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Using Machine Learning to Assess Potential Cases of Transthyretin Cardiac Amyloidosis in Brazil: A Retrospective Database Approach

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Introduction

- Amyloidosis is a group of protein misfolding disorders leading to organ damage due to insoluble amyloid fibril deposits
- The two primary types of cardiac amyloidosis are light-chain amyloid (AL) and transthyretin (TTR) cardiac amyloidosis
- TTR amyloidosis can be hereditary (hATTR) or age-related (wtATTR). It is an often-overlooked cause of heart failure in older adults
- Recent studies reveal its prevalence in various patient groups: up to 13% in HFpEF, 16% in aortic stenosis patients undergoing valve replacement, 7-8% in carpal tunnel release surgery, and 17% in some other contexts
- ATTR-CM is significant in the context of cardiovascular diseases, a leading global cause of death.

Objective

- This study aimed to identify and describe the profile of potential transthyretin cardiac amyloidosis (ATTR-CM) cases in the Brazilian public health system (SUS), using a predictive machine learning (ML) model.

Materials and Methods

- This was a retrospective descriptive database study that aimed to estimate the frequency of potential ATTR-CM cases in the Brazilian public health system (Figure 1) using a supervised machine learning (Figure 2) model, with data extracted from DATASUS outpatient and inpatient datasets from January 2015 to December 2021
- To build the model, a list of ICD-10 codes and procedures potentially related with ATTR-CM was created based on literature review and validated by experts (Figure 3)

Figure 1. Study Design Summary

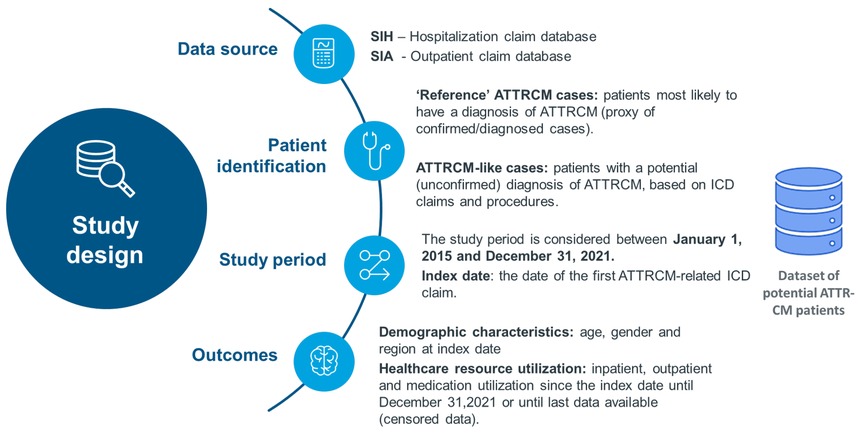


Figure 2. Machine Learning Approach

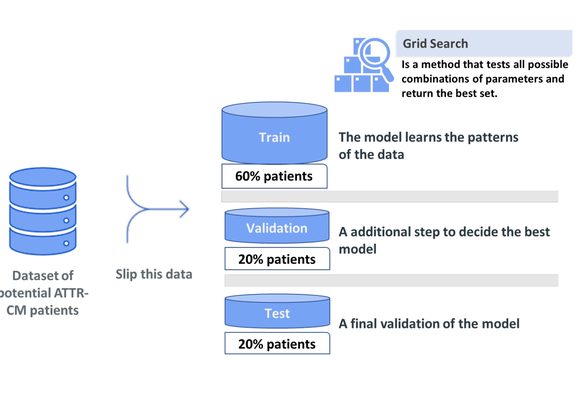
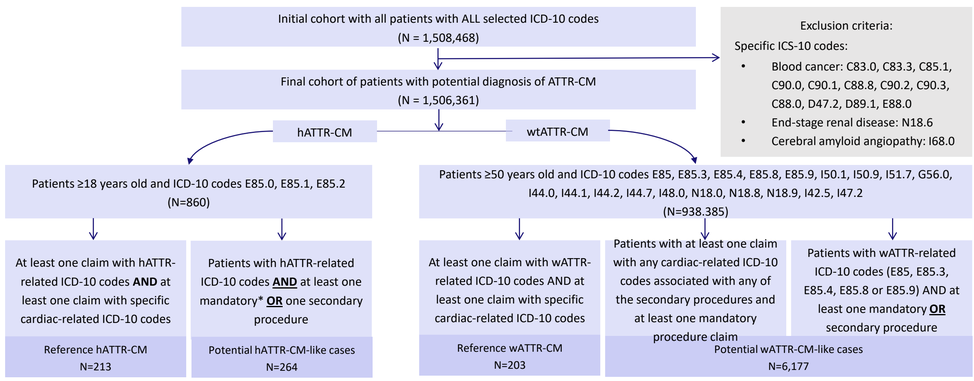


Figure 3. Study Flow Diagram



hATTR-related ICD-10 codes: E85.0, E85.1 ou E85.2
wtATTR-related ICD-10 codes: E85, E85.3, E85.4, E85.8, E85.2
Specific cardiac-related ICD-10 codes: I50.0, I50.1, I50.9, I11.0, I42.0, I42.1, I42.2, I35.0, I44.1, I44.2, I42.5
General cardiac-related ICD-10 codes: I50.0, I50.1, I50.9, I51.7, G56.0, I44.0, I44.1, I44.2, I44.7, I48.0, N18.0, N18.8, N18.9, I42.5, I47.2

Results

- From 2015 to 2021, the ML model classified 262 hATTR-CM (213 reference hATTR-CIM and 49 hATTR-CM-like) and 1,581 wtATTR-CM (203 reference wtATTR-CM and 1,378 wtATTR-CM-like). Overall, the median age of hATTR-CM and wtATTR-CM patients was 66.8 and 59.9 years, respectively
- The ICD-10 codes most presented as hATTR-CM and wtATTR-CM were related to heart failure and arrythmias, with similar procedures performed (Figure 4). Regarding healthcare utilization, hATTR-CM and hATTR-CM-like had similar profiles on proportion of patients with outpatient visits (hATTR-CM 98.0% vs. 92.0% hATTR-CM-like) and different profile related to proportion of hospitalized patients (hATTR-CM 94.4% vs. 32.7% hATTR-CM-like) (Figure 5)
- In wtATTR-CM groups, although both proportions on outpatient visits and hospitalizations were similar, the length of stay (LOS) on hospitalizations was different in wtATTR-CM-like (wtATTR-CM median LOS 5.0 (IQR:2.0 - 10.0] vs. median LOS 7.0 [IQR:3.0 - 14.0])

Figure 4. Most common procedures per cohort

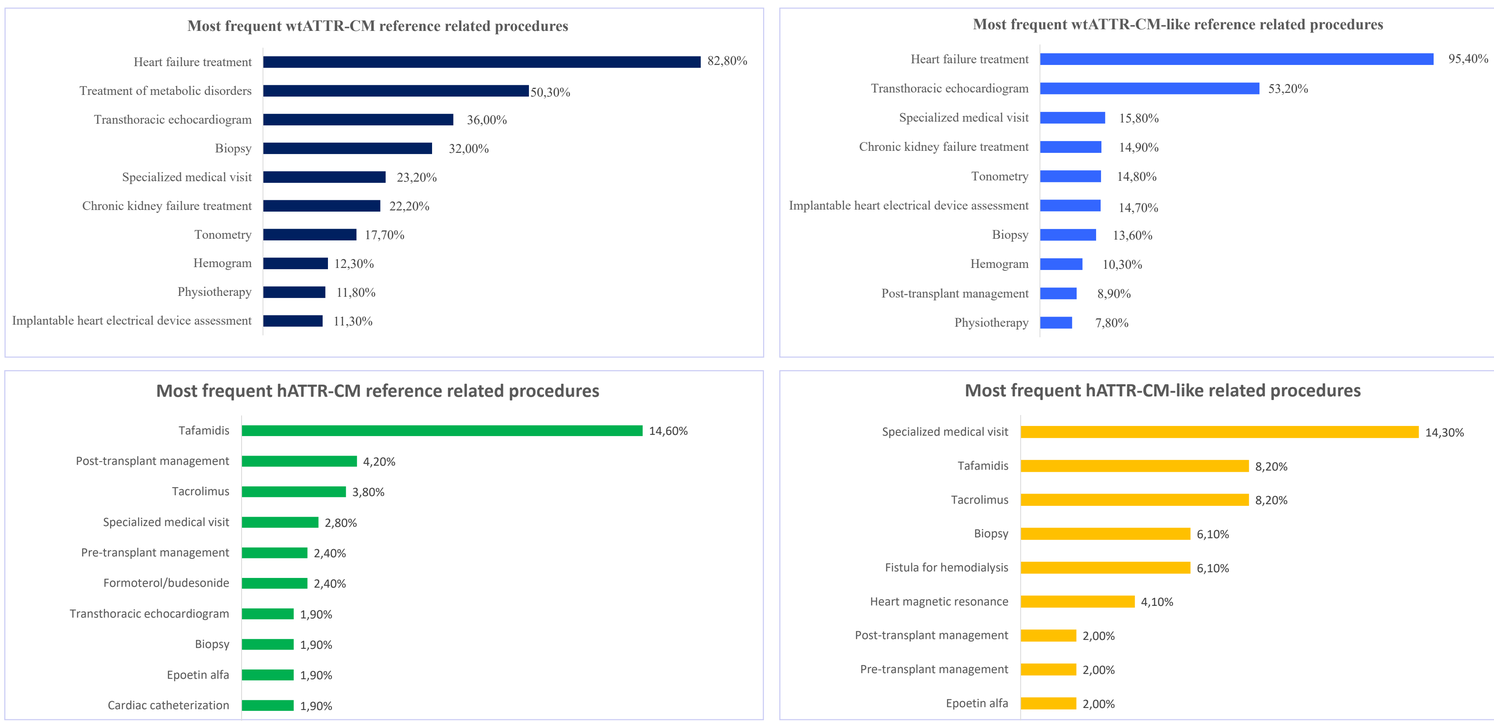
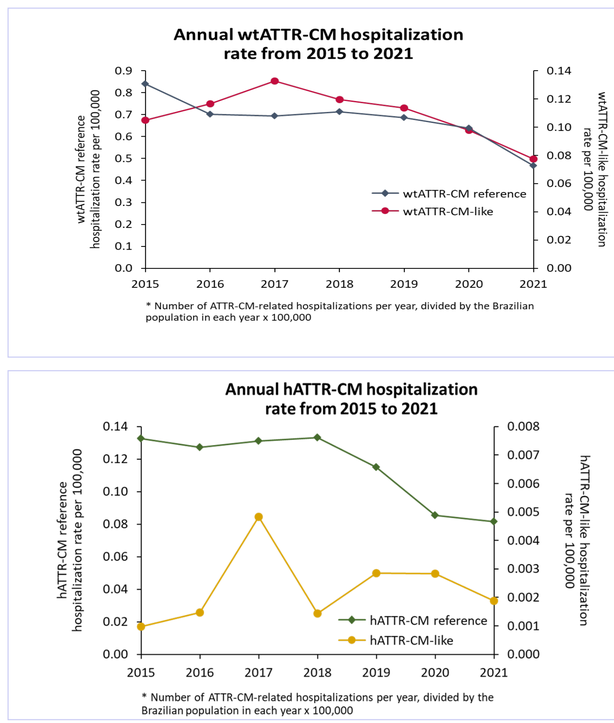


Figure 5. Hospitalization rate for wt- and hATTR-CM



Conclusions

- Our findings may be useful to support decreasing the uncertainties on ATTR-CM population size in Health Technology Assessment appraisals and in the development of healthcare guidelines and policies to address patients' unmet needs and to improve early diagnosis and access to treatment for patients with ATTR-CM in Brazil
- This study puts a spotlight on the ATTR-CM underdiagnosis in Brazil using a machine learning approach, which can be used as an important tool to support diagnosis improvement.

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