibao MD MSCI, Luis E. Okamoto MD, Emily M. Garland PhD MSCI, Alfredo Gamboa MD MSCI, Andre Diedrich MD PhD, Italo Biaggioni MD, David Robertson MD. The Face of Postural Tachycardia Syndrome (POTS): A Cross-Sectional Community-Based Survey. Heart Rhythm 2016; 12 (5S):xx-xx (abstr) [in press].

% Total



“The Big POTS Survey” Common Comorbidities³

- 84% of POTS patients report comorbidities
- Ehlers-Danlos Syndrome: 28%
- Confirmed autoimmune disease: 16%
- Confirmed autoimmune disease within EDS/POTS subjects: 18%
- Some of the comorbidities are likely underdiagnosed

DYSAUTONOMIA INTERNATIONAL



AWARENESS



ADVOCACY



ADVANCEMENT

Thoughts on POTS and Autoimmunity...

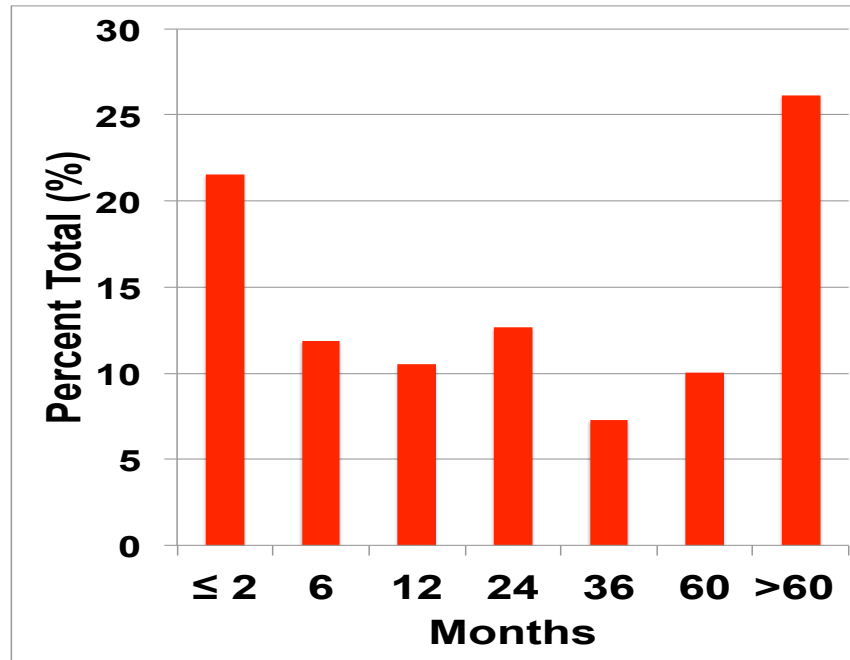
- Initial 1993 description from Mayo suggested partial immune mediated neuropathy.⁴
- Acute onset in 50% of patients, 85-90% female
- Univ. of Oklahoma/Vanderbilt: 100% have adrenergic receptor antibodies (n=14)⁵, larger cohort in progress...
- SUNY Buffalo: 20% have comorbid autoimmune disease, 33% have commonly tested autoimmune markers (n=100)⁶
- Mayo: 33% have thyroid and/or neural antibodies, 11% more than one (n=33)⁷, larger cohort in progress...
- Dysautonomia Intl.: 41% idiopathic dysautonomia patients with dry eyes or dry mouth have novel “early” Sjögren's antibodies (including 6 of 10 POTS patients in the study) (n=95)⁸
- UT Southwestern: muscarinic antibodies in POTS, stay tuned...

POTS and Autoimmunity Citations

- 4 – Schondorf R, Low PA. Idiopathic postural orthostatic tachycardia syndrome: an attenuated form of acute pandysautonomia? *Neurology*. (1993)43(1);132-137.
- 5 - Li H, Yu X, Liles C, Khan M, et al. Autoimmune Basis for Postural Tachycardia Syndrome. *Jour Amer Heart Assn*. (2014).
- 6 - Blitshteyn S. Autoimmune markers and autoimmune disorders in patients with postural tachycardia syndrome (POTS). *Lupus*. (2015)0;1-6.
- 7 - Singer W, Klein CJ, Low PA, Lennon V. Autoantibodies in the postural tachycardia syndrome. *Clin Auton Res*. (2014)24:214.
- 8 - AAN Annual Meeting abstract published online (2016): Gudesblatt M, Wissemann K, Stiles L, Xian S, et al. Autoimmunity & Autonomic Impairment: Preliminary Characterization of a Clinical Syndrome with Sjögren's Features Associated with Novel Organ Specific Antibodies. P5.108.

“The Big POTS Survey” – Diagnostic Delay

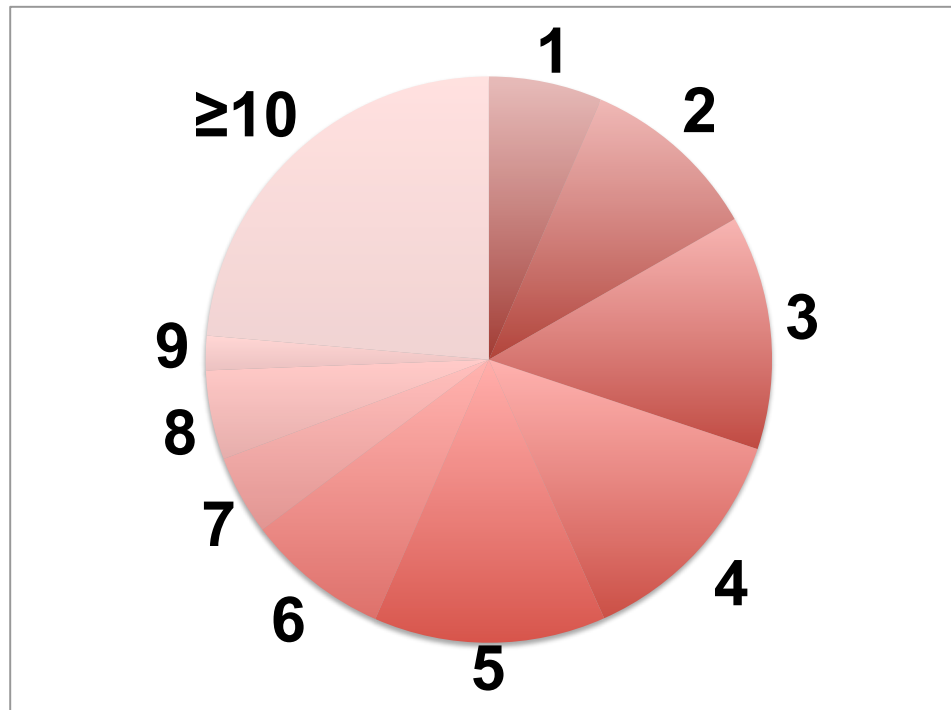
- Avg. diagnostic delay = 50 months (4 years, 2 months)³



- Dysautonomia International’s ~790 patient survey of same online community found an average diagnostic delay of 71 months in 2013.⁹
- Reduction in diagnostic delay by 1 year, 9 months.
- **AWARENESS MATTERS!**

“The Big POTS Survey” – Diagnostic Delay

- Average number of doctors seen prior to diagnosis = 7³



- 24% of respondents saw ≥ 10 physicians prior to diagnosis.³
- “The doctor bounce” is not always the patient’s fault.

Misdiagnosis

Patient Blaming 101

You're just hungover.

Maybe you weren't cut out to be a lawyer. Go home and enjoy a glass of red wine to relax.

You're doing this because you're 31 and don't have babies yet, and you're just trying to get attention from your husband.

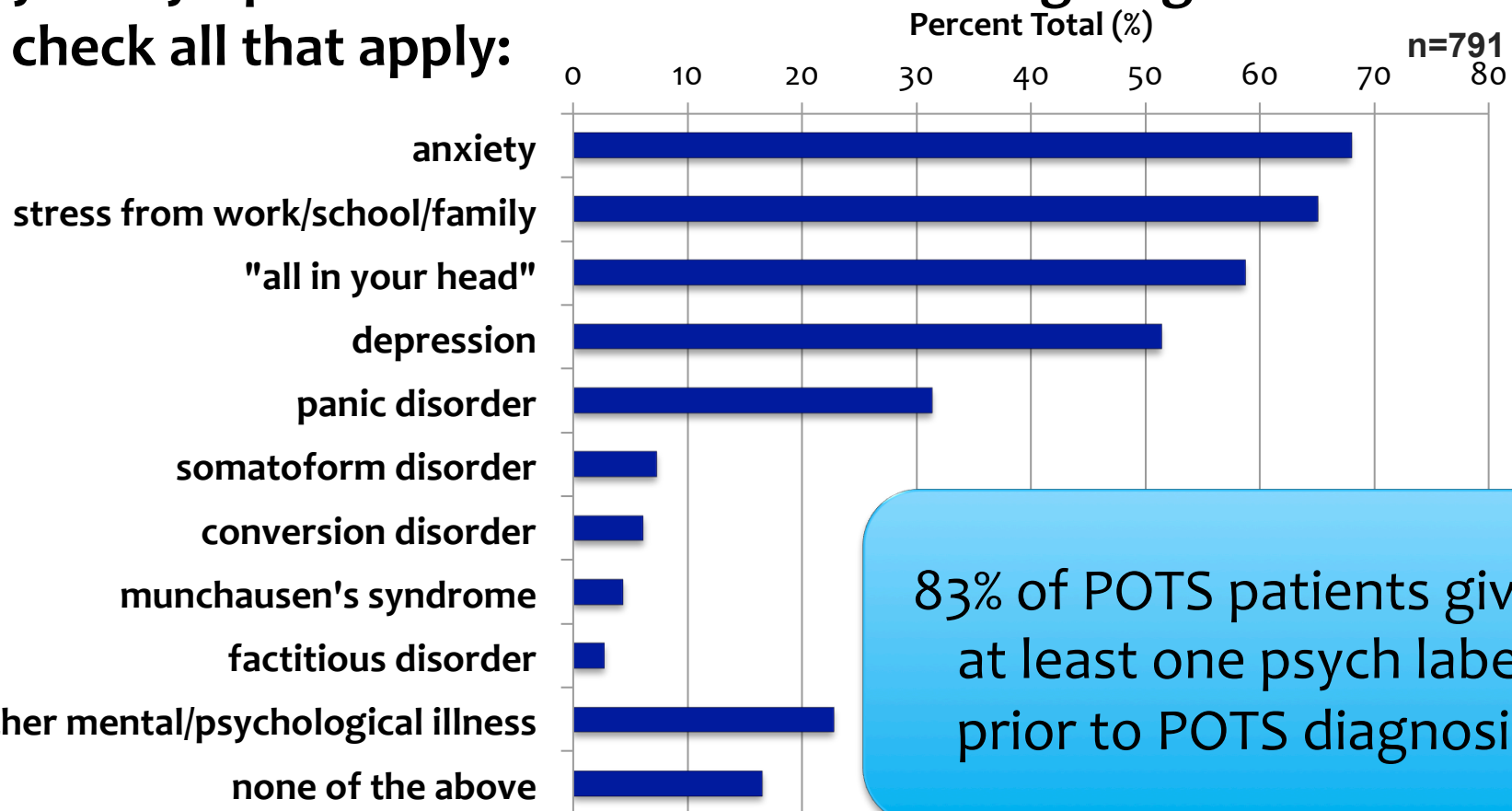
You're deconditioned because lawyers sit at their desks all day.

Sitting at my desk all day...



Misdiagnosis

BEFORE your POTS diagnosis, were you ever told by a doctor that your symptoms were due to the following diagnoses? Please check all that apply:



83% of POTS patients given at least one psych label prior to POTS diagnosis

Misdiagnosis

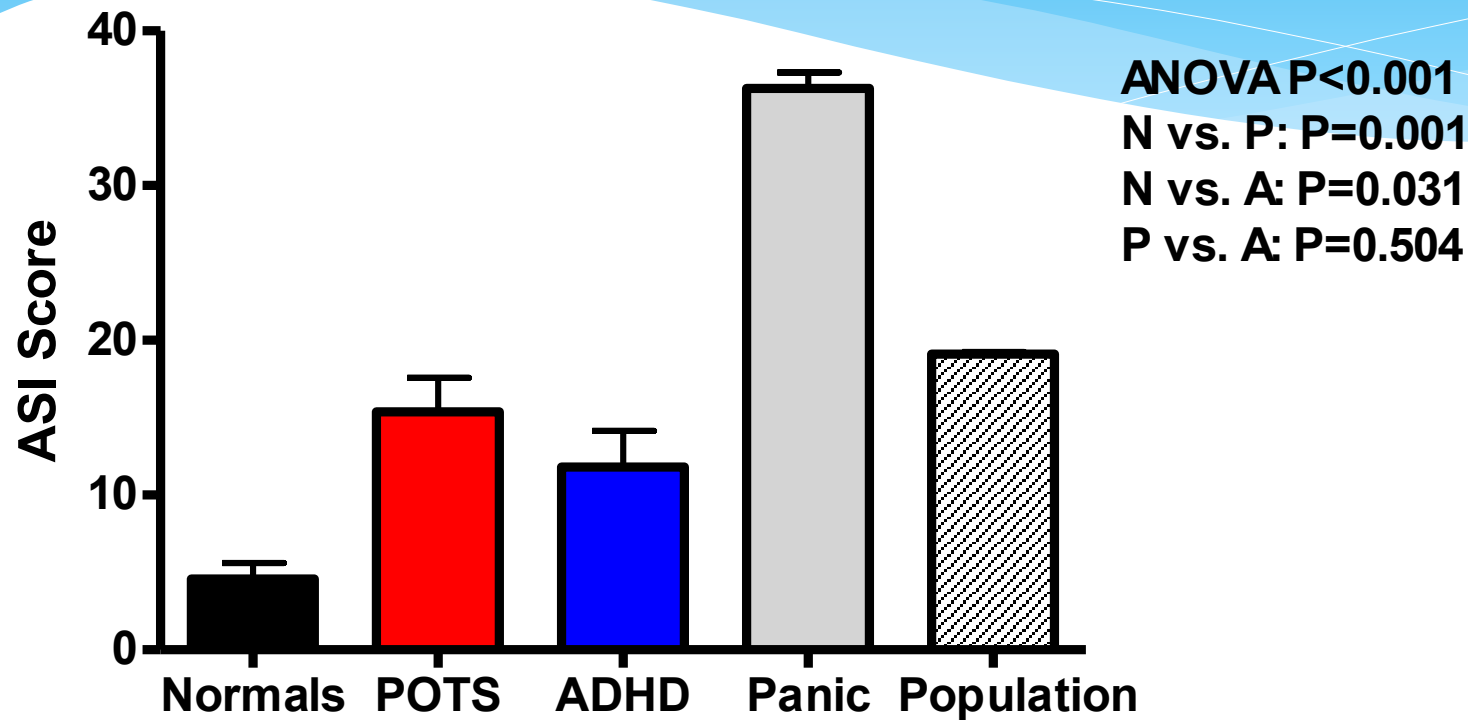
Other data suggests this is largely misdiagnosis...

- Mayo 1992: Psychological domains similar amongst healthy controls, CHF, COPD and POTS.¹
- Vanderbilt 2009: POTS patients slightly less anxious than general population, slight increase in mild depression.¹¹
- Mayo 2016: mean mental composite score normal in pediatric POTS.¹²

When psychological comorbidity is present, it should be addressed. Patients are often afraid to ask for help.

Misdiagnosis

Anxiety (ASI) Scores



* Total score 0-64

* Population 19.1 ± 9.1

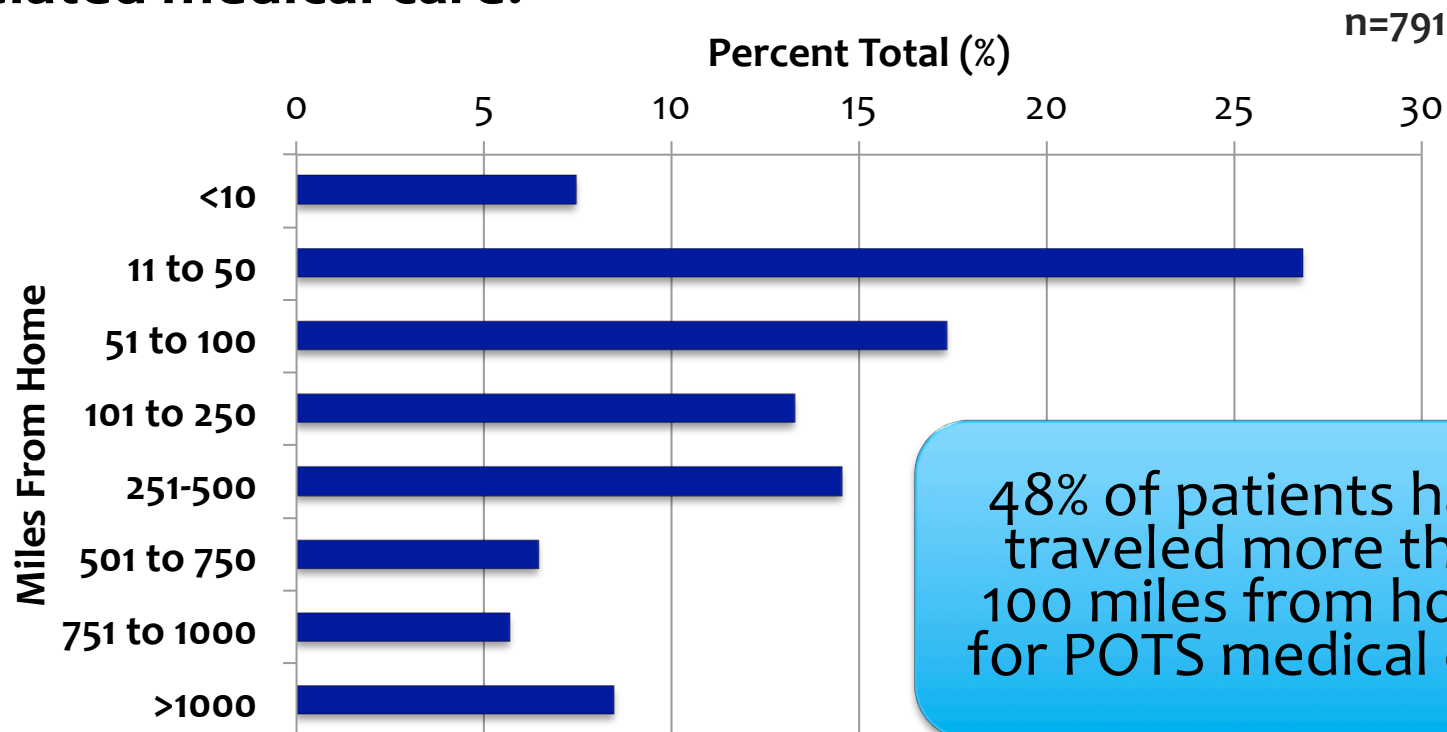
* Panic disorder 36.4 ± 10.3

■ POTS vs. pop: $P = 0.07$

■ ADHD vs. pop: $P = 0.001$

Lack of Access to Care

How many miles from home have you traveled to obtain POTS related medical care? ¹⁰



- Significant financial burden on families, and many can't afford to travel to see a specialist

Disbelief

“You’re too young/pretty to be this sick.”

“You look fine.”

“Are you better yet? I had the flu once too.”

“If you can put on make-up and earrings, you must be better.”

“I saw you at the movies last weekend, so you must be faking.”

**Living with POTS is like being stuck
on a rollercoaster you can't get off of.**



POTS - looking great while feeling awful...



Don't judge a book by it's cover, or a patient by her lipstick!

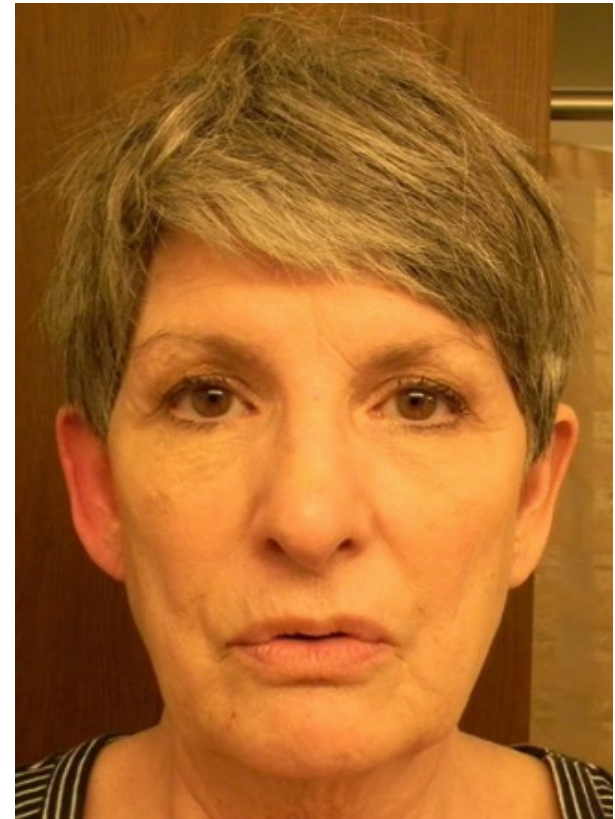
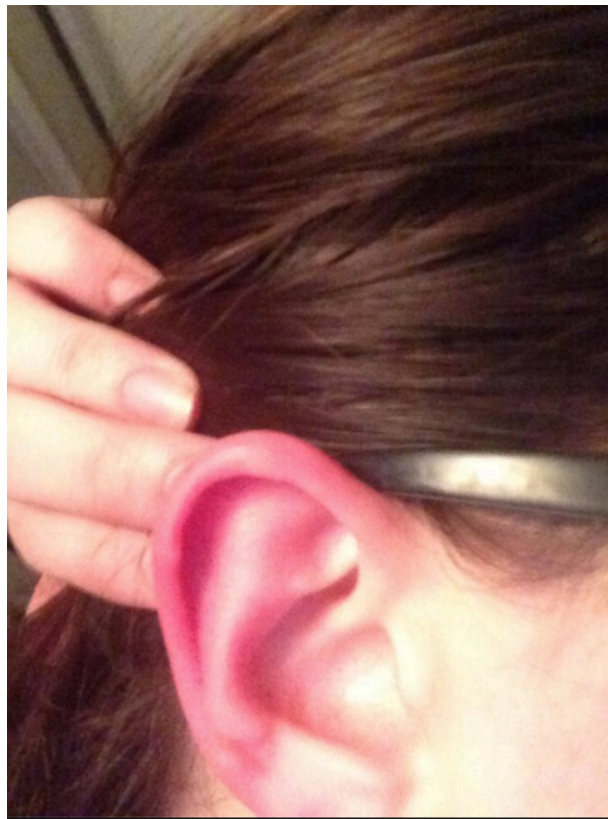
Not really an “invisible illness”

Acrocyanotic
“POTS legs”



Not really an “invisible illness”

“Red Ear Syndrome” – typically unilateral



Not really an “invisible illness”

Lots and lots of flushing...



Misconceptions – Is POTS rare?

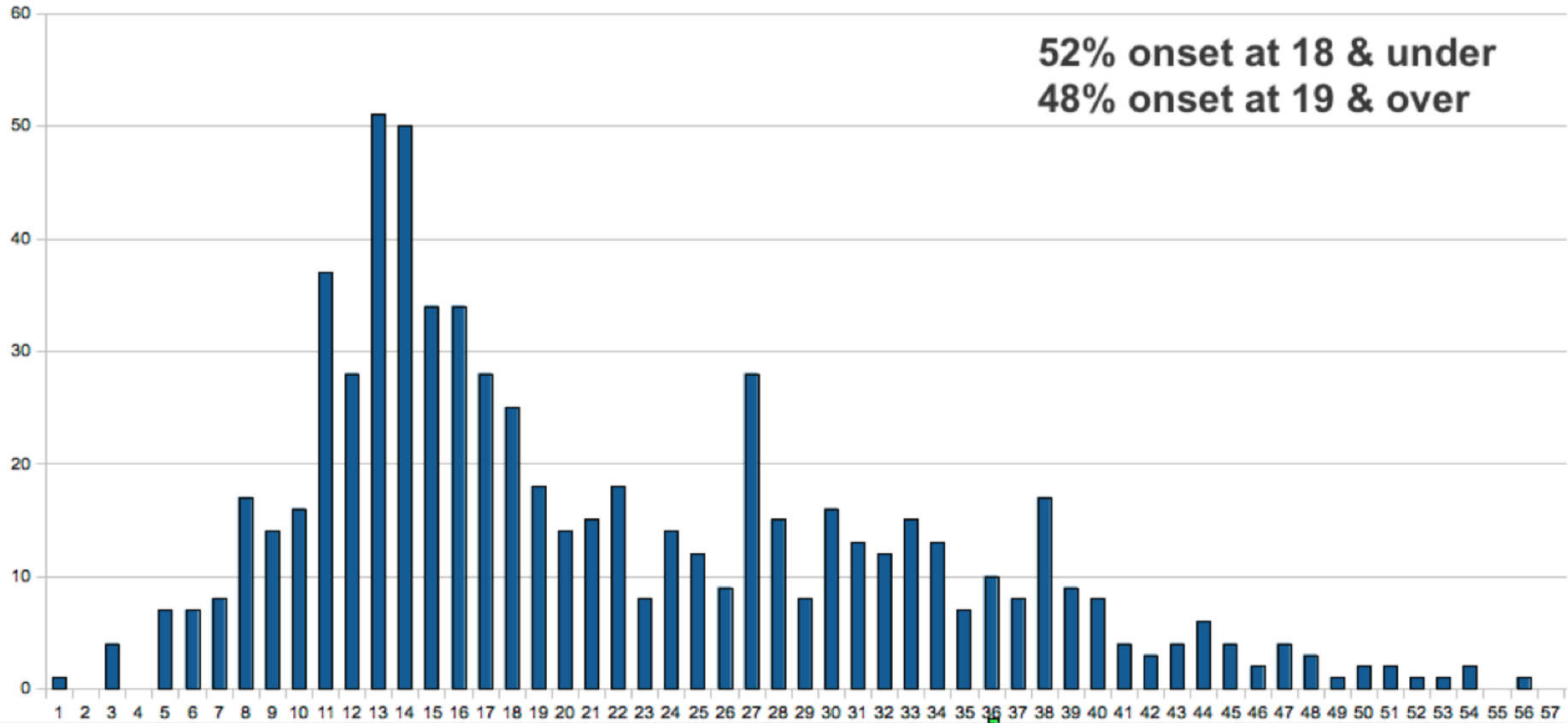
- → → ***POTS is not rare!*** ← ←
- One of the most common orthostatic disorders.
- Peer-reviewed estimates range from 500K-3M in the US alone.
- For comparison: multiple sclerosis – 400K
 Parkinson's – 1M
- Mayo Clinic estimates 1 out of 100 teens¹²
(about 500K teens based on last US census).
- Big POTS Survey found 59% of individuals develop POTS symptoms after age 18.³ Similar data from 2013 Dysautonomia International survey.¹⁰

Not just a “teenage syndrome”

Patient Reported Age at Onset of POTS Symptoms

10

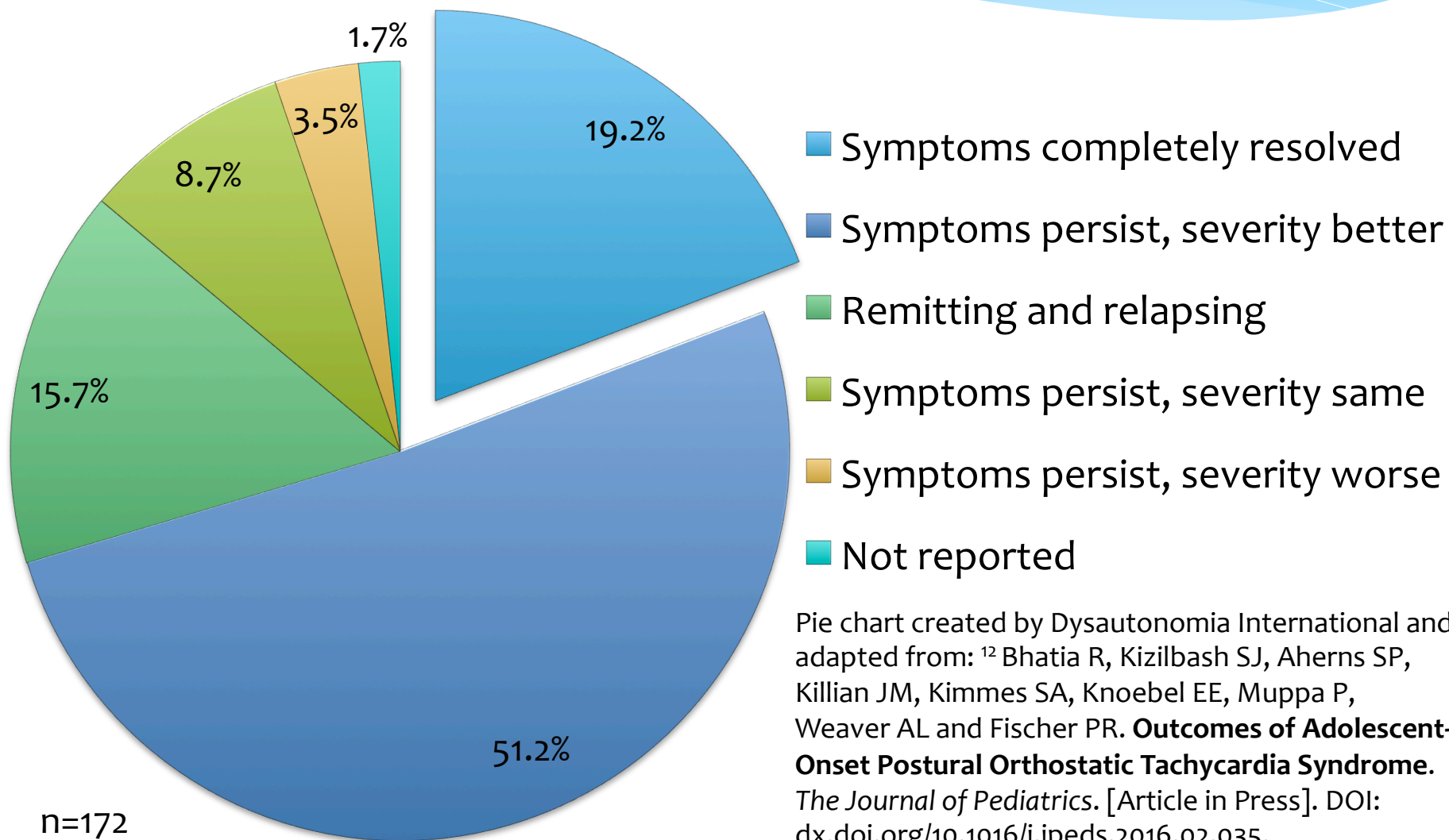
N=697



Outcomes of Adolescent-Onset POTS¹²

- 172 Mayo Clinic peds POTS patients
- Ages 13-18, seen between 2003-2010
- Mean duration from diagnosis to survey: 5.4 years
- Mean age at time of survey 21.8
- 84% female
- Mean physical composite score significant lower than norm (36.6 vs 50)
- Mean mental composite score normal (50.1 vs. 50)
- 86% report improved symptoms
- Patients with persistent symptoms have more physical than mental health concerns

Everyone doesn't "grow out of it"



Pie chart created by Dysautonomia International and adapted from: ¹² Bhatia R, Kizilbash SJ, Aherns SP, Killian JM, Kimmes SA, Knoebel EE, Muppa P, Weaver AL and Fischer PR. **Outcomes of Adolescent-Onset Postural Orthostatic Tachycardia Syndrome.** *The Journal of Pediatrics*. [Article in Press]. DOI: [dx.doi.org/10.1016/j.jpeds.2016.02.035](https://doi.org/10.1016/j.jpeds.2016.02.035).



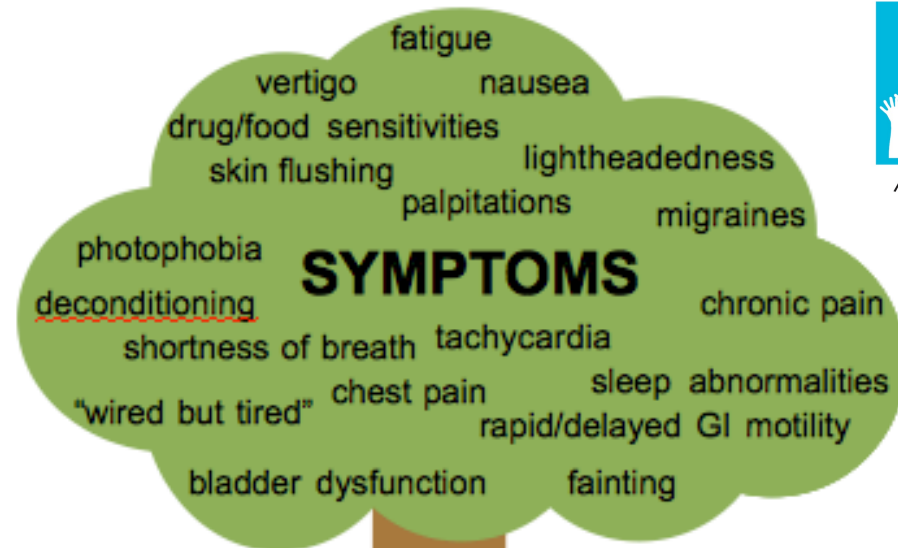
AWARENESS



ADVOCACY



ADVANCEMENT



POTS

ROOT CAUSES

genetic

environmental

autoimmunity?

immunogenetics

infections

concussions

other

pregnancy

surgery

other

mito

NET

EDS



AWARENESS



ADVOCACY



ADVANCEMENT

**1940s thinking:
“When you hear hoofbeats,
think of horses, not zebras.”**



Not every horse is a textbook case. Be on the lookout for horses of a different color.



1 out of 10 Americans has a rare disease. You're going to see a lot of zebras.



Thank you!

Connect with us...

www.dysautonomiainternational.org

www.facebook.com/dysautonomiainternational

www.vimeo.com/dysautonomia

Email: lstiles@dysautonomiainternational.org

Twitter: [@Dysautonomia](https://twitter.com/Dysautonomia)

DYSAUTONOMIA INTERNATIONAL



AWARENESS



ADVOCACY



ADVANCEMENT