# The Patient Voice: Development and Results of a Pilot Patient Experience Data (PED) Survey

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**Poster PLB014**

## BACKGROUND

- Amyloidosis is a rare, systemic disease that is characterized by variable pattern of nonspecific symptoms and affected organs.[1]
  - These characteristics of amyloidosis are common among rare diseases and make them difficult to diagnose and treat.[1,2]
- The 2 main forms of cardiac amyloidosis are light-chain (AL) amyloidosis and transthyretin (ATTR) amyloidosis.
- One way to improve diagnosis is to increase awareness among physicians.
  - It is well established that physicians prefer peer-reviewed articles as their primary source for new information.[3]
  - However, this preference raises a barrier because rare diseases are collectively poorly represented in medical literature.[4]
  - The research also shows that, until recently, patient and caregiver voices were practically absent, regardless of the rare disease.[4]
  - Encouraging signs include some recent publications providing perspectives of patients with amyloidosis.[5,6]
- In the absence of a robust number of publications in rare diseases, patients and caregivers often become experts in their rare disease.[7,8]
- Regulatory agencies are increasingly seeking patient experience data (PED), in addition to clinical and statistical data from trials, as they make decisions on the utility of drugs and interventions, especially in rare diseases such as amyloidosis.[9-11]
- Consequently, it is becoming essential that patient and caregiver voices are heard and incorporated as best practices in delivering health care.

## OBJECTIVE

- To field a pilot survey as the first step in developing a validated survey that captures PED for patients with amyloidosis.

## METHODS

- Based on prior research, we developed a 62-question pilot survey to better understand patients' experiences with amyloidosis.
- The survey focused on 3 categories affected by amyloidosis:
  - Diagnostic journey
  - Interpersonal relationships
  - Attitude toward themselves
- The survey was designed with 16 sets of questions to determine internal consistency of responses by each responder.
- The survey was fielded for 1 week from January 6-12, 2022, inclusive, on www.oneAMYLOIDOSISvoice.com, an online community of patients, advocates, caregivers, researchers, physicians, and other stakeholders.
- The survey was limited to self-identified patients with amyloidosis.
  - Responses from those who completed <50% of the survey were censored as they could possibly skew the data.
- The data are reported as descriptive statistics.

## RESULTS

- The self-reported demographics, diagnosis, and disease characteristics of the responding patients are given in **Table 1**.

**Table 1. Self-Reported Patient Characteristics**

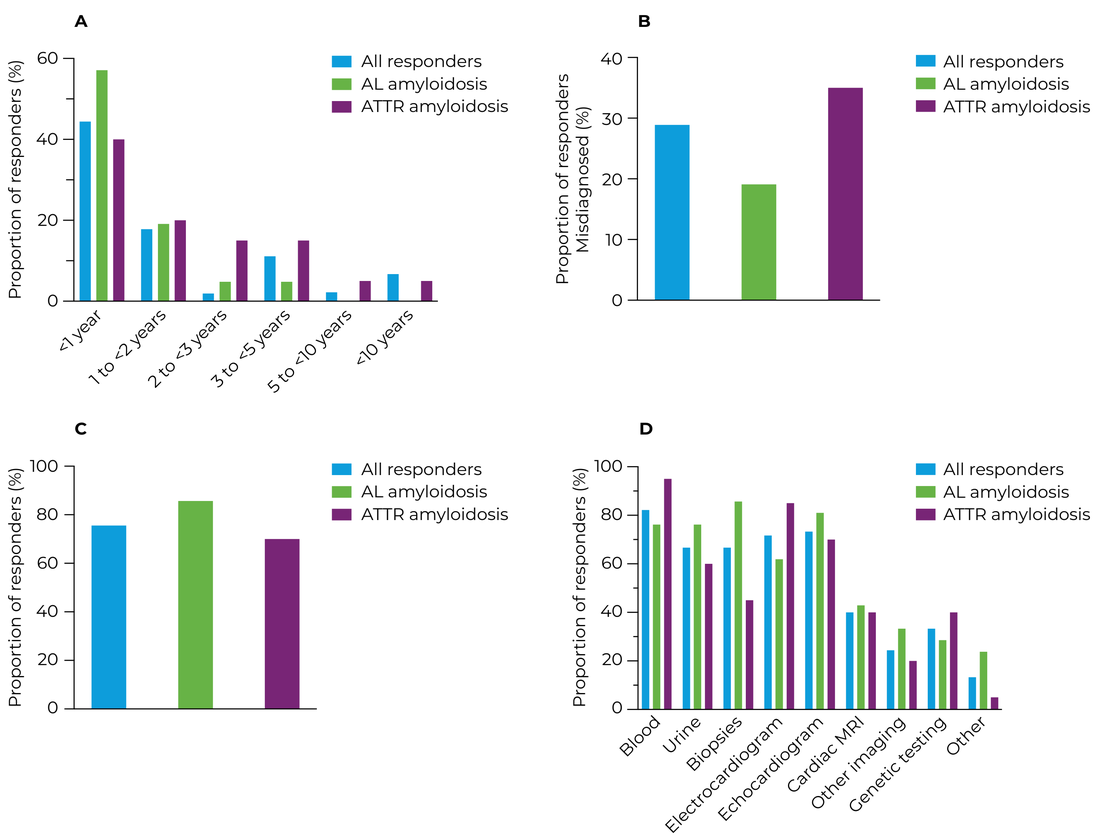
| | Total (N = 45) | AL amyloidosis (n = 21) | ATTR amyloidosis (n = 20) |
|---|---|---|---|
| Median age category at diagnosis, y | 61-80 | 61-80 | 61-80 |
| Type of amyloidosis, n (%) | | | |
| AL amyloidosis | 21 (46.7) | 21 (100) | |
| ATTR amyloidosis | 20 (44.4) | | 20 (100) |
| Other | 4 (8.9) | | |
| Family history of amyloidosis, n (%) | 6 (13.3) | 0 | 5 (25.0) |
| Comorbidities, n (%) | 30 (66.7) | 11 (52.4) | 16 (80.0) |

Abbreviations: AL, light-chain; ATTR, transthyretin amyloidosis.

### DIAGNOSTIC JOURNEY

- Although 28 of 45 (62.2%) patients in our survey were diagnosed in <2 years, there were 3 (6.7%) patients for whom it took more than 10 years (**Figure 1A**).
- Overall, 13 of 45 (28.9%) of patients reported being misdiagnosed prior to correct diagnosis (**Figure 1B**).
- More than 75% of patients reported requiring multiple tests prior to diagnosis (**Figure 1C**).
  - However, only 15 of 45 (33%) of patients reported disliking having to undergo multiple testing.
- The most common tests experienced were blood tests (82.2%), echocardiograms (73.3%), electrocardiograms (71.1%), and urine tests and biopsies (both 66.7%; **Figure 1D**).
  - Biopsies were more common among patients with AL amyloidosis and electrocardiograms were more common among patients with ATTR amyloidosis.

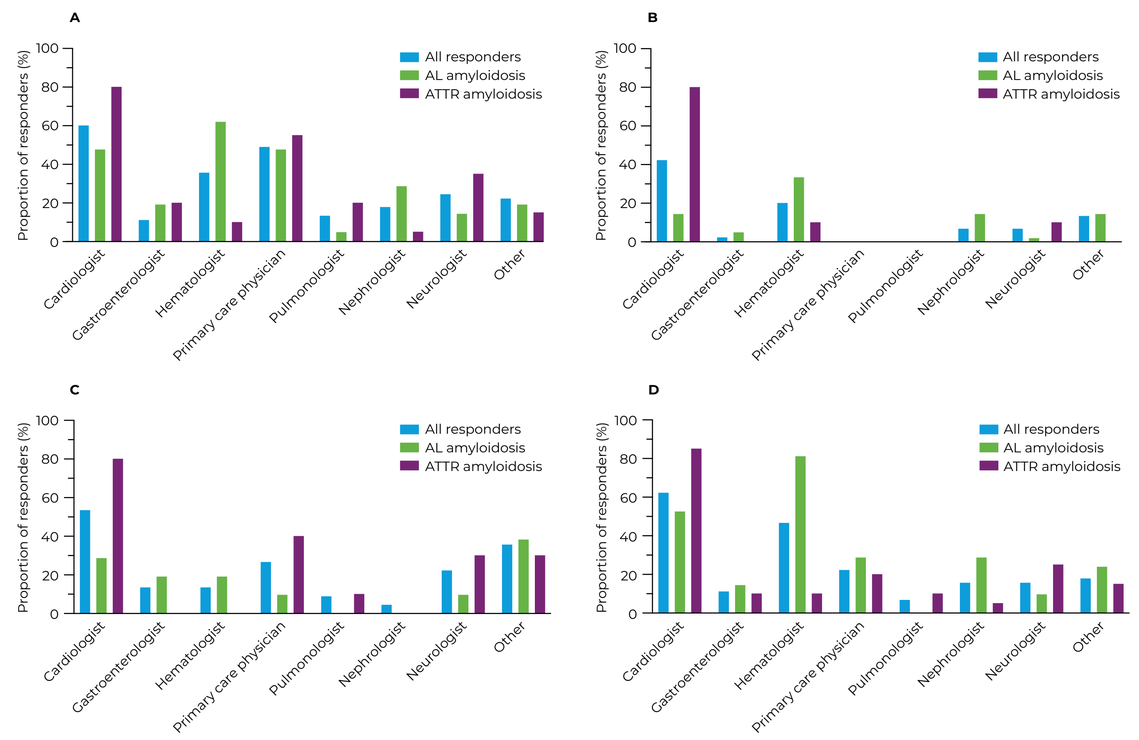
**Figure 1. Diagnostic Journey: (A) Time to Diagnosis, (B) Misdiagnoses, (C) Multiple Testing, and (D) Types of Tests**




Abbreviations: AL, light-chain; ATTR, transthyretin amyloidosis; MRI, magnetic resonance imaging.

- Overall, 27 of 45 (60.0%) patients reported visiting a cardiologist prior to diagnosis, followed by primary care physicians (48.9%) and hematologists (35.6%; **Figure 2A**).
- Overall, cardiologists were identified by 19 of 45 (42.2%) patients as most often determining their amyloidosis diagnosis, followed by hematologists (**Figure 2B**).
  - 33% of patients with AL amyloidosis identified hematologists as the most often diagnosing physicians, whereas 80% of patients with ATTR amyloidosis identified a cardiologist (**Figure 2B**).
- Interestingly, cardiologists also were identified as the leading specialist to misdiagnose amyloidosis (**Figure 2C**).

**Figure 2. Physicians Involved: (A) Prior to Diagnosis, (B) in Correctly Diagnosing, (C) in Misdiagnosing, and (D) in Treating Amyloidosis**




### TREATMENT

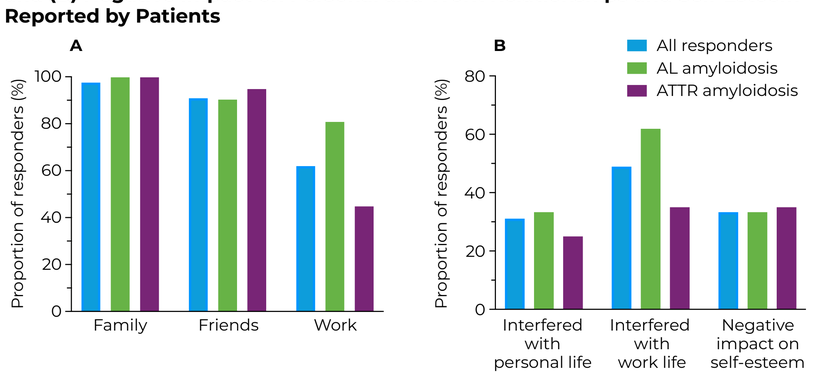
- Overall, 29 of 45 (64.4%) patients were being treated by up to 3 physicians. However, 6 patients reported receiving treatment from ≥6 specialists, with 2 patients receiving treatment from >10 specialists.
- Cardiologists (62.2%) and hematologists (46.7%) were the 2 most common specialists involved in treating patients in our survey (**Figure 2D**).

### PSYCHOSOCIAL IMPACT

- Overall, only 8 of 45 (17.8%) patients were relieved to receive the diagnosis.
- While almost all patients reported being willing to inform their family and friends, a substantial proportion of patients did not inform their coworkers of their diagnosis (**Figure 3A**).
- A substantial proportion of patients reported negative impact on their self-esteem and in their personal and work relationships (**Figure 3B**).

**IT IS IMPORTANT TO UNDERSTAND THE PSYCHOSOCIAL IMPACT OF AMYLOIDOSIS ON PATIENTS AND THEIR CAREGIVERS FROM THEIR PERSPECTIVE**

**Figure 3. A) Proportion of Patients Informing Others About Their Diagnosis and (B) Negative Impact on Personal and Work Relationships and Self-Esteem Reported by Patients**




- Although most patients reported having a supportive family and that the diagnosis strengthened family relationships, there were concerns nevertheless, especially regarding passing on the disease to their children (**Table 2**).
- Most patients reported having supportive friends and that their relationship did not change due to a diagnosis of amyloidosis (**Table 3**).
  - Nevertheless, some patients reported that the disease has made it difficult for them to have fun as they used to.
- Although those patients who informed their coworkers of their diagnosis indicated a supportive work environment, 35.6% chose not to inform them (**Table 3**).
  - A small proportion of patients were also concerned about losing their jobs.

**Table 2. Impact on Relationships With Family**

| Feelings reported by patients | Total (N = 45) | AL amyloidosis (n = 21) | ATTR amyloidosis (n = 20) |
|---|---|---|---|
| Response to diagnosis, n (%) | | | |
| Relieved | 8 (17.8) | 3 (14.3) | 3 (15) |
| Sad or unhappy | 19 (42.2) | 11 (52.4) | 8 (40) |
| Family relationships, n (%) | | | |
| Relieved to inform family | 17 (37.8) | 8 (38.1) | 8 (40.0) |
| Supportive family | 25 (55.6) | 12 (57.1) | 13 (65.0) |
| Worried or stressed informing | 5 (11.1) | 4 (19.1) | 1 (5.0) |
| Worried passing disease to children | 7 (15.6) | 3 (14.3) | 3 (15) |
| Worried to be a financial burden | 1 (2.2) | 1 (4.8) | 0 |
| Felt guilty or ashamed | 1 (2.2) | 0 | 1 (5) |

Abbreviations: AL, light-chain; ATTR, transthyretin amyloidosis.

**Table 3. Impact on Relationships With Friends and Coworkers**

| Feelings reported by patients | Total (N = 45) | AL amyloidosis (n = 21) | ATTR amyloidosis (n = 20) |
|---|---|---|---|
| Relationships with friends, n (%) | | | |
| Relieved to inform friends | 14 (31.1) | 9 (42.9) | 5 (25.0) |
| Supportive friends | 23 (51.1) | 12 (57.1) | 11 (55.0) |
| Worried or stressed informing | 2 (4.4) | 1 (4.8) | 1 (5.0) |
| Felt guilty or ashamed | 0 | 0 | 0 |
| Relationships with coworkers, n (%) | | | |
| Relieved to inform coworkers | 8 (17.8) | 6 (28.6) | 2 (10.0) |
| Supportive coworkers | 8 (17.8) | 5 (23.8) | 3 (15.0) |
| Worried or stressed informing | 2 (4.4) | 2 (9.5) | 0 |
| Worried about losing job | 3 (6.7) | 1 (4.8) | 1 (5.0) |

Abbreviations: AL, light-chain; ATTR, transthyretin amyloidosis.

## DISCUSSION AND CONCLUSIONS

- Our data on the diagnostic journey are consistent with previously reported data on patients with AL and ATTR amyloidosis.[12,13]
  - There seems to be a consistent pattern emerging from the different studies and surveys that awareness and suspicion of amyloidosis among cardiologists needs to be improved.
- Despite the diagnostic journey, <10% of patients reported being relieved to receive a diagnosis.
- Although patients were comfortable sharing their diagnosis with family and friends, more than one-third reported not sharing it with their coworkers.
  - These data potentially suggest a lack of confidence in obtaining support in their work environment, as exemplified by patients expressing a fear of losing their jobs.
- Understandably, some patients were worried that they would pass on or had passed on the disease to their children.
- Financial burden and feelings of guilt were factors in family relationships, but not in relationships with friends or coworkers.
- Understanding the psychosocial impact of amyloidosis, as with other rare diseases, on patients and their caregivers from their perspectives is equally as important as understanding the clinical outcomes of the disease for both humanitarian reasons and commercial success.

### ACKNOWLEDGMENTS

The authors thank the patients who took the time to respond to this survey.

### DISCLOSURES

The authors have no conflicts to declare.

Presented at the XVIII International Symposium on Amyloidosis, 4th – 8th September 2022, Heidelberg, Germany.

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