Male monozygotic twins, age 18 years, were both recognized to have clinical findings of hypertrophic cardiomyopathy (HCM). Both were asymptomatic and had been active in sports; there was a family history of 3 HCM-related sudden deaths at ages 18, 32, and 52 years. The twins showed a striking similarity in phenotypic expression of HCM, in which distribution and magnitude of left ventricular hypertrophy was virtually identical. In both patients, hypertrophy was predominant and virtually confined to the posterior portion of ventricular septum and contiguous posterior (inferior) free wall; maximum wall thickness was 18 mm in twin A and 20 mm in twin B (Figure). Anterior ventricular septum (the region most commonly hypertrophied in HCM), as well as anterolateral and posterior free wall and apex were all largely spared from the hypertrophic process. Left ventricular end-diastolic cavity dimensions (42 mm and 40 mm, respectively) and left atrial size (30 mm and 34 mm) were very similar. Relatively mild systolic anterior motion of the mitral valve, preferentially involving the posterior leaflet (a finding regarded as specific for HCM), was present in both twins without mitral-septal contact; continuous wave Doppler showed normal outflow tract velocities (<1.5 m/s) with no evidence of outflow obstruction. The 12-lead ECG did show some differences in precise pattern; there was T-wave inversion in inferior leads II, III, and AVF for twin A, and T-wave inversion in V1 to V3 with prominent Q waves in III, V4, and V5 in twin B.

This opportunity to assess phenotypic expression in monozygotic twins with HCM is unique. The virtually identical distribution of left ventricular hypertrophy in both twins supports the view that genetic background represents the primary determinant influencing the HCM phenotype, and by inference suggests that environmental factors may have limited impact on the morphological expression of HCM. Hypertrophy predominantly involving posterior ventricular septum is rare in HCM (1% of cases), substantiating the claim that the phenotypic similarity evident in the twins is not likely to be coincidental.

Parasternal short-axis echocardiograms obtained from twins A and B. Distribution of hypertrophy is the same in both and is virtually confined to the posterior and inferior segment of ventricular septum (pvs; arrows). Maximum wall thickness is 18 mm in twin A and 20 mm in twin B. Note that anterior ventricular septum (avs) is predominantly of normal thickness in both patients. fw indicates left ventricular free wall. Calibration marks are 10 mm apart.
Hypertrophic Cardiomyopathy in Monozygotic Twins
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