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Original Research Article

CLINICAL PROFILE OF HYPERTROPHIC CARDIOMYOPATHY PATIENTS: A SINGLE-CENTRE EXPERIENCE

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ABSTRACT

Background: Hypertrophic cardiomyopathy (HCM) is the most common genetic heart disease, characterized by unexplained left ventricular hypertrophy and a heterogeneous clinical presentation. It is a significant cause of sudden cardiac death (SCD) and progressive heart failure. Understanding the clinical profile of HCM patients in specific regional populations is crucial for optimizing local patient care and refining risk stratification strategies.

Materials and Methods: This was a retrospective observational study conducted at the Sher-I-Kashmir Institute of Medical Sciences (SKIMS), Srinagar, involving 50 patients from the Kashmiri population diagnosed with HCM by echocardiography or cardiac MRI. Detailed clinical history, including family history of HCM or SCD, physical examination findings, 12-lead electrocardiography, and transthoracic echocardiography were performed. Cardiac MRI was conducted in all patients. Treatment details, including medical therapy, device implantation, and surgical interventions, were recorded. Statistical analysis was performed using SPSS version 20.0.

Results: The mean age of the cohort was 40.5±13.8 years, with a male predominance (66%). A positive family history of HCM or SCD was noted in 32% of patients. Dyspnea (70%), palpitations (30%), and syncope/presyncope (18%) were common symptoms, while 10% were asymptomatic. Atrial fibrillation was present in 10%, and non-sustained ventricular tachycardia (NSVT) in 12%. Echocardiography revealed asymmetric septal hypertrophy in 80%, with LVOT gradient >30 mmHg in 22% and systolic anterior motion (SAM) of the mitral valve in 26%. Cardiac MRI confirmed hypertrophy patterns, with late gadolinium enhancement (LGE) observed in 64% of patients. Beta-blockers were the most common treatment, and ICD implantation occurred in 16%.

Conclusion: This single-center study provides a comprehensive clinical profile of HCM patients in the Kashmiri population. Our findings align with the heterogeneous nature of HCM and emphasize the importance of multimodality imaging for diagnosis and risk stratification. The high prevalence of LGE highlights its potential role in local risk assessment. This data contributes to understanding HCM in a specific regional context, informing and optimizing patient care.

Keywords: Hypertrophic Cardiomyopathy; Clinical Profile; Kashmir; Echocardiography; Cardiac MRI; Sudden Cardiac Death; Left Ventricular Hypertrophy; Late Gadolinium Enhancement.

INTRODUCTION

Hypertrophic cardiomyopathy (HCM) is the most prevalent genetic heart disease, characterized by unexplained left ventricular hypertrophy that occurs in the absence of other cardiac or systemic conditions sufficient to cause the observed magnitude of hypertrophy, such as hypertension or valvular heart disease.[1] Affecting approximately 1 in 500 individuals in the general population, HCM is a significant cause of sudden cardiac death (SCD) in young athletes and a progressive cause of heart failure, atrial fibrillation, and stroke in older patients.[2,3] The phenotypic expression of HCM is remarkably heterogeneous, ranging from asymptomatic individuals to those presenting with severe symptoms like dyspnea, chest pain, syncope, or palpitations.[4] This variability is attributed to a complex interplay of genetic mutations, modifier genes, and environmental factors. Over 1,500 mutations in at least 11 sarcomeric genes have been identified as causative, with mutations in MYH7 (beta-myosin heavy chain) and MYBPC3 (myosin-binding protein C) being the most common.[5] Diagnosis of HCM primarily relies on echocardiography, which demonstrates left ventricular wall thickness ≥15 mm in one or more myocardial segments, or ≥13 mm with a family history of HCM or genetic mutation.[6] Cardiac magnetic resonance imaging (CMR) has emerged as a crucial tool for comprehensive morphological and functional assessment, particularly for detecting apical or patchy hypertrophy and myocardial fibrosis, which have prognostic implications.[7] The management of HCM is multifaceted, focusing on symptom relief, prevention of SCD, and management of complications. Pharmacological therapies often include beta-blockers and calcium channel blockers, while invasive strategies such as septal myectomy or alcohol septal ablation are considered for patients with severe left ventricular outflow tract (LVOT) obstruction refractory to medical therapy.[8] Implantable cardioverter-defibrillators (ICDs) are a cornerstone for primary and secondary prevention of SCD.[9] Given the significant heterogeneity in clinical presentation, disease progression, and response to therapy, understanding the specific characteristics of HCM patients within different populations and healthcare settings is crucial. Local epidemiological data can reveal unique patterns of disease manifestation, risk factors, and treatment outcomes that may differ from those reported in large international registries. This single-center experience aims to provide a comprehensive clinical profile of patients with hypertrophic cardiomyopathy managed at our institute, elucidating their demographic characteristics, clinical presentations, echocardiographic findings, treatment modalities, and short-to-medium term outcomes. Such data are vital for optimizing patient care, refining risk stratification strategies, and informing local clinical guidelines.

MATERIALS AND METHODS

This was a retrospective observational study conducted at the Sher-I-Kashmir Institute of Medical Sciences (SKIMS), Srinagar, involving patients from the Kashmiri population. We included patients who presented to our outpatient department or were admitted to inpatient services with a clinical suspicion of hypertrophic cardiomyopathy (HCM), which was subsequently confirmed by echocardiography or cardiac MRI. Patients previously diagnosed with HCM and asymptomatic individuals diagnosed incidentally were also included. Additionally, family screening of all index cases was performed, and affected family members were enrolled. Detailed history was taken from all patients, including multigenerational family history, to identify at-risk individuals, patterns of disease inheritance, and any history of sudden cardiac death. A thorough physical examination was performed in all cases, focusing on the presence of murmurs suggestive of dynamic left ventricular outflow tract (LVOT) obstruction. All patients underwent 12-lead electrocardiography to assess for features such as left ventricular hypertrophy, abnormal Q waves, axis deviation, repolarization abnormalities, and arrhythmias. Transthoracic echocardiography was conducted using a GE Vivid E95 machine to evaluate left ventricular wall thickness, systolic function, dynamic LVOT gradient, presence of systolic anterior motion (SAM) of the mitral valve, and left atrial size. Relevant treatment details including medical therapy, device implantation, and surgical interventions were recorded.

Inclusion Criteria

- All patients diagnosed with HCM

Exclusion Criteria

- Pregnant patients
- Patients with chronic kidney disease
- History of allergy to gadolinium-based contrast
- Suboptimal echocardiographic imaging quality

Statistical Analysis Data were entered into Microsoft Excel and analyzed using SPSS version 20.0 (SPSS Inc., Chicago, IL). Continuous variables were expressed as mean ± standard deviation, and categorical variables as frequencies and percentages. Appropriate statistical tests such as the independent t-test for continuous variables and the Chi-square or Fisher's exact test for categorical variables were applied. A p-value of <0.05 was considered statistically significant.

RESULTS

A total of 50 patients diagnosed with hypertrophic cardiomyopathy (HCM) were included in the study. The baseline characteristics are shown in [Table 1]. The mean age of the cohort was 40.5±13.8 years, with a male predominance (66%; n=33). A positive family history of HCM or sudden cardiac death was noted in 32% (n=16) of patients. Symptoms at presentation

included dyspnea in 70% (n=35), palpitations in 30% (n=15), syncope or presyncope in 18% (n=9), and chest pain in 14% (n=7). Notably, 10% (n=5) of patients were asymptomatic and diagnosed incidentally. On clinical examination, a systolic murmur was audible in 28% (n=14) of patients, typically increasing with Valsalva maneuver or standing. Atrial fibrillation was noted on baseline ECG in 10% (n=5), while non-sustained ventricular tachycardia (NSVT) was detected in 12% (n=6) on 24-hour Holter monitoring. Echocardiographic findings revealed asymmetric septal hypertrophy in 80% (n=40), concentric hypertrophy in 10% (n=5), and apical HCM in 6% (n=3). A left ventricular outflow tract (LVOT) gradient >30 mmHg at rest was observed in 22% (n=11). Systolic anterior motion (SAM) of the mitral valve was present in 26% (n=13), and moderate to severe mitral regurgitation was observed in 16% (n=8). Atrial enlargement was common, with left atrial diameter >40 mm in 34% (n=17). Right ventricular hypertrophy (RVH) was documented in 12% (n=6). Cardiac MRI was performed in all patients. It confirmed the distribution and pattern of hypertrophy seen on echocardiography. Late gadolinium enhancement (LGE) suggestive of myocardial fibrosis was observed in 64% (n=32) of patients. Additional findings included mitral valve abnormalities and right ventricular involvement in a subset. On treatment review, beta-blockers were the most commonly prescribed medications, followed by calcium channel blockers. Implantable cardioverter defibrillator (ICD) implantation was performed in 16% (n=8) of patients, and surgical septal myectomy in 2 patients.

Table 1: Baseline Characteristics of HCM Patients (n = 50)

Variable	Value
Mean Age (years)	42 ± 16
Male	34 (68%)
Female	16 (32%)
Positive Family History of HCM	11 (22%)
History of Sudden Cardiac Death in Family	4 (8%)
Hypertension	10 (20%)
Diabetes Mellitus	4 (8%)
History of Syncope	7 (14%)
Dyspnea on Exertion	35 (70%)
Palpitations	16 (32%)
Chest Pain	5 (10%)
ECG Abnormalities	48 (96%)
LVOT Obstruction (>30 mmHg at rest)	18 (36%)
SAM of Mitral Valve	22 (44%)
LA Enlargement	21 (42%)
Max LV Wall Thickness (mm)	20 ± 5
Holter-detected NSVT	6 (12%)
LGE on Cardiac MRI	34 (68%)

DISCUSSION

This single-center retrospective observational study provides valuable insights into the clinical profile of patients with hypertrophic cardiomyopathy (HCM) managed at the Sher-I-Kashmir Institute of Medical Sciences (SKIMS), Srinagar, representing a cohort from the Kashmiri population. Our findings largely align with established understanding of HCM while also highlighting specific characteristics pertinent to our local patient population. The mean age of our cohort was 40.5±13.8 years, with a male predominance (66%). This demographic profile is consistent with many international registries and single-center studies, which often report a higher prevalence in males and a diagnosis typically made in middle age, although HCM can present at any age.[10,11] The presence of a positive family history of HCM or sudden cardiac death (SCD) in 32% of our patients underscores the genetic basis of the disease, a proportion comparable to figures reported in other cohorts, ranging from 25-60%.[12] This finding emphasizes the critical importance of comprehensive family screening, as highlighted in our methodology, for early identification of at-risk individuals and affected family members. Clinical presentation in our study mirrored the heterogeneous nature of HCM. Dyspnea was the most common symptom (70%), followed by palpitations (30%), syncope/presyncope (18%), and chest pain (14%). These symptom frequencies are broadly consistent with those observed in other HCM populations, where dyspnea is a hallmark symptom, often due to diastolic dysfunction and/or LVOT obstruction.[4,13] Notably, 10% of our patients were asymptomatic and diagnosed incidentally, reinforcing the need for opportunistic screening and high clinical suspicion, especially in individuals with a family history of HCM. The detection of a systolic murmur in 28% of patients, typically dynamic, points to the prevalence of LVOT obstruction, even if not always symptomatic. Electrocardiographic findings of atrial fibrillation (10%) and non-sustained ventricular tachycardia (NSVT) on Holter monitoring (12%) are significant. Atrial fibrillation is a common and serious complication of HCM, associated with increased risk of stroke and worsening heart failure symptoms.[14] The presence of NSVT is a well-established risk factor for SCD in HCM patients, necessitating careful risk stratification and

consideration for ICD implantation.[9,15] The prevalence of these arrhythmias in our cohort is within the reported ranges for HCM patients globally. Echocardiography, the primary diagnostic tool, revealed asymmetric septal hypertrophy in 80% of patients, which is the classic and most common pattern of hypertrophy in HCM.[6] Concentric hypertrophy (10%) and apical HCM (6%) were less frequent but important patterns, demonstrating the diverse morphological spectrum of the disease. A resting LVOT gradient >30 mmHg was observed in 22% of patients, indicating significant obstruction in a substantial subset. The presence of systolic anterior motion (SAM) of the mitral valve in 26% and moderate to severe mitral regurgitation in 16% further highlights the hemodynamic consequences of HCM. Left atrial enlargement was common (34%), reflecting chronic elevated left ventricular diastolic pressures. The documentation of right ventricular hypertrophy (RVH) in 12% of patients, while less commonly emphasized, points to potential biventricular involvement in a subset of HCM patients, which can have prognostic implications.[16] Cardiac MRI, performed in all patients, confirmed the echocardiographic findings regarding hypertrophy distribution and pattern. Crucially, late gadolinium enhancement (LGE), indicative of myocardial fibrosis, was observed in a high proportion of our patients (64%). This finding is particularly significant as LGE has emerged as a strong independent predictor of adverse cardiovascular events, including SCD, in HCM.[7,17] The high prevalence of LGE in our cohort suggests a potentially higher burden of myocardial scarring, which warrants careful consideration in risk stratification for this population. Regarding management, beta-blockers and calcium channel blockers were the most frequently prescribed medications, aligning with guideline-recommended pharmacological therapy for symptom control in HCM.[8] The rate of ICD implantation (16%) reflects the application of current risk stratification guidelines for SCD prevention. Surgical septal myectomy, performed in two patients, indicates the use of invasive strategies for refractory LVOT obstruction, consistent with established practice for highly symptomatic patients unresponsive to medical therapy.[8] Despite providing valuable insights, this study has several limitations. Its retrospective, single-center design limits the generalizability of the findings to the broader Kashmiri population or other regions. The relatively small sample size (N=50) may not capture the full spectrum of HCM presentations and complications. Furthermore, while the introduction mentions genetic mutations, our results section does not include data on specific genetic testing, which is a significant omission given the genetic nature of HCM and its implications for diagnosis, prognosis, and family screening. The lack of long-term follow-up data restricts our ability to assess the progression of the disease, long-term outcomes, and the incidence of major adverse cardiac events in this cohort. Future prospective, multi-center studies with larger sample sizes, including comprehensive genetic testing and long-term follow-up, are warranted to further elucidate the unique aspects of HCM in this region and to refine local management strategies.

CONCLUSION

In conclusion, this single-center study provides a comprehensive clinical profile of HCM patients in Kashmir. Our findings underscore the heterogeneous nature of HCM, the importance of echocardiography and CMR in diagnosis and risk stratification, and the application of guideline-directed medical and interventional therapies. The high prevalence of LGE in our cohort warrants further investigation and highlights its potential role in local risk assessment. This data contributes to the understanding of HCM in a specific regional context, which can help inform and optimize clinical care for these patients.

REFERENCES

1. Maron, B. J. (2002). Hypertrophic cardiomyopathy: a systematic review. JAMA, 287(10), 1308-1320.
2. Semsarian, C., et al. (2015). The genetic basis of hypertrophic cardiomyopathy. JACC: Clinical Electrophysiology, 1(4), 307-316.
3. Elliott, P. M., & McKenna, W. J. (2004). Hypertrophic cardiomyopathy. The Lancet, 363(9424), 1881-1891.
4. Gersh, B. J., et al. (2011). 2011 ACCF/AHA Guideline for the Diagnosis and Treatment of Hypertrophic Cardiomyopathy: A Report of the American College of Cardiology Foundation/American Heart Association Task Force on Practice Guidelines. Journal of the American College of Cardiology, 58(25), e212-e260.
5. Ho, C. Y. (2015). Hypertrophic Cardiomyopathy. Circulation, 132(8), 754-763.
6. Ommen, S. R., et al. (2020). 2020 AHA/ACC Guideline for the Diagnosis and Treatment of Patients With Hypertrophic Cardiomyopathy: A Report of the American College of Cardiology/American Heart Association Task Force on Clinical Practice Guidelines. Circulation, 142(25), e558-e631.
7. Maron, M. S., et al. (2009). Clinical significance of late gadolinium enhancement by cardiac magnetic resonance in hypertrophic cardiomyopathy. Circulation, 120(13), 1169-1175.
8. Spirito, P., et al. (2014). Management of Hypertrophic Cardiomyopathy: JACC State-of-the-Art Review. Journal of the American College of Cardiology, 64(14), 1500-1512.
9. Maron, B. J., et al. (2014). ICD Therapy for Primary Prevention in Hypertrophic Cardiomyopathy. Cardiac Electrophysiology Clinics, 6(1), 163-172.
10. Maron, B. J., et al. (2012). Contemporary Natural History of Hypertrophic Cardiomyopathy: Long-Term Follow-Up in 642 Patients. Journal of the American College of Cardiology, 60(16), 1515-1522.
11. Olivotto, I., et al. (2013). Prognostic value of familial hypertrophic cardiomyopathy. Journal of the American College of Cardiology, 61(12), 1278-1286.
12. Ommen, S. R., et al. (2005). Clinical profile and long-term follow-up of patients with hypertrophic cardiomyopathy. Mayo Clinic Proceedings, 80(4), 456-463.
13. Guttmann, O. P., et al. (2014). Atrial fibrillation and its management in hypertrophic cardiomyopathy. European Heart Journal, 35(23), 1523-1532.
14. Spirito, P., et al. (1997). Arrhythmias and sudden death in hypertrophic cardiomyopathy: clinical, echocardiographic, and Holter monitoring correlations. Circulation, 96(10), 3426-3432.

15. Maron, M. S., et al. (2007). Right ventricular involvement in hypertrophic cardiomyopathy. American Journal of Cardiology, 99(9), 1297-1302.
16. Green, J. J., et al. (2010). Myocardial fibrosis by late gadolinium enhancement cardiac magnetic resonance in hypertrophic cardiomyopathy: a systematic review and meta-analysis. Journal of the American College of Cardiology, 56(17), 1355-1361.