**FAMILIAL AMYLOID CARDIOMYOPATHY: A CASE SERIES**

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**Objective:** Familial Amyloid Cardiomyopathy (FAC) is an autosomal dominant but relatively rare condition that results from deposition of mutant and or/ wild transthyretin (TTR) in the heart. Although the Val 122 lle allele has a high prevalence (4%) in African-Americans, the diagnosis is often missed.

**Case 1:** 74 year old woman, presented with shortness of breath, two-pillow orthopnea, and leg swelling of 2 months duration. She had episodes of palpitations. She had been admitted and treated for congestive heart failure and atrial fibrillation in the past. On examination, patient was in mild respiratory distress. There was jugular venous distention; heart sounds were irregular with 3/6 pansystolic murmur with radiation to the apex, loudest in left lower sternal border; lung was clear and patient had 4+ pitting pedal edema. ECG showed atrial fibrillation, left atrial abnormality, low frontal voltage, ventricular premature contractions, long QT and possible anterior and inferior infarction. ECHO revealed speckled appearing myocardium, mild-moderate left atrial enlargement severe TR and interatrial septal thickening. There was diastolic dysfunction & LVH. Patient was diuresed medically optimised. Genetic analysis later revealed Val122Ile mutation in her TTR gene. Abdominal fat biopsy was done but was not conclusive. Patient subsequently had an endomyocardial biopsy which confirmed cardiac amyloidosis.

**Case 2:** 40 year old man (son of cae 1) presents with snoring, daytime fatigue and occasional shortness of breath with exertion. He denied chest discomfort, orthopnea, palpitations or dizziness. He has no significant medical history. Examination findings were within normal limits. ECHO revealed grade I diastolic dysfunction and trivial tricuspid regurgitation. Patient's genetic analysis revealed Val122Ile TTR mutation.

**Conclusion:**With the advent of new therapeutics, FAC associated morbidity and mortality might be reduced especially if the condition is diagnosed early.