Restrictive Cardiomyopathies

Definition and Epidemiology

RCM is an uncommon form of cardiomyopathy characterized by impaired ventricular filling of nondilated ventricles. RCM can be genetic or acquired. Causes include infiltrative disorders (e.g., amyloidosis, sarcoidosis, Gaucher's disease, Hurler's syndrome, fatty infiltration), storage diseases (e.g., hemochromatosis, Fabry's disease, glycogen storage disease), other disorders (e.g., hypereosinophilic syndrome, carcinoid heart disease), drugs (e.g., serotonin, methysergide, ergotamine), and cancer treatment (e.g., irradiation, chemotherapy).

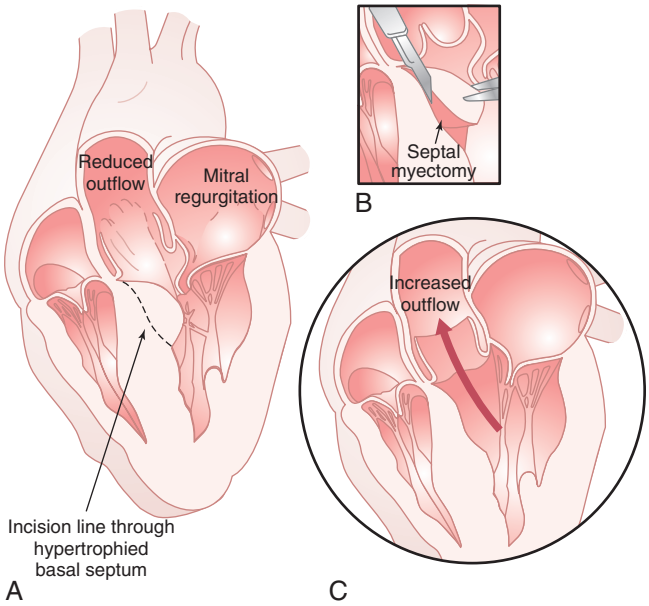

FIGURE 10-3 **A** to **C,** Schematic diagrams of a septal myectomy. (From Nishimura RA, Holmes DR Jr: Clinical practice: hypertrophic obstructive cardiomyopathy, N Engl J Med 350:1320–1327, 2004.)

Pathology

In the purest form of the disease, the atria are disproportionately dilated compared with the normal ventricular size, and the left ventricle has normal or near-normal systolic function in the absence of hypertrophy. Histology is normally nondistinctive and can reveal normal findings or nonspecific degenerative changes, including myocyte hypertrophy, disarray, and degrees of interstitial fibrosis.

Clinical Presentation

Patients often have symptoms and signs of pulmonary and systemic congestion. The most common symptoms include dyspnea, palpitations, fatigue, weakness, and exercise intolerance due to poor cardiac output. As central venous pressure continues to increase in advanced cases, there may be hepatosplenomegaly, ascites, and anasarca. The chest radiograph shows atrial enlargement, pulmonary venous congestion, and pleural effusions.

Diagnosis

The diagnosis of RCM should be considered for patients with predominantly right ventricular heart failure without evidence of cardiomegaly or systolic dysfunction. The correct diagnosis often is not made until months or years after symptom onset. Constrictive pericarditis can mimic RCM, and establishing the correct diagnosis can be challenging. Distinctive features of the two disorders are described in Table 10-3.

TABLE 10-3 DIFFERENTIATION OF RESTRICTIVE CARDIOMYOPATHY FROM CONSTRICTIVE PERICARDITIS

TYPE OF EVALUATION	RESTRICTIVE CARDIOMYOPATHY	CONSTRICTIVE PERICARDITIS
Physical examination	Kussmaul sign present Apical impulse may be prominent Regurgitant murmurs are common	Kussmaul sign may be present Apical impulse usually not palpable Pericardial knock may be present
Electrocardiography	Low QRS voltage (especially in amyloidosis) Pseudoinfarction pattern Bundle branch blocks AV conduction disturbances Atrial fibrillation	Low QRS voltage Repolarization abnormalities
Chest radiography		Calcification of the pericardium may be present
Echocardiography	Marked enlargement of the atria Increased wall thickness (especially in amyloidosis)	Atria usually of normal size Normal wall thickness Pericardial thickening may be seen
Doppler echocardiography	Restrictive mitral inflow (dominant E wave with short deceleration time) No significant variation (<10%) of transvalvular velocities with respiration Reversal of forward flow in hepatic veins during inspiration	Restrictive mitral inflow (dominant E wave with short deceleration time) Increased velocity of RV filling and decreased velocity of LV filling with inspiration; opposite with expiration; variation in velocity exceeds 15% Reversal of forward flow in hepatic veins during expiration
Cardiac catheterization	Prominent atrial *x* and *y* descents (w sign) Dip-and-plateau appearance of ventricular diastolic pressure Diastolic pressures increased but not equalized; LV diastolic pressure higher than RV diastolic pressure	Prominent atrial *x* and *y* descents (w sign) Dip-and-plateau appearance of ventricular diastolic pressure Increase and equalization of diastolic pressures Discordance of RV and LV peak systolic pressures (with inspiration, RV systolic pressure increases and LV systolic pressure decreases)
Endomyocardial biopsy	May reveal specific cause of restrictive cardiomyopathy	No specific findings on endomyocardial biopsy Pericardial biopsy may reveal abnormality
Computed tomography, magnetic resonance imaging		Pericardial thickening

AV, Atrioventricular; LV, left ventricular; RV, right ventricular.