Patient Experience With Hereditary and Wild-type Transthyretin Amyloidosis: A Survey From the Amyloidosis Research Consortium

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BACKGROUND

- Diagnosis of and access to appropriate therapy for patients with hereditary and wild-type transthyretin (TTR) amyloidosis (ATTR) pose significant challenges, and only a paucity of literature depicts the patient's experience
- We conducted a survey to identify the challenges in establishing a diagnosis of hereditary or wild-type amyloidosis and to gain insight into the patient experience

OBJECTIVE

- To gather data on the experience of patients with hereditary or wild-type amyloidosis so as to foster understanding of the challenges physicians face in establishing a correct diagnosis and of the barriers patients face in accessing appropriate care

METHODS

- Patients with all types of amyloidosis, their family members, and their caregivers were invited to participate in an online survey through email and social media channels of the Amyloidosis Foundation (www.amyloidosis.org) and an amyloidosis awareness group on Facebook (www.facebook.com/AmyloidosisFoundation)
- The 16-question survey was developed by the authors and was available online to participants from January 29 to February 5, 2015
- This initial survey covered demographic information, type of amyloidosis, symptoms, organ involvement, diagnosis, amyloidosis education, and clinical trial awareness
- After the initial survey was completed, participants who provided contact information were sent a shorter follow-up survey by email
  - The follow-up survey consisted of 8 questions covering treatments, treatment tolerance, and quality of life before and after treatment

RESULTS

Participants

- The initial survey was completed by 533 participants; 58% identified themselves as patients, 34% as family members, and 8% as caregivers taking the survey on behalf of patients
- Most patients (71.7%) had light chain (AL) amyloidosis. Other types and frequencies were
  - 7.6% hereditary ATTR and 2.7% wild-type ATTR (**Table 1**)
  - 4.8% AA amyloidosis
  - 3.4% non-ATTR
  - Remaining were "other" or "unknown"
- Most respondents were male (62%) and were between 50 and 69 years of age (62%)
- The follow-up survey was completed by 201 participants

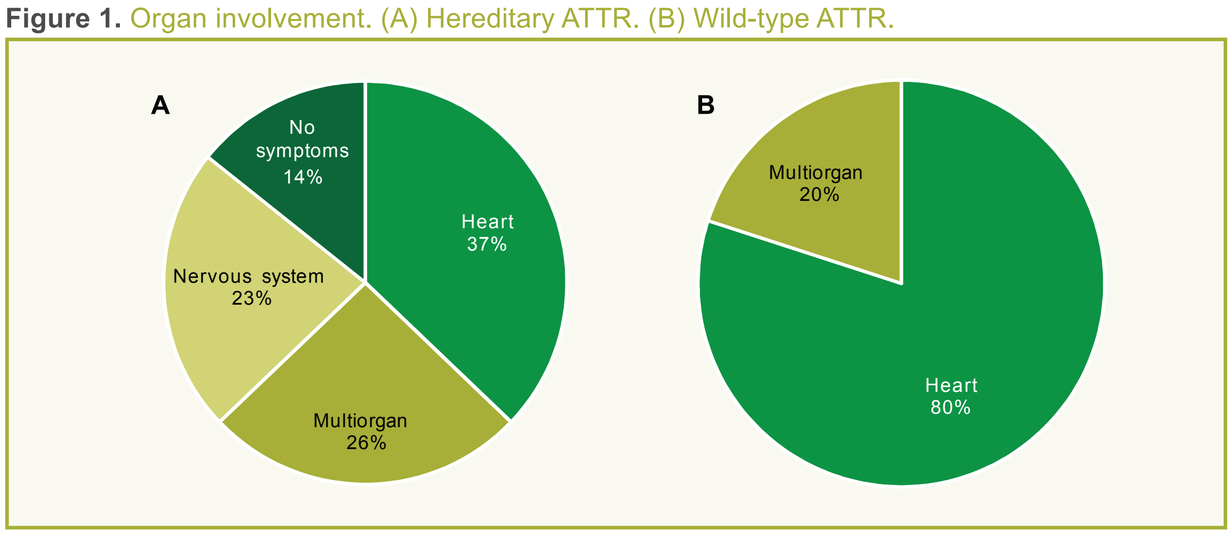
Table 1. Characteristics of Survey Respondents

	Hereditary ATTR	Wild-type ATTR
Respondents, n	37	13
Sex, n (%)		
Female	16 (43.2)	3 (23.1)
Male	21 (56.8)	10 (76.9)
Age, years		
Range	20-73	64-83
Mean at diagnosis	54	71
Time to diagnosis, n (%)	n = 30	n = 11
<6 months	13 (43.3)	5 (45.5)
6 to <12 months	5 (16.7)	3 (27.3)
12 to <18 months	2 (6.7)	1 (9.1)
18 to <2 years	0 (0)	0 (0)
2 to <3 years	1 (3.3)	0 (0)
≥3 years	9 (30.0)	2 (18.2)
Diagnosed by	n = 27	n = 11
Primary care physician	4 (14.8)	0 (0)
Cardiologist	9 (33.3)	8 (72.7)
Hematologist	2 (7.4)	1 (9.1)
Neurologist	8 (29.6)	0 (0)
Ophthalmologist	2 (7.4)	0 (0)
Internist	0 (0)	2 (18.2)
Genetic counselor	2 (7.4)	0 (0)

Amyloidosis Symptoms and Organ Involvement

- Initial symptoms for all respondents were most commonly fatigue, shortness of breath, weakness, neuropathy, and swelling of the legs, tongue, or both

Figure 1. Organ involvement. (A) Hereditary ATTR. (B) Wild-type ATTR.



- Most respondents with hereditary ATTR reported the heart or the nervous system as the major organ affected (**Figure 1**)
- Respondents with wild-type ATTR predominantly had cardiac involvement

Diagnosis of ATTR

- Median age at diagnosis was 54 years for patients with hereditary ATTR and 71 years for patients with wild-type ATTR
- Although some respondents received a diagnosis of hereditary ATTR within 1 year of initial symptoms (60%), diagnosis was delayed for ≥3 years in a substantial proportion (30%) (**Table 1**)
- Patients with wild-type ATTR predominantly received their diagnoses in <12 months (81.8%) (**Table 1**)
- A substantial proportion of respondents reported visiting ≥5 different physicians before receiving the diagnosis of ATTR (**Figure 2**)
- Respondents usually received the correct diagnosis from a cardiologist (47.1%); conversely, however, cardiologists missed most diagnoses (**Figure 3**)

Figure 2. Number of distinct physicians visited to receive diagnosis.

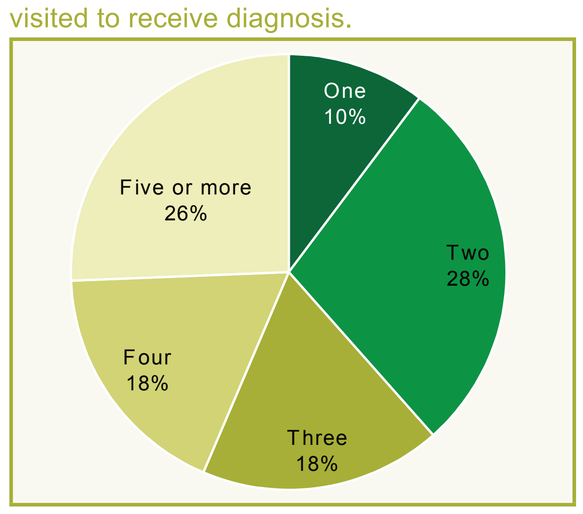
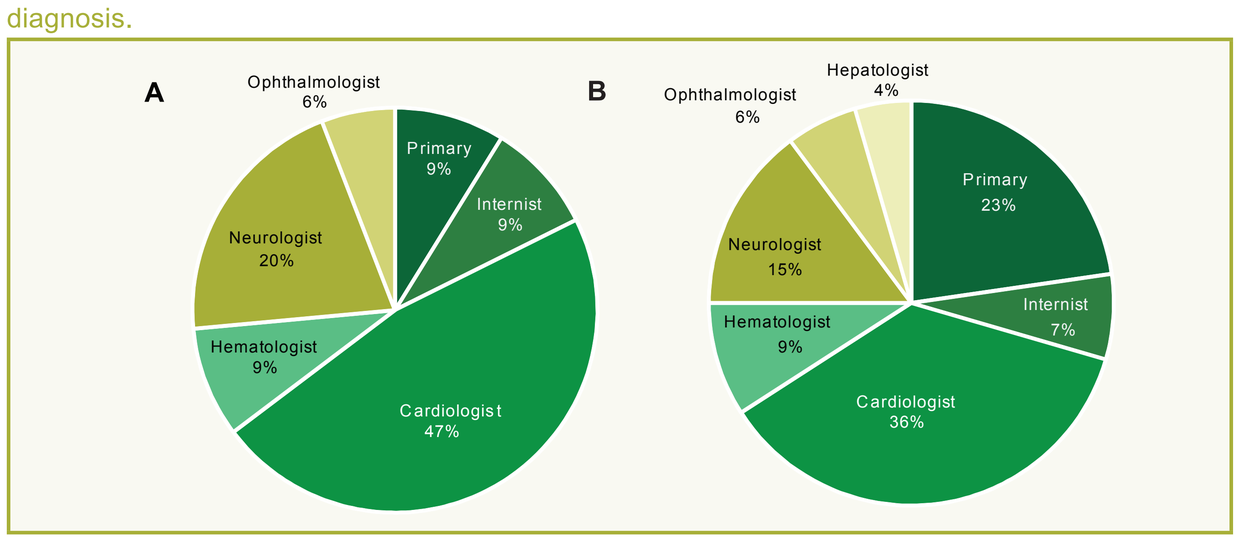


Figure 3. (A) Specialty of physician making diagnosis. (B) Specialty of physician missing diagnosis.



- Evaluation at a core amyloidosis center occurred for 20 patients with hereditary ATTR and 10 patients with wild-type ATTR. Of these, 12 patients with hereditary ATTR and 7 patients with wild-type ATTR received treatment
- When asked how the diagnosis of ATTR made them feel, 48.6% of patients reported feeling frightened, 40.5% depressed, 21.6% numb, 24.3% powerless, 18.9% hopeless, 18.9% relieved, and 21.6% angry (37 respondents; 72 total responses)

Amyloidosis Education

- Among all respondents (including those without ATTR and a total of 427 individuals with any form of amyloidosis responding to this question), at the time of diagnosis of their specific amyloidosis type, 39% of respondents reported receiving the center's own printed handouts, 34% received another organization's disease or treatment literature, 29% received information on support groups, 24% received clinical trial information, and 40% reported receiving no information or educational material

Clinical Trial Awareness

- In the initial survey, participants were asked to score their answers regarding clinical trial knowledge and access on a scale of 1 to 5, from "Not at all" (score 1) to "Absolutely" (score 5)
- Of 36 respondents with either hereditary or wild-type ATTR, 11 had participated in a clinical trial, and 25 had not
- Among patients with hereditary and wild-type ATTR, 31 patients (83.8% of respondents) would consider enrolling in a clinical trial if they were better informed. Although 20 participants (54.1% of respondents) knew how to enroll in a clinical trial, 13 participants (35.1% of respondents) reported having absolutely no idea how to enroll in a clinical trial

CONCLUSIONS

- Many patients with hereditary ATTR were diagnosed in <6 months, probably because some had a known family history; however, a substantial number of patients were not diagnosed for >3 years and after seeing >5 specialists
- Patients with wild-type ATTR were diagnosed more rapidly; in general, diagnosis was made in 6 to 12 months and after seeing an average of 3 specialists
- Survey responses indicate that patient awareness of clinical trials and patient education about the disease can be considerably improved, especially because responders stated a high willingness to participate in clinical trials. A number of opportunities can enhance patient care, such as providing education and information to help patients feel empowered and knowledgeable about their diagnoses and treatment plans and increasing access to support groups and relevant clinical trials
- Data from this study and other studies can help identify areas in which diagnosis is delayed or missed and can illustrate the need for early, accurate diagnosis of amyloidosis to help improve disease management and survival outcomes

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http://bit.ly/1ZDUBgK

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