Echocardiography update for primary care physicians: a review

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ABSTRACT

Echocardiography is a key evaluation tool for the diagnosis, prognosis, and guidance of interventional numerous management of cardiovascular conditions, including ischaemia, heart failure, and structural heart diseases. Recent technological advancements have also seen the exploration of artificial intelligence, intracardiac vortex imaging, and three-dimensional printing in echocardiography. With cardiovascular diseases increasing prevalence worldwide, it is important for clinicians including general practitioners to have updated knowledge of appropriate use of echocardiography. As such, this article reviews the current literature and summarises the latest developments and the general clinical usage of echocardiography.

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Introduction

Cardiovascular diseases are a major burden to healthcare globally and a leading cause of death in industrialised countries.1 In 2015, cardiovascular disorders accounted for 77600 inpatient discharges in Hong Kong, including 6190 deaths, making them the third most common cause of death. The prevalence of cardiovascular disease is increasing and the age-standardised mortality is decreasing.² Thus, patients with heart diseases are increasingly likely to present to primary care physicians as their first point of care or for a follow-up examination: up to 15% of coronary heart disease patients are treated exclusively by primary care physicians.³ Furthermore, a long-term follow-up study has proven the viability of general practitioners providing care for low-risk chronic heart failure patients.⁴ As such, clinical care for patients with heart diseases has become increasingly relevant to primary care physicians.

Echocardiography does not involve any radiation and may be performed as a point-of-care diagnostic tool in both outpatient and inpatient settings. Doppler echocardiography allows haemodynamic assessment of stroke volume, pressure gradients, valvular regurgitations, and intracardiac shunts. The treatment of patients with cardiovascular diseases almost invariably involves the use of echocardiography as a means of initial

and follow-up investigation. Many patients undergo cardiac surgery and/or catheter-based interventions guided by perioperative or intraprocedural echocardiography. Point-of-care echocardiography in primary care settings improves outcomes in some patients,⁵ and may identify otherwise-undiagnosed valvular heart diseases.⁶

Echocardiographic technologies remain a vibrant field of research. The rapid advancement of echocardiographic technologies requires primary care physicians to have an updated understanding of its appropriate use and referral. According to the American College of Cardiology, appropriate echocardiography "likely contributes to improving patient's clinical outcomes" and inappropriate echocardiography may be "potentially harmful to patients and generate unwarranted costs to the healthcare system".7 Recent technological developments have expanded the use of echocardiography dramatically. Therefore, the aim of this study was to review the contemporary literature on clinical indications and emerging technologies of echocardiography and to provide a summary for primary care physicians.

Basic echocardiography and the echocardiographic report

Transthoracic echocardiography (TTE) is non-

invasive, readily available, and is generally the initial imaging option. In contrast, transoesophageal echocardiography (TEE) requires insertion of a probe into the oesophagus and thus involves sedation and risks of complications such as major bleeding (<0.01%) and hoarseness of voice (12%).8 However, TEE offers better resolution, especially of the left heart, mitral and aortic valves, and the aorta. A routine echocardiography involves several methods of imaging, including two-dimensional (2D; previously known as B mode), Doppler, and mode echocardiography. Two-dimensional Μ echocardiography, the most commonly used and intuitive method, is used for visualisation and dimensional measurements of the cardiac structures. Two-dimensional echocardiography is often the initial method of imaging, by which cardiac structures are identified and other methods applied. The left ventricular ejection fraction (LVEF) is also obtainable from 2D echocardiography, with the normal value being 50% to 70%. This is mainly used in the classification of heart failure.

Colour Doppler imaging allows visualisation of the blood flow, the conventional colour scheme being red for flow towards the probe and blue for away. This allows intuitive and easy assessment of valvular assessments, where stenotic valves generally show turbulent flow and often thickened valve leaflets, whereas regurgitating valves show a regurgitant jet. Trace or mild regurgitation is common even in normal, healthy individuals. When an abnormal, turbulent flow is detected by colour Doppler imaging, continuous and pulse wave spectral Doppler techniques should be used to further evaluate the nature and severity of the abnormal flow.

Continuous and pulse wave Doppler imaging provides quantitative measurement of flow velocity, volume, and pressure gradient at operator-defined locations. These allow calculation of stroke volume and haemodynamic assessment of the heart valves. Tissue Doppler provides information about movement of cardiac tissues, such as the diastolic motion of the mitral annulus that reflects left ventricular diastolic function. By means of transmitral flow assessment (ie, E, A, E/A ratio) and tissue Doppler imaging of the mitral annulus (ie, e', a', s', E/e'), filling pressure and diastolic function of the left ventricle can also be assessed.9 Interpretation of these parameters is complex and requires correlation with the clinical picture; in general, the normal E/A ratio is 0.8-2 but may be high in young, fit persons; a E/e' (medial mitral annulus) ratio <8 generally indicates normal left ventricular filling pressure. Further details may be found in the joint recommendation of the European Association for Cardiovascular Imaging and the American Society of Echocardiography for diastolic echocardiographic

超聲心動圖:基層醫療醫生須知的最新發展 陳士楷、謝家偉、趙航、羅秀霞、金春娜、甘嘉豪、范怡婷、 李沛威

超聲心動圖對診斷、預後和計劃多種心血管疾病(包括缺血性心臟 病、心臟衰竭和結構性心臟病)的介入治療非常重要。最近,超聲心 動圖也開始涉獵人工智能、心內渦旋成像以及三維打印等領域。隨着 全球心血管疾病普遍化,認識超聲心動圖的最新發展對基層醫療醫生 等臨床醫生愈加重要。因此,本文回顧當前文獻並總結超聲心動圖的 最新發展及其一般臨床用途。

assessment.10

Lastly, M mode tracks over time the movement of tissues along a straight line extending from the probe and has the advantage of high temporal resolution. The M mode is most commonly used for the assessment of right ventricular function by means of tricuspid annular plane systolic excursion, which is typically ≥ 17 mm. The M mode is also useful in the assessment of constrictive pericarditis, pericardial effusion and cardiac tamponade. hypertrophic obstructive cardiomyopathy, pulmonary hypertension, and left ventricular outflow tract obstruction.¹¹ An echocardiographic report thus contains information about dimensions and functions of the heart chambers and the proximal aorta. Table 1 summarises the typical components of an echocardiographic report.¹² A set of selected standard echocardiographic images is shown in Figure 1.

Common clinical scenarios where echocardiography is indicated

The clinical indications for echocardiography are detailed in the appropriate use criteria from the European Association for Cardiovascular Imaging¹³ and the American College of Cardiology.7 This section summarises the key recommendations for the use of echocardiography in some common chief complaints encountered by clinicians. Key points are summarised in Table 2. In general, echocardiography is indicated for evaluating symptoms or conditions potentially of cardiac aetiology, including chest pain, shortness of breath, palpitations, transient ischaemic attack, stroke, or peripheral embolic event. Echocardiography is also considered appropriate when heart disease or structural abnormality are suspected on prior testing including but not limited to chest X-ray, electrocardiogram (ECG), or cardiac biomarkers, whether the patient is symptomatic or not. As a general principle, echocardiography is accepted as appropriate for initial diagnosis when there is a change in clinical status or when the results of the echocardiography are anticipated to change patient treatment. In contrast, routine testing when

TABLE I. Brief summary of the components of a typical echocardiographic report according to the American Society of
Echocardiography ¹²

Assessed items	Aspects of assessment
Demographics	Name, identification number, age, sex, body height and weight, body surface area
Left ventricle	Size (end-diastolic and end-systolic diameter or volume) Wall thickness (posterior wall and interventricular septum) Ventricular mass Shape (hypertrophy, aneurysm, or pseudoaneurysm) Systolic function • Global: ejection fraction, fractional shortening, fractional area change • Regional: regional wall motion abnormality, septal motion • Evidence of infarction Diastolic function (impaired relaxation, pseudonormal, or restrictive) and left ventricular filling pressure Thrombus Space-occupying lesion Intracardiac shunts: ventricular septal defect
Left atrium	Size Thrombus (within atrial cavity and appendage) Space-occupying lesion Spontaneous echo contrast (indicates low flow state and associated with thromboembolism)
Right ventricle	Size Wall thickness Systolic function Segmental wall analysis Right ventricular systolic pressure
Right atrium	Size Thrombus (within atrial cavity, appendage, and/or extending from inferior vena cava) Space-occupying lesion Catheter or pacemaker wire Spontaneous echo contrast (indicates low flow state and associated with thromboembolism)
Heart valves	 Number of cusps (aortic valve) Chordae (mitral and tricuspid valves; rupture) Leaflet thickening, billowing, prolapse, or flail leaflets Elongated leaflets (mitral and tricuspid valves) Annular calcification or dilatation Space-occupying lesion on annulus or leaflet Mechanical heart valve or bioprostheses (type, function, pressure gradient, dehiscence, paravalvular leak, vegetation/abscess, pannus, or thrombus) Stenosis: severity, peak and mean velocity, peak and mean transvalvular gradient, valve area Regurgitation: severity, jet direction (central/eccentric), regurgitant volume, and effective regurgitant orifice area; vena contracta width (mitral regurgitation)
Pericardium	Thickening Calcification Effusion (size, location [generalised or localised], content) Any haemodynamic compromise (ie, cardiac tamponade and/or contrastive pericarditis) Fat pad Pleural effusion or ascites
Aorta	Ascending, arch and descending aorta: dilatation, aneurysm, plaque, free flap (dissection), coarctation, graft, or transposition of great arteries Sinus of Valsalva (dilatation, aneurysm, pseudoaneurysm or fistula)
Pulmonary artery	Size Thromboembolism Morphology (hypoplasia, branch stenosis)
Veins	Pulmonary veins (size, location, flow pattern, thrombus, mass, and congenital anomalies) Inferior vena cava/hepatic vein (size, respiratory change in dimension [to estimate right atrial pressure], flow pattern, thrombus, mass, pacing wire, venous catheter, and congenital anomalies)

there is no change in clinical status or when test difficult when relying on the clinical history and results are unlikely to lead to changes in treatment are generally considered inappropriate.

Chest pain

physical examination only. The most important diagnosis in patients presenting with acute chest pain is acute coronary syndrome (ACS). As such, 12-lead ECG and troponin assay are the first-line investigations for acute chest pain suggestive of The differential diagnoses of chest pain are ACS. Echocardiography aids or confirms ischaemic broad, and reaching a definitive diagnosis is often diagnoses, and is used to screen for non-ischaemic



FIG I. Selected standard echocardiographic images. (a) Parasternal long-axis view showing an anteroseptal-posterolateral cut of the heart. Parasternal short-axis views showing (b) the papillary muscle, (c) the tip of the mitral valve leaflets, and (d) the aortic valve and the right ventricular outflow tract. (e) Apical 4-chamber view showing all four cardiac chambers. (f) Apical 2-chamber view showing the left atrium and ventricle. (g) Apical 3-chamber view showing the left atrium, left ventricle and proximal aorta. (h) Subcostal view showing all four cardiac chambers. (i) M-mode tracing of the tricuspid annulus for assessment of tricuspid annular plane systolic excursion. (j) Doppler wave tracing at the left ventricular outflow tract. (k) Diastolic function assessment by Doppler wave tracing at the tip of the mitral valve. (I) Tissue Doppler tracing of the septal mitral annulus

structural, aortic, and pericardial pathology in first-line test to assess suspected aortic pathologies, selected patients. For evaluation of acute chest pain with suspected myocardial ischaemia/infarction and non-diagnostic ECG, resting TTE is appropriate and can be performed while the patent has chest pain. The absence of regional wall motion abnormality on 2D TTE during chest pain virtually excludes acute myocardial infarction, with a sensitivity of 93% for any infarct and 100% for ST-elevation myocardial infarction.¹⁴ All patients with chest pain should have initial evaluation of ventricular function by TTE after ACS. After ACS, re-evaluation of ventricular function during the recovery phase is appropriate when results will guide therapy.¹⁵ For detecting other causes of chest pain, including acute aortic syndromes, valvular diseases, pulmonary embolism, and pericarditis, TTE is valuable and can be done in emergency settings. Although TTE can be used as a

TEE allows clearer and more complete assessment of the thoracic aorta, with diagnostic accuracy equivalent to that of cardiac computed tomography (CT) scans or magnetic resonance imaging (MRI).¹⁶ Even though CT is the imaging modality of choice in acute aortic syndromes, TTE and TEE remain valuable for acute assessment and follow-up, with the advantage of not requiring radiation, and especially for hemodynamically unstable patients and situations where CT contrast is contraindicated (eg, severe renal impairment).¹⁷

Shortness of breath

Shortness of breath is a common symptom among patients presenting to primary and emergency care physicians. Differential diagnoses of breathlessness

Chief complaint	Scenario		Appropriate (A)/ inappropriate (I) modality*	Key information to look for	
Chest pain	Inconclusive or unremarkable 12-lead ECG		TTE (A)	Regional strain and wall motion abnormalities Complications of MI	
	Confirmed MI (before discharge)		TTE (A)	Left and right ventricular systolic function, LVEF Valvular abnormalities and complications of MI	
	Suspected acute aortic syndromes		TTE or TEE (A; particularly in unstable patients and/or when CT is contraindicated)	Aortic dissection, aneurysm, intramural haematoma, pseudoaneurysm, or penetrating aortic ulcer	
	Murmur with cardiorespiratory compromise		TEE (A)	Infective endocarditis (±valvular perforation) Acute mitral regurgitation Prosthetic valve dehiscence	
	Significant history of chest trauma/ penetrating cardiac injury		TTE (A)	Cardiac contusion Cardiac tamponade Myocardial perforation	
	Suspected pericarditis		TTE (A)	Pericardial effusion Features of constrictive pericarditis	
Shortness of breath	Severe shortness of breath/desaturation of uncertain or suspected cardiac cause		TTE (A)	Differentiate between cardiac and respiratory causes of hypoxaemia	
	Suspected heart f	ailure	TTE (A)	LVEF for diagnosis and phenotype of heart failure Aetiology of heart failure, such as valvular causes and cardiomyopathy	
	Pulmonary embolism	Haemodynamically stable Haemodynamically unstable	TTE (I) TTE (A)	Thrombus in right heart or pulmonary arteries Right heart dysfunction/other signs of pulmonary embolism (For patients who are haemodynamically unstable and in whom definitive imaging is unsafe, bedside echocardiography may be used to obtain a presumptive diagnosis of pulmonary embolism to justify the administration of potentially life-saving therapies)	
	Respiratory cause is likely	of shortness of breath	TTE (I)	N/A	
	Longstanding pul unexplained deter	nonary diseases with ioration	TTE (A)	Cardiac complications of pulmonary condition, including pulmonary hypertension, right heart failure, cor pulmonale	
Syncope and palpitations	12-lead ECG sugg disease (eg, patho	gestive of structural heart plogical Q waves)	TTE (A)	Underlying structural heart diseases	
	Palpitations with sustained AF, SVT	sustained or non- , or VT	TTE (A)	Underlying structural heart diseases	
	Syncope (with or or or signs of cardio	without other symptoms vascular disease)	TTE (A)	Underlying structural heart diseases	
	Frequent or exerc	ise-induced PVC	TTE (A)	Underlying structural heart diseases LVEF	
	Infrequent PAC or of structural heart	PVC without suspicion diseases	TTE (I)	N/A	
	Asymptomatic isc	lated sinus bradycardia	TTE (I)	N/A	
Murmur or clicks	Flow murmur due	to high flow states	TTE (I)	N/A	
	Initial evaluation c structural heart di	f suspected valvular or sease	TTE (A)	Valvular or congenital heart disease	
	Initial evaluation c endocarditis with	f suspected infective positive blood cultures	TTE (A) TEE (A; if moderate or high clinical suspicion)	Vegetations, abscess, pseudoaneurysm, prosthetic valve dehiscence	
	Re-evaluation of disease with no cl symptoms and sig	nown mild valvular hange in clinical gns	TTE (I)	N/A	
	Transient fever wir bacteraemia or a	thout evidence of new murmur	TTE (I)	N/A	

TABLE 2.	Summary of selected	common clinica	l scenarios in which	n echocardiography	is indicated, ba	ased on the appr	opriate use	criteria by the
European	Association for Cardi	ovascular Imagin	g and the Americar	College of Cardio	logy ^{7,13}			

Abbreviations: AF = atrial fibrillation; CT = computed tomography; ECG = electrocardiogram; LVEF = left ventricular ejection fraction; MI = myocardial infarction; N/A = not applicable; PVC = premature ventricular contractions; SVT = supraventricular tachycardia; TEE = transoesophageal echocardiography; TTE = transthoracic echocardiography; VT = ventricular tachycardia

* Wherever TTE is appropriate, TEE may also be considered when TTE image quality is suboptimal. When TTE is inappropriate, TEE is generally inappropriate

include cardiovascular disease, lung disease, anaemia, obesity, and deconditioning. In any setting, clinical history and examination are the first step in evaluation.13 When heart failure is suspected after initial clinical evaluation, TTE is mandatory to confirm or exclude the diagnosis; to quantify chamber volumes, systolic and diastolic function, and wall thickness; and to identify the aetiology of heart failure (eg, cardiomyopathy, valvular disease, or prior myocardial infarction). The TTE also serves to classify heart failure into heart failure with preserved, mid-range or reduced EF (Fig 2).¹⁸ Such classification guides management of the condition. For patients with heart failure with preserved or mid-range EF, assessment of diastolic function is important as diastolic dysfunction is known to be key in the pathogenesis. Although some patients have resting echocardiographic evidence of diastolic dysfunction, left ventricular filling pressure, which is the main haemodynamic parameter that explains

exertional dyspnoea, may only be elevated during exertion. This may be demonstrated by diastolic stress echocardiography, providing additional haemodynamic information about the patient and aiding diagnosis and treatment.¹⁹

Patients who have undergone chemotherapy or radiotherapy are at risk of developing heart failure, and baseline echocardiography with regular followup examinations are required. The precise algorithm depends on the cardiotoxic agent prescribed. Traditionally, LVEF has been used as the main marker of cardiac function and cancer therapeuticsrelated cardiac dysfunction is defined as a drop in LVEF of \geq 5% in symptomatic patients or a drop in LVEF of \geq 10% to an LVEF of <53% in asymptomatic patients.²⁰ However, significant damage has occurred when a drop in LVEF is detected. Earlier detection can potentially reverse myocardial dysfunction before irreparable damage occurs.²¹ Thus, global longitudinal strain assessment is recommended for



FIG 2. (a, c, e) Apical 4-chamber views and (b, d, f) apical 3-chamber views captured by transthoracic echocardiography. (a, b) Images of a 77-year-old patient with severe aortic stenosis, dilated left ventricle, and left ventricular hypertrophy. The heavy calcification on the aortic valve can be appreciated on the apical 3-chamber image. (c, d) Images of a 70-year-old patient with severe functional mitral regurgitation and heart failure with reduced ejection fraction (left ventricular ejection fraction 29%). The mitral regurgitant jet can be appreciated on colour Doppler imaging in the apical 3-chamber image while both images show dilated left ventricle and left atrium. This patient suffered from moderate-to-severe tricuspid regurgitation as well, with dilated right ventricle and right atrium. (e, f) Images of a 34-year-old healthy individual

these patients (see section "Advanced and emerging imaging technologies").²⁰

In the acute setting of respiratory failure where patients present with acute shortness of breath and hypoxaemia of uncertain aetiology, or with hypotension of uncertain or suspected cardiac aetiology, TTE is useful to confirm or exclude cardiac diseases. However, if pulmonary or other noncardiac causes of respiratory distress are established, TTE is unnecessary. Right ventricular function can be evaluated by TTE to determine prognosis and predict recurrence of pulmonary embolism.^{7,13} However, CT, ventilation-perfusion imaging, and pulmonary angiography remain the imaging modalities of choice in the diagnosis of patients with suspected pulmonary embolism.²² In particular, the triplerule-out CT-which uses contrast enhancement to evaluate the coronary arteries, aorta, and pulmonary arteries-may be used reliably to detect or exclude pulmonary embolism.^{23,24} TTE is not sensitive nor specific for diagnosing acute pulmonary embolism. Conversely, in a haemodynamically compromised patient with high clinical probability of pulmonary embolism, unequivocal signs of right ventricular pressure overload and/or right-heart/pulmonary artery thrombi on bedside TTE may support a presumptive diagnosis of pulmonary embolism and may justify emergency reperfusion treatment if immediate CT angiography is not feasible. Although sudden-onset dyspnoea is the most common presenting complaint in pulmonary embolism (80%), chest pain is also common (49%) and a low threshold of clinical suspicion should be maintained.²⁵

For patients with known chronic pulmonary diseases presenting with acute deterioration, careful physical exam and TTE can identify complicating factors such as pulmonary hypertension and right heart failure (cor pulmonale).

Syncope, dizziness, and cardiac source of embolism

The differential diagnoses for syncope or dizziness include vasovagal and orthostatic syncope, arrhythmias, cardiac structural abnormalities, and neurological causes.²⁶ A TTE is indicated when clinical symptoms, signs, or ECG findings are consistent with a structural cardiac diagnosis known to cause syncope/dizziness, such as aortic stenosis (Fig 2), left ventricular dysfunction, hypertrophic cardiomyopathy, intracardiac tumours, or pericardial effusions. A normal TTE in this setting is still an important finding that helps to direct the clinician in diagnosis and treatment.^{7,13}

In patients with suspected cardioembolic stroke, echocardiography especially TEE is useful for identifying intracardiac thrombi in the left atrial appendage (typically in patients with atrial fibrillation) and aortic atherosclerosis. In

younger patients with cryptogenic stroke, patent foramen ovale, right-to-left interatrial shunt, and/ or atrial septal aneurysm should be looked for on echocardiography with agitated saline contrast at rest and on Valsalva manoeuvre. TEE is also indicated in patients with high clinical suspicion of septic emboli associated with infective endocarditis.²⁷

Palpitation and abnormal electrocardiogram findings

Patients presenting with palpitation should be evaluated with a careful history and physical examination followed by a 12-lead ECG and, where appropriate, ambulatory ECG monitor (eg, Holter, loop recorder). If frequent ventricular premature complexes, atrial fibrillation, supraventricular, or ventricular tachycardia are diagnosed, TTE is appropriate to identify and evaluate underlying structural heart abnormalities.^{7,13,28} However, it is inappropriate to routinely order TTE for evaluation of infrequent atrial premature complexes, ventricular premature complexes without other evidence of heart disease, or asymptomatic isolated sinus bradycardia.¹³

Patients with new right bundle branch block should be assessed with a careful history focusing on potential causes of right ventricular strain (eg, pulmonary embolism, pulmonary hypertension, obstructive sleep apnoea). Echocardiographic examination should be performed if there is suspicion of pulmonary disease potentially impacting the right ventricle. Asymptomatic isolated chronic right bundle branch block generally does not require further evaluation or treatment. In contrast, left bundle branch block needs to be assessed carefully, mostly by history taking and physical examination for any associated disorders, including but not limited to hypertension, coronary artery disease, heart failure, myocarditis, valvular heart disease, and cardiomyopathies. Typically, an echocardiographic examination for LVEF is indicated. A subset of patients may be indicated for further evaluation by stress testing with imaging, depending on the clinical contexts.29

Murmur or clicks

Murmur or clicks on auscultation may suggest the presence of valvular or congenital heart diseases. Initial evaluation with TTE is appropriate when there is a reasonable suspicion of valvular (Fig 2) or structural heart disease.⁷ Conversely, echocardiography is inappropriate for routine evaluation of murmur when there are no other symptoms or signs of valvular or structural heart disease. An innocent murmur is typically a short and soft systolic ejection murmur, with normal S1 and S2, normal cardiac impulse, and no evidence for any haemodynamic abnormality. Hand grip usually decreases the intensity of an innocent murmur but increases that of a regurgitant murmur. Initial evaluation of suspected infective endocarditis with positive blood cultures or a new murmur with TTE is appropriate. Initial evaluation with TEE for suspected endocarditis is appropriate if pretest probability is moderate or high (eg, staphylococcal bacteraemia, fungaemia, prosthetic heart valve, or intracardiac device).^{30,31} However, echocardiography should not be routinely ordered for evaluation of transient fever without evidence of bacteraemia or a new murmur.

Re-evaluation of known valvular heart disease with echocardiography is considered appropriate where there is any change in clinical status or cardiac examination, or to guide therapy (eg, detecting asymptomatic ventricular dysfunction that would prompt early surgical or transcatheter valve intervention). Routinely repeating echocardiography is inappropriate in patients with no or mild valve disease on prior echocardiography with no change in clinical symptoms or cardiac exam. In patients with exercise-induced symptoms but only moderate valvular dysfunction on resting echocardiography, an exercise or pharmacological stress echocardiography may be necessary to reveal the true severity of valve dysfunction during exertion.²⁸ A TEE is indicated in some patients where TTE is inconclusive, or when intervention is planned.³²

Screening asymptomatic patients for hereditary structural heart diseases

This is a complex topic and a full discussion is beyond the scope of this review. Thus, this subsection shall focus on the two most common congenital diseases affecting the heart with the highest inheritability: bicuspid aortic valve (BAV) and Marfan syndrome. Although BAV is not always syndromic, 9% of firstdegree relatives of BAV patients also have BAV,³³ and echocardiographic screening in these patients detects a substantial number of BAV cases.³⁴ In most cases, a single TTE is sufficient to diagnose BAV. Once diagnosed, the patient should be referred for specialist cardiology treatment and follow-up.

In contrast, Marfan syndrome is an autosomal dominant disorder of the fibrillin gene on chromosome 15. Its main effects on the cardiovascular system include aortic aneurysms (especially thoracic) and aortic regurgitation. The diagnosis of Marfan syndrome is made with the Ghent

TABLE 3. Current and emerging echocardiographic technologies

Name	Principle	Current/potential clinical usage	Added clinical benefits
Speckle-tracking echocardiography	Track natural acoustic markers in a defined area, thereby providing non- Doppler information about myocardial deformation	Assessment of heart failure, myocardial ischaemia, and cardiomyopathy Detecting subclinical states likely to progress to heart failure	Early detection of myocardial dysfunction, provide insight into pathophysiology of myocardial dysfunction
Stress echocardiography	Increase myocardial stress by exercise or drugs, and monitor the response by echocardiography	Assessment and risk stratification of myocardial ischaemia and patients at high risk of developing myocardial ischaemia	Non-invasive diagnosis of ischemic heart disease with no radiation and nephrotoxic contrast Detect high-risk patients with myocardial ischaemia, and identify those who will benefit from coronary revascularisation
Contrast echocardiography	Using ultrasound microbubbles to enhance resolution of cardiac structure and assess myocardial perfusion	Suboptimal echocardiographic images, detection of intracardiac shunts, and detect myocardial hypoperfusion	Reduce suboptimal study and the use of downstream expensive imaging techniques
3D echocardiography	Using volumetric matrix imaging to construct 3D cardiac images in real time	Assessment of structural heart diseases, and pre-/intra-/post- operative monitoring of structural heart interventions	Provide 3D views of the heart not obtainable with 2D echo, clearer visualisation and better understanding of anatomy, improve intervention outcomes
Intracardiac vortex imaging	Combining Doppler imaging and speckle tracking technology to visualise blood flow in the heart	Assessment of structural and functional heart disease, postoperative evaluation of patients with structural heart interventions	Potentially earlier detection of myocardial dysfunction and improve performances of structural heart interventions
3D printing	Creating models using 3D images reconstructed from echocardiographic or CT images	Planning of structural heart interventions	Improve intervention outcome by better selection of devices, understanding of anatomy, and allowing preoperative "test runs" of interventions
Artificial intelligence	Using machine learning to automatically measure and interpret echocardiographic images	Chamber quantification and general interpretation of echocardiographic images	Improve efficiency and consistency of interpreting echocardiographic images, and increasing patient volume

Abbreviations: 3D = three-dimensional; CT = computed tomography

nosology, a set of diagnostic criteria last revised in 2010 that includes clinical, laboratory, and imaging criteria.³⁵ The imaging criteria includes significant aortic root dilatation (Z score of \geq 2). The European Society of Cardiology recommends at-risk relatives without a definite diagnosis of Marfan syndrome to be screeened every 5 years until a definitive diagnosis is established or excluded.¹⁷ Once diagnosed, yearly echocardiography is required for the stable patient, as well as baseline and at least 5-yearly cardiac MRI scan. If aneurysmal changes are detected beyond the aortic root, MRI scan should be repeated at least yearly.³⁶

Advanced and emerging imaging technologies

This section summarises advanced echocardiographic technologies that are relevant to common cardiovascular diseases, as well as emerging imaging technologies (Table 3).

Stress echocardiography is valuable for evaluation of suspected coronary artery disease. Myocardial ischaemia causes reduced systolic wall thickening and excursion during stress, constituting stress-induced regional wall motion abnormalities (Fig 3). These may precede angina and ST-T changes on ECG. The sensitivity of exercise and dobutamine stress echocardiography for detection of coronary artery disease are 85% and 80% with specificity of 77% and 86%, respectively.³⁷ Using cardiac MRI as reference standard, threedimensional (3D) echocardiography provides more accurate and reproducible left ventricle volume and ejection fraction measurements than 2D echocardiography.³⁸⁻⁴² The American Society of Echocardiography recommends the use of 3D echocardiography in chamber quantification when such technique is available in the echocardiography laboratory.43 Three-dimensional TEE has brought new insights to the mechanisms of mitral regurgitation,^{44,45} and has become essential in guiding transcatheter structural interventions such as transcatheter edgeto-edge mitral⁴⁶ or tricuspid⁴⁷ repair, transcatheter aortic valve implantation,⁴⁸ and left atrial appendage occlusion (Fig 4).49-51 More recently, 3D printing has been used to create 3D models from 3D TEE images for preprocedural simulation.⁵²⁻⁵⁴ Speckletracking echocardiography allows measurement of myocardial strain (deformation; Fig 3).55,56 Assessment of the global longitudinal strain is useful for detection of chemotherapy-related cardiotoxicity



FIG 3. (a) Dobutamine stress echocardiography demonstrating stress-induced regional wall motion abnormalities of entire inferior segments (arrows) which corresponded to the coronary angiogram finding. (b) Finding of critical stenosis at mid right coronary artery. (c) Speckle-tracking echocardiography measuring circumferential, radial, and longitudinal strain



FIG 4. Photorealistic three-dimensional reconstruction of the mitral valve (green arrow) and a left atrial appendage occluding device (yellow arrow) as viewed from the left atrium. The image was taken by transoesophageal echocardiography

in cancer patients, better than the assessment of $\ensuremath{\text{LVEF}}^{20}$

More recently, echocardiography has been used to visualise intracardiac blood flow.⁵⁷ Intracardiac vortex patterns of various pathological conditions have been shown to differ from those of healthy patients, including heart failure,⁵⁸ dilated and hypertrophic cardiomyopathy,⁵⁹ coronary heart disease,⁶⁰ and valvular diseases.⁶¹

Conclusion

Echocardiography is essential in a wide range of clinical scenarios. Appropriate use of echocardiography improves clinical outcomes by increasing diagnostic accuracy, providing non-invasive or minimally invasive assessment of disease status and risk stratification, and enabling real-time monitoring and guidance of interventional procedures. Technological advancements have extended echocardiography beyond 2D imaging, allowing advanced structural visualisation and functional measurement of the heart. Echocardiography remains a vibrant field of research, and recent developments are promising clinically. Echocardiography will continue to grow as a core clinical investigation for a wide spectrum of disorders, and its relevance to primary care physicians cannot be overstated.

Author contributions

All authors are responsible for writing the article. APW Lee had critical revision for important intellectual content. All authors had full access to the data, contributed to the study, approved the final version for publication, and take responsibility for its accuracy and integrity.

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