Abstracts

The majority of positive mutations identified were in MYBPC3 (20%) and MYH7 (12%) (see figure 1). For all other sarcomeric genes, yield was <3%. Percentage yield varied greatly between studies with a range of 19–90% (mean 44%) and showed no discernible relationship with panel size (see figure 2).

Conclusion Over time, the number of genes included on HCM diagnostic genetic panels has increased but this has not translated to a significant improvement in diagnostic yield. Larger panels may introduce more complexity and uncertainty in the clinical setting and some of the high diagnostic yields may be explained by lax variant interpretation methods.

Conflict of Interest none

124 SURVIVAL WITH VALVULAR HEART DISEASE (OXVALVE-SURVIVE)

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Introduction Valvular heart disease (VHD) occurs commonly in older patients (>65 years) but the majority is mild disease, which is of uncertain importance. Understanding the impact of VHD on mortality in this older group of patients would help determine its relevance and aid the appropriate use of healthcare resources. We determined survival in the OxValve cohort study to investigate whether people with VHD are at increased risk of death.

Methods OxValve is a cohort study in Oxfordshire, involving systematic screening of people aged 65 and over for VHD. Over 4,009 participants were recruited between August 2009 and May 2016 and screened using echocardiography to establish the presence and severity of VHD. The cohort was linked to Office for National Statistics mortality data to obtain date and cause of death. Cox regression was used to investigate the association of any VHD, VHD of significant severity, and VHD subtypes with all-cause and cause-specific mortality, adjusting for potential confounders including age, sex, socioeconomic status, smoking, and comorbidities.

Results Linked mortality data was available for 3,511 OxValve participants up to September 2018 (median 8.5 years follow-up). VHD was present in 2,645 (75.3%) participants and of these 288 (8.2%) had significant (moderate or severe) VHD. In total, 311 (8.9%) participants had died. Cancer was the commonest cause of death (n=135), followed by cardiovascular disease (n=75) and respiratory disease (n=35). After adjustment for age and other covariates, mild VHD was not associated with increased all-cause mortality (HR 1.16, 95% CI: 0.89 to 1.50). However, VHD of significant severity (moderate or severe disease) was associated with a nearly two-fold higher risk of death overall (HR 1.92, 95%CI: 1.38 to 2.67) including increased CVD mortality (HR 2.25, 95% CI: 1.21 to 4.18) – see figure.

Conclusion Mild VHD is very common, but is not associated with increased mortality. Significant (moderate-severe) VHD was however associated with a two-fold reduction in survival. Further research is required to understand the natural history of VHD, how to identify those with progressive disease and when to intervene.

Conflict of Interest None

125 DEFINITIVE HISTOLOGICAL EVIDENCE OF MYOCARDIAL FIBROSIS IN PRIMARY DEGENERATIVE MITRAL REGURGITATION: CAUSATION OF SYMPTOMS?

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Chronic primary mitral regurgitation (MR) exposes the left ventricle (LV) with a volume load that leads to progressive compensatory adjustments; initially LV enlargement and eccentric hypertrophy to progressive structural and functional damage. Identifying the deleterious stages of LV remodelling before the onset of irreversible dysfunction is critical to patient management and prognosis. Myocardial fibrosis with MR is proposed to develop in response to increased wall stress, but data are limited to small echo based imaging biomarkers. The aim of this prospective multi-centre study is to characterise the histological status of patients with chronic primary MR and examine associated changes in cardiac structure, function and symptom burden. 120 patients with primary degenerative MR (67% asymptomatic) were recruited prospectively. Coronary disease was excluded by angiography. All subjects underwent clinical assessment, multiparametric cardiac magnetic resonance (CMR) imaging, symptom assessment with the Minnesota Living with Heart Failure Questionnaire (MLHFQ) and cardiopulmonary exercise testing (CPET). 106 patients were referred for mitral valve surgery with the aim of collecting 3 LV biopsies per patient. Control myocardial