

Perspectives on the Overall Risks of Living With Hypertrophic Cardiomyopathy

Hypertrophic cardiomyopathy (HCM) is the most common genetic heart disease, unique in many respects among cardiovascular conditions, including its extreme clinical heterogeneity. Through the years, HCM has acquired the unfortunate reputation as an unrelenting disease with a highly unfavorable and generally grim prognosis, and largely without effective treatment strategies or acceptable expectation for longevity and quality of life.¹ Many patients with HCM, based on evaluation in general cardiology practices, have come to mistakenly believe that their diagnosis is among the least favorable that could confront them.

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Recent Outcome Data

This highly negative perception, created in part by early reports of cohorts impacted by patient referral bias, persists despite recent major advances in diagnosis and treatment, including advanced imaging, contemporary risk stratification, and implantable cardioverter-defibrillators for sudden death prevention²; surgical septal myectomy, alcohol ablation, and heart transplant to reverse progressive heart failure; prophylactic anticoagulation to prevent embolic stroke; and defibrillation and targeted temperature management for out-of-hospital cardiac arrest.¹⁻³ Current estimates of the natural history of HCM indicate a significant reduction in disease-related mortality to 0.5%/year, no different than in the general population, and similar at any age of presentation, including in children and young adults who often demonstrate the most aggressive clinical course.¹

Prior and Present Misconceptions

Older misconceptions regarding a heterogeneous and complex disease such as HCM are difficult to extinguish.³ Novel ideas about patient management and natural history often penetrate slowly and unpredictably into the practicing cardiovascular community because of an understandable preoccupation with more common conditions, namely, ischemic heart disease. Also, the unfortunate perception endures that powerful treatments known to preserve life in HCM are only to be considered palliative if not directly related to basic molecular mechanisms.⁴ Proposals for precision medicine and gene therapy, as well as genome editing and engineering (reverse genetics) and preimplantation genetic diagnosis, are unlikely to resolve clinical issues relevant to the present generation of patients with HCM.

HCM and Other Risks of Living

In this commentary, we have placed the risks of living with HCM into perspective, with comparisons to several chronic and systemic noncardiac or cardiac (but non-HCM) conditions known to impact survival, as expressed in terms of annual and

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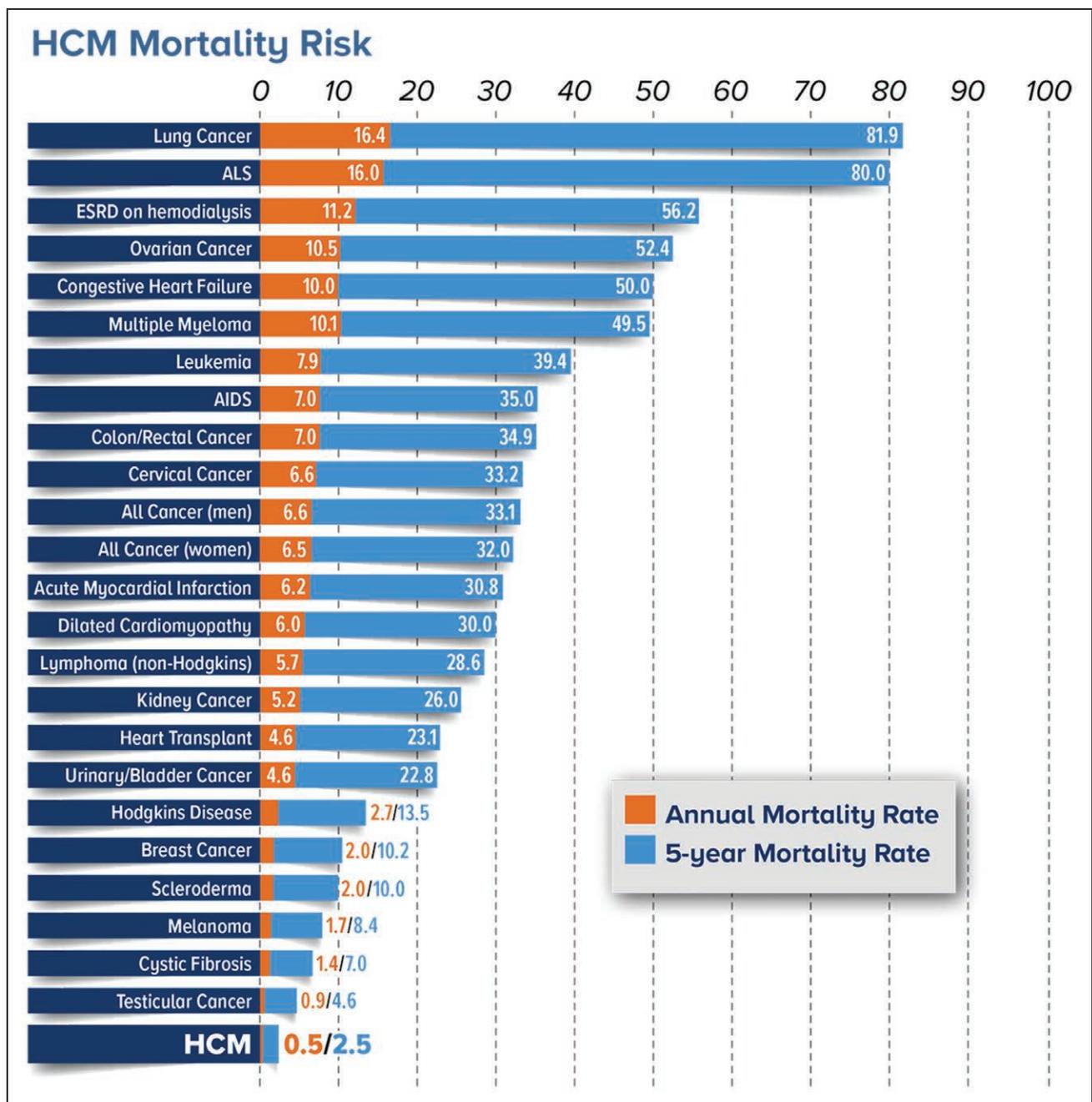


Figure. Annual and 5-year mortality rates (taking into account current standard treatments) for several chronic diseases that impact survival and represent the overall risks of living, as compared with hypertrophic cardiomyopathy (HCM) with contemporary management strategies.

AIDS indicates acquired immune deficiency syndrome; ALS, amyotrophic lateral sclerosis; and ESRD, end-stage renal disease.

5-year mortality rates (Figure). These diseases were selected to characterize the risks of living from the perspective of patients with HCM who otherwise may have been conditioned by misleading information persisting in the practicing community, or on the internet, that exaggerate the consequences of their own disease.

For example, non-HCM forms of congestive heart failure (systolic and diastolic) impact cardiovascular medicine in epic proportions, associated with a 10%/year mortality

(50% over 5 years), 20-fold that of HCM, despite numerous large clinical trials and initiatives with a variety of pharmacological agents and device therapies. Other common cardiovascular diseases such as acute myocardial infarction and dilated cardiomyopathy have mortality rates that also greatly exceed that experienced by patients with HCM when exposed to contemporary treatments (by ~10-fold).

It is notable that despite decades of targeted research and considerable public, private, and govern-

ment support, many cancers continue to be associated with significant mortality rates that far exceed that of HCM (by up to 35-fold), including tumors of lung, breast, and kidney.⁵ Furthermore, many of these diseases retain a predilection for patients >60 years, an age group in which HCM-related deaths are particularly uncommon.¹ Notably, current mortality for all cancers combined is 6%/year,⁵ a rate similar to that previously attributed to HCM, but from data 35 years ago.¹

CONCLUSIONS

Mortality directly attributable to HCM has been substantially reduced because of the application of contemporary treatment options, constituting a paradigm change in disease natural history, and providing many patients with an opportunity to survive to normal or extended longevity with good quality of life. However, the appraisal of HCM by patients is too often impeded by the misconception that prognosis is ominous and no different (if not worse) than many other diseases known to significantly reduce life expectancy.

Therefore, our novel comparisons of mortality rates in major chronic or systemic conditions to that of HCM, place this disease in proper perspective for affected patients by underscoring the substantially more favorable overall prognosis attributable to HCM with current management strategies. These insights should provide a measure of reassurance to many patients with this complex and often misunderstood genetic heart disease.

DISCLOSURES

None.

AFFILIATION

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FOOTNOTES

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