Delays in Diagnosis of Transthyretin Cardiac Amyloidosis after Establishing Cardiac Care

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Background

- Transthyretin cardiac amyloidosis is an underrecognized cause of heart failure.
- With new and evolving treatment options, early diagnosis is critical for patients as degree of cardiac involvement carries worse prognosis.
- The relationship between amyloidosis diagnosis and cardiology initial evaluation has been not been investigated.

Methods

- We identified 70 patients (average age 73.0 years, 10% female, 38.6% black ethnicity) with transthyretin cardiac amyloidosis (CA) seen at our institution (1/2008-11/2019): 36 wild-type amyloidosis (wtATTR), and 34 hereditary amyloidosis (hATTR).
- Of hATTR patients, 74% had the p.Val142Ile mutation.
- Diagnosis was confirmed by cardiac biopsy with mass spectroscopy or technetium pyrophosphate scan.
- Genetic testing was conducted to assess for familial ATTR.
- Charts were reviewed for first clinical contact with cardiology.

Results

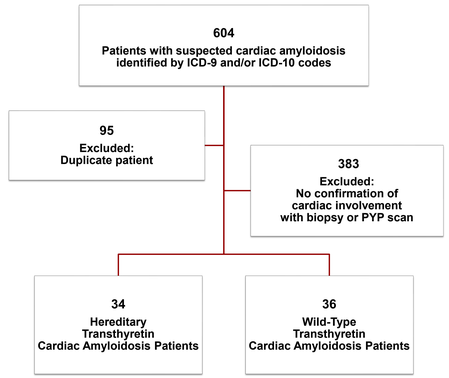

Figure 1: Identification of cardiac amyloidosis patients based on ICD codes: Criterion 1: Diagnosis with wtATTR (E85.82) or hATTR (E85.2), with known cardiac involvement (E85.4). OR Criterion 2: Diagnosis of heart failure (ICD-9: 428.*; ICD-10: I50.*) plus diagnosis of amyloidosis (ICD-9: 277.3*; ICD-10 E85.*).

Cardiac Amyloidosis	hATTR	wtATTR
Number	34	36
Age at diagnosis (years)	70.3	75.5
Female	14.7%	5.5%
Caucasian (%)	26.4%	91.7%
Hypertension	61.8%	69.4%
Hyperlipidemia	58.8%	69.4%
Diabetes Mellitus	23.6%	30.6%
Sleep Apnea	29.4%	33.3%
Obstructive CAD	11.8%	44.4%
Valve Disease	8.8%	22.2%
Arrhythmia	44.1%	69.4%

Figure 2. Baseline characteristics of cardiac amyloidosis patients at time of cardiac amyloidosis diagnosis.

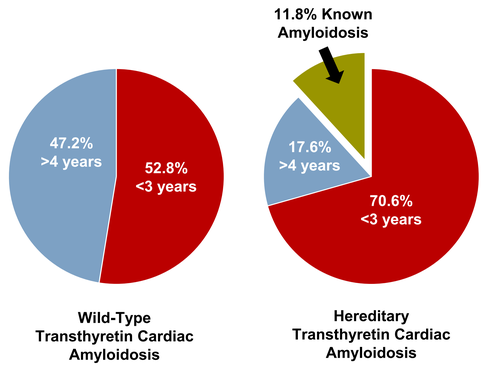

Figure 3. Time range when patients with cardiac amyloidosis initially established care with cardiology prior to diagnosis.

Amyloidosis Type	<3 years of dx	>4 years of dx
Wild-Type Transthyretin	Arrhythmia (36.8%) Dyspnea (31.6%) Heart Failure (21.1%)	CAD (47.1%) Arrhythmia (29.4%) Dyspnea (17.6%)
Hereditary Transthyretin	Dyspnea (54.5%) Heart Failure (27.2%) Edema (18%)	Dyspnea (33.3%)

Figure 4. Most frequent chief complaints at initial cardiology consultation segregated by type of transthyretin amyloidosis and time prior to diagnosis.

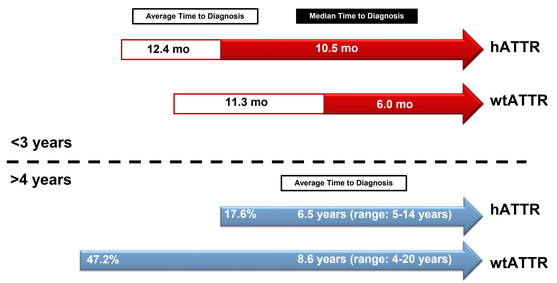

Figure 5. Average time to diagnosis by transthyretin amyloidosis type.

Results

- The majority of transthyretin amyloidosis present to cardiology within three years.
- The most common chief complaint was dyspnea and arrhythmia.
- A quarter of patient presented with heart failure.
- The average time to cardiac amyloidosis diagnosis was 12.4 months for hATTR and 11.3 months for wtATTR.
- Nearly half of patients with wtATTR amyloidosis had established care with cardiology >4 years prior to diagnosis - most often for coronary artery disease.
- Some hATTR family members saw cardiology after a positive genetic test and were found to have cardiac involvement on initial evaluation.

Conclusion

Transthyretin cardiac amyloidosis patients established care with cardiology for months and even years prior to their amyloidosis diagnosis.

Clinical Implementation

This study reveals an opportunity for earlier diagnosis of cardiac amyloidosis by the cardiology field.

Disclosures

Courtney M. Campbell – no financial disclosures