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The Reply

We thank Baviriseaty et al for their interest in hypertrophic cardiomyopathy and our work.[1] However, we wish to clarify some of the uncertainty expressed by Baviriseaty et al in their letter. First, commercial genetic testing is rarely used as a primary diagnostic test for probands with hypertrophic cardiomyopathy, due largely to recognition that disease-causing (pathogenic) sarcomere mutations have proved uncommon in only 20%-30% of consecutive hypertrophic cardiomyopathy cohorts. The standard clinical diagnostic tool in hypertrophic cardiomyopathy to identify otherwise unexplained left ventricular hypertrophy is imaging, most commonly echocardiography over the last 50 years and cardiovascular magnetic resonance (CMR) for the last 10-15 years.

Also, because most patients with hypertrophic cardiomyopathy are asymptomatic when diagnosed, this is itself unhelpful in formulating a primary diagnosis. Of course, as Baviriseaty et al suggest, patients with hypertrophic cardiomyopathy commonly have associated systemic hypertension, which can confuse diagnosis because both diseases are characterized by a wide range of left ventricular wall thickness, or may occasionally coexist with cardiac amyloidosis (about 1%-2% of patients evaluated for hypertrophic cardiomyopathy). Finally, as stated in the Abstract of our article: "while a primary change in the phenotypic expression of hypertrophic cardiomyopathy cannot be excluded by our data, these observations most likely reflect evolving referral practice patterns, including greater diagnostic suspicion for the disease in the community particularly at advanced ages, and/or with less substantial left ventricular hypertrophy."[1]

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Reference

1. Maron BJ, Rowin EJ, Ambe SP, Maron MS. Changing demographics in hypertrophic cardiomyopathy and implications for management: clinical research. *Am J Med* 2022;135(10):1244–6. https://doi.org/10.1016/j.amjmed.2022.05.006.

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