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ABSTRACT TEMPLATE

Title:	**Clinical Profile and Outcomes in Hypertrophic Cardiomyopathy: Findings from the Tayside Inherited Cardiac Condition Clinic (ICC) Study.**
Authors:	**Victoria Lamour*, Arash Dehkordi*[1], Andrew PM Lang, Yi Liew, Mya Win, Ify Mordi, Chim C Lang, Anna Maria Choy *joint first authors**
Affiliations:	**University of Dundee, [1]University of Georgia**

Introduction:

Hypertrophic cardiomyopathy (HCM) is heart muscle disease that is defined by left ventricular (LV) hypertrophy (LVH) in the absence of abnormal cardiac loading and is predominantly caused by autosomal dominant mutations in sarcomeric protein genes. Understanding the spectrum of disease, symptom burden and natural history is critical for effective patient management. This study aims to assess the clinical, genetic and imaging profiles in patients diagnosed with HCM in the ICC registry in Tayside to better understand disease progression.

Methods:

Clinical data from 242 patients (mean age 63.1± 15.1years, 67% male) with phenotypic HCM in the Tayside ICC service were reviewed. Genetic testing results, symptoms, co-morbidities, imaging data and cardiac events were gathered from medical records. Specific phenotypic HCM features of septal wall thickness, apical hypertrophy and outflow tract gradient of >30mmHg at rest or >50mmHg with Valsalva were collected.

Results:

Of the 242 patients with phenotypic HCM, 189 (78%) were index cases. Genotyping was done in 232 patients and pathogenic variants were detected in 85 (37%) patients. Symptom burden was high with 149 (61%) patients reporting at least one symptom of breathlessness, palpitations, chest pain or dizziness. Comorbidity was common, with 60% of patients having more than 1 comorbidity and atrial fibrillation was found in 64(27%) at the time of presentation. Left ventricular outflow tract obstruction was found in only 9% of patients. Gene negative patients were older (mean (SD) 67 (13.2) vs 56 (1.6) P<0.001) and were more symptomatic of dyspnoea (32 vs 18%, P<0.02) and chest pain (30 vs 17%, P<0.02). There were 22 deaths over the 9.5 years of follow up. There were 4 deaths from sudden cardiac death and 4 from heart failure which occurred in the gene positive group. There was a preponderance of non-cardiovascular deaths in the gene negative cohort.

Conclusion:

Our Tayside ICC study reveals that HCM is associated with significant morbidity and mortality and that atrial fibrillation is a frequent finding. In contrast to previous reports, left ventricular outflow tract obstruction is not frequent and does not account for the symptom burden in HCM. HCM associated with pathogenic variants appears to have worse outcomes from sudden death and heart failure.

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