

Mitral valve abnormalities in decedents of sudden cardiac death due to hypertrophic cardiomyopathy and idiopathic left ventricular hypertrophy



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Hypertrophic cardiomyopathy (HCM) is defined as left ventricular (LV) hypertrophy (LVH) in the absence of abnormal loading conditions and characterized by myocyte disarray at histology.¹ After sudden cardiac death (SCD), the sole identification of significant LVH at autopsy may lead to an erroneous diagnosis of HCM. Data suggest that idiopathic LVH (ILVH) and HCM may be separate entities.² We aimed to report the prevalence and nature of mitral valve (MV) abnormalities in SCD victims with postmortem findings consistent with HCM and ILVH. We hypothesized that MV abnormalities are more common in individuals with HCM and considered as additional macroscopic features to differentiate between these 2 entities.

We reviewed 6860 consecutive cases of SCD referred to our specialist cardiac pathology center between 1994 and 2020. SCD was defined as death due to a cardiovascular cause within 12 hours of apparent well-being. All cases underwent detailed autopsy, and a minimum of 10 tissue blocks underwent histological analysis.³ ILVH was defined as unexplained LVH (heart weight >500 g in males and >400 g in females) and LV wall thickness > 15 mm in the absence of myocardial disarray or secondary causes of LVH.² The MV was examined for patency, circumference, thickening, nodularity, ballooning, bulging between cords, perforation,

endocarditis, and the presence of impact lesions in the LV outflow tract and aortic outlet.

Ethical approval was granted for this study (10/H0724/38).

Of the total cases of SCD, 264 (4%) were due to HCM (mean age 41 ± 18 years; 78% males; LV maximal wall thickness 19 ± 6 mm) (Figure 1). Antemortem symptoms were reported in 44 cases (17%), and for the majority ($n = 217$ [82%]), the diagnosis of HCM was established at postmortem. Death was attributed to ILVH in 253 cases (3%; mean age 43 ± 16 years; 80% males; LV maximal wall thickness 18 ± 4 mm). MV abnormalities were found in 58 decedents with HCM (22%; mean age 38 ± 17 years; 72% males) and 13 decedents with ILVH (5%; mean age 55 ± 5 years; 77% males; $P < .001$). Myocardial fibrosis was observed in 162 cases of HCM (61%) and 99 cases of ILVH (39%) ($P < .001$).

Of the 58 cases with HCM and MV abnormalities (22%), 15 (6%) had multiple MV abnormalities. These included impact lesions associated with thickening of the anterior MV leaflet ($n = 39$) and degenerative changes ($n = 34$) such as bulging and ballooning as well as thickening and nodularity. Decedents with HCM exhibiting MV abnormalities were younger than decedents with a normal MV (38 ± 17 years vs 45 ± 19 years; $P = .08$).

Of the 253 decedents with ILVH, 13 (5%) exhibited MV abnormalities, which largely included degenerative changes ($n = 12$). Among decedents with HCM and ILVH exhibiting MV abnormalities, the former was significantly younger (38 ± 17 years vs 55 ± 15 years; $P = .001$).

MV abnormalities were identified in 22% and 5% of decedents of SCD attributed to HCM and ILVH, respectively. Imaging studies, predominantly on cohorts with dynamic LV outflow tract obstruction, have reported mitral MV malformations in up to 70% of patients with HCM.^{4,5} The lower

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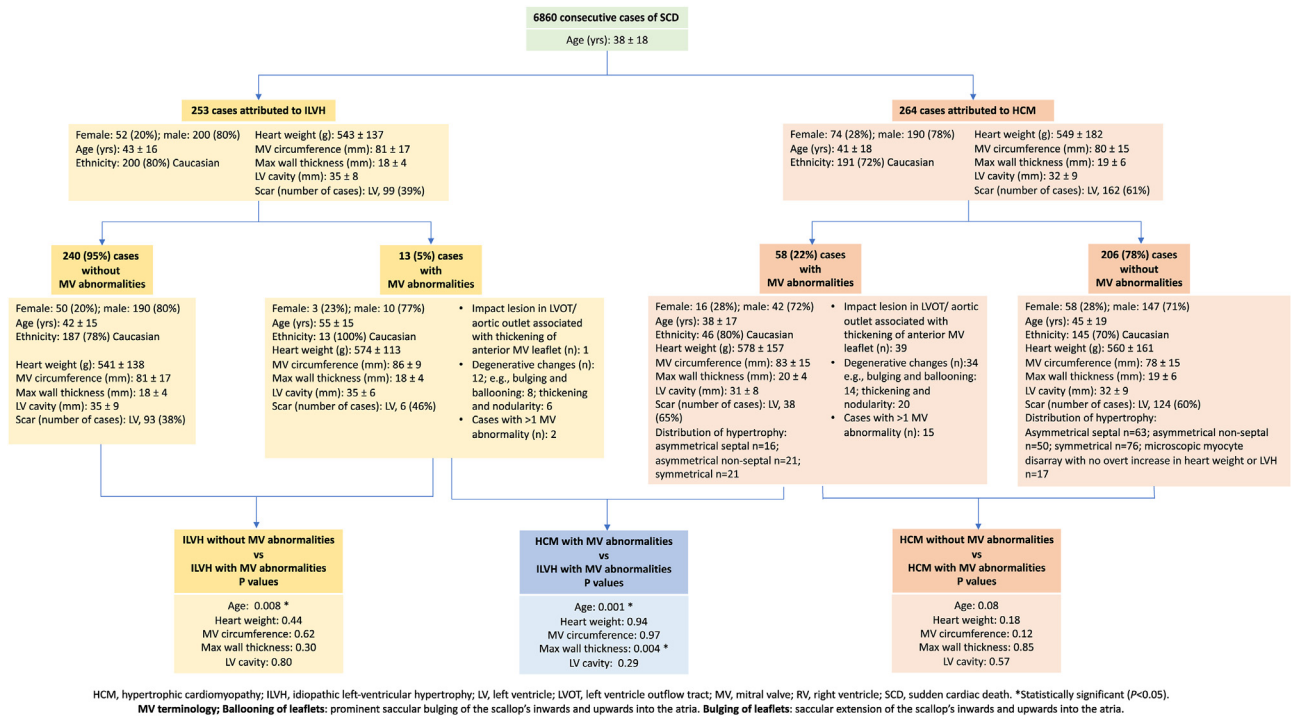


Figure 1 Study overview.

prevalence of significant MV abnormalities in our cohort may be due to a lower proportion of obstructive cases, since most decedents did not have any preexisting cardiovascular symptoms and SCD was the first manifestation. Although LV outflow tract obstruction is regarded as a risk factor for fatal arrhythmias, an impact lesion was observed in 39 of 264 cases of HCM (15%), suggesting that systolic anterior motion of the MV and possible dynamic obstruction were relatively rare in this population. MV abnormalities were rare in ILVH and were found in older individuals, suggesting an underlying mechanism that is unrelated to sarcomeric disease. A limitation of our study is the absence of molecular autopsy findings. Furthermore, the inherent descriptive terminologies used when assessing the MV support a greater emphasis on the standardization and quantification of MV abnormalities as part of the autopsy, including the sub-MV apparatus and geometrical arrangements of papillary muscles, the presence of papillary muscle hypertrophy, and abnormal chordae tendineae arrangements.

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