

Proportions and temporal trends of diseases leading to a hypertrophic cardiomyopathy phenotype. A prospective single-center cohort study

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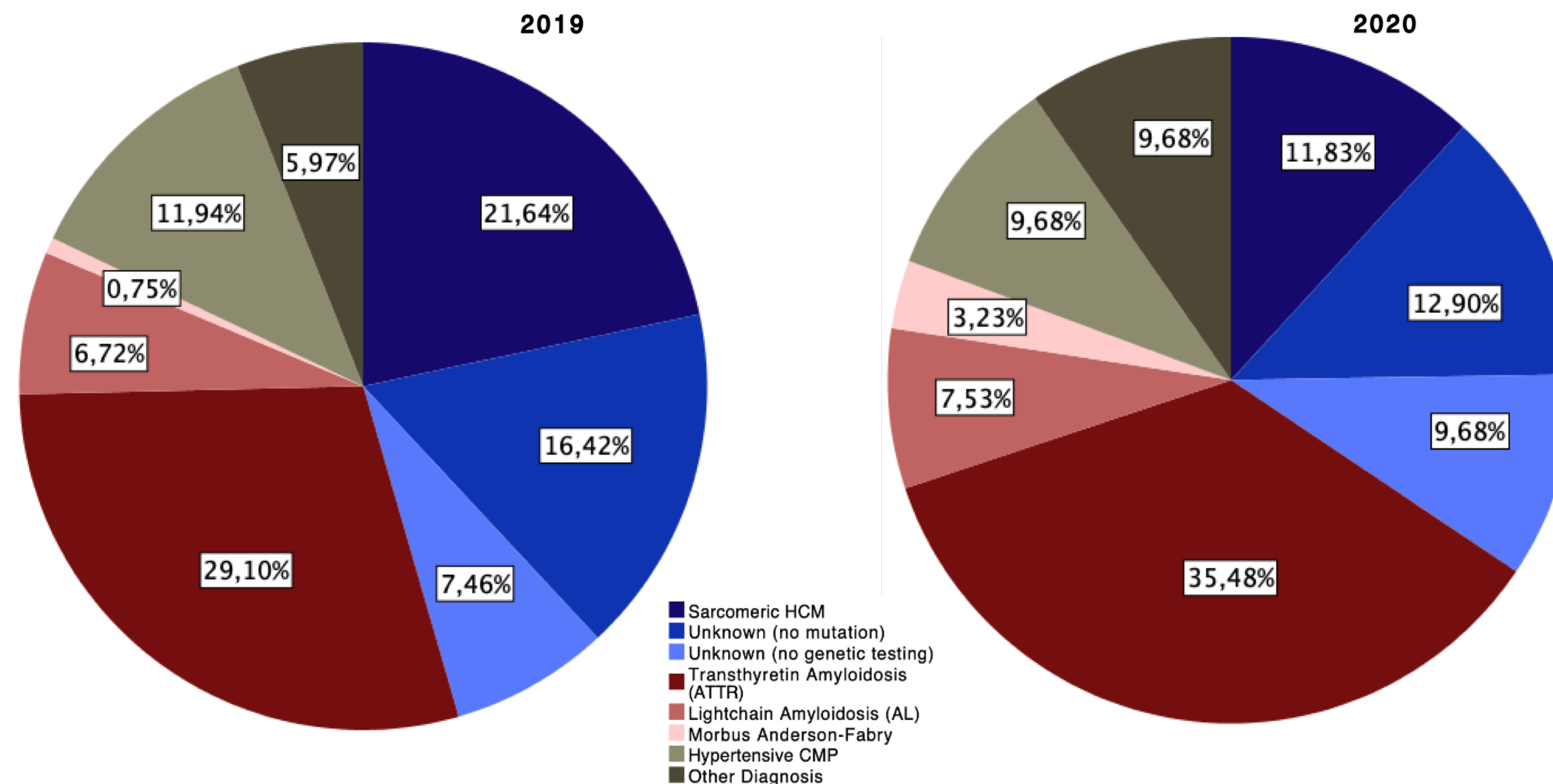
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Introduction

Hypertrophic cardiomyopathies (HCM) are caused by genetic and non-genetic diseases leading to increased left ventricular wall thickness. Previous studies described sarcomere protein gene mutations as the most common aetiology, whereas cardiac amyloidosis was rated as a rare cause affecting an insignificant minority of HCM patients. Due to recent improvements of non-invasive diagnostic tools and novel therapeutic options, earlier epidemiological data on HCM may be outdated. We therefore aimed to investigate proportions and trends of HCM aetiologies using data derived from the Graz HCM Registry.

Methods

The Graz HCM Registry is a prospective, single-center cohort study that has been initiated in February 2019. All patients who consult our HCM outpatient clinic are invited for participation. For the present analysis, we included all participants who fulfilled echocardiographic HCM criteria. Patients underwent cardiovascular imaging and genetic testing according to international guidelines and local SOPs. Cardiac amyloidosis was diagnosed and subtyped either noninvasively using bone scintigraphy and monoclonal gammopathy assessment or invasively using endomyocardial or non-cardiac biopsy.



Results

Within 2 years 175 patients were enrolled. Mean age was 62.8 ± 16.6 years, 58 were females. Median LVEF was 56.1 ± 11 % and mean E/e' was 12.7 ± 5.6 . Most common aetiologies were sarcomeric HCM in 38 patients (21.7%), cardiac amyloidosis in 59 patients (33.7%; 51 with ATTR and 8 with AL), hypertensive cardiomyopathy in 18 patients (10.2%), and Anderson-Fabry disease in 4 patients (2.3%). Due to restrictions associated with the Covid-19 pandemic, recruitment rates were lower in 2020 compared to 2019 (106 vs 69 patients). To investigate the trending of proportions, data collected in 2019 and 2020 was compared. The proportion of patients diagnosed with cardiac amyloidosis and Anderson-Fabry disease increased from 35.8% to 43.1% and 1% to 3%, respectively. On the other hand, there was a decline of sarcomeric HCM from 21.6% to 11.8%. Proportions of hypertensive cardiomyopathy and other diseases with low prevalence did not change considerably.

Demographics (n=175)

Age, years	62.8 ± 16.6
Females, n (%)	58 (33)
LVEF, %	56.1 ± 11
E/e'	12.7 ± 5.6

Conclusion

Our data suggest that TTR amyloidosis has emerged as the most common cause of HCM-phenotype in a tertiary care setting. Moreover, its prevalence appears to increase further. Vice versa, sarcomeric HCM, as the previously most common aetiology underlying HCM, is only diagnosed in approximately one of five HCM patients.