Cardiomyopathies and their resultant systolic and diastolic heart failure remain a major cause of cardiovascular morbidity and mortality. The prevalence of heart failure continues to increase and it remains a major public health threat, particularly in the elderly. The overall annual healthcare expenditure for heart failure continues to increase. While a new diagnosis of heart failure is associated with substantial risk of death within one year, the institution of appropriately guided pharmacologic treatment has led to substantial reductions in cardiovascular mortality. Identification of potential candidates for such treatment can be facilitated through use of echocardiography.

In many patients the diagnosis of a cardiomyopathy is made after the onset of heart failure symptoms, atrial or ventricular arrhythmias, or a stroke. These complications of the underlying cardiomyopathy represent major causes of cardiovascular morbidity and mortality and frequently result in referral for echocardiography. Echocardiography provides an assessment of systolic and diastolic function as well as an estimation of left and right heart filling pressures. In addition, specific echocardiographic features allow the clinician to determine more accurately the aetiology of the cardiomyopathy. Integration of clinical and echocardiographic features now allows for a better assessment of both immediate risk and long term prognosis in patients with a cardiomyopathy.

DEFINITION AND CLASSIFICATION OF CARDIOMYOPATHIES

Cardiomyopathies are defined by the World Health Organization as diseases of the myocardium which result in cardiac dysfunction. The WHO classification of cardiomyopathies includes: hypertrophic, dilated, restrictive, and arrhythmogenic right ventricular cardiomyopathies. Although isolated non-compaction of the ventricular myocardium has not yet been identified as a distinct cardiomyopathy by WHO, it will also be discussed in this review. It is a rare but important cardiomyopathy with distinctive echocardiographic features. The echocardiographic features of specific cardiomyopathies are presented in table 1.

Echocardiographic features of arrhythmic right ventricular dysplasia include focal dilatation, thinning, and hypokinesis of the right ventricle.

GENERAL PRINCIPLES REGARDING USE OF ECHOCARDIOGRAPHY IN PATIENTS WITH CARDIOMYOPATHY

Systolic function

When using echocardiography to measure indices of global systolic function it is important to remember that most of the measures commonly assessed by echocardiography are load dependent. These measures include left ventricular (LV) ejection fraction, dP/dt, and stroke volume. The change in pressure over change in time or dP/dt can be measured by quantifying the slope of the Doppler profile of the mitral regurgitant jet. This is accomplished by measuring the time required for the mitral regurgitant profile to increase from 1 m/s to 3 m/s and is represented by the formula:
There are several less load dependent measures of global systolic function now in clinical use that allow a more comprehensive, less load dependent assessment of ventricular function (table 2). These include measurement of elastance \( (e_{max}) \), which can be derived from the end systolic pressure-volume or pressure-dimension relationships, cyclic variation of integrated backscatter, and LV strain/strain rate. Myocardial strain is measured using data obtained through colour Doppler myocardial imaging and provides non-invasive quantification of myocardial deformation. Strain and strain rate (which represents the velocity of deformation) appear to be less affected by cardiac motion and segmental myocardial tethering.

Though it has recognised limitations, the most common measure of systolic function derived from echocardiography is the LV ejection fraction. There are numerous approaches to measuring ventricular volumes and most make assumptions about the geometry of the ventricle. The biplane Simpson’s method utilising two apical echocardiographic views incorporates much of the shape of the ventricle in its calculation of volume; this is particularly important in the myopathic heart with its alterations in shape.

### DIASTOLIC FUNCTION

The transmitral and pulmonary venous spectral Doppler and mitral annular tissue velocity Doppler profiles can be used to identify patterns of impaired ventricular relaxation and restriction to filling. The propagation of blood flow in the LV cavity can be quantified by colour M Mode imaging. The flow propagation velocity is inversely proportion to  \( \tau \) (time constant of isovolumic relaxation).

\[
dP/dt = \frac{[4(3)^3] - [4(1)^3]}{\text{Time interval}} = \frac{32}{\text{Time interval}}
\]

Mitrail annular Doppler velocity is a relatively new method for quantifying the base to apical longitudinal motion of the ventricle through the cardiac cycle. The ratio of transmitral E velocity to mitral annular E velocity \( (E/E_a) \) is related to left atrial pressure.

Impaired ventricular relaxation is characterised by a decreased transmirtal Doppler E/A ratio, a prolonged transmirtal Doppler E wave deceleration time \( (>240\, ms) \), and a pulmonary venous Doppler systolic/diastolic velocity ratio \( >1 \). A progressive increase in the LV end diastolic pressure can alter this pattern of delayed relaxation and such a “pseudonormal” transmitral Doppler pattern will have a normal E/A ratio, a pulmonary venous Doppler systolic/diastolic wave velocity ratio \( <1 \), but a reduced tissue Doppler (TDI) annular wave (\( E_a \)) velocity \( (<10\, cm/s) \). With further increases in left atrial pressure, the transmitral E wave deceleration time decreases and the E/A ratio increases resulting in a restrictive pattern with E wave deceleration time \( <150\, ms \), E/A ratio \( >2 \), and mitral annular TDI \( E_a \) velocity \( <10\, cm/s \).

### VALVAR REGURGITATION

Tricuspid and mitral regurgitation are common findings in the heart failure patient. In some patients, these volume overload lesions, along with aortic insufficiency, are the aetiology for a dilated cardiomyopathy and can be quantified with echocardiography and Doppler. However, in those with dilated and ischaemic cardiomyopathy, the atrioventricular valve incompetence is typically “functional” and reflects geometric distortions of the chambers, which displace the normal valvar and subvalvar closing mechanisms. The closure of the valve leaflets tends to be displaced into the ventricle and is known as incomplete valve closure. This functional mitral regurgitation is typically a marker of adverse LV remodelling and increased sphericity of the chamber, while functional tricuspid regurgitation is a
marker of both primary right ventricular (RV) systolic dysfunction, RV dilation, and pulmonary hypertension. Since LV remodelling and pulmonary hypertension have prognostic value in heart failure patients it is easy to see that the presence of functional mitral and tricuspid regurgitation assessed by echocardiography also has additive prognostic value.

**DILATED CARDIOMYOPATHY**
Dilated cardiomyopathy is characterised with echocardiography by the presence of a dilated left ventricle with impaired ventricular systolic function. The aetiology of this form of cardiomyopathy may be idiopathic, familial, viral, ischaemic or immunological. Echocardiography may be utilised to determine the degree of impairment of LV systolic function and to characterise diastolic function. Specific echocardiographic features when present are helpful in identifying the aetiology of the dilated cardiomyopathy. The presence of isolated wall motion abnormalities which correlate with a specific coronary artery distribution suggests the presence of underlying coronary artery disease.

RV function is an important predictor of prognosis in individuals with a dilated cardiomyopathy. Not only do individuals with biventricular dysfunction have a lower New York Heart Association (NYHA) functional class, they also have more severe LV dysfunction and worse long term prognosis.7

In addition to LV size and systolic function, specific features of dilated cardiomyopathy which can be assessed by echocardiography include the degree, if any, of RV dilation and systolic dysfunction, an estimation of RV systolic pressures derived from the tricuspid regurgitation Doppler velocities, the presence of LV thrombus and an assessment of left atrial (LA) and LV end diastolic pressure (LVEDP). In the presence of adequate aortic and mitral regurgitant Doppler velocity profiles, and assuming there is no obstruction to left heart inflow or outflow, left atrial and LV end diastolic pressures can be estimated using the simultaneous systolic and diastolic blood pressures using the simplified Bernoulli equation:

\[
\Delta P = 4v^2, \quad \text{where } \Delta P \text{ represents the instantaneous pressure gradient in mm Hg, and } v \text{ represents the instantaneous velocity in m/s.}
\]

- LA pressure = (systolic blood pressure) – (peak LV to LA pressure gradient from mitral regurgitation).
- LVEDP = (diastolic blood pressure) – (maximal AV to LV gradient at end diastole from the aortic regurgitant velocity).

The presence of a restrictive LV Doppler filling pattern results in a tripling of the mortality rate compared to patients with a non-ischaemic dilated cardiomyopathy without this marker of increased risk.8

**HYPERTROPHIC CARDIOMYOPATHY**
Hypertrophic cardiomyopathy is an autosomal dominant disorder associated with a high risk of sudden death in otherwise healthy young individuals, particularly athletes.9 It is characterised by variable degrees of LV hypertrophy and diastolic dysfunction. While genetic screening studies can be useful in predicting risk of sudden cardiac death, echocardiography remains the most useful diagnostic method. Characteristic echocardiographic features of hypertrophic cardiomyopathy include variable degrees of RV and/or LV hypertrophy. Several forms of the disease are seen with echocardiography: hypertrophic non-obstructive cardiomyopathy, hypertrophic obstructive cardiomyopathy (HOCM), and apical variant of hypertrophic cardiomyopathy. In those with HOCM, systolic anterior motion of the mitral valve and the presence of left ventricular outflow tract (LVOT) obstruction are noted on echocardiography. LV wall thickness in excess of 13 mm without apparent cause, or a ratio of the septal to posterior wall thickness of > 1.3, is diagnostic of HOCM. Systolic anterior motion of the mitral valve and LVOT obstruction occurs in approximately 25% of individuals with hypertrophic cardiomyopathy. This obstruction occurs as anterior motion of the anterior mitral leaflet during systole results in a narrowing of the LVOT. The presence of a resting LVOT gradient of > 30 mm Hg in individuals with hypertrophic cardiomyopathy is predictive of an increased risk of death related to the hypertrophic cardiomyopathy, progression to NYHA class III or IV heart failure, or death from heart failure or stroke.9 Right ventricular outflow tract obstruction occurs less frequently.

Echocardiography can be utilised to tailor treatment for individuals with hypertrophic cardiomyopathy. The response to medical treatment for hypertrophic cardiomyopathy, including a decrease in an outflow tract gradient or improvement in diastolic function, can be evaluated by serial echocardiographic studies. The decision to proceed with non-medical therapeutic options, including surgical septal reduction, percutaneous alcohol septal reduction, or DDD pacing, can also be guided by echocardiography. The presence and degree of LVOT obstruction detected by echocardiography aids the clinician in determining whether or not to perform septal reduction. Surgical septal reduction is recommended in individuals with symptoms refractory to medical treatment in the presence of a resting gradient of > 50 mm Hg. Indications for percutaneous alcohol septal reduction include a septal thickness > 18 mm Hg, the presence of a resting LVOT gradient of > 30 mm Hg, or an inducible gradient of > 60 mm Hg in the presence of NYHA functional class III or IV symptoms unrelated by maximal medical treatment.10 Echocardiographic guidance during alcohol septal ablation, using intravenous contrast selectively into septal coronary vessels, allows the operator to identify the correct vessel supplying the appropriate myocardial territory, thus decreasing the likelihood of infarction of other portions of the heart and reducing the need for permanent pacing.11

In addition to characterising the degree of ventricular hypertrophy in affected individuals, echocardiography can also be used to screen asymptomatic relatives of affected individuals. Most recently, tissue Doppler imaging has been used to identify abnormal diastolic function in individuals with genotypes for hypertrophic cardiomyopathy before the development of significant ventricular hypertrophy. The sensitivity and specificity of the mitral annular early diastolic (Ea) velocity varies in the different reports and this may relate to the genotype studied.12

**RESTRICTIVE CARDIOMYOPATHY**
While restrictive cardiomyopathies are less common than dilated and hypertrophic cardiomyopathies, they are associated with greater morbidity and mortality. As the name implies, restrictive cardiomyopathies are associated with impaired ventricular filling and increased LV end diastolic pressure. Echocardiographic features include biatrial dilatation, hypertrophied ventricles with decreased compliance, initially small LV cavities, and normal to depressed systolic
function. Infiltrative processes and metabolic storage diseases including amyloidosis (which will be discussed separately), haemochromatosis, sarcoidosis, Fabry’s disease, and glycogen storage diseases are the most frequent causes of restrictive cardiomyopathies. Less common forms of restrictive cardiomyopathies include endocardial fibrosis associated with the hypereosinophilic syndrome (Loeffler’s cardiomyopathy) and idiopathic restrictive cardiomyopathy. The echocardiographic features of endocardial fibrosis associated with hypereosinophilia include haemodynamic evidence of restriction and obliteration of the ventricular apices caused by deposition of thrombus and eosinophilic cationic protein. The regional myocardial motion adjacent to this deposition remains normal (fig 1).

While the clinical distinction between a restrictive cardiomyopathy and pericardial constriction in a patient with symptoms of systemic congestion can be challenging, echocardiography can non-invasively separate these two distinct entities. This distinction is extremely important since the treatments are dramatically different; pericardial constriction can be successfully treated with pericardial stripping while the treatment of restrictive cardiomyopathy is largely aimed at improvement in symptoms. The constraining effect of a thickened pericardium leads to rapid early diastolic dilation and flow reversal, and two distinct septal motions which are unrelated to contraction. One of these septal motions is high amplitude and low frequency, and reflects differential filling of the ventricles during the phases of the respiratory cycle which is due to ventricular interdependence. The second is a low amplitude, high frequency diastolic septal motion which reflects differences in timing of the filling of the ventricles (the “septal bounce”). While the restrictive pattern of the transmitral E wave is similar in both constriction and restriction, there is no significant respiratory change in the transmitral inflow pattern associated with restrictive cardiomyopathy. Restrictive processes are also much more likely to be associated with pulmonary hypertension and the presence of diastolic mitral regurgitation. Tissue Doppler imaging can also distinguish between constriction and restriction. While the early mitral annular velocity (Ea) is notably decreased in restriction reflecting a myocardial abnormality, it remains normal in the presence of pericardial constriction since in this disease the myocardium typically remains normal.

Idiopathic restrictive cardiomyopathy is a rare entity distinguished from the other forms of restrictive cardiomyopathy by the presence of normal ventricular wall thickness. This disease is seen in individuals where other aetiologies for cardiomyopathy have been excluded such as connective tissue disease, carcinoid syndrome, amyloidosis, haemochromatosis, eosinophilic syndrome, malignancy, radiation exposure, cardiotoxic drug exposure, or history of alcohol abuse. In addition these individuals do not have a history of ischaemic heart disease, treated hypertension for more than five years, or organic valvar, pericardial or congenital diseases.

CARDIAC AMYLOIDOSIS

Myocardial infiltration by amyloid fibrils can occur in primary, familial, secondary, and senile amyloidosis. The degree of involvement is variable depending upon the type of amyloidosis. Two dimensional echocardiography remains the ideal method for identifying and following individuals with cardiac amyloidosis.

Characteristic two dimensional and Doppler echocardiographic features have been described in individuals with cardiac amyloidosis. Two dimensional features include thickening of the LV walls, increased reflectivity of these walls (the “speckled” or “granular” myocardium), biventricular enlargement, thickening of the interatrial septum, thickening and regurgitation of the mitral and tricuspid valves, and the presence of a small pericardial effusion (fig 2). Transmitral Doppler flow patterns in patients with amyloidosis exhibit evidence of diastolic dysfunction. Characteristic transmural Doppler patterns representative of early impaired relaxation and later restrictive filling have been demonstrated in this population. An increase in both the pulmonary venous Doppler atrial reversal duration and the ratio of this atrial reversal duration to the transmitral A wave duration have been observed in these patients and reflect increased LA pressure. Cardiac amyloidosis should be considered in individuals in whom several of these echocardiographic features are observed. The coexistence of increased thickening of the LV walls on echocardiography yet low voltage on electrocardiography is highly suggestive of amyloid infiltration of the myocardium.

Differentiating cardiac amyloidosis from hypertrophic cardiomyopathy can be difficult on echocardiography. Asymmetric septal thickening can result from focal amyloid deposition and can mimic hypertrophic cardiomyopathy. The presence of decreased LV function would argue strongly against hypertrophic cardiomyopathy. The presence of highly reflective myocardium can also be seen in individuals with primary and secondary causes of LV hypertrophy, ventricular fibrosis, and other infiltrative processes. The integration of clinical, echocardiographic, and electrocardiographic data is essential when making the diagnosis of cardiac amyloidosis.

While the long term prognosis is substantially worse in patients with primary amyloidosis, the echocardiographic distinction between familial, primary, and secondary amyloidosis can be difficult. Impaired LV systolic function is more common in primary amyloidosis than in the other forms.
Doppler tissue echocardiography is helpful in differentiating amyloid patients from those with similar two dimensional echocardiographic features but without amyloidosis. Abnormally low tissue Doppler diastolic velocities are present in individuals with cardiac amyloid compared with control patients.16

ARRHYTHMIC RIGHT VENTRICULAR DYSPLASIA

Arrhythmic right ventricular dysplasia (ARVD), also known as arrhythmic right ventricular cardiomyopathy, is an idiopathic cardiomyopathy that appears to be genetically transmitted. ARVD is associated with RV fibrosis, fatty infiltration, and dysfunction. It can be complicated by symptoms of heart failure, heart block, and ventricular arrhythmias, and is associated with an increased risk of sudden cardiac death. Pathologic features include loss of RV myocardium, fibrofatty tissue replacement, and resultant aneurysmal dilation of the “triangle of dysplasia” (diaphragmatic, infundibular, and apical RV regions). While RV involvement is universal, the left ventricle is involved less frequently and the degree of involvement is less severe.

The diagnostic criteria for ARVD include a family history of ARVD or sudden cardiac death, imaging (echocardiographic, magnetic resonance imaging, or ventriculography), evidence of characteristic RV dysfunction, typical electrocardiographic features (electrocardiographic depolarisation and repolarisation abnormalities), and pathologic evidence of fibrofatty replacement of myocardium on tissue obtained by endomyocardial biopsy. The electrocardiographic abnormalities include right precordial T wave inversion, the presence of an epsilon wave (small amplitude potential occurring after the QRS complex), and prolonged QRS duration.

Echocardiographic features are variably present and include RV and RV outflow tract dilation, RV wall thinning, aneurysms of the posterior RV wall or RV free wall, and highly reflective moderator band and trabecular disarray. The relative value of various echocardiographic features is currently being examined in the multicentre study of arrhythmogenic RV dysplasia.

ISOLATED LV NON-COMPACTION

While isolated ventricular non-compaction (IVNC) remains an “unclassified” form of cardiomyopathy, its echocardiographic and clinical features have recently been characterised and IVNC is now being recognised with increasing frequency. This disease, which results from an interruption of the normal process of embryologic myocardial compaction, is associated with a high risk of systolic dysfunction, systemic embolisation, and ventricular arrhythmias. Accurate recognition requires knowledge of the echocardiographic features which include the presence of a thin (compacted) epicardium and a thick, spongy endocardial (non-compacted) surface with extensive trabeculation and sinusoid formation. Communication between the deep intertrabecular spaces and the ventricular cavity can be demonstrated by colour Doppler imaging. In the majority of cases the non-compacted myocardium involves the mid lateral, apical, or inferior walls. A ratio of non-compacted to compacted myocardium of > 2:1 is diagnostic of this entity. Additionally, both regional and generalised LV hypokinesis has been observed in this condition. RV involvement has been described in some patients with this condition. The hypokinetic segments can occur in both the affected area and the surrounding normally compacted myocardial segments.17

Numerous studies have been performed demonstrating the prognostic value of echocardiographic indices of cardiac size and function in heart failure patients. Regardless of the aetiology of the heart failure, the findings suggest that these echocardiographic measures provide prognostic value and should be integrated in the assessment of these patients. Echocardiographic measures with documented prognostic value in heart failure patients include LV systolic function, RV systolic function, LV diastolic function, LV size, RV size, LA size, severity of mitral regurgitation, severity of tricuspid regurgitation, RV systolic pressure, ventricular synchrony, and measures of contractile reserve.

LV ejection fraction has long been the primary index used as a marker of risk in heart failure and recently the strength of this marker has been demonstrated even in the elderly.18 While most studies have focused on ejection fraction as a marker of prognosis in chronic heart failure, the same relation holds in acute heart failure and cardiogenic shock. Particularly in cardiogenic shock the value of ejection fraction has been shown to remain present regardless of type of treatment.

While initial measures of diastolic filling and function by echocardiography were shown to be of value in patients with symptoms of heart failure but preserved systolic function, the prognostic value of these diastolic indices remain even when the ejection fraction is low.19 These measures appear to be additive in value to the ejection fraction and may even be stronger measures of prognosis than LV ejection fraction. The transmitral pulse wave Doppler deceleration time has been shown to be a powerful predictor of functional capacity and correlates with maximum oxygen consumption. Thus it is no surprise that measures of diastolic filling have prognostic value in heart failure patients.

For example, in symptomatic congestive heart failure patients the restrictive filling pattern of transmitral Doppler, especially a deceleration time < 140 ms, has been shown to be the single best predictor of cardiac death in patients with ischaemic and dilated cardiomyopathy.20 For those with cardiac amyloid diastolic function has been shown to be a stronger predictor of cardiac death than LV wall thickness or systolic function regardless of symptom status.21

The pattern of transmitral Doppler velocity profiles are highly dependent on preload and, as described above,
pseudonormal patterns can be produced. Since the diastolic measures with prognostic value are indirect measures of LA and LV diastolic pressure and the pseudonormal patterns are a response to increased diastolic pressures, it is logical to ask if pseudonormal patterns have prognostic value in heart failure patients. In fact the pseudonormal transmitral Doppler filling pattern has been shown to identify patients with an intermediate prognosis. Specifically those with abnormal relaxation patterns appear to have the lowest all cause death and rehospitalisation for congestive heart failure compared to those with pseudonormal filling or restrictive filling. In both ischaemic and idiopathic dilated cardiomyopathy, the prognostic value of tricuspid regurgitation has been demonstrated. Additional for patients with impaired systolic function increasing degrees of mitral regurgitation have a direct impact on survival. While the majority of investigations have focused on the value of valvar regurgitation in assessing prognosis in chronic heart failure, similar relations are seen with acute heart failure shock where the one year survival has been shown to depend on the extent of mitral regurgitation assessed at presentation with shock. Contractile reserve and myocardial viability are important prognostic factors in ischaemic cardiomyopathy. Viable myocardium identified by dobutamine echocardiography identifies the subset of ischaemic cardiomyopathy patients who benefit most from revascularisation. Specifically those with myocardial viability who undergo revascularisation have a threefold lower long term mortality than those ischaemic cardiomyopathy patients without viability or those with viability who do not undergo revascularisation.

Recently investigators have focused on LV synchrony as a marker of prognosis and shown that those with widened QRS duration have a poorer long term survival than those with coordinated wall motion and dilated cardiomyopathy.

**SUMMARY**

Echocardiography is a powerful tool for assessing systolic and diastolic heart failure, the specific aetiology of the cardiomyopathy, assessing response to treatment, and categorising prognosis. It is non-invasive, relatively low cost, does not expose patients to ionising radiation, and serial studies can be done at the bedside. Thus echocardiography continues to have advantages over competing technologies.

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Utility of echocardiography in the evaluation of individuals with cardiomyopathy

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