CARDIAC AMYLOIDOSIS in LATIN AMERICA: CASE COMPENDIUM
AMILO-LATAM STEERING COMMITTEE

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• **Case 1:** 76-year-old male, retired, presents with exertional dyspnea NYHA Class II that has worsened in the last 8 months
• **Case 2:** 79-year-old male, no history of heart disease, with dyspnea and periods of chest discomfort over the last year and one recent episode of syncope

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• **Case 3:** 78-year-old male with hypertension and dyslipidemia presents with mild shortness of breath and fatigue
• **Case 4:** 83-year-old female of African American descent complains of increased fatigue, shortness of breath on exertion, and worsening ankle edema

Jorge Camilletti, MD and Juan Erriest, MD
• **Case 5:** 86-year-old male came to the ER following 2 weeks of progressive dyspnea and functional class impairment, currently NYHA Class IV
• **Case 6:** 69-year-old male with decompensated heart failure of unknown cause presents with progressive lower limb edema associated with dyspnea and class functional impairment, NYHA Class III and increased abdominal circumference, decrease in diuretic rate, refractory to oral diuretic treatment

Erick Alexanderson, MD and Isabel Carvajal-Juarez, MD
• **Case 7:** 39-year-old asymptomatic female, carrier of hATTR (Ser50) 10 years ago, treated with tafamidis 20mg/day for the last five years with no cardiac imaging evaluation
• **Case 8:** 77-year-old male with multiple hospitalizations for decompensated heart failure despite optimal treatment, dyspnea with minimal effort, and lower limb edema
<table>
<thead>
<tr>
<th>Abbreviation</th>
<th>Full Form</th>
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<tbody>
<tr>
<td>AF</td>
<td>Atrial fibrillation</td>
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<tr>
<td>AL</td>
<td>Light chain</td>
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<tr>
<td>ANT</td>
<td>Anterior</td>
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<tr>
<td>AS</td>
<td>Aortic stenosis</td>
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<tr>
<td>ASA</td>
<td>Aspirin</td>
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<tr>
<td>ATTR</td>
<td>Transthyretin amyloidosis</td>
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<tr>
<td>ATTR CA</td>
<td>Transthyretin cardiac amyloidosis</td>
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<tr>
<td>AV</td>
<td>Atrioventricular</td>
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<td>CA</td>
<td>Cardiac amyloidosis</td>
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<tr>
<td>CMR</td>
<td>Cardiac magnetic resonance</td>
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<tr>
<td>ECG</td>
<td>Electrocardiogram</td>
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<td>ECHO</td>
<td>Echocardiography</td>
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<td>ECV</td>
<td>Extracellular volume</td>
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<td>EF</td>
<td>Ejection fraction</td>
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<td>ESR</td>
<td>Erythrocyte sedimentation rate</td>
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<td>GLS</td>
<td>Global longitudinal strain</td>
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<td>hATTR</td>
<td>Hereditary transthyretin amyloidosis</td>
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<tr>
<td>Hb</td>
<td>Hemoglobin</td>
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<td>H/CL</td>
<td>Heart to contralateral wall ratio</td>
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<td>HCM</td>
<td>Hypertrophic cardiomyopathy</td>
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<tr>
<td>HF</td>
<td>Heart failure</td>
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<td>HFrEF</td>
<td>Heart failure with preserved EF</td>
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<td>HU</td>
<td>Hounsfield units</td>
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<tr>
<td>JVP</td>
<td>Jugular venous pressure</td>
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<td>LA</td>
<td>Left atrium</td>
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<td>LAO</td>
<td>Left anterior oblique</td>
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<tr>
<td>LL</td>
<td>Left lateral</td>
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<tr>
<td>LV</td>
<td>Left ventricle</td>
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<tr>
<td>LVEF</td>
<td>Left ventricular ejection fraction</td>
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<td>LVH</td>
<td>Left ventricular hypertrophy</td>
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<td>LVMI</td>
<td>Left ventricular mass index</td>
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<tr>
<td>MBq</td>
<td>Megabecquerel</td>
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<tr>
<td>mCI</td>
<td>Millicurie</td>
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<tr>
<td>MGUS</td>
<td>Monoclonal gammopathy of undetermined significance</td>
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<td>NYHA</td>
<td>New York Heart Association functional classification</td>
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<tr>
<td>NT-ProBNP</td>
<td>Amino terminal pro B-type natriuretic peptide</td>
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<td>RA</td>
<td>Right atrium</td>
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<td>RBBB</td>
<td>Right bundle branch block</td>
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<td>RV</td>
<td>Right ventricle</td>
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<tr>
<td>RVEF</td>
<td>Right ventricular ejection fraction</td>
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<tr>
<td>sFLC</td>
<td>Serum free-light chain assay</td>
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<tr>
<td>sIFE</td>
<td>Serum immunofixation electrophoresis</td>
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<tr>
<td>SOB</td>
<td>Shortness of breath</td>
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<tr>
<td>uIFE</td>
<td>Urine immunofixation electrophoresis</td>
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<tr>
<td>TAVI</td>
<td>Transcatheter aortic valve implantation</td>
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CASE 1

Patient History
- 76-year-old male, retired
- Ex-smoker, hypertension treated with enalapril 10 mg/12 h
- Chronic AF treated with rivaroxaban and carvedilol
- Exertional dyspnea NYHA II that has worsened in the last 8 months
- No chest pain; no syncope

ECG
- AF, fast ventricular response (122 bpm)
- Premature or aberrantly conducted ventricular complexes
- RBBB
- Possible anterolateral infarction

2-D Trans Thoracic Echocardiogram
- Severe wall thickness increase (septum 20 mm, posterior wall 19 mm)
- Severe biatrial dilatation
- Abnormal GLS with apical preservation
- Tri leaflet aortic valve, mildly calcified area = 1 cm2
- Severe decrease in volume/beat (18 mL/m2)
- Medium gradient <25 mmHg
- LVEF = 40%
CASE 1 - continued

Conclusion
- Hemodynamically severe AS with low flow/low gradient profile
- Differential diagnosis with severe aortic pseudo-stenosis
- Associated CA is suspected
- Suggested follow-up: calcium score, $^{99m}$Tc-PYP scintigraphy, sFLCs

Coronary Calcium Score
- Aortic valve score = 930 HU (not consistent with severe AS)
- Coronary score = 2211 HU

Reason for Test
ATTR CA in patients with apparently severe AS is highly prevalent
Patient also has several “red flags”:
- Age >65
- Male
- Symptoms of HF (dyspnea)
- Relatively preserved LVEF
- Increased wall thickness
- Abnormal GLS with apical sparing
- No LVH on ECG, AF, RBBB

Imaging Protocol
- $^{99m}$Tc-PYP intravenously (18 mCi = 666 MBq)
- Images 1-h post-injection, dual-head gamma camera
- Thorax planar views: ANT, LAO, LL
  - 256 x 256 matrix, no zoom, >750K cts/view
  - H/CL ratio
- SPECT 360°, detectors in 180°
  - 128 x 128 matrix, no zoom, 120 steps (60/detector), 20 sec/step
  - Iterative reconstruction, no attenuation or scatter correction
Interpretation and Key Points to Report

- Positive $^{99m}$Tc-PYP study
- Intense, diffuse myocardial radiotracer uptake in LV and RV
- No equivocal findings at one hour; imaging at 2–3 hours not performed
- sFLC results normal; AL amyloidosis excluded
  - Kappa = 13.7 mg/L (ref. 3.3 to 19.4 mg/L)
  - Lambda = 22.1 mg/L (ref. 5.7 to 26.3 mg/L)
  - K/L ratio = 0.62 (ref. 0.26 to 1.65)

Final diagnosis: ATTR CA – patient referred for treatment

**Note:** Up to 20% of patients with AL can exhibit myocardial $^{99m}$Tc-PYP uptake (usually Perugini Grade 1-2). Final diagnosis requires exclusion of AL amyloidosis.
CASE 2

Patient History
- 79-year-old male
- No previous history of heart disease
- Dyspnea and episodes of chest discomfort >1 year
- One episode of syncope; referred for evaluation

ECG
- AF, slow ventricular response (55 bpm)
- Possible inferior infarction
- Poor R progression V1–V4

2-D Trans Thoracic Echocardiogram
- Normal LV cavity (LVDD 40 mm)
- Increased wall thickness (S 15 mm, PW 14 mm)
- Severe biatrial dilatation (LA 52 mL/m², RA 50 mL/m²)
- Abnormal GLS with apical sparing
- Severe diastolic dysfunction
- Aortic valve 1.3 cm²
- LVEF = 55%
**Conclusion**
- CA suspected
- Suggested follow-up: biomarkers, CMR, $^{99m}$Tc-PYP scintigraphy, sFLCs

**Cardiac Magnetic Resonance (CMR)**
- Normal LV volume
- LVEF = 58%
- Increased wall thickness
- LV mass 90 g/m² (Normal: 56–89 g/m²)
- Severe biatrial dilatation
- Diffuse subendocardial late gadolinium enhancement
- Mild RV dysfunction

![CMR Image](image)

**Reason for Test**
- Male >65 years old
- HFpEF
- ECHO and CMR findings suggestive of ATTR CA
- ECG compatible

**Imaging Protocol**
- $^{99m}$Tc-PYP intravenously (19 mCi = 703 MBq)
- Images at 1- and 3-hours post-injection, dual-head gamma camera
- Thorax planar views: ANT, LL, LAO
- 256 x 256 matrix, no zoom, >750K cts/view
- SPECT 360°, detectors in 180°
- 128 x 128 matrix, no zoom, 120 steps (60/detector), 20 sec/step
- Iterative reconstruction, no attenuation or scatter correction
CASE 2 - continued

Planar Images

1-h post-injection
Perugini Grade 3

3-h post-injection
Perugini Grade 3

H/CL = 1.85 at 1-hr
H/CL = 1.76 at 3-hrs
SPECT Images

**Interpretation and Key Points to Report**

- Positive $^{99m}$Tc-PYP study for ATTR CA provided sFLCs are negative
- There is intense, diffuse uptake in LV walls
- Significant RV uptake also noted
- 3-hour imaging could have been omitted because study was positive at 1 hour, but it was done for a research protocol
- Biomarkers and sFLC results:
  - Troponin T = 43 ng/L (ref. <50 ng/L)
  - Troponin I = 0.0540 ng/mL (ref. <0.0300 ng/mL)
  - BNP = 479.4 pg/mL (ref. 1–400 pg/mL)
  - Kappa = 33.43 mg/L (ref. 3.3–19.4 mg/L)
  - Lambda = 38.24 mg/L (ref. 5.7–26.3 mg/L)
  - $\kappa:\lambda$ ratio = 0.874 (ref. 0.26–1.65)
The patient needs further investigation because of abnormal kappa and lambda values, although $\kappa:\lambda$ ratio is normal.

**Teaching Points**

- Although kappa and lambda are above-referenced values, the K/L ratio is within normal limits, and AL is unlikely.
- Increased kappa and lambda with an average K/L ratio can be due to MGUS, which is a frequent condition in elderly patients and probably not related to amyloidosis.
- $^{99m}$Tc-PYP RV uptake is present in 12–50% of patients with ATTR CA.
- RV uptake has a very high positive predictive value (>97%) for ATTR CA.
- No solid evidence links RV uptake with a worse prognosis.
- Current investigations focus on the relationship between RV uptake and cardiac dysfunction, as well as its prognostic significance.
CASE 3

Patient History
- 78-year-old male, hypertension, and dyslipidemia
- Complains of mild shortness of breath and fatigue

Physical Exam
- No acute distress
- Weight: 82 kg; Height: 172 cm
- BP: 150/90 mmHg; HR = 88 bpm
- No orthostatic hypotension
- Regular rhythm, no murmurs
- No hepatojugular reflux
- No rales
- 1+ bilateral ankle edema
- No evidence of peripheral neuropathy

Doppler Echocardiogram
- LA diameter = 4.5 cm
- EF = 48%
- Septum: 12 mm
- Posterior wall: 12 mm
- Moderate diastolic dysfunction and mild impairment of global systolic function

Laboratory Tests for AL Exclusion
- Urinary protein immunofixation: negative
- Serum protein immunofixation: absence of monoclonal component

Reason for Test
- Red flags: Gender, age, 12mm septum, diastolic dysfunction

Imaging Protocol
- 18 mCi (666 MBq) of $^{99m}$Tc-PYP was injected intravenously
- Three hours after injection, planar and SPECT/CT images of the thorax were acquired
CASE 3 - continued

Study Images

Interpretation and Key Points to Report
- There is diffuse $^{99m}$Tc-PYP uptake in the cardiac area on the planar images.
- SPECT images demonstrate residual blood pool in the LV cavity.
- There is no myocardial uptake.
- Semi-quantitative uptake grade 0.
- $^{99m}$Tc-PYP scan is not suggestive of ATTR CA.

Teaching Points
- Always perform SPECT to differentiate myocardial uptake from residual blood pool activity.
- If no myocardial uptake is present on SPECT, the visual grade is 0 and the scan does not suggest ATTR CA.
CASE 4

Patient History
- 83-year-old female of African American descent
- Previously healthy: ex-smoker
- History of long-lasting hypertension treated with telmisartan 80 mg
- Diagnosed with a poorly differentiated squamous cell lung carcinoma
- Sent to cardiology for pre-op assessment
- Complains of increased fatigue, SOB on exertion, and worsening ankle edema

Physical Exam
- No acute distress
- Weight: 67.8 kg; Height: 160 cm
- BP: 116/70 mmHg; HR = 92 bpm
- No orthostatic hypotension
- Regular rhythm, no murmurs
- Hepatojugular reflux +; estimated JVP: 9 cm/H₂O
- No rales
- 3+ bilateral ankle edema
- No evidence of peripheral neuropathy

Labs and ECG
- NT-ProBNP = 644 pg/mL
- Hb = 12.1 g/dL
- Creatinine: 0.97 mg/dL
- Urea = 49 mg/dL
- Na = 140 mEq/L
- K = 5.3 mEq/L
- HbA1c = 5.5%
- TSH = 1.2 mUI/L
- Free T4 = 1.0 ng/dL
- Low QRS voltage

Labs for AL Exclusion
- Urinary protein immunofixation: negative
- Serum protein immunofixation: absence of monoclonal component
- IgG = 900 mg/dL (700–1600 mg/dL normal)
- IgA = 283 mg/dL (80–350 mg/dL normal)

Doppler Echocardiogram
- LA diameter = 4.5 cm
- EF = 51%
- Septum: 19 mm
- Posterior wall: 15 mm
- E/e’ relation = 18

Note: Increased parietal thickness with increased refringence, moderate diastolic dysfunction, and mild impairment of global systolic function.
CASE 4 - continued

Cardiac MRI
- End diastolic volume: 179 mL
- End systolic volume: 116 mL
- Ejection volume: 62 mL
- LVEF: 35%.
- T1 map shows altered values (~1087–1115 ms)
- T2 map values are within the normal range (~47–49 ms)
- Extracellular volume calculated at 52%
- Diffuse delayed enhancement compromising the myocardium

Reason for Test
- Red flags: Age, African American descendant, HF symptoms, ECHO and CMR findings

Imaging Protocol
- 18 mCi (666 MBq) of ⁹⁹ᵐTc-PYP injected intravenously
- 3-hours after injection, planar and SPECT/CT images of the thorax were acquired

Study Images

Planar

Recent guidelines consider the H/CL ratio of limited value to evaluate CA. The LAO image shows rib fractures on the right side and these findings can affect the ratio.
Interpretation and Key Points to Report
- Semi-quantitative myocardial uptake grade 3 (greater than rib uptake)
- $^{99m}$Tc-PYP scan is strongly suggestive of ATTR CA

Teaching Points
- Be aware of red flags.
- Discordance between QRS voltage on an ECG and wall thickness on imaging.
- Always exclude monoclonal gammopathy.
- Always perform SPECT or SPECT/CT if available.
- After a $^{99m}$Tc-PYP positive scan, order a genetic test to confirm or exclude hereditary ATTR.

Genetic Testing for TTR Amyloidosis

Second Visit
- Confirmed diagnosis of TTR hereditary amyloidosis.
- Started on tafamidis.
CASE 5

Patient History
- 86-year-old male
- Hypothyroidism
- Hypertension, dyslipidemia
- 2017: Coronary invasive angiography performed without obstructive lesions
- 2019: ECHO: LVEF=68%, LV hypertrophy, no motility disorders
- Medications: levothyroxine, ASA, valsartan, hydrochlorothiazide, rosuvastatin
- 2022: Came to the ER following 2 weeks of progressive dyspnea, functional class impairment, currently NYHA IV

Physical Exam
- BP: 160/100 mmHg
- Heart rate: 88 bpm
- Breathing frequency: 20 cpm
- R1 and R2 present in 4 foci, hypophonic, free silences. Jugular engorgement without inspiratory collapse. Edema in the lower limbs 2/4
- Good respiratory mechanics, slightly decreased vesicular murmur, few bilateral wheezes, and bibasal crackles

ECG
Chest X-ray

- Anterior view—portable X-ray, off-center. Increased cardio thoracic index.
- Prominent aortic button and marked left second cardiac arch. Veiled left and right diaphragmatic costal sinuses (pleural effusion). Congestive lung ileus, signs of flow redistribution.

2-D Trans Thoracic Echocardiogram

Mitral valve (ANT and posterior leaflets) and aortic valve (non-coronary leaflets and right leaflet) thickened with signs of infiltration vs. degenerative sclerosis due to history of HTN and age. Signs of thickening of the septum and the posterior wall. In a patient without severe valvular disease, think about true hypertrophy due to HTN vs. infiltration; for example, amyloid proteins in the extracellular space. Dynamic obstruction in the outflow tract is ruled out. LVEF = 63%

Increased hyper refringence of the interatrial and interventricular septum and LV and RV walls. Dilation of both atria.
CASE 5 - continued

Reason for Test

Red Flags:
- HFpEF, age >60 years, LV wall thickness (>12 mm), diastolic dysfunction. AV block
- Suggestive ECHO findings
- Elevated biomarkers for BNP and troponins
- Renal insufficiency

Imaging Protocol
- 20 mCi (740 MBq) of $^{99m}$Tc-PYP was injected intravenously
- One and three hours after injection, planar and SPECT images of the thorax were acquired

Study Images

Planar images at 1 hour: Perugini Grade 3

Planar images at 3 hours: Perugini Grade 3
Reason for Test

Red Flags:
- HFpEF, age >60 years, LV wall thickness (>12 mm), diastolic dysfunction.
- AV block
- Suggestive ECHO findings
- Elevated biomarkers for BNP and troponins
- Renal insufficiency

Imaging Protocol
- 20 mCi (740 MBq) of 99mTc-PYP was injected intravenously
- One and three hours after injection, planar and SPECT images of the thorax were acquired

Study Images

Lab Tests for AL Exclusion: Normal
- Isotype κ = 12.58 (normal 3.3–19.4 mg/L)
- Isotype λ = 9.25 (normal 5.7–26.3 mg/L)
- κ:λ ratio = 1.36 (normal 0.26–1.65)

Interpretation and Key Points to Report

- Clinical suspicion
- Suggestive ECG and echocardiogram
- Positive 99mTc-PYP scan
- AL amyloidosis ruled out

Teaching Points

- Important to utilize the published diagnostic algorithms.
- See position statement from the European Society of Cardiology.

CASE 6

Patient History
- 69-year-old male
- Decompensated HF of unknown cause
- Chronic renal failure (creatinine clearance 1 mL/min)
- Hypertension
- 2015: AF (CHA2DS2-VASC 3; HASBLED 3) + dyspnea, ECHO: LVH with predominance of the interventricular septum with preserved EF
- 2018: Worsening of HF symptoms. Coronary invasive angiography showed no angiographically significant lesions
- 2022: Consultation due to progressive lower limb edema associated with dyspnea and class functional impairment, NYHA III + increase in abdominal circumference, decrease in diuretic rate, refractory to oral diuretic treatment

Physical Exam
- Regular general status, lucid, hemodynamically stable, without angor, with dyspnea (NYHA III/IV), does not tolerate the decubitus anasarca.
- BMI 33
- Vital signs: BP: 110/60 mmHg, HR: 80 bpm, Breathing rate: 20 cpm, S0₂: 94%
- Cardiovascular: R1 and R2, normophonic, no puffs. Lower limb edema, sacral edema, IY 3/3 without respiratory collapse, good peripheral perfusion
- Abdomen: Congestive hepatomegaly, ascites

Hospital Admission Day
Labs
- Hb 10%; hematocrit 29.5%; urea 1.46 g/L; creatinine 2.27 mg% (creatinine clearance: 41 mL/min); ESR:15 mm/h; total proteins 60 g/L; ProBNP > 9000 pg/mL
**ECG and Chest X-ray**

**2-D Trans Thoracic Echocardiogram**
Biventricular infiltrative cardiomyopathy, normal LVEF, HCM, LVMI (361 g/m²), without motility alterations. Monophasic wave in mitral pattern. Severely dilated LA, dilated RA, increased filling pressures, depressed RVF, normal PSAP, moderate pericardial effusion. IVC with collapse <50%.

**Cardiac MRI**
- Preserved LVEF and RVEF
- Increase in LV thickness, predominantly septal (24 mm), increase in LV mass
- Dilation of both atria
- Early annulment of the blood pool and diffuse
- Subendocardial enhancement, affecting the LV, the RV, and both atria, compatible with non-ischemic necrotic fibrosis, suggestive of CA
CASE 6 - continued

Lab Tests for AL Exclusion
- Normal immunoglobulin quantification
- Normal serum electrophoretic proteinogram
- Normal immunofixation in blood and urine
- Negative Bence Jones proteins, normal protein profile
  → Ruling out monoclonal gammopathy

Reason for Test
- Clinical suspicion: HFpEF
- Suggestive ECHO and CMR
- AL amyloidosis ruled out

Imaging Protocol
- 20 mCi (740 MBq) of $^{99m}$Tc-PYP was injected intravenously
- One and three hours after injection, planar and SPECT images of the thorax were acquired

Study Images

Planar images at 1 hour
- Intense uptake of the radiotracer in the myocardium (Grade 3)
- H/CL ratio is 1.86
- Highly suggestive of ATTR CA

SPECT at 1 hour
- Intense uptake of radiotracer in the myocardium
- Highly suggestive of ATTR CA

Follow-up
- Patient was taken off inotropes, started on furosemide and tafamidis
- Discharged with improvement of symptoms
**Interpretation and Key Points to Report**

- In every patient admitted to the hospital with HFpEF, ATTR CA should be considered a differential diagnosis.
- Although ECHO, CMR, and $^{99m}$Tc-PYP are suggestive of ATTR CA, it is essential to rule out AL amyloidosis using sFLCs.

**Teaching Points**

- Seven years passed from the patient’s first clinical manifestations to diagnosis, longer than reported in the literature (5 years).
- It is important to know the evolution of symptoms as well as the presence of red flags, to make a timely diagnosis.
CASE 7

Patient History
- 39-year-old female
- Identified as a carrier of hATTR (Ser50) 10 years ago
- Father diagnosed with hATTR due to polyneuropathy at 53 years of age
- Has been on treatment with tafamidis 20 mg/day for last 5 years
- Asymptomatic
- Never had cardiac imaging evaluation

Physical Exam
- Normal

Reason for Test
- Screening of first-degree family members with the antecedent of hATTR is important for early detection of the disease, even in asymptomatic patients.
- ECHO could be the study of choice to evaluate cardiac involvement due to its availability and costs. Nevertheless, when suggestive findings are found on the ECHO, the disease is likely already established.

Imaging Protocol
- 20 mCi (740 MBq) of $^{99m}$Tc-PYP was injected intravenously
- One and three hours after injection, planar and SPECT images of the thorax were acquired
- Whole-body image acquired at 3 hours

Images at 3 Hours
- Perugini Grade 3
- H/CL 1.76
- Biventricular Uptake
Interpretation and Key Points to Report

- Diagnosis of ATTR CA can be made in asymptomatic cardiovascular patients when intentionally screened.
- $^{99m}$Tc-PYP scintigraphy is the study of choice to evaluate cardiac involvement of ATTR due to its availability, costs, and high diagnostic performance.

Teaching Points

- Early diagnosis is key, allowing for improved patients outcomes, particularly with the availability of novel therapeutic options.
CASE 8

Patient History
- 77-year-old male
- Hypertension
- One year of functional class impairment, NYHA III
- Multiple hospitalizations for decompensated HF despite optimal treatment: sacubitril/valsartan, bisoprolol, furosemide
- Dyspnea with minimal effort
- Lower limb edema

2D Trans Thoracic Echocardiogram
- LVEF = 42%, 16mm septum, LVH, GLS with Japan flag sign

Reason for Test
- Confirm the suspected diagnosis of ATTR CA

Imaging Protocol
- 20 mCi (740 MBq) of $^{99m}$Tc-PYP was injected intravenously
- One and three hours after injection, planar and SPECT images of the thorax acquired

Images at 3 Hours

Planar

Perugini Score 0
H/CL 1.1

SPECT
Interpretation and Key Points to Report

- Despite having a negative $^{99m}$Tc-PYP scan, the diagnosis of ATTR CA cannot be ruled out.
- Given the high suspicion due to clinical symptoms and two suggestive imaging studies (ECHO and CMR), it is necessary to perform an endomyocardial biopsy.
- Endomyocardial biopsy was performed and confirmed the diagnosis of ATTR CA (positive congo red).
- It must be complemented with genetic typing to determine hereditary ATTR CA non-avid $^{99m}$Tc-PYP variant.

Teaching Points

- When there is high clinical suspicion and there is a negative $^{99m}$Tc-PYP scan, false negatives must be considered.
- Hereditary variants with low $^{99m}$Tc-PYP uptake include: Leu75pro, ApoA, Ser77Tyr, Phe64Leu.
Selected References


