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 worst 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

For wtATTR CA patients, 52.3% established care with cardiology less than 3 years prior to diagnosis. The median time to diagnosis was 6 months (average 11.3 months). The most common complaint was arrhythmia (36.8%), dyspnea (31.6%), and heart failure (21.1%). Cardiology care was established for greater than 4 years (average 8.6 years, range 4-20 years) for 47.2% of wtATTR patients prior to diagnosis. The most common complaints were coronary artery disease (47.1%), arrhythmia (29.4%), and dyspnea (17.6%). For hATTR CA patients, 64.7% patients established cardiology care less than 3 year prior to diagnosis. The median time to diagnosis was 10.5 months (average 12.4 months). The most common complaints were dyspnea (54.5%), heart failure (27.2%), and edema (18%). Cardiology care was established for greater than 4 years (average 6.5 years, range 5-14) for 17.6% of hATTR patients with most common complaint of dyspnea (33.3%). After positive genetic testing, 11.8% hATTR patients established cardiology care based on family member recommendation.

Conclusion

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

Clinical Implications

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