



Hypertrophic cardiomyopathy in the developing world: focus on India

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Introduction

Atherosclerotic coronary artery disease (CAD) and peripheral arterial disease are global public health and socio-economic issues associated with important treatment advances that affect millions of lives every year. Hypertrophic cardiomyopathy (HCM), although less common, has also become a treatable form of heart disease with demonstrable reduction in mortality and increased survival,^{1–4} entering a modern era with the inception of high resolution imaging, widely available genetic testing and counselling, implantable cardioverter defibrillators (ICDs), heart transplant, surgical myectomy, and alcohol septal ablation.⁴ Much has changed for HCM patients who historically have too often been considered ‘interesting’ or as having an ‘exotic’ disease, making them ideal subjects for research studies, but with prognosis considered largely grim. Effective treatment advances, and recognition that many (if not most) patients affected by HCM may not require major therapeutic interventions, permits a realistic aspiration for normal or extended longevity and good quality of life.¹ However, most interest in HCM has resided disproportionately in North America, Europe, portions of Asia (largely Japan), Israel, and Australia. Hence, it is patients in those countries who benefit most from the technological and management advances applicable to HCM.

Hypertrophic cardiomyopathy throughout the world

Hypertrophic cardiomyopathy can now be regarded as a global disease, recognized in >50 countries,⁵ with patients exposed to the intricacies of a wide variety of healthcare systems. This includes countries with developing economies and healthcare which involves many other medical and non-medical priorities that dominate their resources and logistics, and inadvertently direct attention from less common complex genetic heart diseases (such as HCM). In this respect, India can be considered a model of the medical dilemma in which important diseases such as HCM are in danger of being over-run and obscured by the mass of patients with valvular and

atherosclerotic CAD, at a time when cardiovascular diseases have become the number one cause of death in the country.⁶

Indeed, India is currently the second most populous country in the world, with 1.23 billion people, soon to surpass China in this regard. This fact alone poses obvious obstacles to the delivery of specialized healthcare services, and particularly for less common diseases. India has 4000 cardiologists, but the ratio to cardiac patients is unfavourable, with an estimated 9.2 million productive years of life lost to cardiovascular diseases in 2000, a number that is expected to rise to 18 million by 2030 (10 times the rate in the USA).⁶ This point underscores the overwhelming burden on practitioners in terms of time, energy, and resources legitimately devoted to a variety of cardiac problems, other than those posed by diseases such as HCM. Inevitably, patients with HCM become ‘lost’ within general cardiology practice.

In addition, there is the practical challenge faced by cardiologists and other practitioners in keeping abreast of advances in diagnosis and management for such a heterogeneous disease distinctly different from CAD, and which occupies an essentially separate literature. This is a particular concern considering the heavy workload and long week to which practising Indian cardiologists are routinely exposed.⁶ Indeed, in HCM, there is seemingly ever-changing information (including formal practice guidelines)^{7,8} that can often generate controversy, even among the relatively small group of investigators dedicated to this disease.

Significance of hypertrophic cardiomyopathy

Hypertrophic cardiomyopathy is now recognized as a relatively common cardiac disease, although it continues to be viewed as rare in much of the medical community. Based on a number of epidemiological studies throughout the world, HCM occurs in ~1:500 people in the general population,⁹ which translates to ~700 000 affected Americans and up to 2 million people in India or China.¹⁰ This may even underestimate the true prevalence, given that available data are based on HCM probands with clinically expressed disease and does not account entirely for the familial nature of this autosomal

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dominant disease; many (if not most) genetically affected individuals, with or without the disease phenotype, probably remain unrecognized. Indeed, the clinical HCM patient population actively under surveillance represents only the 'tip of the iceberg' relative to the overall disease population.¹

Clinically, HCM is notable as the most common cause of sudden cardiac death in the young, is responsible for progressive heart failure and disability at a wide range of ages, and is also associated with significant risk for atrial fibrillation and embolic stroke.^{1,7,8,10,11} Although less commonly encountered in clinical practice than either valvular heart disease or CAD, as a group HCM patients are no less deserving of contemporary management, particularly given the mortality reduction and enhanced survival that can be anticipated employing currently available major treatment strategies.^{1–4,7,8} In an ethical context, this is only a matter of fairness to a substantial group of patients.

Focused hypertrophic cardiomyopathy programmes

Reliable delivery of expert care to HCM patients requires preferential referral to centres of excellence focused specifically on this disease (usually within academic centres).^{7,12,13} Over the last 15 years, the principle of segregating HCM patients into defined programmes dedicated to their disease within divisions of cardiology has attracted considerable interest. Initiatives, such as the 'Tufts Experiment' (at Tufts Medical Center, Boston),¹² and long-standing HCM centres at the Mayo Clinic, Cleveland Clinic, and Toronto General, as well as European centres in Italy (Genoa, Florence, and Rome) and in London, and also Sydney, have demonstrated the feasibility of targeting HCM patients in this way, promoting more effective clinical care and enhanced survival. These initiatives are based on the principle that each individual patient with this disease should have access to (and the opportunity for) all appropriate treatments.^{1,2,4,7,8,12,13}

A formalized multidisciplinary HCM centre^{12,13} will include: contemporary diagnosis with advanced imaging,¹⁴ genetic testing and counselling,¹⁵ risk stratification for sudden death (SD) with the option for ICD therapy,¹⁶ both expert surgical myectomy^{17,18} and alcohol septal ablation,¹⁹ and also transplant programmes for end-stage heart failure²⁰ (Figure 1). Indeed, only in such a referral environment can operative candidates be sufficiently clustered to accumulate the critical threshold of patient volume which enables myectomy surgeons to achieve and sustain the necessary level of expertise specifically with this operation.^{7,12} Importantly, such centres require a clinical cardiology director, knowledgeable in the HCM clinical spectrum, who is responsible for the flow of patients within the programme. Furthermore, such multidisciplinary specialty centres of excellence targeting HCM patients can also stimulate subspecialty programmes for other less common diseases such as pulmonary hypertension or adult congenital heart disease.²¹

Hypertrophic cardiomyopathy and the Indian healthcare system

Spectacular, rapid advances in the technology and business sectors have not yet been translated with similar effectiveness to certain

facets of healthcare delivery in India. For example, at this time, expertise in HCM is underdeveloped and there are no established multidisciplinary centres in India specifically designed for the systematic assessment of patients with this disease. However, in recognition of that deficit, there are emerging initiatives, i.e. Amrita Institute in Kochi (Kerala) and Osmania University (Hyderabad) in south India, as well as Kalra Hospital (New Delhi), the All India Institute of Medical Sciences (New Delhi), and the Post Graduate Institute of Medical Education and Research (Chandigarh) in the north (Figure 1). To date, most Indian research activity in HCM has been in basic molecular science, with identification of pathogenic mutations,^{22–24} echocardiography,^{25,26} or interventional (percutaneous) reduction of outflow obstruction,^{27,28} largely in lieu of the surgical myectomy option.

Obstacles to accessibility for contemporary treatment strategies

Only 0.08% of Indian citizens have health insurance (compared with 84% in the USA).²⁹ The absence of some form of widely available private, governmental, or socialized medical insurance programme in populous countries (such as India and China¹⁰) creates a major healthcare delivery barrier for cardiac patients who require access to longitudinal care and expensive technology (including open heart surgery and ICDs). Without mature insurance programmes, care for patients with HCM is highly dependent on the economic resources of individual patients and families.

The cost of medical devices or surgical/interventional treatments for diseases such as HCM, including ICDs or myectomy, is clearly prohibitive for all but a small minority of patients. Indeed, 70% of ICDs in India are paid for directly by patients and families; only 20% are under federal/state governmental programmes, and 10% are covered by private insurance, including employer-sponsored plans.²⁹ Consequently, contemporary management strategies for HCM, now shown to be responsible for a reduction in disease-related mortality to <1%/year,^{3,4} remains beyond the access of vast numbers of deserving patients. This is particularly notable when considering that potentially there are 2-fold more patients with HCM available for diagnosis and treatment in the most populous countries of India and China than in all of North America and Europe.^{5,9}

In addition, other obstacles in less developed healthcare systems, such as in India (and China) are responsible for underserving the needs of patients with genetic diseases such as HCM. These include: (i) the massive volume of patients with CAD in such populous countries, with a focus on interventional cardiology; (ii) the paradox of recognizing the risk of SD in HCM, but with limited access to ICDs due to underdeveloped governmental or private medical insurance programmes; (iii) the necessity for systematic, longitudinal outpatient surveillance as an integral part of an overall management strategy, which may not be adequately built into the current system; (iv) over-reliance on percutaneous intervention (i.e. alcohol septal ablation) or pharmacological treatment, but with insufficient access to surgeons experienced with the myectomy operation; (v) lack of established patient registries as part of academic centre programmes dedicated to HCM; (vi) limited

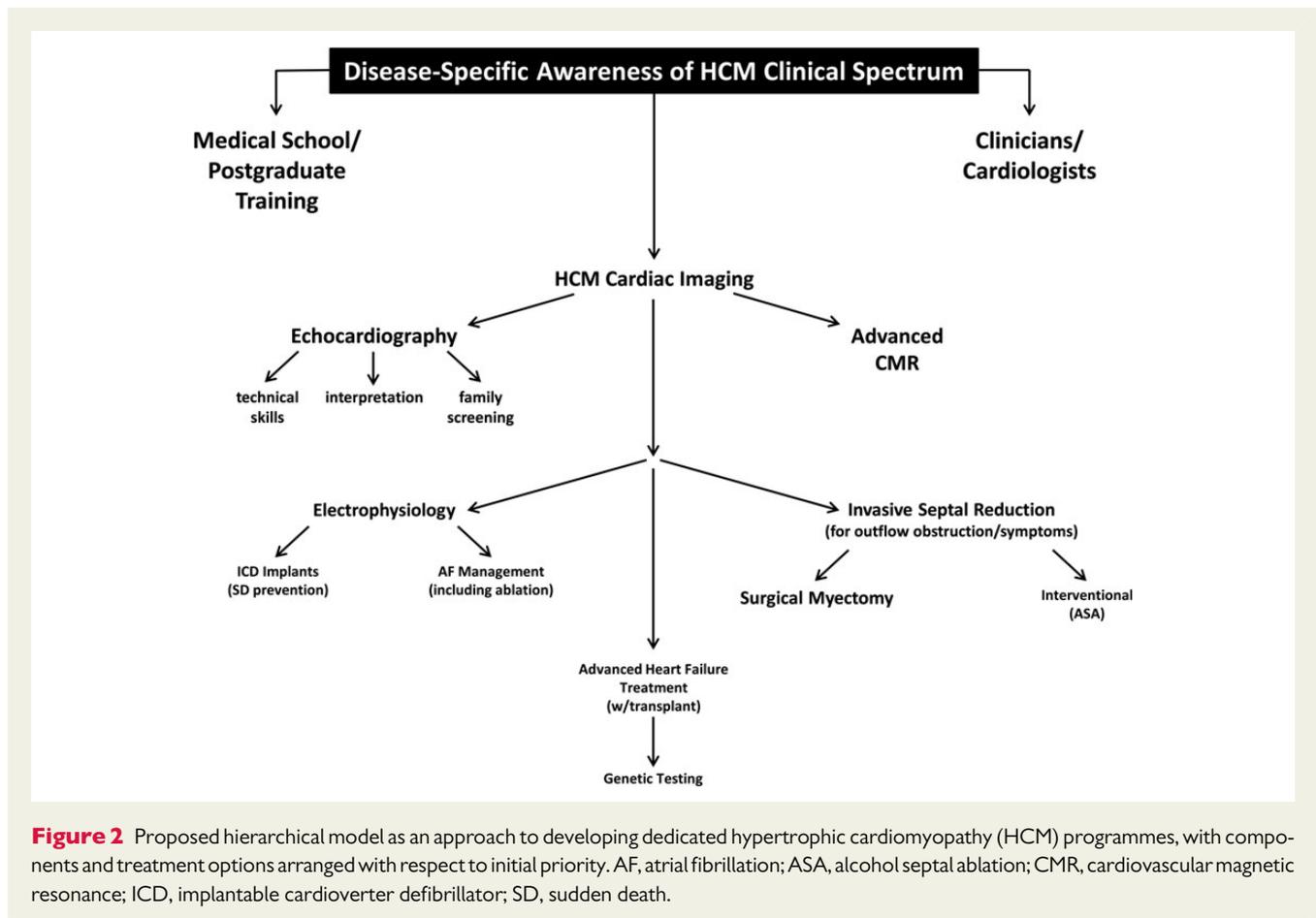


Figure 1 Map of India showing the location of centres currently with a demonstrated interest in hypertrophic cardiomyopathy.

referral to specialized centres due to under-diagnosis of HCM in the primary practising community; (vii) general lack of standardized cardiovascular disease training programmes and uniform board-certification processes; (viii) continuing influence of indigenous treatments in some towns and villages; (ix) mistrust in the patient–doctor relationship due to commercialization of medicine, associated with a substantial degree of patient distrust in implanted devices; and (x) corruption in the system with an acknowledged practice of kickbacks.

Recommendations

There is growing interest in HCM in the cardiology communities of India^{22–28} and China.^{30–35} However, it is an unavoidable reality that the priority for the available healthcare resources in these countries is necessarily with the most prevalent cardiovascular conditions such as CAD. Therefore, it may be difficult to immediately provide the highest level of care to all HCM patients, such as now demonstrated by comprehensive disease-specific



programmes available in some Western institutions.^{12,13} In such societies, the available resources have permitted progress, heretofore largely unexplored in the most populous countries.

However, rather than anticipating an abrupt short-term conversion to the highest standard, a more gradual and attainable hierarchical strategy represents a more realistic approach to contemporary HCM management (Figure 2). For example, in India it is most important initially to develop a contemporary public and physician awareness and focus on the broad clinical spectrum of HCM,¹ by education and training and integrating this disease into clinical teaching programmes in accordance with existing consensus guidelines.^{7,8} A critical element is developing advanced HCM-specific cardiac imaging with expert technical and interpretative echocardiography³⁶ and cardiovascular magnetic resonance.¹⁴ Other early important initiatives involve access to adequate numbers of trained medical and paramedical personnel to support expanding defibrillator implant programmes, as well as the identification of surgeons experienced in septal myectomy.^{7,12,35} Also, the increasing impact of advanced heart failure and the option of transplant²⁰ now have greater relevance to the multidisciplinary approach for HCM.⁴ Conversely, in terms of priority, laboratory-based genetic testing has less of a direct clinical impact on HCM management since specific mutations do not reliably predict prognosis,^{4,7,15} and an acceptable level of family screening can be achieved with standard imaging and clinical evaluation.^{1,4,14,15}

Conclusions

Hypertrophic cardiomyopathy is a relatively common disease, occurring world-wide, with increasing recognition as a public health issue in less developed countries, such as India. Given the effective management strategies that are now available to extend life in HCM patients, it is important to elevate the clinical visibility of this disease in highly populous countries with evolving healthcare systems. Numerically, most individuals affected by genetic diseases such as HCM live in countries where available healthcare resources and trained personnel may be limited, and are directed toward more common diseases. Consequently, HCM has not been a priority, and these patients have been underserved. Solutions are challenging and will take time, but can be best achieved by emphasizing HCM-related education and training, and also by developing multidisciplinary programmes dedicated to this disease within academic referral centres. Enhancing medical expertise in HCM requires commitment and willingness to invest in time and money, as well as the development of an expanded and accessible medical insurance programme (with adequate reimbursement provisions). These considerations can create a more global perspective of care for this relatively common and important genetic heart disease.

Conflict of interest: none declared.

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