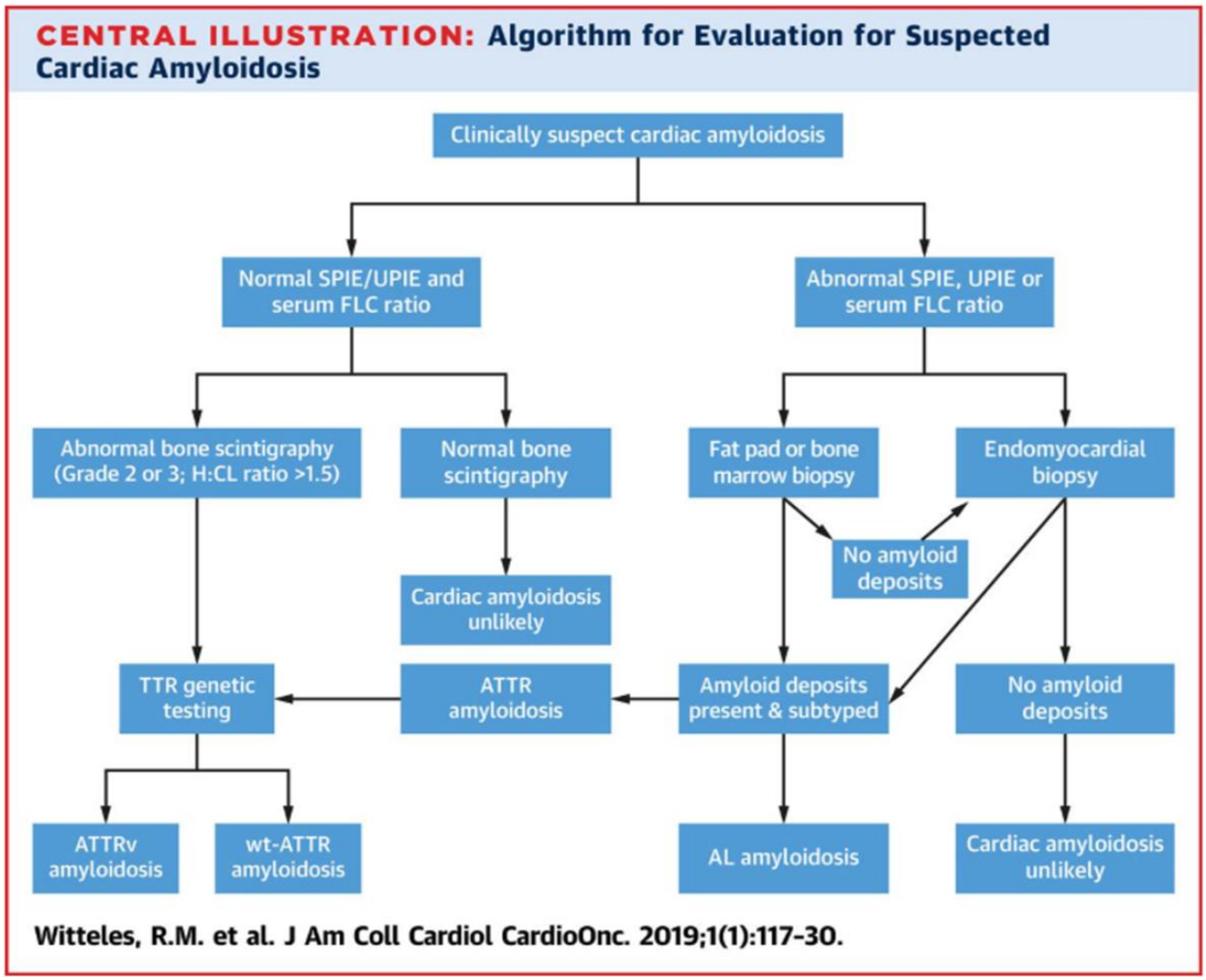
CME

Carpal Tunnel Syndrome and ATTR Cardiomyopathy

Case Presentation by Dr. Rola Khedraki

Transcript edited for brevity and clarity

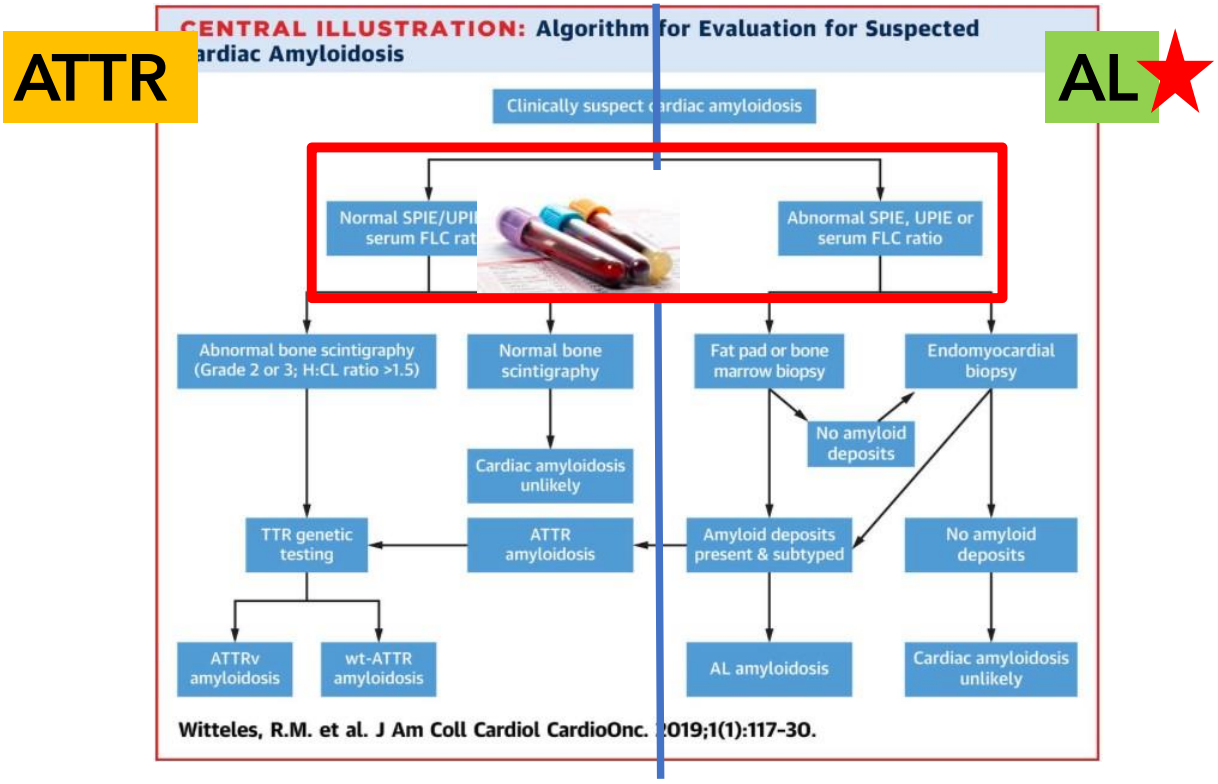


Case 1

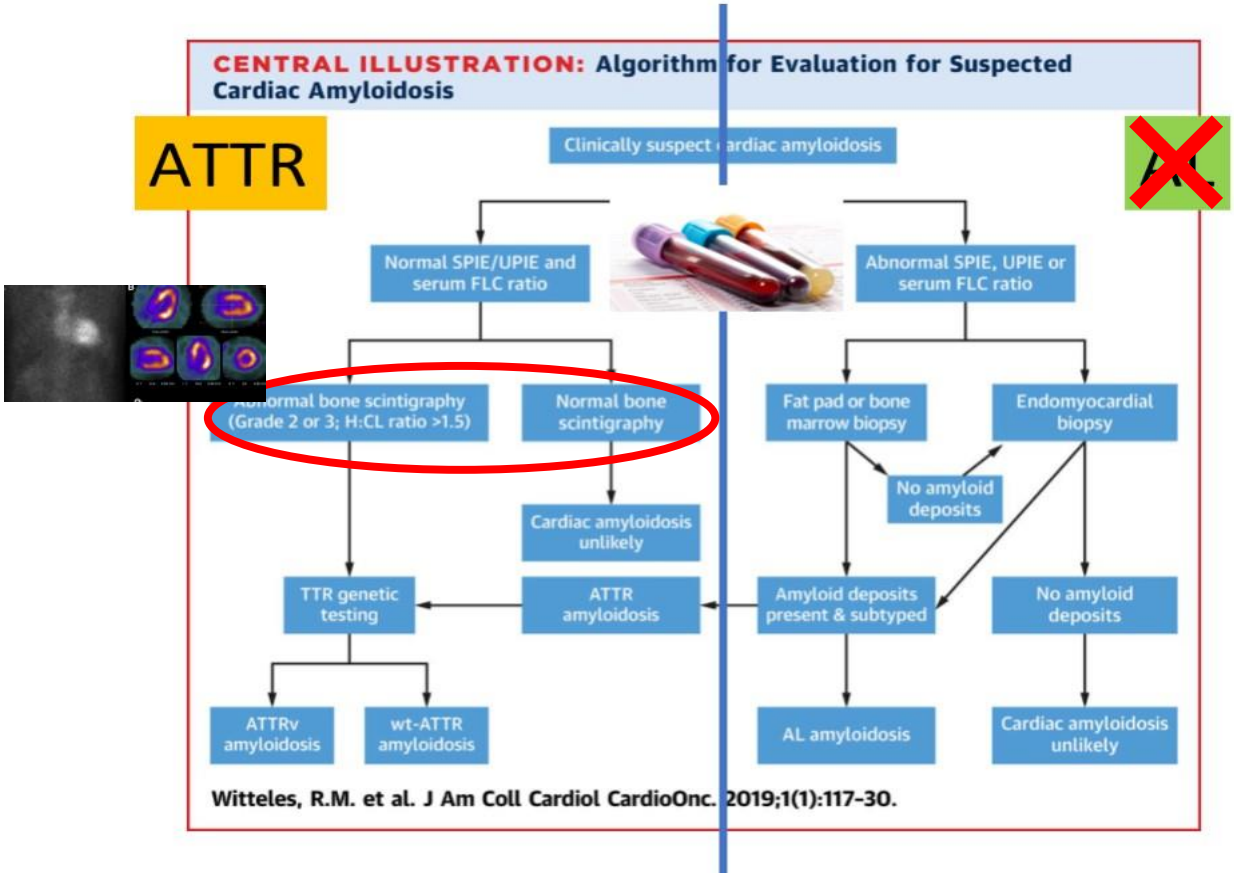
A 52-year-old African American male with bilateral carpal tunnel syndrome and long-standing cardiomyopathy due to "hypertensive heart disease"

Echo: Marked thickening of the walls and classical apical sparing pattern. **EKG**: Poor R-wave progression and a voltage that is out of proportion to the degree of mass seen on echo.

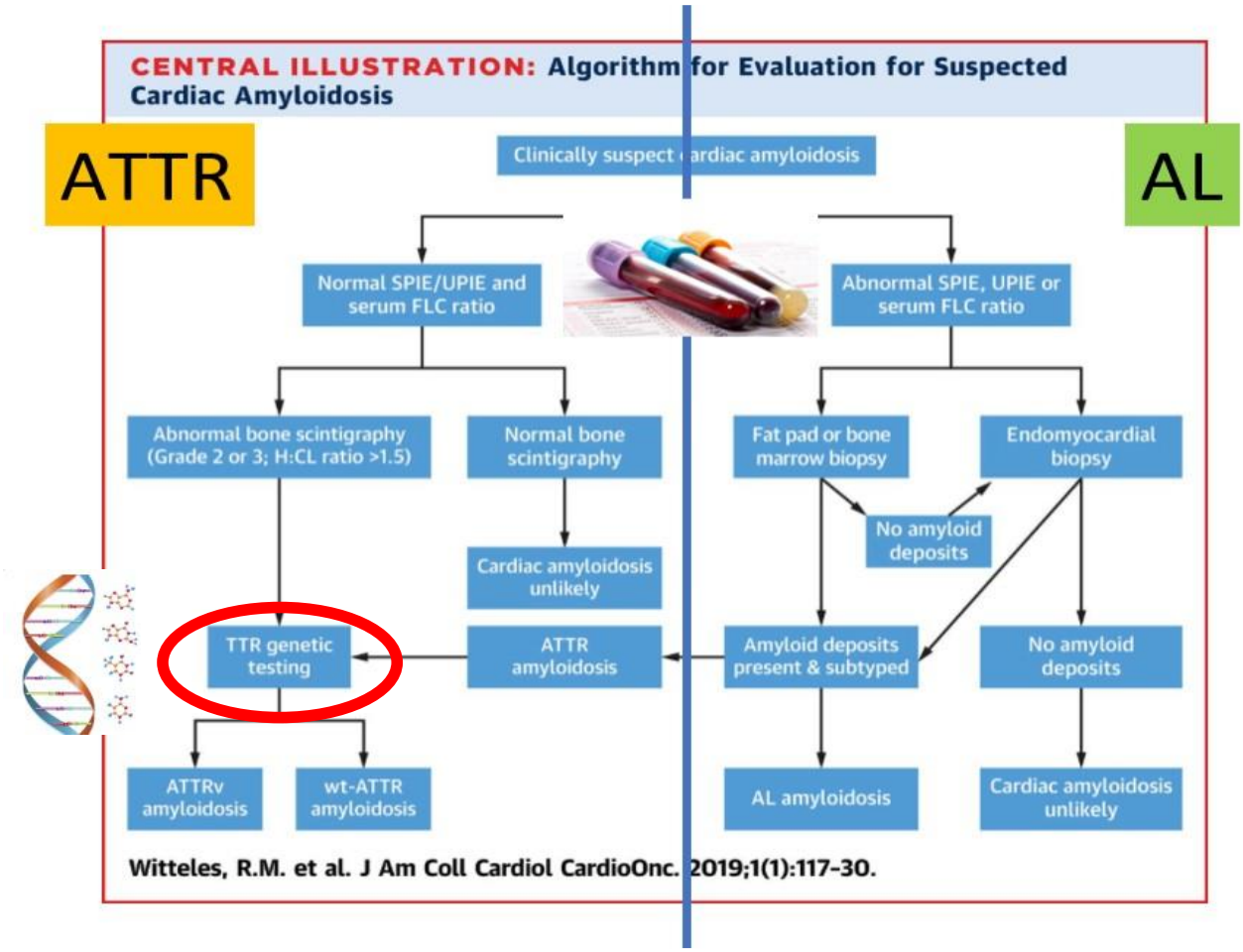
Next step: The clinical suspicion for cardiac amyloidosis was raised based on history and findings from EKG and Echo. The workup of cardiac amyloidosis **always starts with the AL side** (see below). We did **screening labs for AL**. The results came back negative. So, we ruled out AL.



Next step: The next is to do a **bone scintigraphy** or **technetium pyrophosphate scan (PYP scan)**. This is on the ATTR side (see below). This patient did have myocardial uptake. He was diagnosed with ATTR.



Next step: The next is to do **genetic testing** to differentiate between hereditary and wild type of ATTR (see below). Sure enough, this patient had AV1 22 mutation. Remember that this affects 3 to 4% of those of African descent.

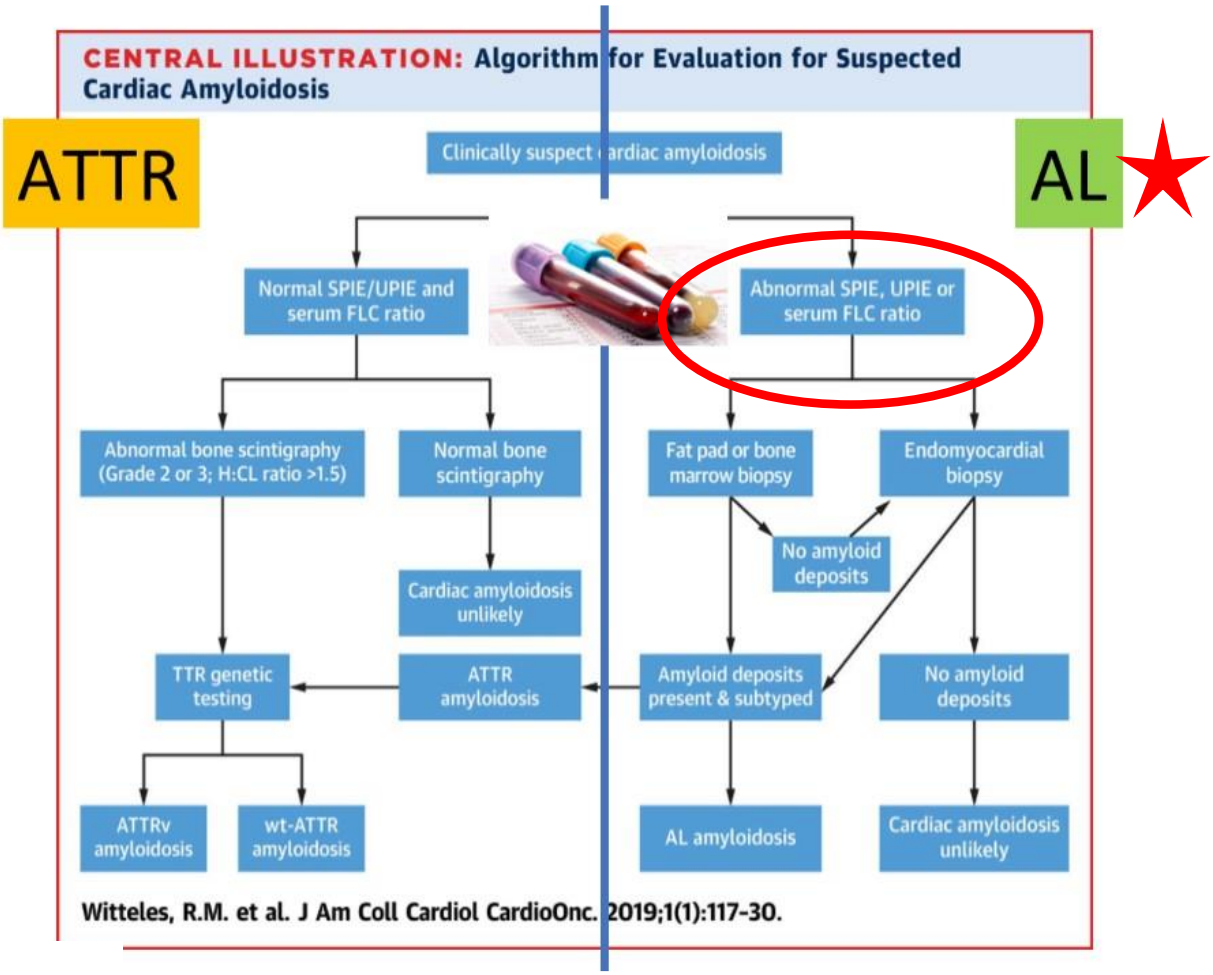


Outcome: Unfortunately, this patient had a delay in his diagnosis because he was assumed to have hypertensive heart disease. It was too late to alter the trajectory of his disease with medications. He ended up having to undergo a heart transplant.

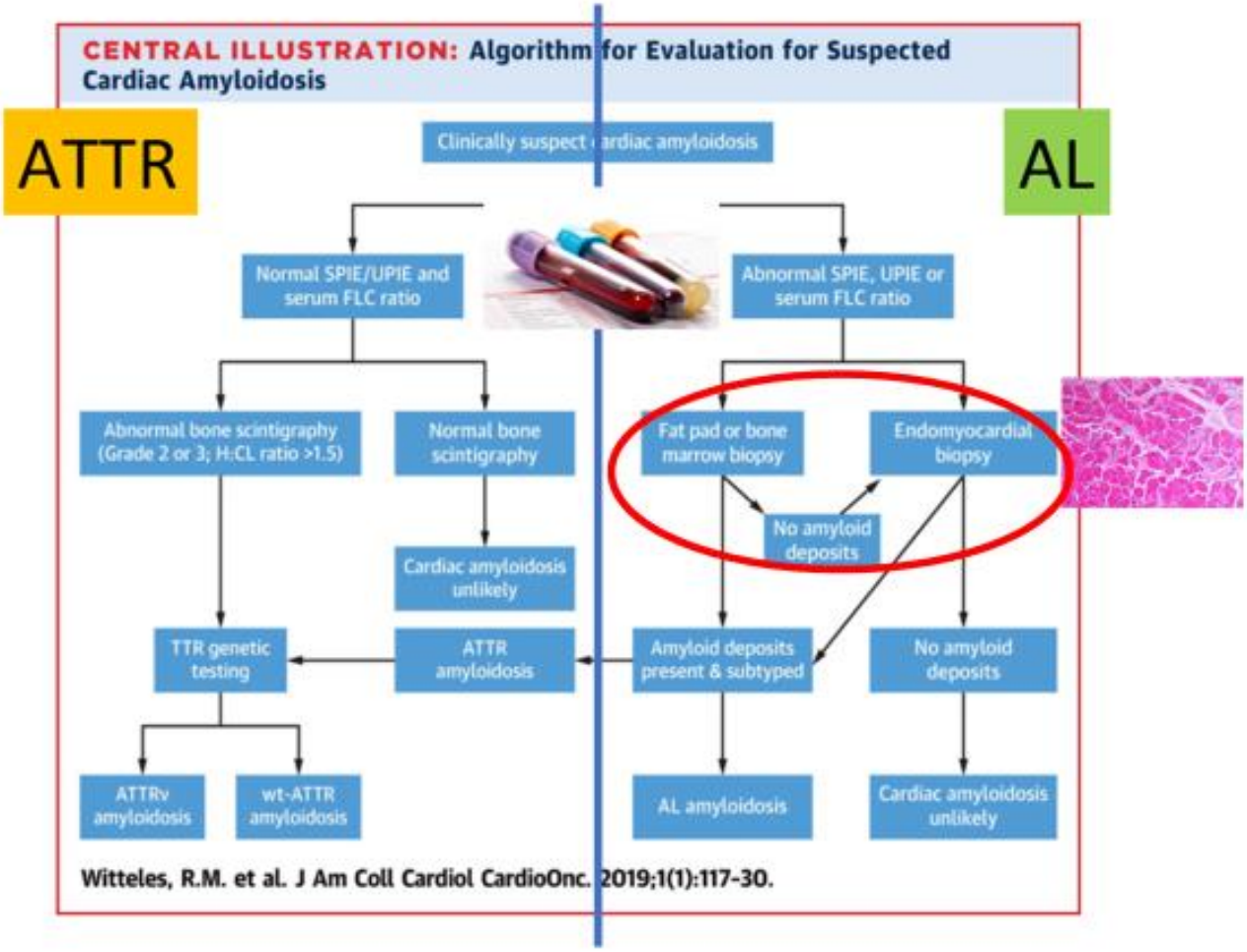
Case 2

A 74-year-old Caucasian male with surgeries for bilateral carpal tunnel syndrome and spinal stenosis. Thickened walls was found on his echo.

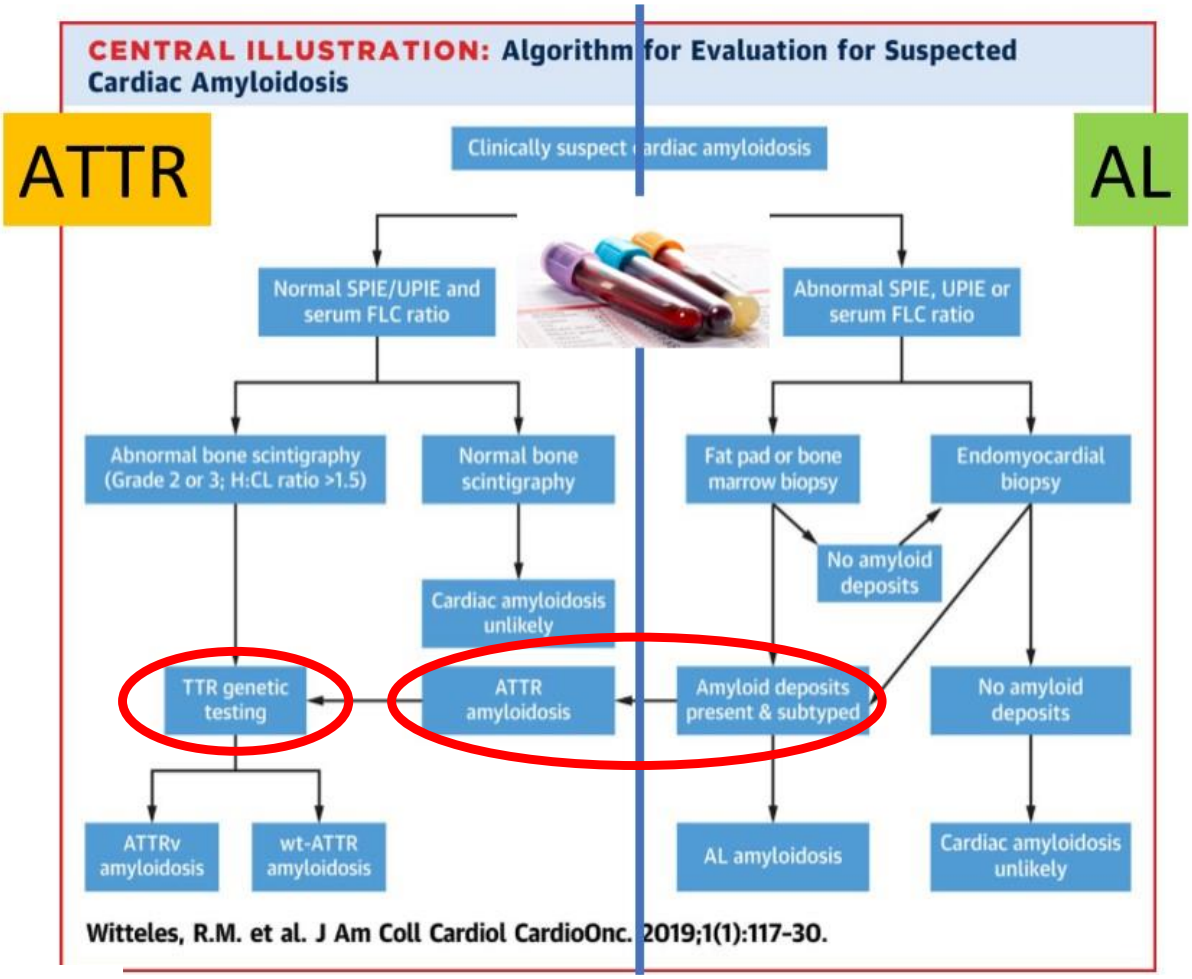
Screening lab for AL: This patient does have elevated Kappa light chains and a Kappa monoclonal protein on his immunofixation. Screening lab for AL is positive.



Next step: Now we want to do a **tissue biopsy** since screening lab is positive. This patient underwent a cardiac biopsy that showed positive amyloid staining (see below). He also underwent a bone marrow biopsy and was found to have incidental finding of Waldenström's.



Next step: We thought that we were going to be on the side of AL, but the positive biopsy actually now pushed us over to the **ATTR side** (see below). **Genetic testing** result was negative for any mutations. He was diagnosed as having wild type cardiac amyloid.


CENTRAL ILLUSTRATION: Algorithm for Evaluation for Suspected Cardiac Amyloidosis
ATTR
AL
Clinically suspect ardiac amyloidosis
Normal SPIE/UPIE and serum FLC ratio
Abnormal SPIE, UPIE or serum FLC ratio
Abnormal bone scintigraphy (Grade 2 or 3; H:CL ratio >1.5)
Normal bone scintigraphy
Fat pad or bone marrow biopsy
Endomyocardial biopsy
No amyloid deposits
Cardiac amyloidosis unlikely
TTR genetic testing
ATTR amyloidosis
Amyloid deposits present & subtyped
No amyloid deposits
ATTRv amyloidosis
wt-ATTR amyloidosis
AL amyloidosis
Cardiac amyloidosis unlikely
Witteles, R.M. et al. J Am Coll Cardiol CardioOnc. 2019;1(1):117-30.