

Patient and family experience of ATTR Amyloidosis: Results of two focus groups

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Background:

Patients with transthyretin amyloidosis (ATTR) manage a chronic, life-threatening condition that severely affects their quality of life. Although the primary symptoms and diagnostic criteria for transthyretin amyloid cardiomyopathy (ATTR-CM) and transthyretin amyloid polyneuropathy (ATTR-PN) are well established in the medical literature,¹ very little has been published on the patient and family experiences of these life-altering conditions. Two focus groups of ATTR patients and their family members, one for ATTR-CM and one for ATTR-PN, were convened. Each focus group was asked to describe the diagnostic process, symptoms, and impact on quality of life that they experienced related to their illness.

Methods:

In collaboration with two amyloidosis patient organizations, we convened a focus group for ATTR-CM patients and family members in San Francisco, and a second focus group for ATTR-PN patients and their family members in Buenos Aires. The ATTR-CM group included 4 male patients with wild-type ATTR with cardiomyopathy (ATTRwt-CM), 2 male patients and 1 female patient with hereditary ATTR with cardiomyopathy (ATTR-CM), and family members. The ATTR-PN focus group included 10 patients and 5 family members. No patient-specific identifying data were collected, as such the ages of the participants are not reported. An exemption of IRB approval was issued by the Western Institutional Review Board (IRB). We developed discussion guides for semi-structured focus groups, which queried a number of specific areas including: the path to diagnosis, symptoms experienced, and the impact on patient and family. Participants in each group also participated in an exercise in which they were asked to list the symptoms of ATTR that affected their physical health and quality of life and choose the top three that had the most impact on their lives.

We conducted a content analysis of the transcripts, using techniques described by Miles, et al.² Three themes most closely related to symptoms and quality of life were identified as: 1) diagnostic odyssey; 2) symptoms and impact; 3) the family system.

Results:

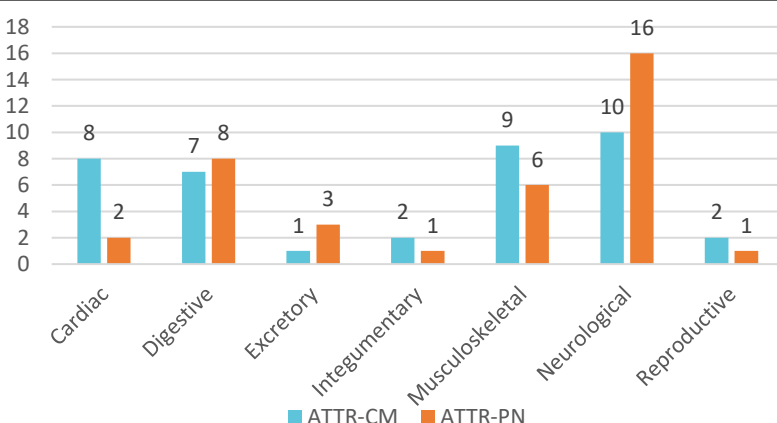
Patients in both ATTR groups often experienced a long and difficult diagnostic odyssey characterized by misdiagnoses, inappropriate treatment and delay in finding the specialized treatment they needed. Collectively, patients with ATTR-CM reported 26 different symptoms and patients with ATTR-PN reported 24 different symptoms. The ATTR-CM group identified intolerance to activity, inability to exercise, insomnia and fatigue as the most impactful symptoms. The ATTR-PN group identified fatigue, gastrointestinal symptoms and sensory symptoms as the most impactful symptoms. The illness was enormously stressful for both patients and their families. Spouses experienced considerable anxiety. Patients with ATTR-PN were stressed not only by the physical effects of the illness, but also by its effects on their relatives, most notably the potential for genetic inheritance in subsequent generations. Yet family members were also a source of coping, motivation, inspiration and support.

Discussion:

To our knowledge, this is the first study that reports on the lived experience of ATTR patients and their families (see also the Amyloidosis Research Consortium Patient-Focused Drug Development Meeting Report).³ Patients with ATTR-CM and ATTR-PN experienced a wide range of debilitating symptoms that had a profound effect on their ability to function and participate in activities of daily life. Patients with ATTR typically underwent a long, confusing and uncertain diagnostic odyssey in which they experienced symptoms with no explanation and inadequate treatment.

The experience reported by these patients and families serves to highlight the multifaceted burden of disease within the entirety of the family unit and its implications. First, more education is needed for primary care physicians and specialists who care for ATTR patients, through the various organ systems impacted (e.g., cardiologists, gastroenterologists, and urologists). This could reduce the likelihood that patients undergo a protracted diagnostic odyssey or at least shorten its duration. Second, patients with ATTR need ready access to mental health services and informal sources of support such as support groups and patient advocates. Third, patient advisory and focus groups are needed to help guide clinical trials and other treatment options. Such groups help hold professionals accountable for intervening in ways most attuned to patients' needs.

ATTR-CM & ATTR-PN Reported Symptoms by Organ System:



Conclusion & Recommendations:

Structured and detailed reporting provided from patients and family members living with and impacted by ATTR is essential for understanding how best to respond to the illness, for developing quality of life measures, and for creating conceptual illness models. A full conceptual model should consist of biological and physiological factors, symptoms, functioning, general health perceptions, and overall quality of life. Patient reporting of the impact of ATTR-related symptoms on their lives and the lives of their families is an important component of such a model. One such conceptual model of light chain amyloidosis (AL) was developed by Lin, et al.⁴ and the current results can form the basis of a model for ATTR. Quality of life measurements in development should be able to account for the most troubling symptoms as reported in these results. Finally, additional studies examining the experience of ATTR patients can help health care providers gain a greater understanding of this still inadequately understood disease and inform institutions about methods to address patients' needs and lessen patients' stress.

Conclusion & Recommendations:

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3. Amyloidosis Research Consortium. *Voice of the Patient.* 2016. 40 p. <https://arci.org/wp-content/uploads/2018/05/Voice-of-the-Patient.pdf>
4. Lin HM, Seldin D, Hui A-M, Berg D, Dietrich CN, Flood E. The patient's perspective on the symptom and everyday life impact of AL amyloidosis. *Amyloid.* 2015 Nov 20;22(4):244-251.