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

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INTRODUCTION / OBJECTIVES

- Transthyretin amyloidosis (ATTR) is a progressive, multisystem disease caused by gene variants (ATTRv) or age-related wild-type transthyretin (ATTRwt) amyloidosis.
- Patients are benefitting from recent advances in treatment and increased disease awareness; however, their diagnostic journey is often lengthy and challenging.
- This analysis examined the diagnostic journey of patients recently diagnosed with ATTR amyloidosis in a cross-sectional study.

MATERIAL / METHODS

- In 2022 and 2023, the Amyloidosis Research Consortium (ARC) conducted multi-country online surveys of amyloidosis patients.
- Data collected included demographic, clinical characteristics, symptoms, and diagnostic journey.
- Patients were categorized as newly diagnosed with ATTRv or ATTRwt if they reported receiving a diagnosis between January 2021 – May 2022 in the 2022 survey or between June 2022 – November 2023 in the 2023 survey.

RESULTS

- Of the 1025 total respondents with ATTR amyloidosis, 228 (22%) were recently diagnosed (115 and 113 patients in the 2022 and 2023 surveys, respectively).
- Of those, 176 (77%) were diagnosed with ATTRwt amyloidosis and 52 (23%) with ATTRv amyloidosis. (Table 1).
- Mean (Standard Deviation [SD]) time from symptom onset to ATTRv amyloidosis diagnosis was 3.3 (3.2) and 2.2 (3.2) years in the 2022 cohort and 2023 cohort, respectively.
- Following a similar trend, the mean (SD) time from symptom onset to ATTRwt amyloidosis diagnosis was 3.3 (6.6) and 2.4 (3.4) years in the 2022 cohort and 2023 cohort, respectively.
- Less than 10% of ATTRwt patients in both 2022 and 2023 surveys were not currently on amyloidosis treatment (Figure 1).
- The majority of ATTRv patients were also on treatment; 5 (20%) and 4 (15%) patients were not currently on treatment in 2022 and 2023, respectively.
- Among ATTRwt patients, the most common cardiac symptoms appearing prior to diagnosis were fatigue, shortness of breath and irregular heart rate or heart palpitations in 2022 and 2023 (Figure 2).
- Lower back pain (spinal stenosis) appeared prior to diagnosis in 20 (13%) and 28 (21%) ATTRwt patients in 2022 and 2023, respectively.
- Over half of ATTRv patients, 13 (52%) reported numbness/tingling or pain in hands or feet prior to diagnosis in the 2022 survey; in 2023, it was reported among 8 (30%) of patients prior to diagnosis.
- The second most common initial symptom prior to diagnosis of ATTRv amyloidosis was fatigue and was reported in 4 (16%) and 11 (41%) patients with ATTRv amyloidosis in 2022 and 2023, respectively.

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

These findings suggest that while the time to diagnosis from symptom onset is still lengthy, it may be improving for ATTR amyloidosis patients.

RESULTS (continued)

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

	ATTRv Patients		ATTRwt Patients	
	2022 Survey, N = 25	2023 Survey, N = 27	2022 Survey, N = 90	2023 Survey, N = 86
Region				
Africa	**0** (0%)	**0** (0%)	**0** (0%)	**1** (1.2%)
Asia-Pacific	**1** (4.0%)	**4** (15%)	**2** (2.2%)	**2** (2.3%)
Europe	**6** (24%)	**2** (7.4%)	**2** (2.2%)	**3** (3.5%)
North America	**18** (72%)	**20** (74%)	**86** (96%)	**80** (93%)
South America	**0** (0%)	**1** (3.7%)	**0** (0%)	**0** (0%)
Live in United States	**18** (72%)	**19** (70%)	**83** (92%)	**79** (92%)
Gender (Male)	**12** (48%)	**15** (56%)	**82** (92%)	**78** (91%)
Age, mean (SD)	**67.92** (9.92)	**62.44** (11.97)	**77.68** (8.10)	**76.12** (7.36)
White Race	**22** (88%)	**21** (78%)	**87** (97%)	**85** (99%)
College Education	**19** (76%)	**15** (56%)	**75** (83%)	**71** (83%)
Retired	**14** (56%)	**15** (56%)	**74** (82%)	**65** (76%)
Time from Symptom Onset to Diagnosis, mean (SD)	**3.27** (3.19)	**2.21** (3.16)	**3.31** (6.57)	**2.41** (3.40)
Heart Impacted by Amyloidosis	**18** (72%)	**22** (81%)	**84** (93%)	**78** (91%)
Nervous System Impacted by Amyloidosis	**19** (76%)	**21** (78%)	**21** (23%)	**29** (34%)
Gastrointestinal System Impacted by Amyloidosis	**7** (28%)	**13** (48%)	**8** (8.9%)	**11** (13%)
TTR Genetic Variant				
T60A	**4** (17%)	**11** (42%)	-	-
V30M	**7** (29%)	**1** (3.8%)	-	-
V122I	**3** (13%)	**2** (7.7%)	-	-
Not sure	**6** (25%)	**4** (15%)	-	-
Not typed	**0** (0%)	**1** (3.8%)	-	-
Other	**4** (17%)	**7** (27%)	-	-

RESULTS (continued)

Figure 1: Current Treatment of ATTRv and ATTRwt Amyloidosis Patients Recently Diagnosed by Survey Year

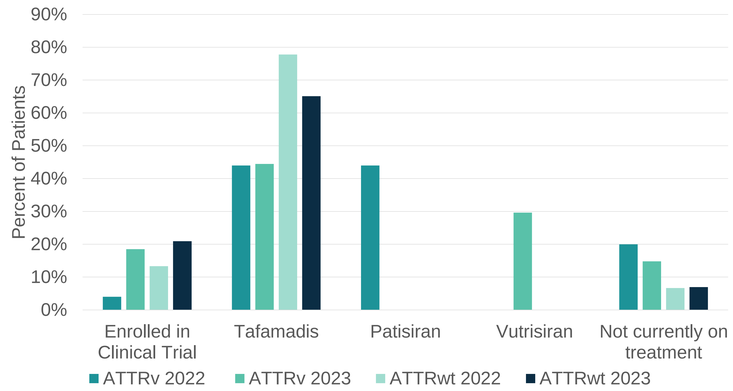
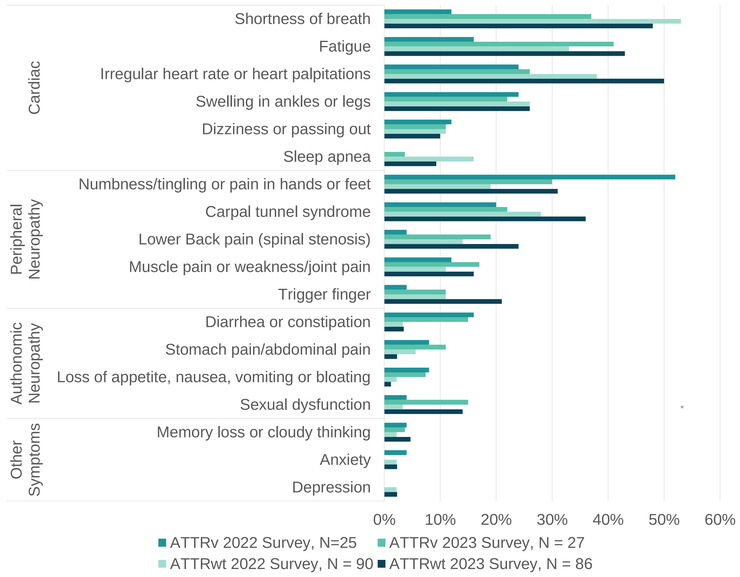


Figure 2: Number of Patients with Initial Presenting Symptoms Prior to Diagnosis of ATTR Amyloidosis in the 2022 and 2023 Survey



SUMMARY / CONCLUSION

- Further studies are needed to evaluate the impact of increased disease awareness on earlier diagnosis and treatment interventions on patient outcomes.

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