Quantifying the POTS Patient Experience

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Dysautonomia International
Awareness
Advocacy
Advancement

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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

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

Disability

Score

SF36 Sub-Scores

Physical

Mental

POTS
Back Pain
Dialysis

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

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
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


“The Big POTS Survey” – Diagnostic Delay

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

![Bar chart showing diagnostic delay distribution]

• 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!
• Average number of doctors seen prior to diagnosis = 7

• 24% of respondents saw ≥10 physicians prior to diagnosis.

• “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...
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:

- Anxiety
- Stress from work/school/family
- "All in your head"
- Depression
- Panic disorder
- Somatoform disorder
- Conversion disorder
- Munchausen's syndrome
- Factitious disorder
- Other mental/psychological illness
- None of the above

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

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.
Anxiety (ASI) Scores

- Total score 0-64
- Population 19.1±9.1
- Panic disorder 36.4±10.3

ANOVA P<0.001
N vs. P: P=0.001
N vs. A: P=0.031
P vs. A: P=0.504

POTS vs. pop: P=0.07
ADHD vs. pop: P=0.001

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

- 48% of patients have traveled more than 100 miles from home for POTS medical care.

- Significant financial burden on families, and many can’t afford to travel to see a specialist.
“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...
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 teens\(^1\) (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

N=697

52% onset at 18 & under
48% onset at 19 & over
Outcomes of Adolescent-Onset POTS

- 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”
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!

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