Journal of *Personalized Medicine*

MDPI

*Supplementary Materials*

# Genomic screening identifies individuals at high risk for hereditary transthyretin amyloidosis

**Emily R. Soper, MS[1,2#], Sabrina A. Suckiel, MS[1,2#], Giovanna T. Braganza, BS[1], Amy R. Kontorovich, MD, PhD[2,3], Eimear E. Kenny, PhD[1,2,4], Noura S. Abul-Husn, MD, PhD[1,2,4*]**

[1] The Institute for Genomic Health, Icahn School of Medicine at Mount Sinai, New York, NY, USA
[2] Department of Medicine, Icahn School of Medicine at Mount Sinai, New York, NY, USA
[3] The Zena and Michael A. Wiener Cardiovascular Institute, Icahn School of Medicine, New York, NY, USA
[4] Department of Genetics and Genomic Sciences, Icahn School of Medicine at Mount Sinai, New York, NY, USA
[#] These authors contributed equally to this work.

* Correspondence: noura.abul-husn@mssm.edu

**Table S1.** Presence of hATTR-related systemic features and related symptoms in 32 V142I variant positive individuals.

**Table S2.** Presence of hATTR-related systemic features by age, sex, and self-reported ancestry after follow up with recommended specialists.

**Citation:** Soper, E.R.; Suckiel, S.A.; Braganza, G.T.; Kontorovich, A.R.; Kenny, E.E.; and Abul-Husn, N.S. Genomic screening identifies individuals at high risk for hereditary transthyretin amyloidosis. *J. Pers. Med.* **2021**, *11*, 49. https://doi.org/10.3390/jpm11010049

Received: 21 December 2020
Accepted: 09 January 2021
Published: 15 January 2021

**Table S1.** Presence of hATTR-related systemic features and related symptoms in 32 V142I variant positive individuals.

| Phenotype | Systemic feature / *Related symptom* | No. (%) at result disclosure | No. (%) after specialist follow-up | No. (%) with family history* |
|---|---|---|---|---|
| Cardiac | Heart failure | 4 (13) | 5 (16) | 6 (19) |
| | Cardiomyopathy | 1 (3) | 1 (3) | 1 (3) |
| | Atrial fibrillation | 0 (0) | 1 (3) | 3 (9) † |
| | *Shortness of breath* | 16 (50) | 19 (59) | - |
| | *Dizziness* | 8 (25) | 12 (38) | - |
| | *Syncope* | 4 (13) | 6 (19) | - |
| | *Palpitations* | 9 (28) | 12 (38) | - |
| | *Edema* | 9 (28) | 10 (31) | - |
| Peripheral neuropathy | Carpal tunnel syndrome | 10 (31) | 10 (31) | 8 (25) |
| | Spinal stenosis | 10 (31) | 10 (31) | 0 (0) |
| | *Carpal tunnel-related symptoms* | 13 (41) | 15 (47) | - |
| | *Gait/Balance problems* | 11 (34) | 13 (41) | - |
| | *Muscle weakness* | 13 (41) | 13 (41) | - |
| | *Numbness/tingling* | 17 (53) | 20 (63) | - |
| | *Pain in extremities* | 14 (44) | 14 (44) | - |
| | *Back pain* | 16 (50) | 18 (56) | - |
| | *Temperature sensitivity* | 0 (0) | 4 (13) | - |
| | *Joint pain* | 18 (56) | 19 (59) | - |
| Autonomic neuropathy | Autonomic dysfunction# | 1 (3) | 1 (3) | 0 (0) |
| | Incontinence | 4 (13) | 5 (16) | 0 (0) |
| | Sexual dysfunction/Impotence | 3 (9) | 4 (13) | 0 (0) |
| | *Nausea/vomiting* | 6 (19) | 7 (22) | - |
| | *Diarrhea* | 8 (25) | 8 (25) | - |
| | *Constipation* | 12 (38) | 15 (47) | - |
| | *Loss of appetite/early satiety* | 9 (28) | 10 (31) | - |
| Presence of any systemic feature or related symptom | | 30 (94) | 31 (97) | - |
| Presence of any systemic feature only | | 18 (56) | 19 (59) | 15 (47) |

* Family history was assessed for presence of hATTR-related systemic features only.
† Family history was assessed broadly for arrhythmia and not specifically for atrial fibrillation.
# Autonomic dysfunction includes gastroparesis and orthostatic hypotension.

**Table S2.** Presence of hATTR-related systemic features by age, sex, and self-reported ancestry after follow up with recommended specialists.

| **Demographic variable** | **Presence of any hATTR-related systemic feature No. (%)** | ***P*-value** |
|---|---|---|
| <60 years (N=18) | 9 (50) | 0.29 |
| ≥60 years (N=14) | 10 (71) | |
| Female (N=26) | 16 (62) | 0.67 |
| Male (N=6) | 3 (50) | |
| African American/African (N=17) | 9 (53) | 0.49 |
| Hispanic/Latinx (N=15) | 8 (68) | |