The Cardiomyopathies, a Review for the Primary Physician

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The cardiomyopathies constitute a group of diseases with direct involvement of the heart muscle itself, and is a significant cause of morbidity and mortality. The World Health Organization (WHO) and the International Society and Federation of Cardiology (ISFC) have promulgated a classification taking into consideration the etiology and pathophysiology, which includes dilated cardiomyopathy, hypertrophic cardiomyopathy, arrhythmogenic right ventricular cardiomyopathy, restrictive cardiomyopathy, and unclassified cardiomyopathies. Over 25 causes are associated with the development of the cardiomyopathies. The classification of the diseases affecting the heart and causing the cardiomyopathies is presented including the highlights of the most important data for diagnosis and management of each one.

Key words: Cardiomyopathy, Dilatedc Cardiomyopathy, Hypertrophic cardiomyopathy, Arrhythmogenic right ventricular cardiomyopathy, Restrictive cardiomyopathy, Unclassified cardiomyopathy

The term *cardiomyopathy* is used to describe a group of conditions associated with myocardial dysfunction where the predominant feature is the involvement of the cardiac muscle itself (1). According to the World Health Organization (WHO) and the International Society and Federation of Cardiology (ISFC), there are five categories in which the cardiomyopathies could be classified: dilated, hypertrophic, restrictive, arrhythmogenic right ventricular and unclassified cardiomyopathy (Table 1) (2).

Multiple conditions could present as a cardiomyopathy (specific cardiomyopathies), including cardiovascular, metabolic, infectious/inflammatory, toxic, genetic, tachycardia, and pregnancy. Ischemic cardiomyopathy is the most frequent condition in developed countries and frequently the final result is the development of a heart failure syndrome. In several diseases there may be features of more than one type of cardiomyopathy (restrictive and dilated), and prognosis is variable depending of the type of cardiomyopathy present.

Signs and symptoms are the same as in heart failure including shortness of breath, progressive dyspnea on exertion, orthopnea, leg edema, paroxysmal nocturnal dyspnea, jugular venous distension, systolic murmurs of mitral and tricuspid valve regurgitation, enlarged and pulsatile liver, S3 or S4 gallop, displaced apical impulse, and accentuated pulmonary component of S2 (if pulmonary hypertension is associated). The presence of conduction disturbances is common and there is an increased risk of sudden death due to malignant ventricular arrhythmias. Progressive deterioration is common, with worsening heart failure and death. For a better understanding of the different cardiomyopathies, each condition will be discussed separately in the next sections of this review.

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Definition and Classification of Cardiomyopathies

I. Dilated Cardiomyopathy

This condition (previously known as congestive cardiomyopathy) is defined by the presence of a dilated left ventricle (end-diastolic size more than 115% of predicted for age and body surface area) and an ejection fraction of less than 40%. Both ventricles could be dilated with impaired contraction. Mechanical dysfunction is the result of combined myocyte apoptosis and necrosis with increased myocardial fibrosis after exposure to multiple biological insults, which could be toxic (alcohol, cocaine) or mechanical (chronic exposure to volume overload). Cardiac chambers dilate due to myocyte failure and

Table 1. Basic Scheme for the Classification of the Cardiomyopathies

I. Dilated cardiomyopathy	Ischemic cardiomyopathy Idiopathic dilated cardiomyopathy Toxic cardiomyopathy Hypertensive heart disease Infective cardiomyopathy Valvular heart disease Peripartum cardiomyopathy Tachycardia-induced cardiomyopathy Cardiomyopathy secondary to metabolic conditions
II. Hypertrophic cardiomyopathy (3-4)	Familial hypertrophic cardiomyopathy/ Hypertrophic obstructive cardiomyopathy
III. Arrhythmogenic Right Ventricular Cardiomyopathy	Arrhythmogenic right ventricular dysplasia
IV. Restrictive Cardiomyopathy (5)	Idiopathic restrictive cardiomyopathy Löffler endocarditis Endomyocardial fibrosis Amyloid heart disease Sarcoidosis Radiation carditis Carcinoid heart disease Hemochromatosis Glycogen storage diseases (Gaucher's, Hurler's, and Fabry's disease)
V. Unclassified Cardiomyopathy	Noncompacted myocardium Systolic dysfunction with minimal dilatation Mitochondrial cardiomyopathy Endocardial fibroelastosis

cytoskeletal uncoupling, a process known as "adverse ventricular remodeling" (6). As presented in table 1, the complete list of dilated cardiomyopathies is extensive. The highlights of some of the most important and most frequently encountered specific conditions within the dilated cardiomyopathies will be presented.

a. Ischemic cardiomyopathy. More than 60% of patients with dilated cardiomyopathy and heart failure have an ischemic etiology in which myocardial dysfunction comes from loss of contractile function secondary to localized myocyte necrosis with resultant scar formation after myocardial infarction. Myocytes distal to the infarcted myocardium have increased wall stress and undergo adverse remodeling with development of chamber dilatation that increases the myocardial dysfunction. Mitral valve regurgitation is common due to papillary muscle dysfunction and failure of mitral valve leaflets to coapt in the presence of a dilated ventricle. Systolic function decreases and myocardial oxygen demand increases secondary to the overload state. Atrial fibrillation and ventricular arrhythmias are common, with further deterioration of heart function as a consequence (6).

b. Idiopathic dilated cardiomyopathy. Most of the patients with non-ischemic dilated cardiomyopathy fall in this group. It is 3 times more common in males and blacks, and the estimated incidence is 5-8 per 100,000 per year. About a fourth of the causes of congestive heart failure in the United States are due to this condition. Disease course is one of progressive deterioration and long-term prognosis is poor (25% die within 1 year of diagnosis, 50% die in 5 years, 25% improve spontaneously) (1). The coronary arteries are usually normal. Cardiac valves are normal and intracavitary thrombi are common. Several mechanisms proposed for the development of idiopathic dilated cardiomyopathy involves that it could be secondary to an acute myocarditis (most likely of viral etiology) and that cytotoxic insults and immunological abnormalities could be involved. Arrhythmias and sudden cardiac death are common and can occur at any stage of this condition.

c. Toxic cardiomyopathies. Two samples of this condition will be summarized. Among these are the alcoholic cardiomyopathy and the therapeutic or illicit drug-induced cardiomyopathies.

Alcoholic cardiomyopathy. Is the second most common cause of non-ischemic dilated cardiomyopathy. It accounts for approximately 4% of all cardiomyopathies. If alcohol consumption is stopped at an early stage, progression of disease could be decreased and in several cases LV dysfunction could be reversed. Cardiomyopathy could be developed secondary to a direct toxic effect of alcohol or its metabolites, or as a consequence of nutritional effects and dietary deficiencies (thiamine deficiency, beriberi heart disease), or secondary to the toxic effect of additives in certain beverages (cobalt used in beer back in the mid 1960's) (1). Hypertension and coronary artery disease is also commonly seen in heavy alcohol use. Biventricular failure may be present. Management should be directed to total abstinence of alcohol intake, treatment of heart failure and thiamine supplement as needed.

Therapeutic and illicit drugs. Include other known toxic agents to the heart such as cocaine and

amphetamines (including ecstasy) that could cause a dilated cardiomyopathy. It is believed to be secondary to direct myocyte toxicity, infarction, tachycardia-induced, or hypertension. Doxorubicin and trastuzumab are chemotherapeutic agents known to cause cardiomyopathy and monitoring with BNP level during treatment with these agents could be useful in these patients (6).

d. Hypertensive heart disease. The initial presentation of this condition is usually a preserved systolic function in the presence of left ventricular hypertrophy and diastolic dysfunction. As remodeling progresses the hypertrophied left ventricle dilates and systolic dysfunction develop. It is a common cause of heart failure in elderly women and atrial fibrillation is frequently seen.

e. Infective cardiomyopathies. There are multiple infectious organisms that could cause an acute myocarditis. Dilated cardiomyopathy could be a consequence of a previous infection with these infectious agents, including HIV and Hepatitis C virus. Also parasitic agents like Trypanosoma cruzi, Toxoplasma gondii and Trichinella spiralis may cause a myocarditis with further development of cardiomyopathy.

HIV cardiomyopathy. Occurs in 15.9 cases/1000-infected patients. Dilated cardiomyopathy is strongly associated with CD4 counts of less than 100 cells/ml. Proposed mechanisms for development of cardiomyopathy includes myocardial infection with HIV itself, an autoimmune response to the viral infection, cardiotoxicity to therapeutic or illicit drugs, nutritional deficiencies, cytokine over expression, and opportunistic infection. Rapid onset of CHF in HIV patients has a grim prognosis with more than half of patients dying within 6-12 months of presentation. Besides the treatment of heart failure, management includes treatment of opportunistic infections (1).

Trypanosomiasis (Chaga's Disease) - is frequent in Central and South America. Chaga's disease and infection is transmitted to humans through the bite of *reduviig* bug (found in armadillos, raccoons, skunks, domestic dogs and cats). Approximately 30% of infected patients develop the chronic condition 20 years after the infection was acquired. The proposed mechanisms for development of the cardiomyopathy are an autoimmune response with antibody formation against myocyte sarcoplasmic reticulum, laminin, and other structures, or a parasympathetic denervation. Anatomical findings include dilatation and hypertrophy of all chambers and aneurysm formation in the apex of the heart. Extensive fibrosis and right atrial thrombus formation is also seen. Heart failure is often with right-sided symptoms and tricuspid valve regurgitation. Angina, arrhythmias, syncope and sudden death are common. Parasites could be detected in the blood of infected patients and the complement-fixation test developed by Machado and Guerreiro could be useful in the diagnosis of chronic disease. Prognosis is not good and once decompensation occurs, the progression to death is the rule. Cardiac transplantation has not provided good results at this moment (1).

f. Valvular Heart Disease. In the presence of significant valvular disease, adverse remodeling occurs secondary to pressure overload and volume overload states. Along with increase in valvular deterioration comes a process of myocyte hypertrophy, chamber dilatation, and myocardial fibrosis. Sub endocardial ischemia and myocyte necrosis occurs with further deterioration in heart function. Concomitant coronary artery disease and arrhythmias are common (atrial fibrillation in mitral valve regurgitation or stenosis) (6).

g. Peripartum cardiomyopathy. Occurs during the last month of pregnancy or within the next 5 months postpartum (most cases 75% occurs within 2 months postpartum). Risk factors identified include age > 30 years, multiparity, twin pregnancy, African descent, and family history of peripartum cardiomyopathy. Recovery in 6 months occurs in 50% of patients. Further pregnancies should be discouraged (6).

h. Tachycardia-induced cardiomyopathy. Myocardial dysfunction can occur after prolonged exposure to elevated heart rates. Atrial fibrillation that is persistent or permanent induces remodeling of the atria and dilated cardiomyopathy could occur secondary to adverse remodeling. Rate control and restoration of sinus rhythm could improve the systolic function in these patients (6).

i. Cardiomyopathy secondary to metabolic conditions. Several endocrine disorders could induce cardiomyopathy. Adrenal insufficiency, thyrotoxicosis, hypothyroidism, acromegaly, and pheochromocytoma are some of these. A diabetic cardiomyopathy has also been recently described. Malnutrition and vitamin deficiencies are also associated to the development of dilated cardiomyopathy (Vitamin B deficiency, carnitine, selenium, calcium, and phosphate).

Hypertrophic Cardiomyopathy

Hypertrophic cardiomyopathy is a genetic cardiac disorder characterized by myocardial hypertrophy that is out of proportion to the hemodynamic load to the heart. As most of these patients do not have evidence of an outflow gradient or obstruction, it is now preferred to use

the term hypertrophic cardiomyopathy. It is caused by a missence mutation in 1 of at least 10 genes encoding the proteins of the cardiac sarcomere. Familial hypertrophic cardiomyopathy is an autosomal dominant disease in at least 50% of cases. Approximately 1 in 500 (0.2%) adults in the general population have phenotypic expression of this condition including massive hypertrophy involving the ventricular septum (3). Many patients are asymptomatic, but some present with limiting symptoms including dyspnea, angina, and syncope. The initial presentation could be sudden death. Several features of this condition are dynamic left ventricular outflow tract obstruction (present in 30-50% of patients), diastolic dysfunction (in contrast to dilated cardiomyopathy in which the predominant feature is systolic dysfunction), mitral valve regurgitation, cardiac arrhythmias, and myocardial ischemia. It is the most common cause of sudden death among young athletes. Obstruction in hypertrophic cardiomyopathy is not always evident and has to be provoked by changing cardiac hemodynamics. With increasing obstruction of the left ventricular outflow tract, the systolic pressure in the left ventricle increases, causing increased left ventricular diastolic pressure, myocardial ischemia, and a decreased cardiac output. Common complications of this condition include infective endocarditis, arrhythmias, and heart failure. A systolic ejection murmur that increases during maneuvers that decrease the preload, and the presence of left ventricular hypertrophy on ECG or two-dimensional echocardiography are common findings. First-degree family members should be screened with echocardiography and prophylaxis against infective endocarditis should be recommended to these patients (3).

The initial treatment recommended is beta-blockers, enhancing the diastolic filling time, and blocking the catecholamines that could increase the outflow tract obstruction. If beta-blockade is insufficient, disopyramide could be added due to its negative inotropic effect (4). If beta-blockers are contraindicated, a calcium-channel blocker could be used instead. Options for the treatment of hypertrophic cardiomyopathy include surgical septal myectomy, alcohol-induced septal ablation, and dual-chamber pacing. At this moment, surgical septal myectomy is the treatment of choice when medical therapy fails to improve symptoms of obstruction (3).

Atrial fibrillation is poorly tolerated in patients with hypertrophic obstructive cardiomyopathy, for which prompt cardioversion is recommended. If atrial fibrillation is paroxysmal or chronic, anticoagulation is recommended for the prevention of thromboembolic events. Prevention of sudden death in patients with high-risk features may need the implantation of an automatic defibrillator.

Arrhythmogenic Right Ventricular Cardiomyopathy

This type of cardiomyopathy is rare, also known as arrhythmogenic right ventricular dysplasia (1). Primary mechanism is the patchy myocardial cell in the right ventricle, and to a lesser extent in the left ventricle, with the replacement of cells by adipose and fibrous tissue (fatty infiltration). It has male predominance and is familial in 33-50% of patients with an autosomal dominant inheritance. Usually the patient presents with palpitations and syncope. Re-entrant ventricular tachycardia is common. There is an increased risk of sudden death. Right ventricle is dilated and poorly contractile. Treatment with beta-blockers, sotalol, and amiodarone is useful for arrhythmias, and radio frequency ablation may also be effective. ICD implants are reserved for recalcitrant cases. In selected cases transplantation is an alternative treatment.

Restrictive Cardiomyopathy

Restrictive cardiomyopathy is defined by an impaired ventricular filling in the presence of normal or decreased diastolic volume of one or both ventricles (5). A hallmark of this condition is diastolic dysfunction. The ventricular walls are excessively rigid causing an impairment of ventricular filling. Systolic function usually is preserved. This condition has similar hemodynamics as constrictive pericarditis, and distinction between these two conditions is very important since constrictive pericarditis could be cured surgically. Only a very small percent of the population develop this condition (less than 5%).

Principal findings in restrictive cardiomyopathy include signs and symptoms of right and/or left sided heart failure. Commonly patients present with elevated jugular venous pressure, peripheral edema, and ascites. Shortness of breath and pulmonary edema are present when left sided failure is present. Atrioventricular blocks and arrhythmias are common with atrial fibrillation been poorly tolerated. Some of these cardiomyopathies are familial and could be related to skeletal myopathy.

Cardiomyopathies with restrictive physiology could be classified in 3 subheadings: *primary* (idiopathic restrictive cardiomyopathy, Löffler endocarditis, endomyocardial fibrosis), *secondary* if another primary condition is present such as infiltrative diseases (amyloidosis, sarcoidosis, radiation carditis), and *storage diseases* (hemochromatosis and the glycogen storage disorders, including Gaucher's disease, hurler's disease, and Fabry's disease) (6).

Primary Restrictive Cardiomyopathies

a. Idiopathic restrictive cardiomyopathy - It is sometimes familial with autosomal dominant inheritance and is associated to distal skeletal myopathy. Symptoms appear during the third-fourth decade of life. This condition could also occur without the distal myopathy. When restrictive cardiomyopathy presents in childhood, the prognosis is worse than in adults. There is increase in cardiac weight, bi-atrial enlargement and thrombi formation in the atrial appendages. Also common is the finding of patchy endocardial fibrosis (5).

b. Löffler endocarditis (Hypereosinophilic syndrome) - Men in their 40's that live in a temperate climate and have eosinophilia (1500 eosinophils/mm^3) for at least 6 months are more likely to develop this condition. There is biventricular involvement with mural endocardial thickening of the inflow portion of the apex in both ventricles. Cardiac dysfunction is present in most patients and systemic embolism is frequent. Treatment includes corticosteroids during the acute myocarditis, and combination with cytotoxic drugs, interferon, digitalis, diuretics, anticoagulation, and afterload reducing agents. Palliative surgery is possible during the fibrotic stage (1).

c. Endomyocardial fibrosis - It is most commonly seen in tropical and subtropical Africa (Uganda and Nigeria). It is most common in blacks. Fibrous endocardial lesions of the inflow of the right and left ventricles characterize it and often have involvement of the atrioventricular valves. The right atrium is dilated and pericardial effusion may be present. One half of patients have biventricular disease and symptoms vary depending of the ventricle involved. The presence of atrial fibrillation is a poor prognostic sign. Digitalis is useful for the heart rate control and surgery is palliative (1).

Secondary Restrictive Cardiomyopathies

a. Cardiac amyloidosis - Cardiac involvement is more common in primary amyloidosis caused when a monoclonal population of plasma cells produced immunoglobulin light chains, due to multiple myeloma (most cases) (5). The ventricular walls are firm and rubbery. There is also focal thickening or deposits on cardiac valves (amyloid could also be deposited in the intramural coronary arteries and veins). In 10% of patients, orthostatic hypotension may be present. Atrial fibrillation, sick sinus syndrome, syncope and sudden death are common.

The echocardiogram shows an increased wall thickness with granular sparkling texture, small ventricular chambers, dilated atria, thickened septum and pericardial effusion. Also scintigraphy with technetium-99m pyrophosphate could be used in the evaluation of amyloid heart disease. Diagnosis is done by abdominal fat aspirate (most commonly), biopsy of rectum, gingiva, bone marrow, liver, kidney, or endomyocardial biopsy. Management includes anticoagulation, insertion of a permanent pacemaker if indicated, and digitalis. Calcium channel blockers exacerbate heart failure and cardiac transplantation has poor outcomes (1).

b. Cardiac sarcoidosis - Interstitial inflammation results in diastolic impairment, and subsequent injury leads to fibrosis and systolic dysfunction. The myocardial wall shows diffuse hypokinesis but the apex is usually spared. Right-sided failure is the result of pulmonary involvement. Cardiac conduction defects are common, including heart block, ventricular arrhythmias, and sudden death. Aneurysm formation is seen. Permanent pacing may be helpful in some patients and ICD placement may be necessary. The treatment of cardiac sarcoidosis includes corticosteroids, hydroxychloroquine, methotrexate and possibly cyclophosphamide.

c. Radiation carditis - Clinically significant radiation-induced cardiac dysfunction secondary to myocardial and endocardial fibrosis occurs in a minority of patients, long after the radiation therapy. Cardiac damage is related to the dose of radiation, and amount of myocardium exposed to radiation. The pericardium is the most commonly affected site and patients develop chronic pericardial effusion or pericardial constriction. Left-sided valvular regurgitation and stenosis may be seen. Heart block and arrhythmias are also common (1).

d. Carcinoid heart disease - Only when metastasis reaches the liver will result in carcinoid heart involvement. There is preference for the right side of the heart with fibrous plaques that involve the tricuspid valve, pulmonary valve, endocardium, the intima of the vena cavae, pulmonary artery and coronary sinus. The fibrotic tissue disrupts the valve anatomy causing stenosis and regurgitation. Management includes digitalis, diuretics, and somatostatin analogs. Invasive therapy consists of balloon valvuloplasty of right-sided valves or surgical replacement, which carries a high mortality (35%) (1).

Storage Disorders

a. Hemochromatosis. 'Bronze diabetes" results from the excessive deposition of iron in the parenchymal tissue of several organs including the heart. It could be idiopathic or inherited as a familial autosomic-recessive disorder. The ventricular walls are thickened and the heart is dilated. There is common involvement of the conduction system with frequent arrhythmias. Finding elevated iron levels, markedly elevated ferritin levels, and normal or low total

iron binding capacity may do diagnosis. Endocardial biopsy could confirm diagnosis. Treatment with serial phlebotomy is used (1).

b. Gaucher's disease. A deficiency of the enzyme beta-glucocerebrosidase results in the accumulation of cerebrosides in several organs. Clinical features include left ventricular dysfunction, hemorrhagic pericardial effusion, increased myocardial wall mass, and thickening of the left atrioventricular valves (1,6).

c. Hurler's syndrome. This syndrome results in a restrictive cardiomyopathy due to the deposition mucopolysacchararide in the myocardial interstitium, the cardiac valves, and the coronary arteries (6).

d. Fabry's disease (Angiokeratoma corporis, diffusum universalis). This is an X-linked disorder of the glycosphyngolipid metabolism secondary to the deficiency of lysosomal enzyme alpha-galactosidase. There is intracellular accumulation of neutral glycolipid with involvement of the myocardium, vascular endothelium, conducting tissues, and cardiac valves. Patients could present with angina and myocardial infarction. Left ventricular wall thickness is increased, with resulting left ventricular dysfunction and heart failure symptoms. Mitral regurgitation mat be seen. Systemic hypertension and mitral valve prolapse are common findings (1).

Unclassified Cardiomyopathies

Noncompacted myocardium. Noncompacted myocardium occurs as a result of an arrest in endomyocardial morphogenesis and presents in early childhood. The right ventricle is usually involved and occasionally it could have biventricular involvement. Patients present with systolic and diastolic dysfunction, ventricular arrhythmias, and thromboembolism. Mortality in this condition is high (6).

Systolic dysfunction with minimal dilatation. Characterized by an ejection fraction of less than 30% in the presence of preserved left ventricular dimensions and no evidence of restrictive physiology. There is myofibrillar loss at the histological level. Some patients have a family history of dilated cardiomyopathy and prognosis is poor (6).

Mitochondrial cardiomyopathy. This condition shows maternal inheritance and is secondary to mutations in the mitochondrial DNA with resultant impaired oxidative phosphorylation. Arrhythmias are common. There is progressive hypertrophy and dilatation. Skeletal muscle biopsy could be used for diagnosis (6).

Endocardial fibroelastosis. More than 80% of cases occur before 1 year of age. There is a suspected association with mumps infection. Thickening of the left ventricular endocardium characterize it. Most patients have a dilated left ventricle with thickened wall and systolic dysfunction. Some patients could present a restrictive cardiomyopathy. Diagnosis is commonly done on autopsy (1).

Diagnosis of Cardiomyopathy

A detailed history and physical examination are important in the diagnosis of a cardiomyopathy (figure 1). It is important to obtain information about the family history, exposure of the patient to cardio-toxins, and the presence of previous protracted "flu-like illness" or respiratory tract infection that may suggest previous myocarditis. While evaluating the patient with a cardiomyopathy, 5 important items need to be established, including the onset of symptoms, conventional risk factors (smoking, hypertension, diabetes, previous cardiac events), family history of heart disease, exposure to toxins (alcohol, cocaine, amphetamines), and any past or current illness. When indicated, physical examination, ECG, and echocardiography are the preferred methods for the screening of family relatives (6).

Physical examination is focused on palpation of a dilated heart, the presence of heart murmurs (systolic murmur of mitral/tricuspid valve regurgitation), and the presence of additional heart sounds (S3 or S4). The most useful clinical signs to establish the severity of the cardiomyopathy are the general appearance (dyspnea at rest, cachexia), hypotension, tachycardia, elevated jugular venous pressure, and a displaced left ventricle point of maximal impulse.

Useful tests done during the evaluation of these patients include an ECG, chest radiography, and an echocardiogram. Blood testing include CBC, electrolytes, lipids, fasting blood sugar, thyroid and liver panels. Serology for HIV is ordered when indicated. Viral titers have not proven to be useful. BNP is now a useful test available in many institutions and is a very useful marker for diagnosis, severity, and prognosis in heart failure.

The most common findings on ECG are pathologic Q waves, diffuse ST-segment abnormalities, LBBB or any intraventricular conduction delay, atrial fibrillation, and abnormal p waves. The chest radiograph findings most commonly seen are cardiomegaly, interstitial edema, pleural effusions, wires from previous sternotomy, or the chest radiograph may be normal if heart failure is secondary to diastolic dysfunction.

The two-dimensional echocardiogram is very useful in the evaluation of a patient with cardiomyopathy. Increased chamber dimensions, decreased left ventricular ejection fraction or fractional shortening, mitral and tricuspid valve regurgitation, regional wall motion abnormalities, and

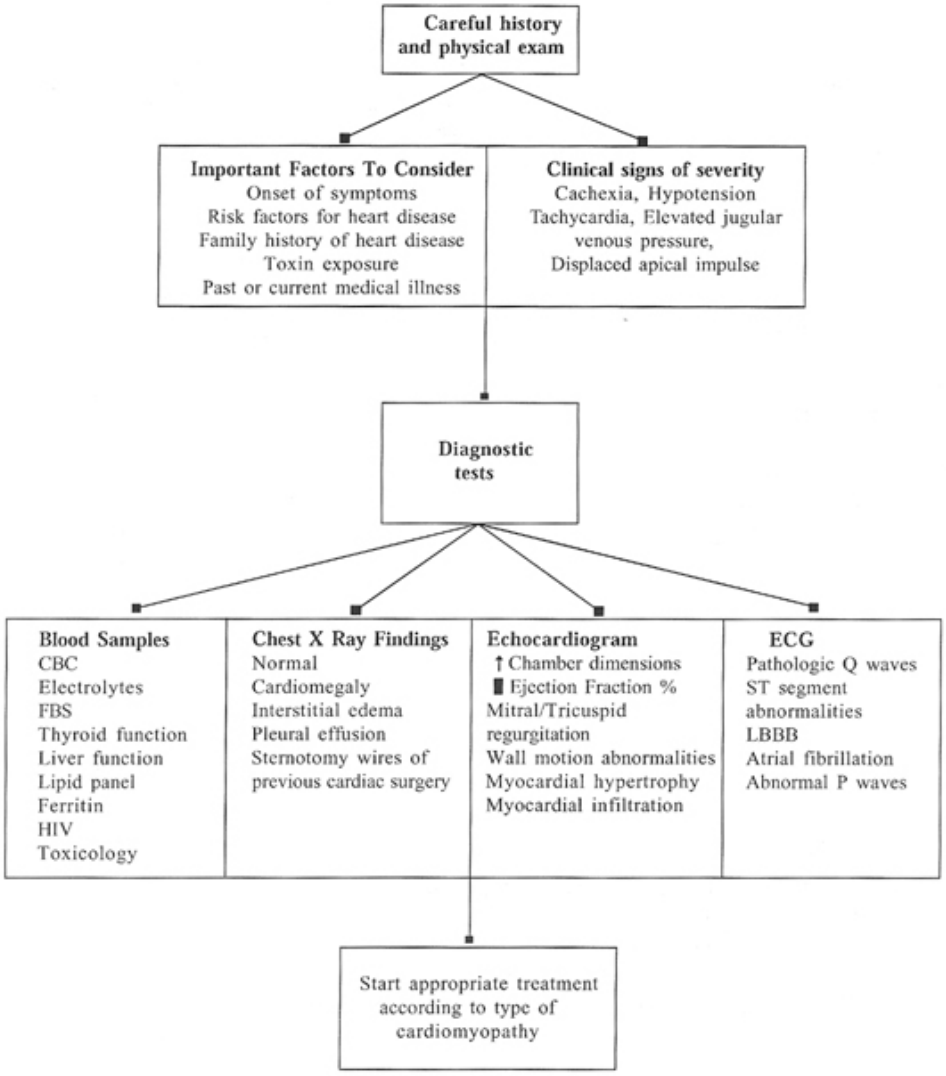
myocardial thickening are common findings. If Holter monitoring is used, it will most likely show the presence of premature atrial/ventricular complexes, atrial fibrillation, nonsustained ventricular tachycardia, and first- or second-degree atrioventricular block.

Non-invasive studies like dobutamine stress echocardiography, positron emission tomography, or magnetic resonance are used to rule out ischemia as the cause of ventricular dysfunction. Invasive studies include right and left heart catheterization, and coronary angiography. Endomyocardial biopsy is not performed on a routine basis since most findings are non-specific and will not affect medical management. Biopsy is useful for the diagnosis of giant cell myocarditis, anthracycline toxicity, and to differentiate constrictive from restrictive etiology (6).

Management of Cardiomyopaties

Generally, the treatment of a cardiomyopathy is the same as in heart failure. Common medications used include diuretics, digitalis, ACE inhibitors, spironolactone, and

Figure 1. Algorithm for Diagnosis and Management of Cardiomyopathies



beta-blockers. Neither steroids nor intravenous immunoglobulins have proven to be useful in the management of this condition. The overall outcome when no reversible etiology is found is poor with a 5-year survival rate of 50%. Costs are elevated from the polypharmacy and multiple admissions to the hospital. A peak VO2 uptake of less than 14 ml/kg/min is a marker of severity and is accepted criteria for indicating heart transplantation. The median survival in heart transplant is approximately 10 years (6).

Every patient with a suspected or diagnosed cardiomyopathy should be referred to a cardiologist, since these patients have a poor prognosis and treatment for this condition is complex. Aggressive efforts should be done to ascertain the etiology of the cardiomyopathy and to start treatment with optimized therapy early after diagnosis.

Resumen

Las cardiomiopatías son un grupo de enfermedades que se caracterizan por el envolvimiento directo del músculo cardiaco y son una causa significativa de morbilidad y mortalidad. La Sociedad Mundial de la Salud en conjunto con la Sociedad Internacional y Federación de Cardiología han promovido una clasificación tomando en cuenta la etiología y patofisiología de estas condiciones, incluyendo 5 grupos: cardiomiopatía dilatada, cardiomiopatía hipertrófica, cardiomiopatía arritmogénica del ventrículo derecho, cardiomiopatía restrictiva y cardiomiopatía no-clasificadas. La cardiomiopatía dilatada es la que más frecuentemente se observa, y a su vez la cardiomiopatía isquémica es la más importante de este grupo. El tratamiento de estas condiciones es muy similar al tratamiento de fallo cardiaco, pero hay ciertas cardiomiopatías que requieren tratamientos adicionales o diferentes. Para poder establecer un diagnóstico certero se ha presentado un algoritmo de diagnóstico y manejo con pruebas específicas para dirigir la evaluación del paciente. El historial médico es fundamental en todos los casos. Dentro de las pruebas diagnósticas más útiles están la radiografía de pecho, el electrocardiograma, el ecocardiograma y los laboratorios de sangre específicos. Todo paciente con una cardiomiopatía debe ser referido a un cardiólogo, ya que estas condiciones son progresivas, y pueden terminar en la muerte temprana del paciente de no ser tratadas a tiempo y de forma agresiva.

References

1. Braunwald E, Zipes D, Libby P. Heart Disease sixth edition A Textbook of cardiovascular medicine, Philadelphia: WB Saunders Company; 2001, p.1751-1806.
2. Richardson P, McKenna W, Bristow, Maisch B, Mautner B, O'Connel JB,et al. Report of the 1995 WHO/ISCF Task Force on the definition and classification of cardiomyopathies. Circulation 1996;93:841-842.
3. Nishimura RA, Holmes Jr, DR. Hypertrophic obstructive cardiomyopathy. New Engl J Med 2004;350:1320-1327.
4. Spirito P, Seidman CE, McKenna WJ, Maron BJ. The Management of hypertrophic cardiomyopathy. New Engl J Med 1997;336:775-785.
5. Kushwaha SS, Fallon JT, Fuster V. Restrictive Cardiomyopathy. New Engl J Med 1997;336:267-276.
6. O'Neil JO, Silverman CB. Dilated and Restrictive cardiomyopathies. Cleveland Clinic Review March 12,2003.
7. Kelly DP, Strauss AW. Inherited cardiomyopathies. New Engl J Med 1994;330:913-919.