Patient-reported Diagnostic Journey of Patients Recently Diagnosed with Light Chain Amyloidosis: Data from the Amyloidosis Research Consortium's 2022 and 2023 Amyloidosis Community Surveys

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INTRODUCTION / OBJECTIVES

- Light chain amyloidosis (AL) is a progressive, multisystemic disease caused by the buildup of misfolded immunoglobulin light chains.
- AL amyloidosis patients are benefitting from recent advances in treatment and increased disease awareness; however, the time to diagnosis and treatment is often lengthy.
- This analysis examined the diagnostic journey of recently diagnosed AL amyloidosis patients in a cross-sectional study.

MATERIAL / METHODS

- In 2022 and 2023, the Amyloidosis Research Consortium conducted online multi-country surveys of amyloidosis patients.
- Data collected included demographics, clinical characteristics, and diagnostic journey.
- Patients who responded to the 2022 survey were considered recently diagnosed if they reported receiving an AL amyloidosis diagnosis between Jan 2021-May 2022.
- Patients who responded to the 2023 survey were considered recently diagnosed if they reported receiving an AL amyloidosis diagnosis between diagnosis from June 2022 – November 2023.

RESULTS

- Of 846 total respondents with AL amyloidosis, 112 (13%) were recently diagnosed with AL amyloidosis (59 and 53 patients in the 2022 and 2023 surveys, respectively).
- The majority of patients lived in the United States, 41 (69%) patients in 2022 and 45 (85%) patients in the 2023 survey (Table 1).
- Heart impact was reported among 40 [68%] and 33 (62%) patients in 2022 and 2023, respectively; kidney involvement was reported among 32 (54%) and 34 (64%) patients in 2023 and 2023, respectively.
- The most common symptoms appearing prior to diagnosis of AL amyloidosis were cardiac, specifically fatigue, shortness of breath and swelling in ankles/feet (Figure 1).
- Fatigue was reported in 28 (47%) and 26 (49%) of patients in the 2022 and 2023 survey, respectively. Shortness of breath was reported in 25 (42%) and 23 (43%) patients in 2022 and 2023, respectively, and swelling of legs/ankles was reported in 21 (36%) and 19 (36%) [patients in 2022 and 2023, respectively.
- Treatment was started within a month of diagnosis for approximately half of patients in the 2022 survey (29 [52%]) and over two-thirds in the 2023 survey (35 [69%]). (Figure 2).
- Mean (Standard Deviation [SD]) time from symptom onset to diagnosis was 1.7 (4.1) and 1.7 (2.4) years in 2022 survey and 2023 survey, respectively. (Table 2).
- The number of patients seeing more than 2 physicians before being diagnosed with AL amyloidosis was 35 (64%) and 30 (59%) in 2022 survey and 2023 survey, respectively.

Patients with AL amyloidosis experience a variety of symptoms before being diagnosed which can be years after initial onset.

These annual findings suggest the journey of patients to a diagnosis of AL amyloidosis remains unchanged in both the average time from symptom onset to a diagnosis and the number of physicians seen in search of a diagnosis. However, the time to initiate treatment after diagnosis may be improving.

RESULTS (continued)

Table 1: Patient Demographic and Clinical Characteristics of Patients Recently Diagnosed with AL Amyloidosis by Survey Year

	2022 Survey, N = 59	2023 Survey, N = 53
Region of Residence		
Asia-Pacific	**9** (15%)	**1** (1.9%)
Europe	**6** (10%)	**4** (7.5%)
North America	**44** (75%)	**48** (91%)
Live in United States	**41** (69%)	**45** (85%)
Gender (Male)	**31** (53%)	**22** (42%)
Age, mean (SD)	**66.32** (10.82)	**64.58** (10.69)
Race/Ethnicity		
Asian	**2** (3.4%)	**3** (5.7%)
Black or African	**2** (3.4%)	**0** (0%)
Hispanic or Latino	**0** (0%)	**3** (5.8%)
White	**55** (93%)	**47** (89%)
College Education	**42** (71%)	**36** (69%)
Work Status (Retired)	**40** (68%)	**28** (53%)
Organ Involvement		
Heart	**40** (68%)	**33** (62%)
Kidney	**32** (54%)	**34** (64%)
Nervous System	**15** (25%)	**17** (32%)
Gastrointestinal System	**18** (31%)	**12** (23%)

RESULTS (continued)

Figure 1: Initial Presenting Symptoms Prior to AL Amyloidosis Diagnosis in 2022 and 2023 Survey Years

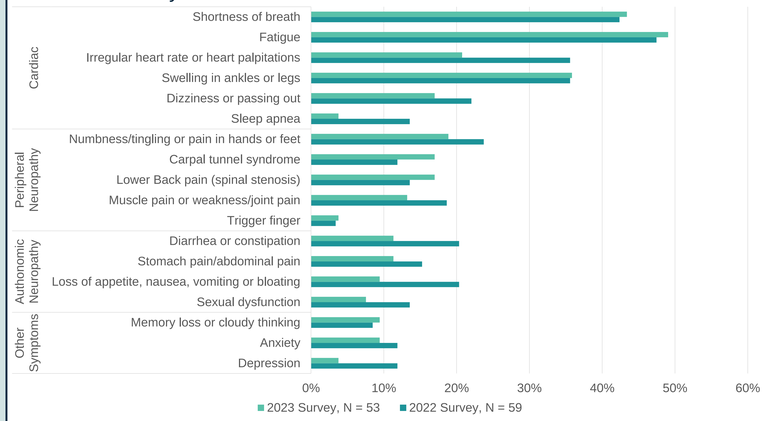

Figure 2: Time to Start Amyloidosis Treatment After AL Amyloidosis Diagnosis in 2022 and 2023 Survey Years

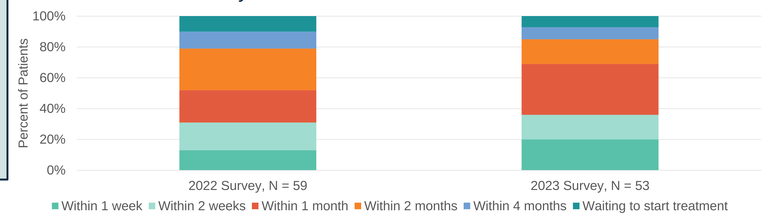

Table 2: Journey to Diagnosis of AL Amyloidosis in 2022 and 2023 Survey Years

	2022 Survey, N = 59	2023 Survey, N = 53
Years from Symptom Onset to Diagnosis, mean (SD)	**1.70** (4.05)	**1.70** (2.41)
Age at Diagnosis, mean (SD)	**65.54** (10.82)	**64.00** (10.80)
Number of MDs Seen Before Diagnosis		
1	**12** (22%)	**9** (18%)
2	**8** (15%)	**12** (24%)
More than 2	**35** (64%)	**30** (59%)
Imaging Performed as Part of Diagnostic Workup	**35** (59%)	**26** (49%)
Biopsy Performed as Part of Diagnositc Workup	**31** (53%)	**21** (40%)
Diagnosing Physician		
Cardiologist	**9** (16%)	**8** (16%)
Hematologist/Oncologist	**22** (40%)	**16** (32%)
Other (Nephrologist, Gastroenterologist, Orthopedist, Primary Care, etc)	**24** (44%)	**26** (52%)

SUMMARY / CONCLUSION

- These annual findings suggest that while average time from symptom onset to diagnosis of AL amyloidosis remains unchanged, average time to initiation of treatment after diagnosis may be improving.
- Further research is needed to confirm this trend and to include all demographic groups.

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