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

• 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

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