Cập nhật 2018 về Bệnh Cơ Tim Phì Đại

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Đại cương

- BCTPĐ: tần suất cao nhất trong bệnh tim mạch do di truyền; nam = nữ
- Biểu hiện lâm sàng đa dạng
- Sinh lý bệnh duy nhất
- Tiến triển tự nhiên thay đổi

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TL: Maron BJ. Braunwald's Heart Disease 9th ed, 2012 Elsevier Saunders 1582-1594

Định nghĩa và tần suất

- BCTPĐ:
  - ✓Dầy nhưng không dãn thất trái
  - ✓Không do bệnh hệ thống hoặc bệnh tim khác (TD: hẹp van ĐMC, THA…)
- Tần suất 1/500 quần thể chung (# 600.000 người Mỹ)
- Bệnh toàn cầu
- Ca đầu tiên 1958 (Brock, Teare)

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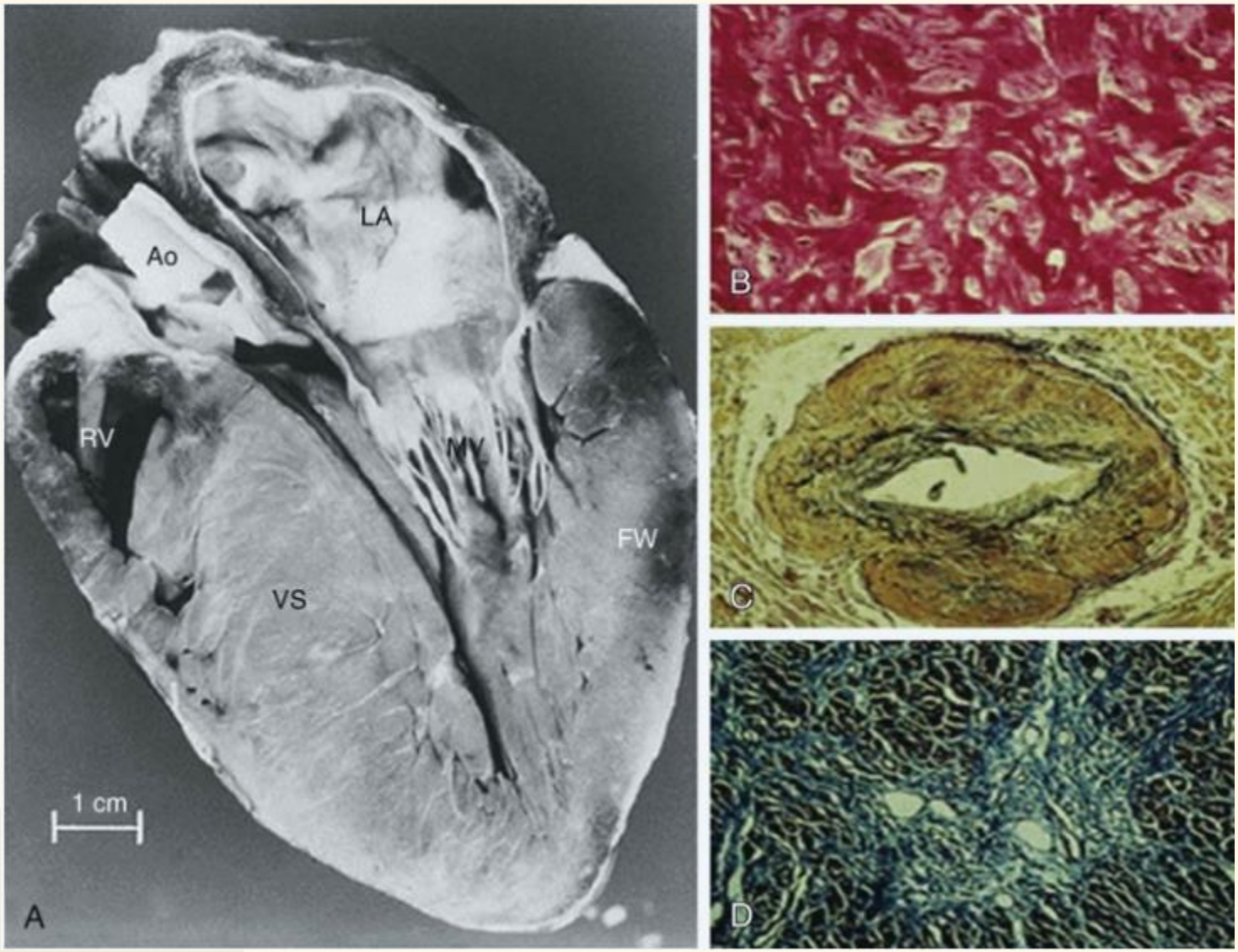
Gross morphology and histopathology of HCM



B: xáo trộn sắp xếp tế bào cơ tim
C: lòng ĐMV hẹp do thành mạch dầy
D: sẹo ở vách liên thất

TL: Maron BJ, Olivotto I. Heart Disease 2015, 10th ed, p 1574-1587

Di truyền BCTPĐ

- Di truyền NST thường, trội (autosomal dominant)
- Đã xác định:
  - 11 genes liên quan đến cardiac sarcomere: chuỗi nặng beta-myosin và protein C
  - 9 gene: troponin T, I, alpha tropomyosin, tritin, chuỗi nặng alpha- myosin, myosin chuỗi nhẹ
  - Không xác định được số genes và số đột biến BCTPĐ

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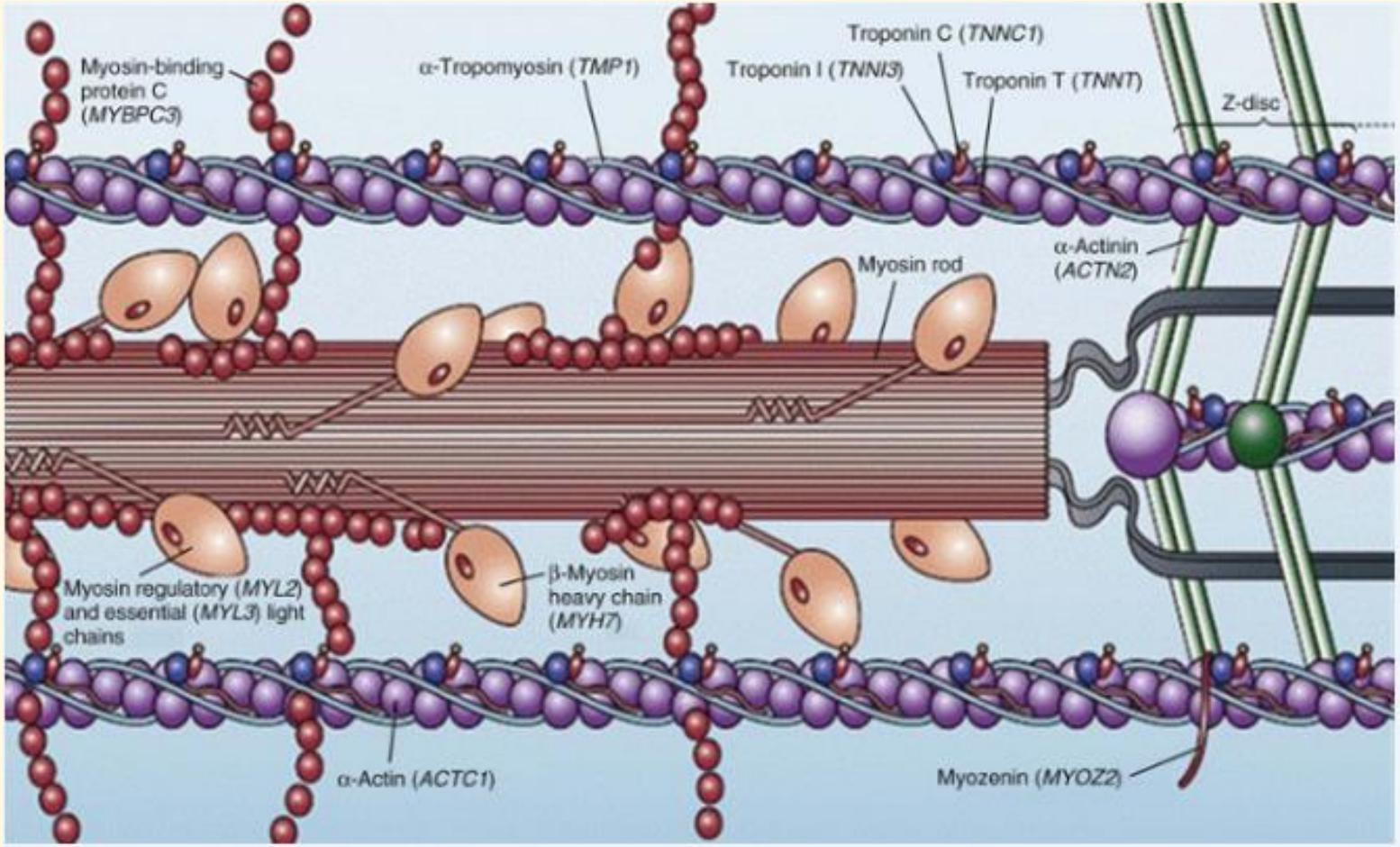
Location of genes within the cardiac sarcomere to cause HCM



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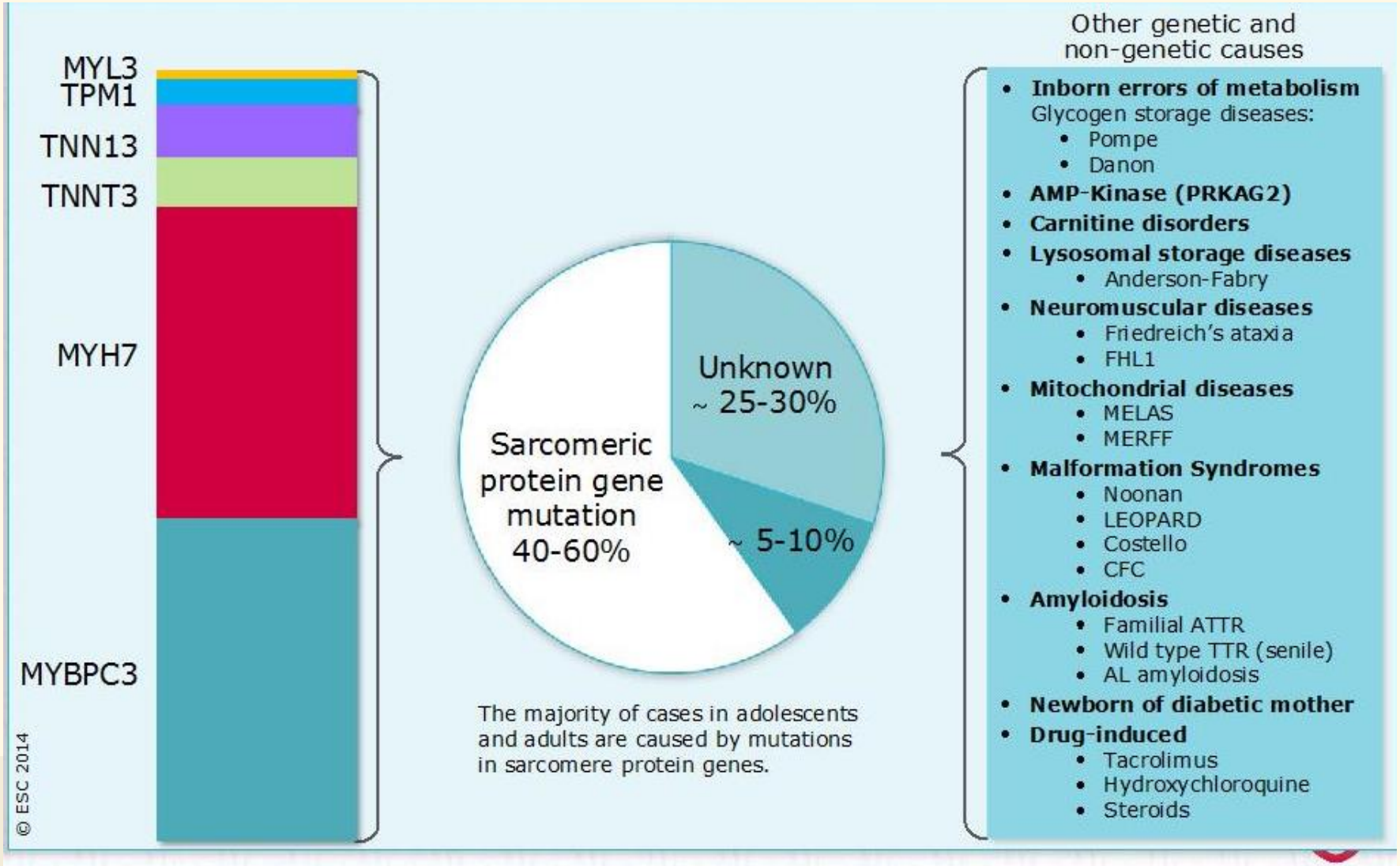
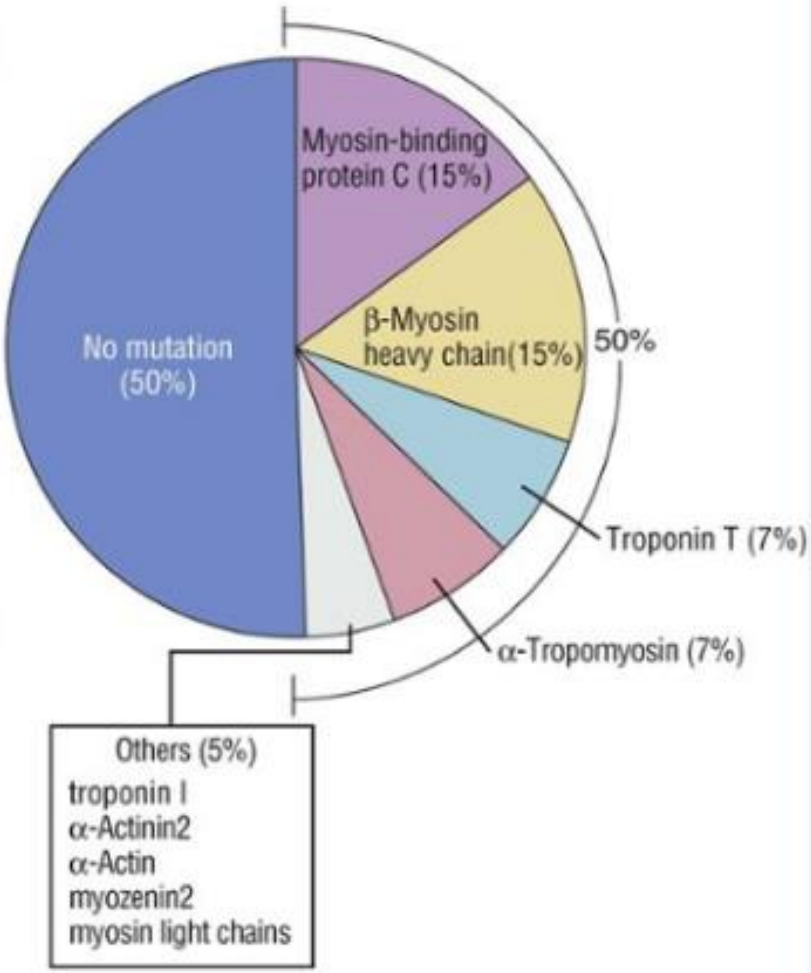
TL: Maron BJ, Maron MS. Lancet 381: 242, 2013

Diverse aetiology of hypertrophic cardiomyopathy



Genetic substrate of HCM

Molecular Substrate of HCM	
Strongest evidence of pathogenicity	
Thick filament	
1. β-Myosin heavy chain	*MYH7*
2. Regulatory myosin light chain	*MYL2*
3. Essential myosin light chain	*MYL3*
Thin filament	
4. Cardiac troponin T	*TNNT2*
5. Cardiac troponin I	*TNNI3*
6. Cardiac troponin C	*TNNC1*
7. α-Tropomyosin	*TPM1*
8. α-Cardiac actin	*ACTC*
Intermediate filament	
9. Cardiac myosin-binding protein C	*MYBPC3*
Z-disc	
10. α-Actinin2	*ACTN2*
11. Myozenin2	*MYOZ2*
Lesser evidence of pathogenicity	
Thick filament	
12. α-Myosin heavy chain	*MYH6*
13. Titin	*TTN*
Z-disc	
14. Muscle LIM protein	*CSRP3*
15. Telethonin	*TCAP*
16. Vinculin/metavinculin	*VCL*
Calcium handling	
17. Cal sequestrin	*CASQ2*
18. Junctophillin 2	*JPH2*



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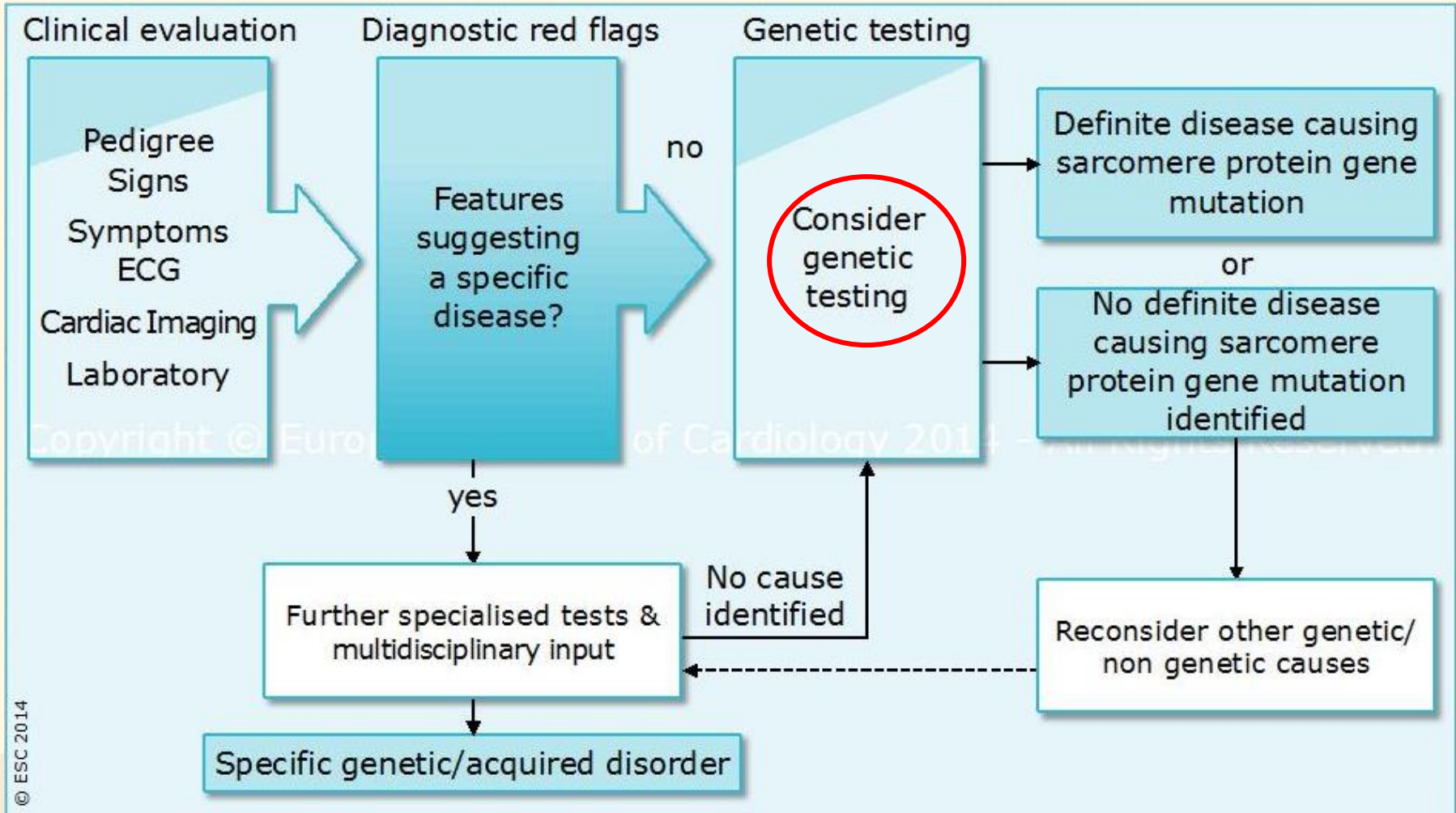
TL: Maron BJ, Maron MS. Lancet 381: 242, 2013

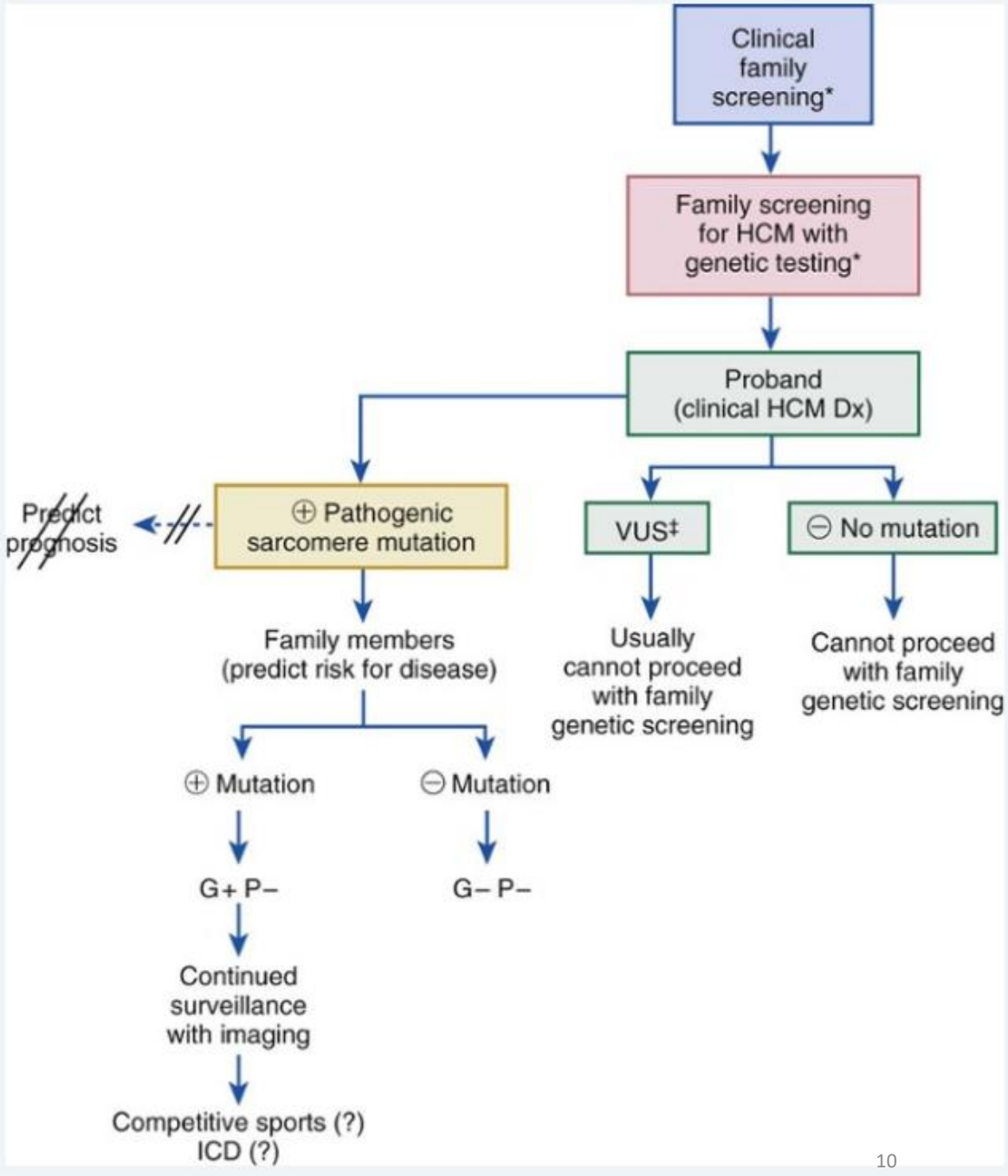
Schematic summarizing the general approach to the diagnosis of hypertrophic cardiomyopathy



Notes: 1. Counselling is essential before and after testing for genetic disease. 2. Genetic testing is recommended in patients fulfilling diagnostic criteria for HCM to enable cascade genetic screening of their relatives. 3. For recommendations on individual investigations see relevant sections.

European Heart Journal (2014);35:2733–2779 - doi:10.1093/eurheartj/ehu284

Role of genetic testing in family screening strategy



TL: Maron BJ et al. J. Am Coll Cardiol 60: 705, 2012

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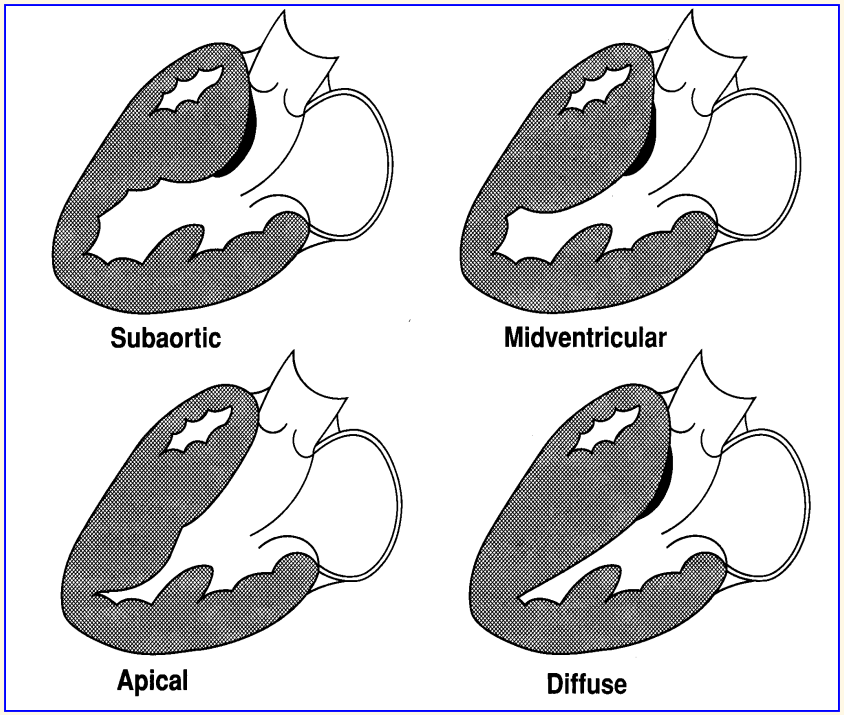
Giải phẫu bệnh

❖**Các thể BCTPĐ**

- Phì đại bất đối xứng thất trái, chủ yếu ở vách liên thất và vách trước thất trái xảy ra ở 70% trường hợp.
- Phì đại VLT phần đáy xảy ra ở 15-20% trường hợp.
- Phì đại vùng mỏm , hiếm gặp ở phương Tây (<2%) ; nhiều hơn ở Nhật (25%). Thể phì đại này thường có biểu hiện sóng T khổng lồ và đảo ngược ở các chuyển đạo ngực trái.



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Bệnh cơ tim phì đại do đột biến gene $LAMP_2$

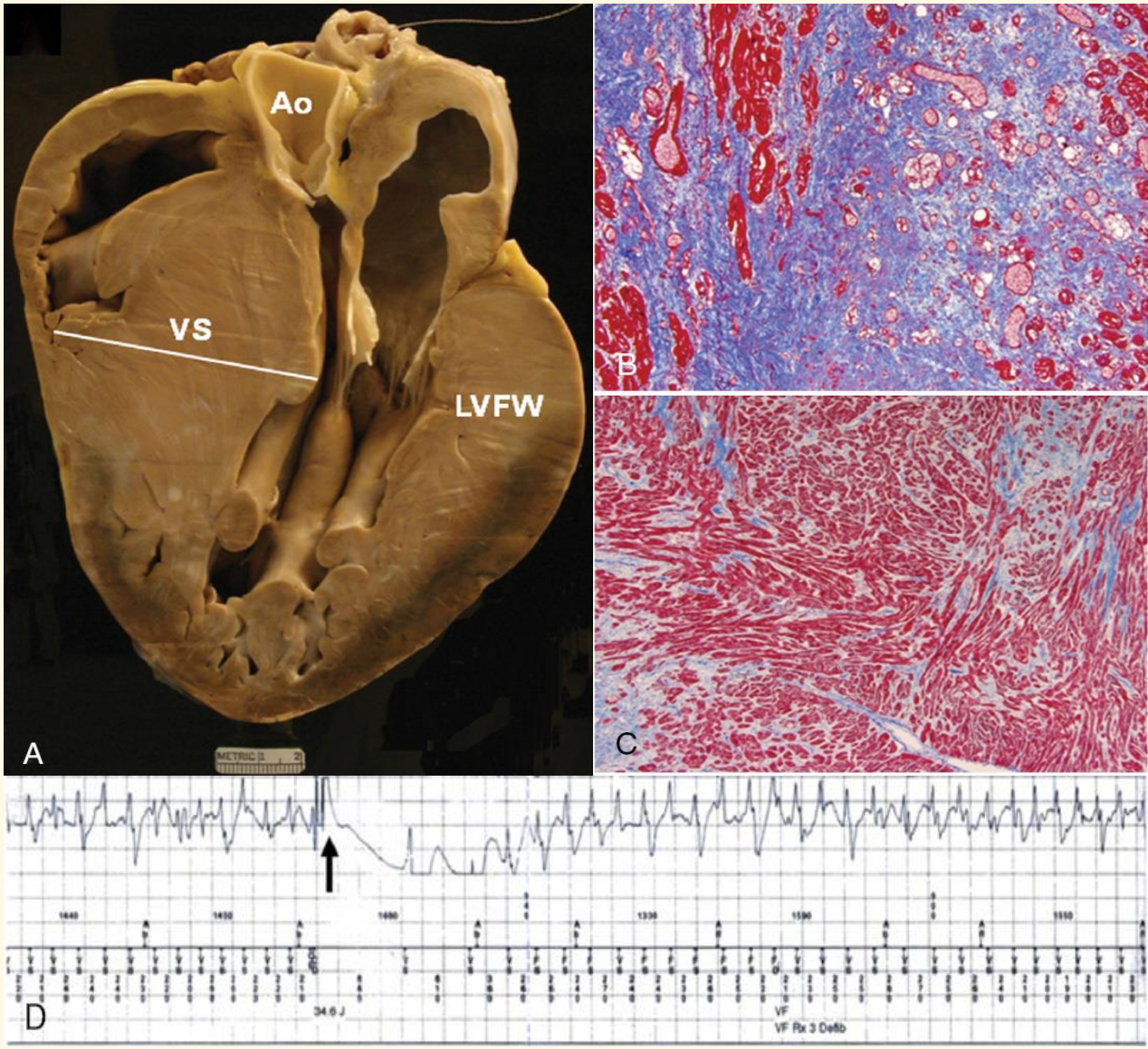
- Bé trai 14 tuổi, đột tử
- VLT 65 mm (tim nặng 1425g)

Ao: động mạch chủ; VS: vách liên thất

B: tế bào cơ tim (myocyts) kèm vacuolated sarcoplasm (màu đỏ) nằm trong sẹo (màu xanh)

C: xáo trộn sắp xếp myocytes, đặc thù của sarcomeric BCTPĐ

D: ECG trong tim, ICD tạo sốc tim thất bại (rung thất 280 nhát/ph)



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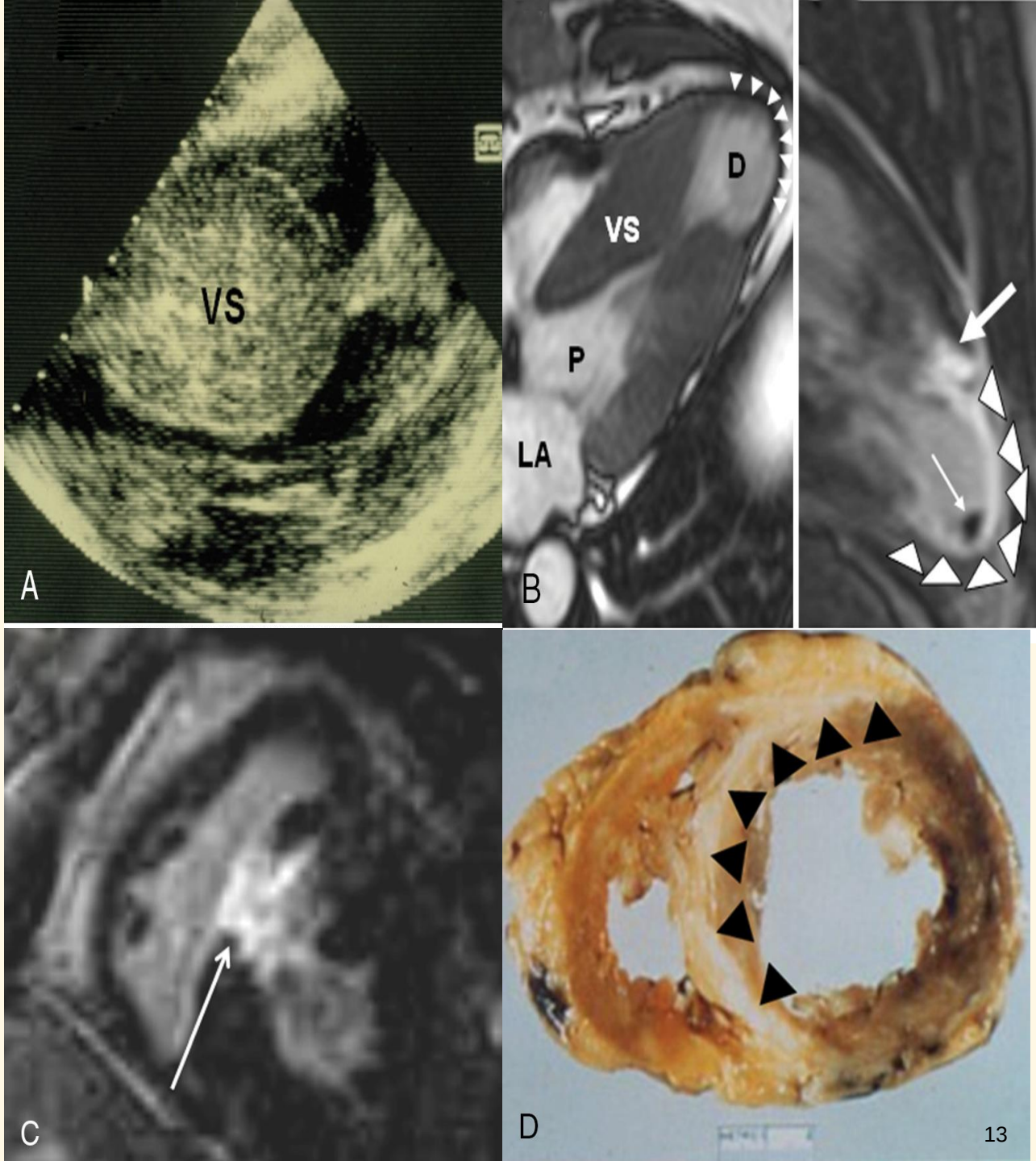
TL: Maron BJ. Braunwald's Heart Disease 9th ed, 2012 Elsevier Saunders 1582-1594

Dạng thất trái/ BCTPĐ có kèm nguy cơ loạn nhịp thất và đột tử



A: VLT phì đại nặng, dầy 55 mm

B: Túi phình mõm TTr kèm phì đại cơ tim vùng giữa/ BCTPĐ

C: Sẹo lớn xuyên thành VLT do huỷ cơ tim bằng cồn

D: BCTPĐ giai đoạn cuối: sẹo xuyên thành VLT, lan đến vách trước TTr

VLT: vách liên thất
TTr: thất trái
TL: Valeti US et al. J. Am Coll Cardiol 2007; 49: 350

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Các đặc điểm hình thái học và cấu trúc cần quan tâm/ BCTPĐ

- Dầy thất trái
- Bộ máy van 2 lá: van 2 lá dài gấp đôi bình thường hoặc dãn lớn từng vùng lá van trước hay lá van sau
- Mô bệnh học cơ tim: xáo trộn sắp xếp sợi cơ tim
- Bất thường ĐMV trong cơ tim: thành dầy (nội mạc và trung mạc), lòng hẹp

Sinh lý bệnh

- Nghẽn buồng tống thất trái (≥ 30 mmHg): yếu tố mạnh dẫn đến suy tim và tử vong tim mạch
- Tương quan yếu giữa mức độ nghẽn BTTTr và đột tử do tim
- Nghẽn dưới ĐMC: do vận động tới trước van 2 lá kỳ tâm thu (SAM) sát vào VLT kỳ tâm thu
- Độ chênh áp BCTPĐ: có tính động học
- TMCB cơ tim/ BCTPĐ: do rối loạn chức năng vi mạch
- Rối loạn chức năng tâm trương: 80% BCTPĐ → suy tim, khó thở gắng sức

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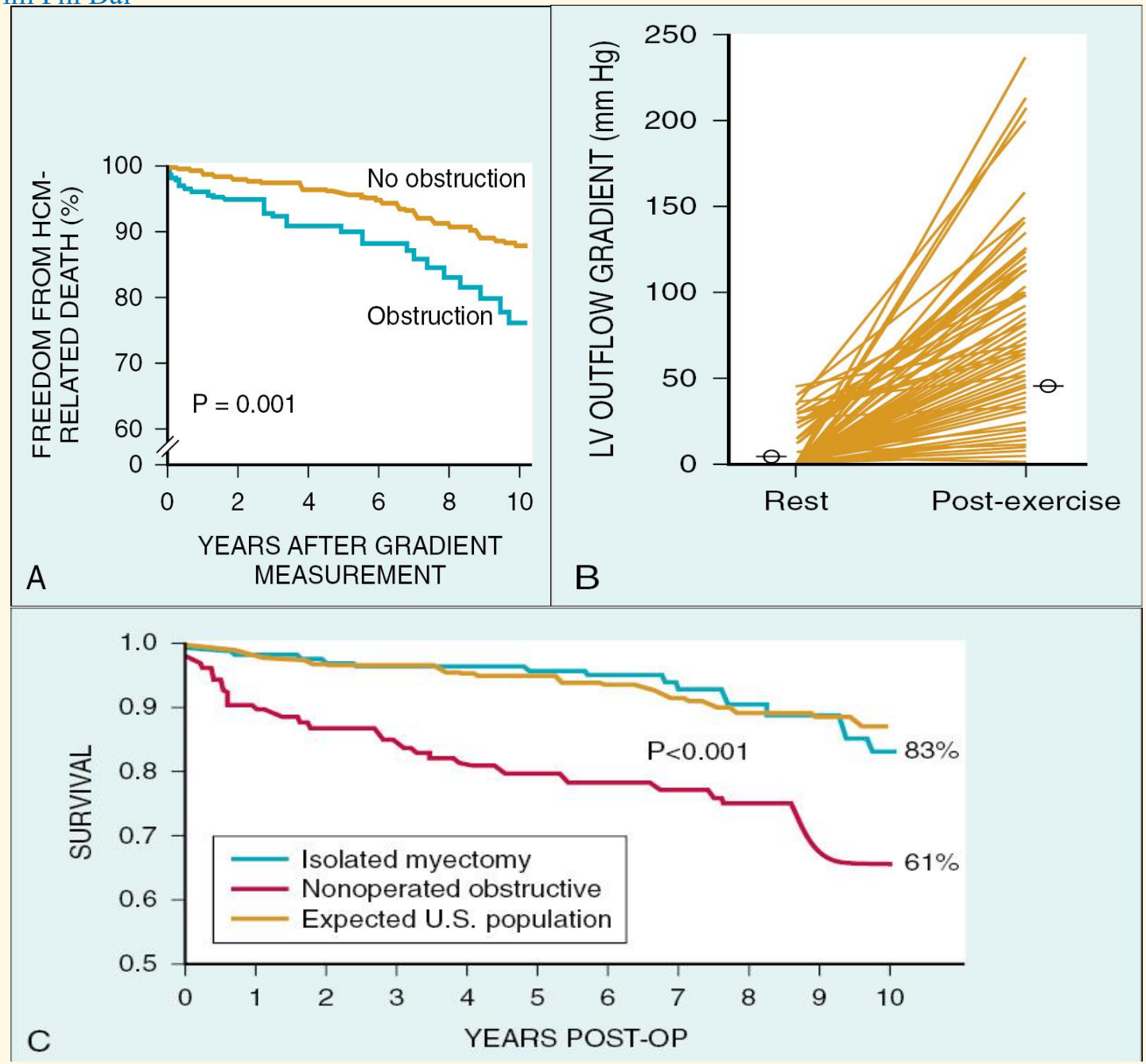
SAM: systolic anterior motion

Ảnh hưởng lên sống còn của nghẽn buồng tống thất trái



C: sau phẫu thuật cắt bớt cơ tim vách thất (septal myoctomy): sống còn tương đương quần thể chung

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TL: Maron BJ. Braunwald's Heart Disease 9th ed, 2012 Elsevier Saunders 1582-1594

Chẩn đoán BCTPĐ

- Lâm sàng
- Cận lâm sàng:
  - ECG, XQ ngực, siêu âm tim QTN, siêu âm tim QTQ, SAT Dobutamin
  - MSCT, MRI, thông tim chụp mạch
- Chẩn đoán các bệnh có phì đại cơ tim: Noonan, Danon, Fabry…

Examples of signs and symptoms suggestive of specific diagnoses

Symptom/sign	Diagnosis
Learning difficulties, mental retardation	• Mitochondrial diseases • Noonan/LEOPARD/Costello syndrome • Danon disease
Sensorineural deafness	• Mitochondrial diseases (particularly with diabetes) • Anderson-Fabry disease • LEOPARD syndrome
Visual impairment	• Mitochondrial diseases (retinal disease, optic nerve atrophy) • TTR-related amyloidosis (cotton wool type vitreous opacities) • Danon disease (retinitis pigmentosa) • Anderson-Fabry disease (cataracts, corneal opacities)

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Examples of signs and symptoms suggestive of specific diagnoses *(Cont.)*

Symptom/sign	Diagnosis
Gait disturbance	• Friedreich's ataxia
Paraesthesia/sensory abnormalities/neuropathic pain	• Amyloidosis • Anderson-Fabry disease
Carpal tunnel syndrome	• TTR-related amyloidosis (especially when bilateral and in male patients)
Muscle weakness	• Mitochondrial diseases • Glycogen storage disorders • FHL1 mutations • Friedreich's ataxia
Palpebral ptosis	• Mitochondrial diseases • Noonan/LEOPARD syndrome • Myotonic dystrophy
Lentigines/café au lait spots	• LEOPARD/Noonan syndrome
Angiokeratomata, hypohidrosis	• Anderson-Fabry disease

FHL1 = four and a half LIM domains 1; LEOPARD = lentigines, ECG abnormalities,ocular hypertelorism, pulmonary stenosis, abnormal genitalia, retardation of growth and sensorineural deafness; TTR = transthyretin.

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Electrocardiographic abnormalities suggesting specific diagnoses or morphological variants

Finding	Comment
Short PR interval/ pre-excitation	Pre-excitation is a common feature of storage diseases (Pompe, PRKAG2, and Danon) and mitochondrial disorders (MELAS, MERFF). A short PR interval without pre-excitation is seen in Anderson-Fabry disease.
AV block	Progressive atrioventricular conduction delay is common in mitochondrial disorders, some storage diseases (including Anderson-Fabry disease), amyloidosis, desminopathies and in patients with PRKAG2 mutations.
Extreme LVH (Sokolow score ≥50)	Extremely large QRS voltage is typical of storage diseases such as Pompe and Danon disease, but can be caused by pre-excitation alone.
Low QRS voltage (or normal voltages despite increased LV wall thickness)	Low QRS voltage in the absence of pericardial effusion, obesity and lung disease is rare in HCM (limited to cases with end-stage evolution) but is found in up to 50% of patients with AL amyloidosis and 20% with TTR amyloidosis. Differential diagnosis between HCM and cardiac amyloidosis is aided by measuring the ratio between QRS voltages and LV wall thickness.

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Electrocardiographic abnormalities suggesting specific diagnoses or morphological variants (Cont.)

Finding	Comment
Extreme superior ("North West") QRS axis deviation	Seen in patients with Noonan syndrome who have severe basal hypertrophy extending into the RV outflow tract.
Giant negative T wave inversion (>10 mm)	Giant negative T wave inversion in the precordial and/or inferolateral leads suggests involvement of the LV apex.
Abnormal Q waves ≥40 ms in duration and/or ≥25% of the R wave in depth and/or ≥3 mm in depth in at least two contiguous leads except aVR	Abnormally deep Q waves in the inferolateral leads, usually with a positive T wave, are associated with an asymmetrical distribution of LVH. Q waves of abnormal duration (≥40 ms) are associated with areas of replacement fibrosis.
Coved ST-segment elevation in lateral chest leads	Some patients with apical or distal hypertrophy develop small apical aneurysms, sometimes associated with myocardial scarring. These may only be detectable on CMR, ventriculography or contrast echo, and are occasionally associated with ST-segment in the lateral chest leads.

MELAS =mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes; MERFF =myoclonic epilepsy with ragged red fibres; PRKAG2 = gamma-2 subunit of the adenosine monophosphate-activated protein kinase.

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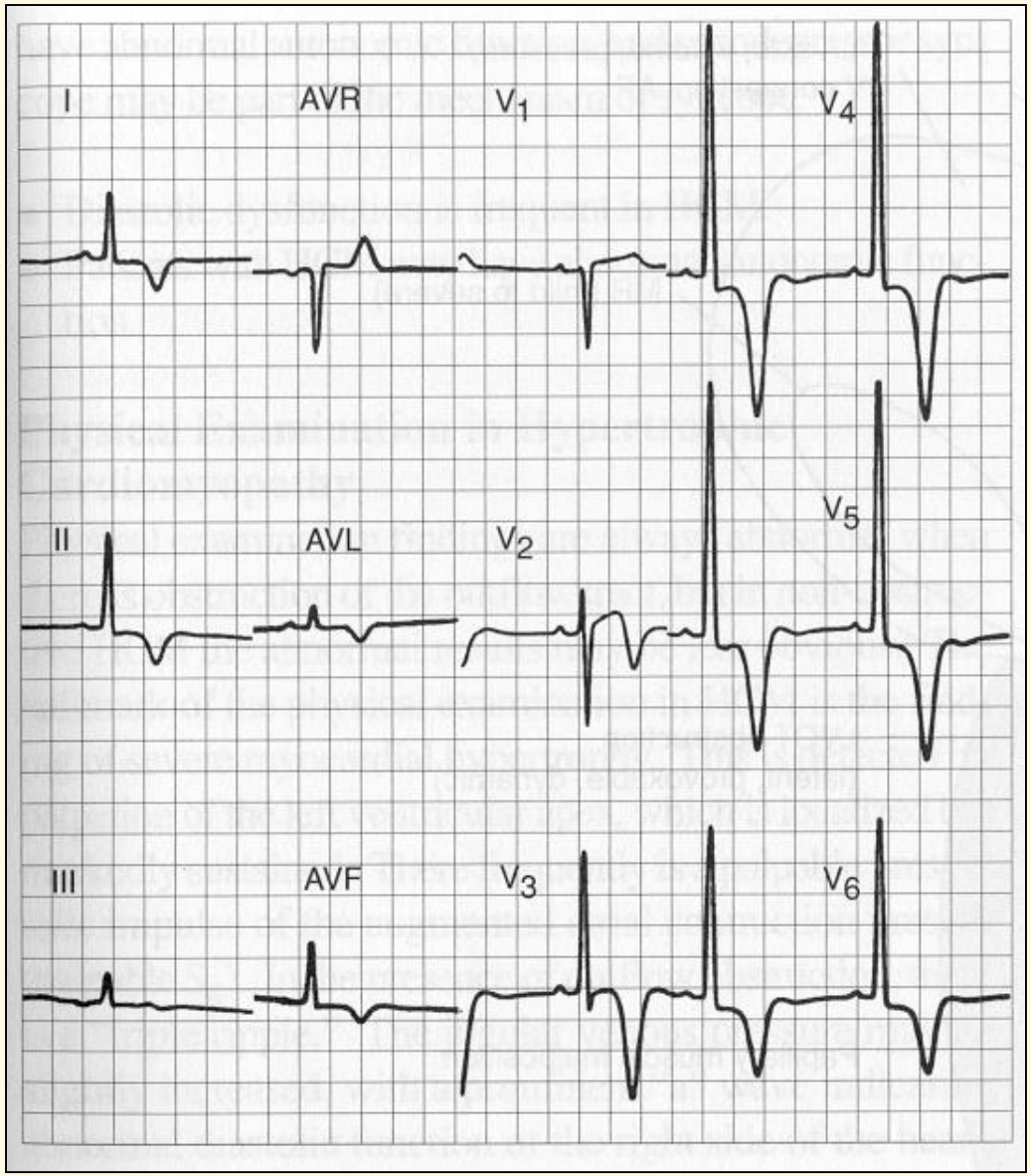
Cận lâm sàng: ECG

- Nhịp xoang
- Thường có loạn nhịp: NNTT, NTT thất, NNT, rung nhĩ
- Dầy thất trái



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Echocardiographic features that suggest specific aetiologies

Finding	Specific diseases to be considered
Increased interatrial septum thickness	Amyloidosis
Increased AV valve thickness	Amyloidosis; Anderson-Fabry disease
Increased RV free wall thickness	Amyloidosis, myocarditis, Anderson-Fabry disease, Noonan syndrome and related disorders
Mild to moderate pericardial effusion.	Amyloidosis, myocarditis
Ground-glass appearance of ventricular myocardium on 2-D echocardiography	Amyloidosis
Concentric LVH	Glycogen storage disease, Anderson-Fabry disease, PRKAG2 mutations
Extreme concentric LVH (wall thickness ≥30 mm)	Danon disease, Pompe disease
Global LV hypokinesia (with or without LV dilatation)	Mitochondrial disease, TTR-related amyloidosis, PRKAG2 mutations, Danon disease, myocarditis, advanced sarcomeric HCM, Anderson-Fabry disease
Right ventricular outflow tract obstruction	Noonan syndrome and associated disorders

PRKAG2 = gamma-2 subunit of the adenosine monophosphate-activated protein kinase;

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Cận lâm sàng: siêu âm tim

Các dấu hiệu lâm sàng:

* Phì đại không đối xứng vách liên thất (ASH)
* Chuyển động tới trước van 2 lá kỳ tâm thu (SAM)
* Nghẽn buồng tống thất trái : ±

ASH (Asymmetric Septal Hypertrophy)

$$\text{Tỷ lệ } \frac{\text{VLT}}{\text{Vách sau thất trái}} \geq 1.3 : 1$$

ASH còn thấy được ở :

* Trẻ sơ sinh đến 1, 2 tuổi
* Tổn thương làm tăng áp lực thất phải lâu ngày (TD: Hẹp van ĐMP, tăng áp ĐMP nguyên phát, CVĐĐM)
* Hẹp van ĐMC, tăng huyết áp,
* Sarcomas, amyloidosis, Freidreich' s ataxia, phù niêm (myxedema)

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Transoesophageal echocardiography

Recommendations	Class	Level
Perioperative TOE is recommended in patients undergoing septal myectomy, to confirm the mechanism of LVOTO, to guide the surgical strategy, to assess post-surgical complications and to detect residual LV outflow tract obstruction.	I	C
TOE should be considered in patients with LVOTO if the mechanism is unclear, or when assessing the mitral valve apparatus before a septal reduction procedure, or when severe mitral regurgitation, caused by intrinsic valve abnormalities, is suspected.	IIa	C
TOE with intracoronary contrast injection of the candidate septal perforator artery should be considered to guide septal alcohol ablation when transthoracic windows are insufficient for proper visualization of echo-contrast within the myocardium.	IIa	C

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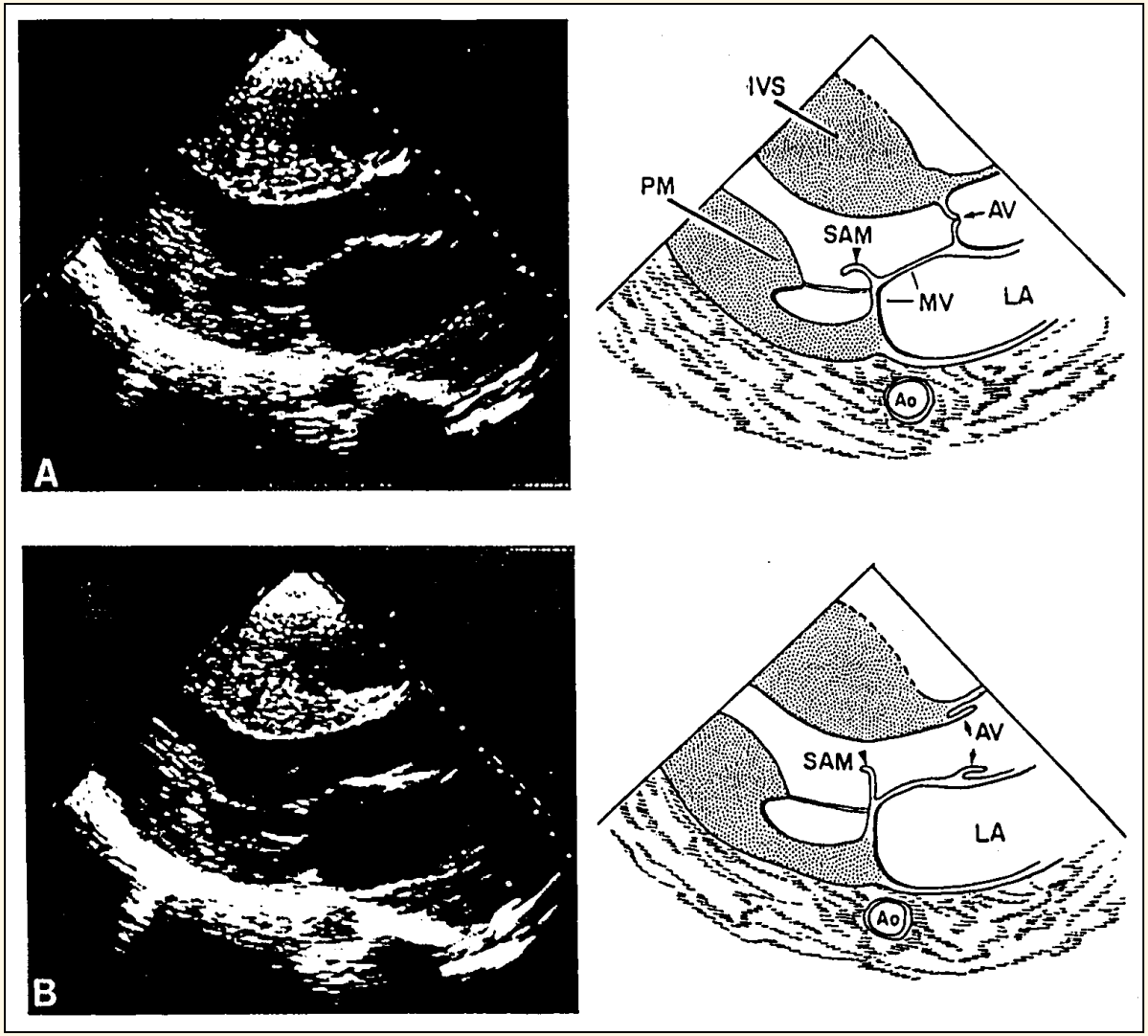
Siêu âm tim

ISV : Vách liên thất
PM : Cột cơ
MV : Van 2 lá
LA : Nhĩ trái
AV : Van ĐMC
SAM : Systolic anterior motion (vận động tới trước kỳ tâm thu)



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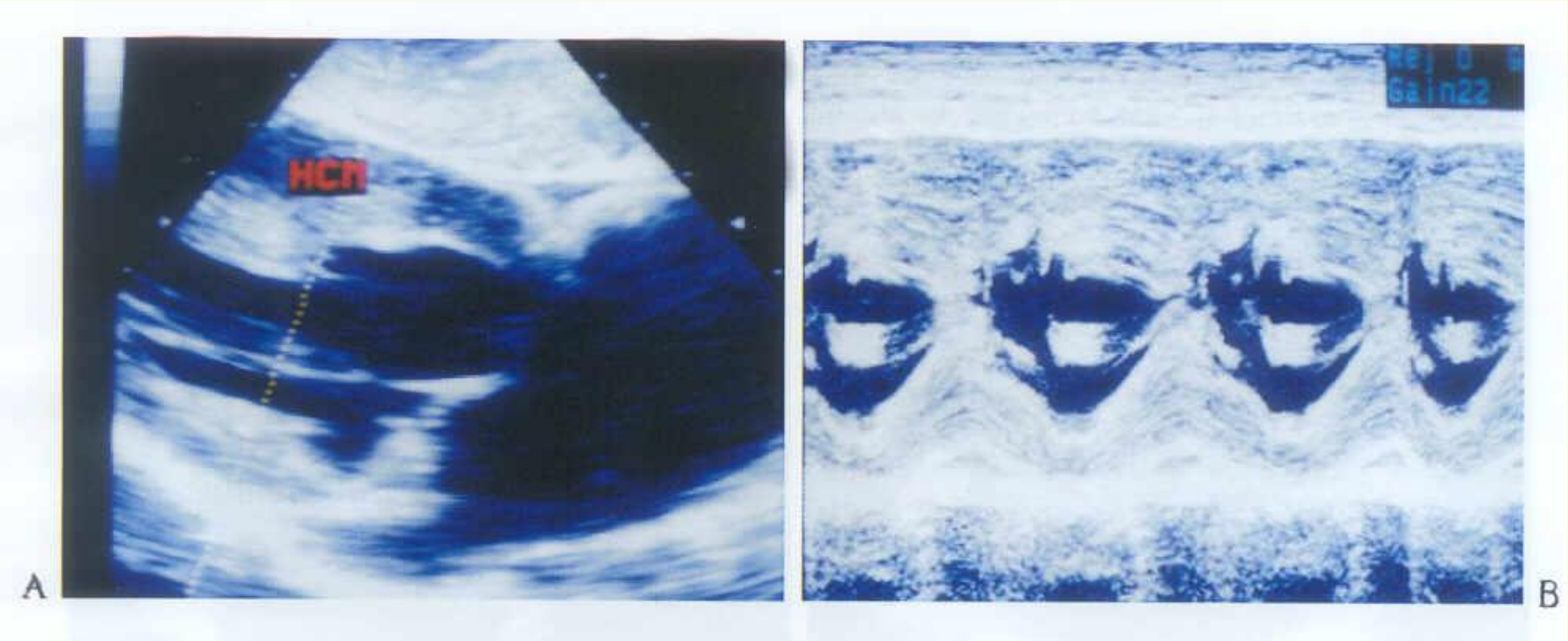
Siêu âm tim: bệnh nhân nam 8 tuổi/ BCTPĐ



Mặt cắt cạnh ức trục dọc ngang qua thất trái: hình ảnh bệnh cơ tim phì đại thể tắc nghẽn trên bệnh nhân nam 8 tuổi. Thất trái dầy đồng tâm; bề dầy vách liên thất là d1 = 13,5mm; bề dầy thành sau thất trái là d2 = 7mm. Tỷ lệ VLT/thành sau thất trái = 1,9

TL: Phạm Nguyễn Vinh và c/s: Atlas siêu âm tim 2D và Doppler màu. NXBYH 2002

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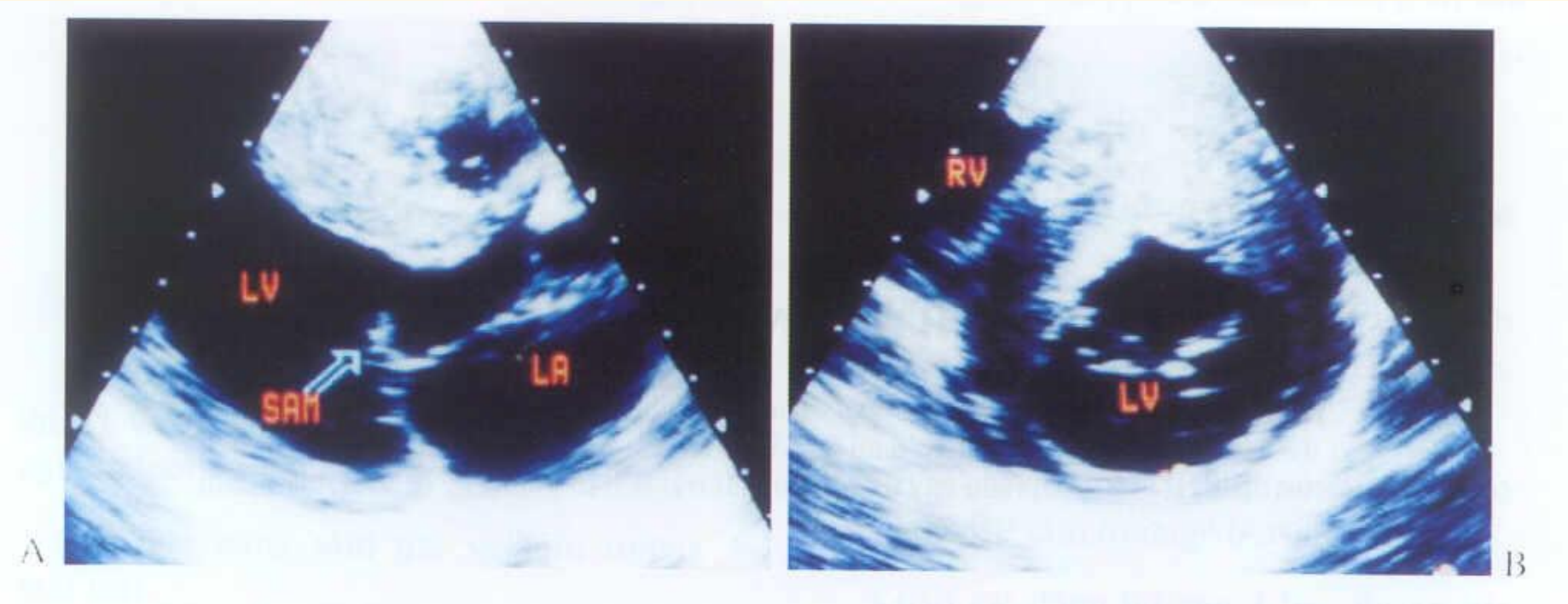
Siêu âm tim: bệnh nhân nam 18t/ BCTPĐ



Mặt cắt cạnh ức trục dọc: hình ảnh bệnh cơ tim phì đại trên bệnh nhân nam 18 tuổi. Hình ảnh SAM lá van trước van 2 lá (A). Mặt cắt cạnh ức trục ngang - ngang 2 thất, chiều dầy vách liên thất là 24mm, chiều dầy thành sau thất trái là 8mm, tỷ lệ VLT/thành sau thất trái = 3 (B)

TL: Phạm Nguyễn Vinh và c/s: Atlas siêu âm tim 2D và Doppler màu. NXBYH 2002

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Cardiovascular magnetic resonance evaluation in hypertrophic cardiomyopathy

Recommendations	Class	Level
It is recommended that CMR studies be performed and interpreted by teams experienced in cardiac imaging and in the evaluation of heart muscle disease.	I	C
In the absence of contra-indications, CMR with LGE is recommended in patients with suspected HCM who have inadequate echocardiographic windows, in order to confirm the diagnosis.	I	B
In the absence of contra-indications, CMR with LGE should be considered in patients fulfilling diagnostic criteria for HCM, to assess cardiac anatomy, ventricular function, and the presence and extent of myocardial fibrosis.	IIa	B
CMR with LGE imaging should be considered in patients with suspected apical hypertrophy or aneurysm.	IIa	C
CMR with LGE imaging should be considered in patients with suspected cardiac amyloidosis.	IIa	C
CMR with LGE may be considered before septal alcohol ablation or myectomy, to assess the extent and distribution of hypertrophy and myocardial fibrosis.	IIb	C

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Nuclear scintigraphy; cardiac CT

Recommendations	Class	Level
Bone scintigraphy (particularly with 99mTc-DPD) should be considered in patients with symptoms, signs and non-invasive tests consistent with TTR-related amyloidosis.	IIa	B
Cardiac CT should be considered in patients who have inadequate echocardiographic imaging and contra-indications for CMR.	IIa	C

- TTR-related amyloidosis: age > 65 yo; history of bilateral carpal tunnel S; absent family history of HCM; features consisting with cardiac amyloidosis in ECG and cardiac imaging
- TTR derived fibrils: avidity for bone tracers

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Laboratory tests in adult patients with hypertrophic cardiomyopathy (HCM)

Test	Comment
Haemoglobin	• Anaemia exacerbates chest pain and dyspnoea and should be excluded whenever there is a change in symptoms.
Renal function	• Renal function may be impaired in patients with severe left ventricular impairment. • Impaired GFR and proteinuria may be seen in amyloidosis, Anderson-Fabry disease and mitochondrial DNA disorders.
Liver transaminases	• Liver tests may be abnormal in mitochondrial disorders, Danon disease and ß-oxidation defects.
Creatine phosphokinase	• Serum creatine phosphokinase is raised in metabolic disorders such as Danon and mitochondrial disease.

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Genetic counselling

Recommendations	Class	Level
Genetic counselling is recommended for all patients with HCM when their disease cannot be explained solely by a non-genetic cause, whether or not clinical or genetic testing will be used to screen family members.	I	B
Genetic counselling should be performed by professionals trained for this specific task working within a multidisciplinary specialist team.	IIa	C

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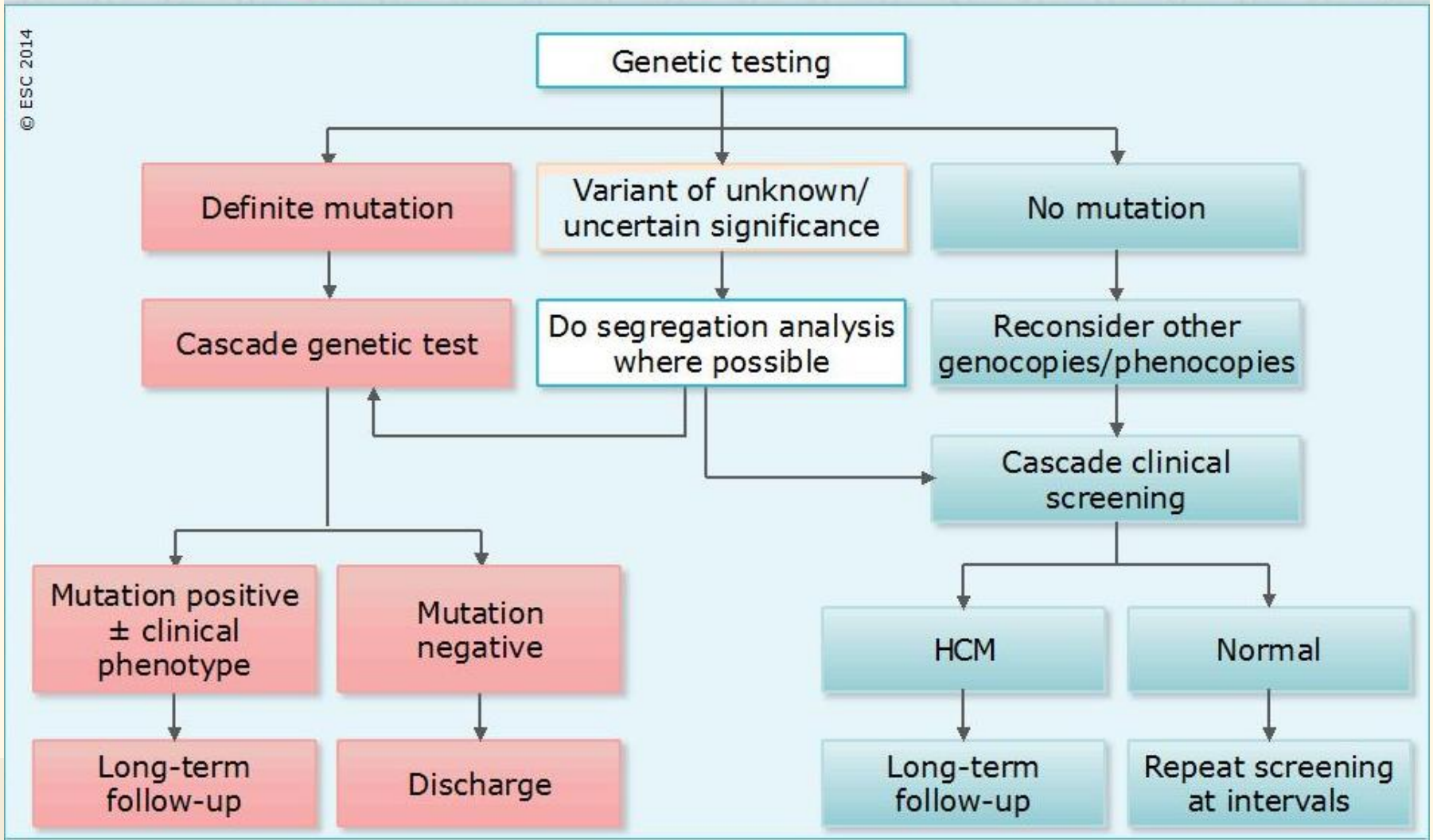
Flow chart for the genetic and clinical screening of probands and relatives



HCM = hypertrophic cardiomyopathy.
Cascade genetic test = screening of first degree relatives of patients already diagnosed with HCM.

Segregation analysis: identify the factors responsible for familial aggregation

Genetic and clinical screening in children

Recommendations	Class	Level
The children of patients with a definite disease-causing mutation should be considered for predictive genetic testing—following pre-test family counselling—when they are aged 10 or more years and this should be carried out in accordance with international guidelines for genetic testing in children.	IIa	C
In first-degree child relatives aged 10 or more years, in whom the genetic status is unknown, clinical assessment with ECG and echocardiography should be considered every 1–2 years between 10 and 20 years of age, and then every 2–5 years thereafter.	IIa	C

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Coronary angiography

Recommendations	Class	Level
Invasive coronary angiography is recommended in adult survivors of cardiac arrest, in patients with sustained ventricular tachyarrhythmia and in patients with severe stable angina (Canadian Cardiovascular Society (CCS) Class ≥3).	I	C
Invasive or CT coronary angiography should be considered in patients with typical exertional chest pain (CCS Class <3) who have an intermediate pre-test probability of atherosclerotic coronary artery disease based on age, gender and risk factors for atherosclerosis, or a history of coronary revascularization.	IIa	C
In all patients aged 40 years or more, invasive or CT coronary angiography should be considered before septal reduction therapy, irrespective of the presence of typical exertional chest pain.	IIa	C

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Medical treatment of left ventricular outflow tract obstruction

Recommendations	Class	Level
Non-vasodilating ß-blockers, titrated to maximum tolerated dose, are recommended as first-line therapy to improve symptoms in symptomatic patients with resting or provoked LVOTO.	I	B
Verapamil, titrated to maximum tolerated dose, is recommended to improve symptoms in symptomatic patients with resting or provoked[a] LVOTO, who are intolerant or have contra-indications to ß-blockers.	I	B
ß-Blockers or verapamil may be considered in children and *asymptomatic* adults with resting or provoked[a] LVOTO, to reduce left ventricular pressures.	IIb	C

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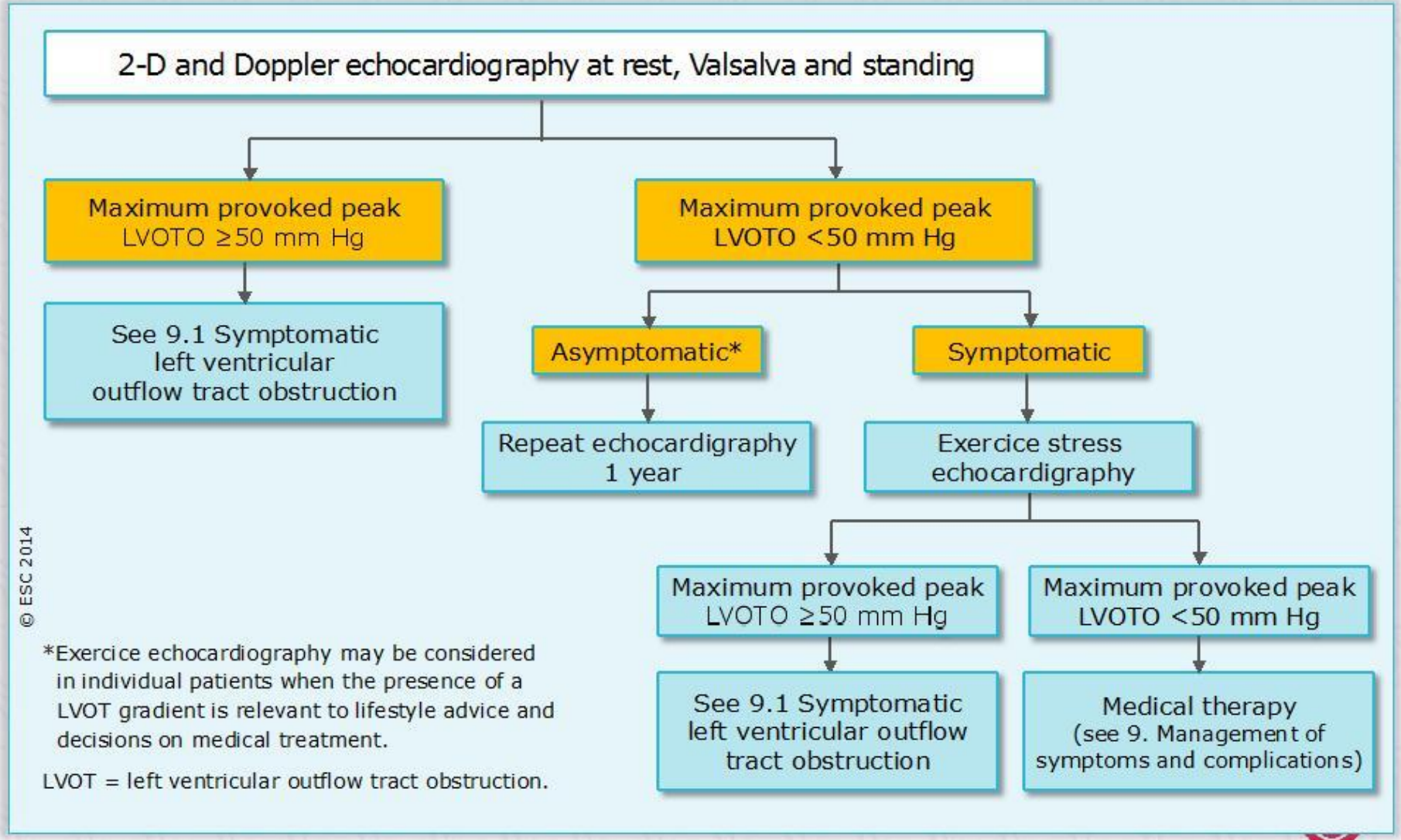
Protocol for the assessment and treatment of left ventricular outflow tract obstruction



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Septal reduction therapy

Recommendations	Class	Level
It is recommended that septal reduction therapies be performed by experienced operators, working as part of a multidisciplinary team expert in the management of HCM.	I	C
Septal reduction therapy to improve symptoms is recommended in patients with a resting or maximum provoked LVOT gradient of ≥50 mm Hg, who are in NYHA functional Class III–IV despite maximum tolerated medical therapy.	I	B
Septal reduction therapy should be considered in patients with recurrent exertional syncope caused by a resting or maximum provoked LVOTO gradient ≥50 mm Hg despite optimal medical therapy.	IIa	C
Septal myectomy, rather than SAA, is recommended in patients with an indication for septal reduction therapy and other lesions requiring surgical intervention (e.g. mitral valve repair/replacement, papillary muscle intervention).	I	C
Mitral valve repair or replacement should be considered in symptomatic patients with a resting or maximum provoked LVOTO gradient ≥50 mm Hg and moderate-to-severe mitral regurgitation not caused by SAM of the mitral valve alone.	IIa	C
Mitral valve repair or replacement may be considered in patients with a resting or maximum provoked LVOTO gradient ≥50 mm Hg and a maximum septal thickness ≤16 mm at the point of the mitral leaflet–septal contact or when there is moderate-to-severe mitral regurgitation following isolated myectomy.	IIb	C

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Pre-assessment check list for patients being considered for invasive septal reduction therapies

Are there alternative/additional explanations for symptoms?

- Obesity
- Respiratory Disease
- Coronary artery disease
- Anaemia
- Thyroid disease
- Arrhythmia (e.g. AF)
- Drug side-effects
- Systemic disease (e.g. amyloid)
- RVOT obstruction

What is the mechanism of obstruction?

- SAM-related
- Mid-cavity
- Sub-aortic membrane
- Aortic stenosis
- Anomalous papillary muscle insertion
- Accessory mitral valve tissue

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Pre-assessment check list for patients being considered for invasive septal reduction therapies *(Cont.)*

Assess mitral valve anatomu/function

- Mitral prolapse
- Other instrinsic MV abnormality

Assess distribution and severity of hypertrophy

Minimum anterior septal thickness 17 mm

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Patients with heart failure and preserved LV ejection fraction (≥50%)

Recommendations	Class	Level
In patients in NYHA functional Class II–IV with an LVEF ≥50% and no evidence for resting or provocable LVOTO, ß-blockers, verapamil or diltiazem should be considered, to improve heart failure symptoms.	IIa	C
Low-dose loop and thiazide diuretics should be considered in patients in NYHA functional Class II–IV with an EF ≥50%, and no evidence for resting or provocable LVOTO, to improve heart failure symptoms.	IIa	C

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Patients with heart failure and reduced LV ejection fraction (<50%)

Recommendations	Class	Level
An ACE-inhibitor (or ARB if ACE inhibitor not tolerated) should be considered, in addition to a ß-blocker, for patients without LVOTO who have an LVEF <50%, to reduce the risk of HF hospitalization and risk of premature death[a].	IIa	C
A ß-blocker should be considered, in addition to an ACE-inhibitor (or ARB if ACE-inhibitor not tolerated), for patients without LVOTO who have an LVEF <50% to improve symptoms and reduce the risks of HF hospitalization and premature death[a].	IIa	C
Low-dose loop diuretics should be considered for symptomatic patients in NYHA functional Class II–IV with an LVEF <50%, to improve symptoms and reduce the risk of HF hospitalization[a].	IIa	C
For all patients with persisting symptoms (NYHA functional Class II–IV) and an LVEF <50%—despite treatment with an ACE-inhibitor (or an ARB if an ACE-inhibitor is not tolerated) and a ß-blocker—a mineralocorticoid receptor antagonist (MRA) should be considered, to reduce the risks of HF hospitalization and premature death[a].	IIa	C
Low-dose digoxin may be considered for patients without LVOTO who are in NYHA functional Class II–IV and have an LVEF <50% and permanent atrial fibrillation to control heart rate response.	IIb	C

[a]In the absence of randomized trials in HCM, the benefit on hospitalization, symptoms and mortality is assumed but unproven.

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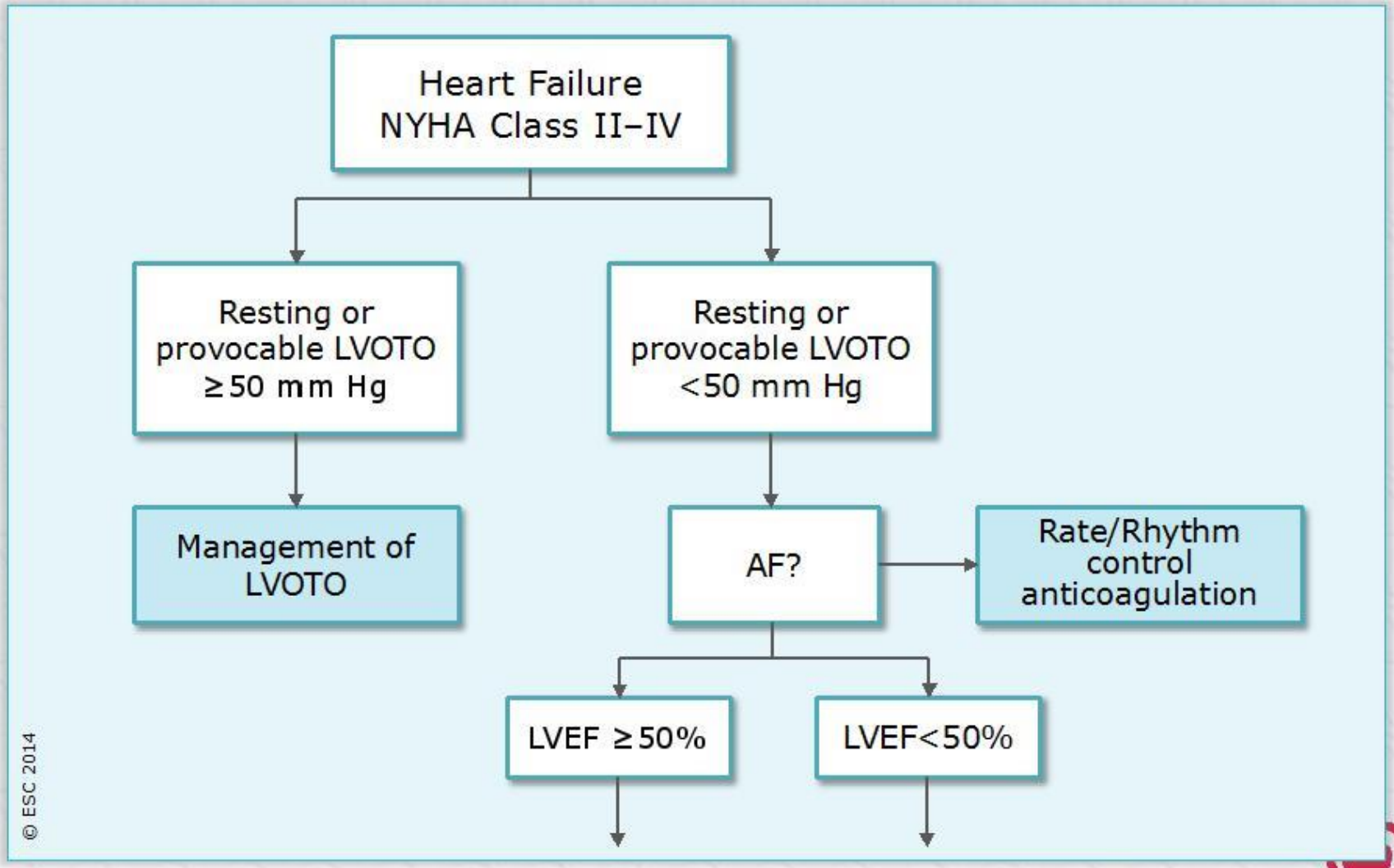
Algorithm for the treatment of heart failure in hypertrophic cardiomyopathy



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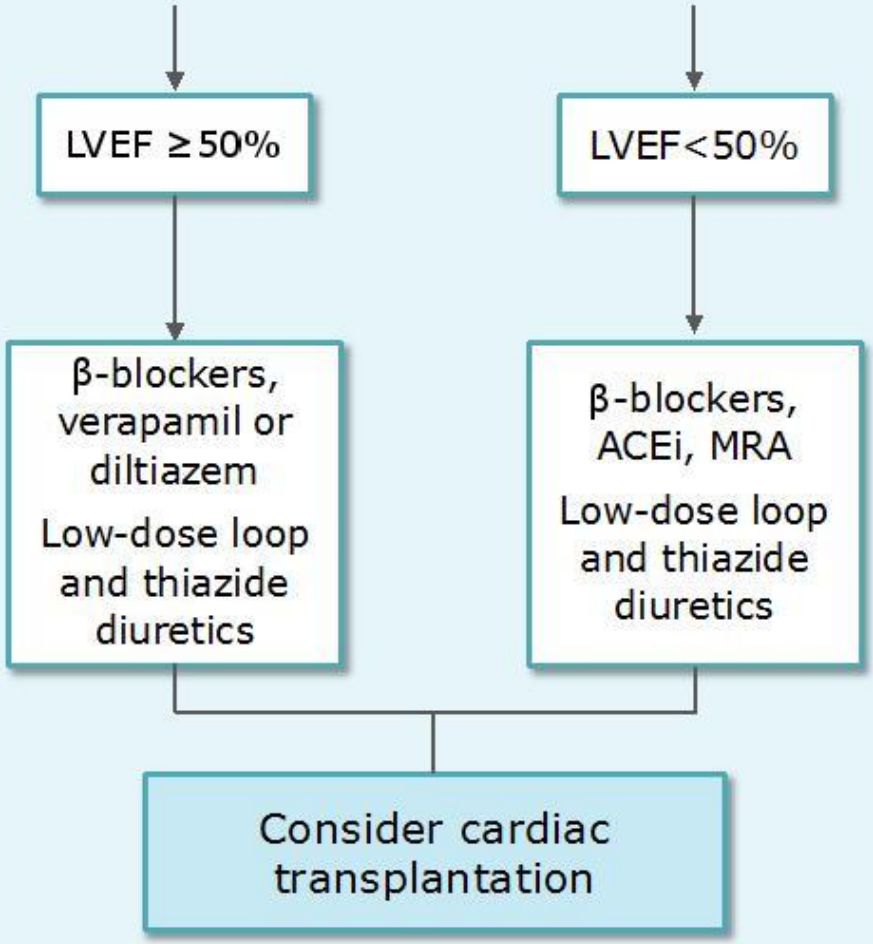
Algorithm for the treatment of heart failure in hypertrophic cardiomyopathy *(Cont.)*



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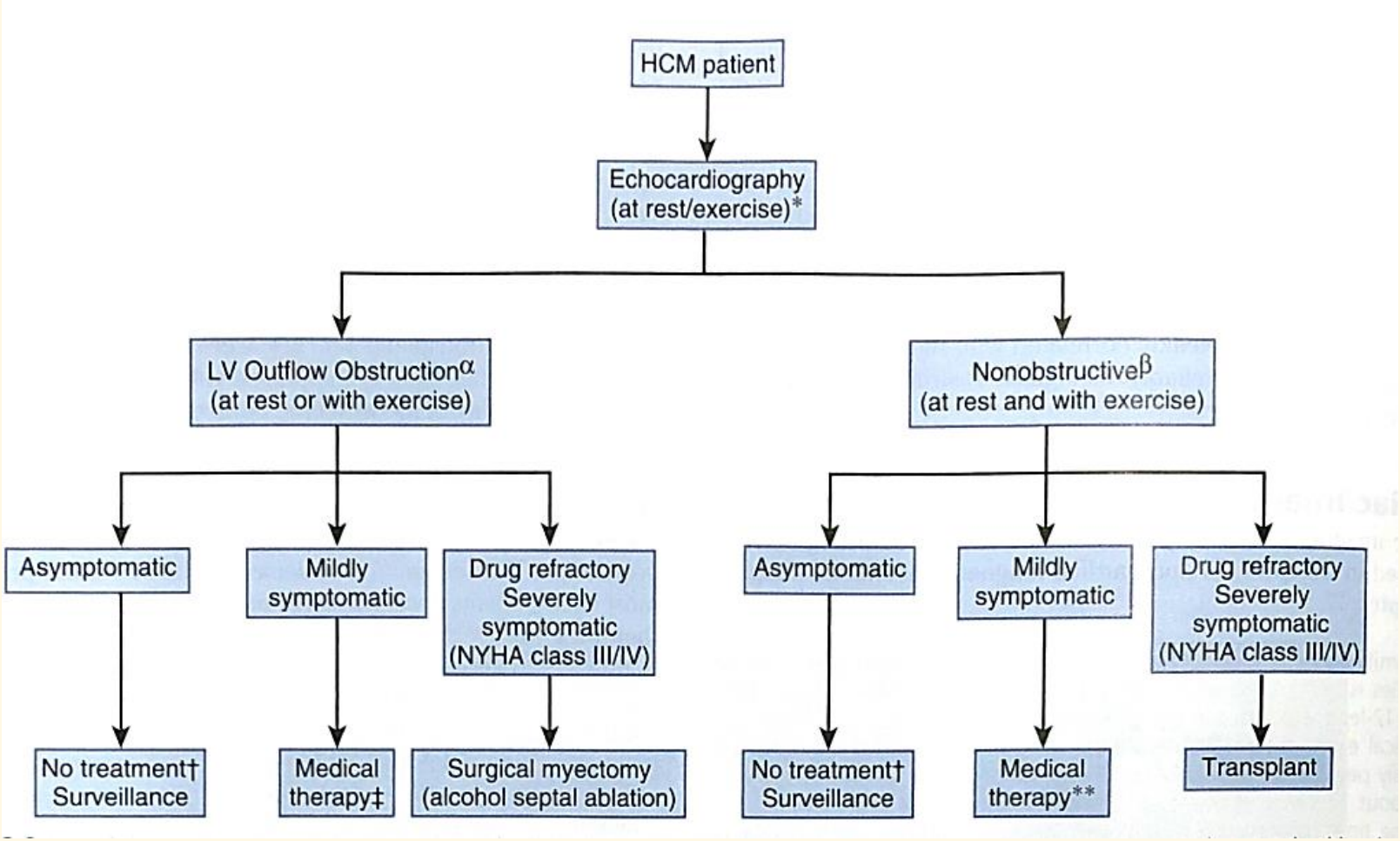
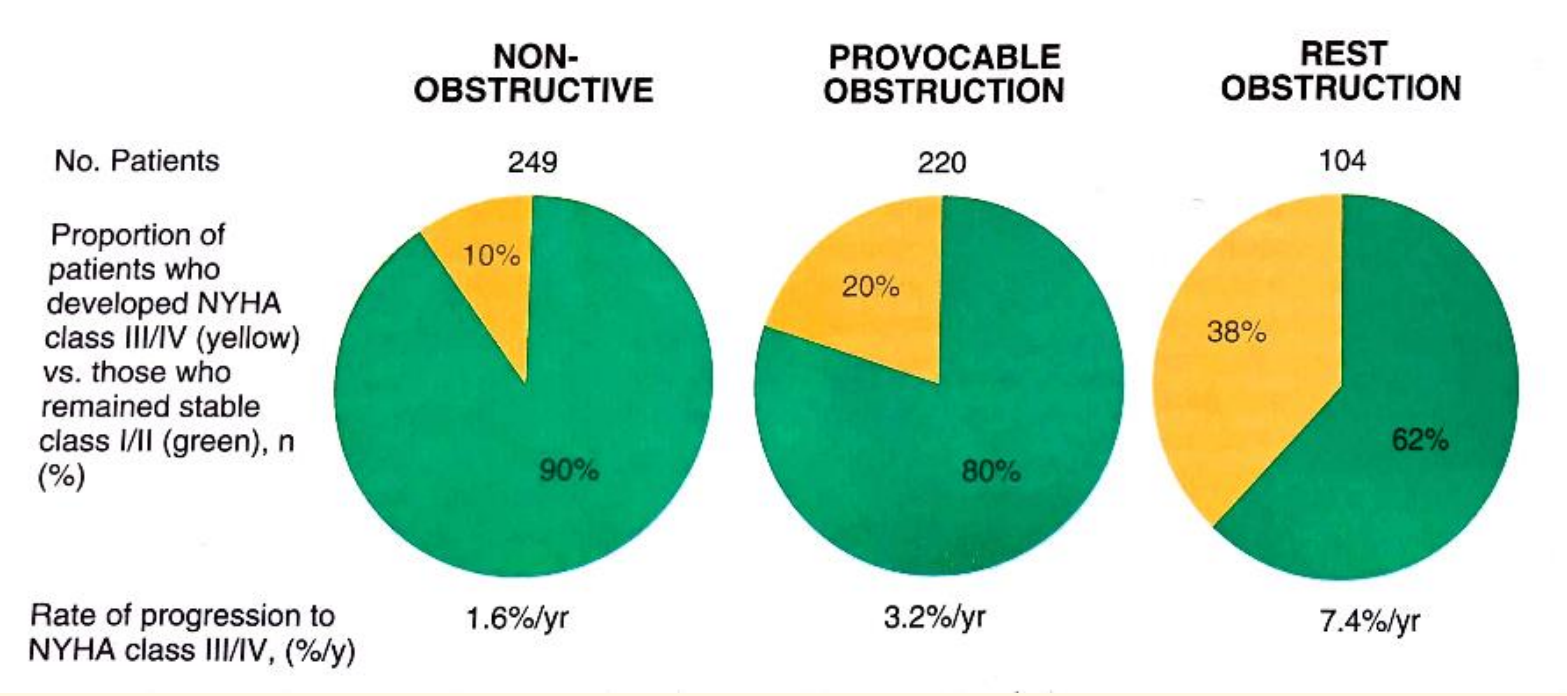
Chiến lược điều trị suy tim bắt đầu từ SÂ tim



Pham Nguyen Vinh

TL: Maron BJ et al. Braunwald's Heart Disease 11th ed, 2018 Elsevier Saunders, p. 1602-1615

Tiến triển đến suy tim trên 3 thể huyết động khác nhau của BCTPĐ



Pham Nguyen Vinh

TL: Maron BJ et al. Braunwald's Heart Disease 11th ed, 2018 Elsevier Saunders, p. 1602-1615

Risk factors for sudden death in HCM

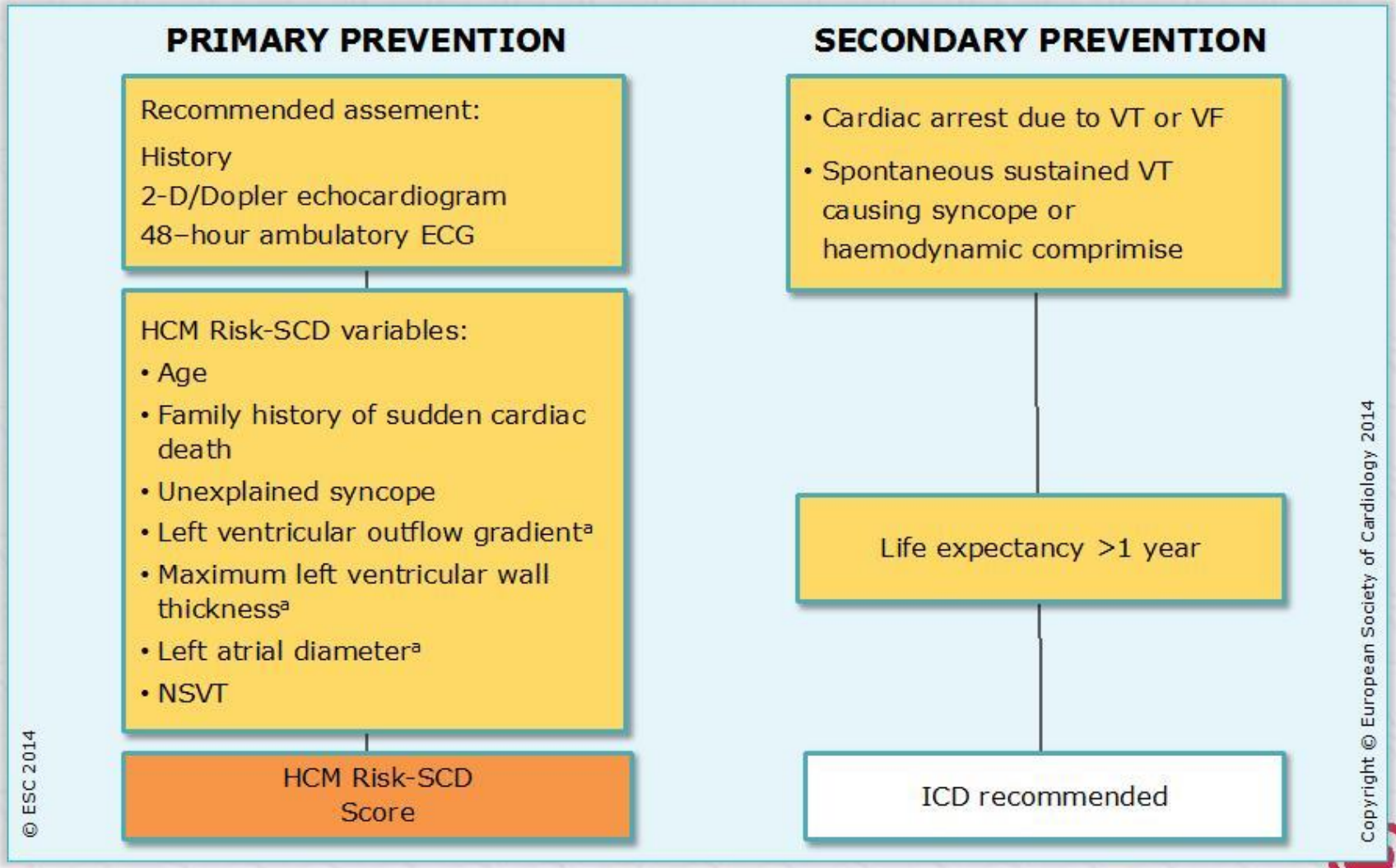
Secondary Prevention
• Cardiac arrest or sustained VT
Conventional Primary Prevention Risk Markers
• Family history of sudden death due to HCM • Unexplained recent syncope • Multiple/repetitive nonsustained VT (on ambulatory ECG tracing) • Hypotensive or attenuated blood pressure response to exercise • Massive LV hypertrophy (wall thickness, ≥30 mm*) • Extensive/diffuse late gadolinium enhancement (contrast CMR)
Potential High-Risk Subsets for Primary Prevention
• End-stage phase (ejection fraction <50%) • LV apical aneurysm and scarring
Potential Arbitrators for Primary Prevention†
• Substantial LV outflow gradient at rest • Alcohol septal ablation (infarct) • Multiple sarcomere mutations • Modifiable—e.g., intense competitive sports, coronary artery disease

VT = ventricular tachycardia.

Pham Nguyen Vinh

TL: Maron BJ, Maron MS. Lancet 381: 242, 2013

Flow chart for ICD implantation



[a]Use absolute values for LVOT gradient, MLVWT and left atrial dimension.

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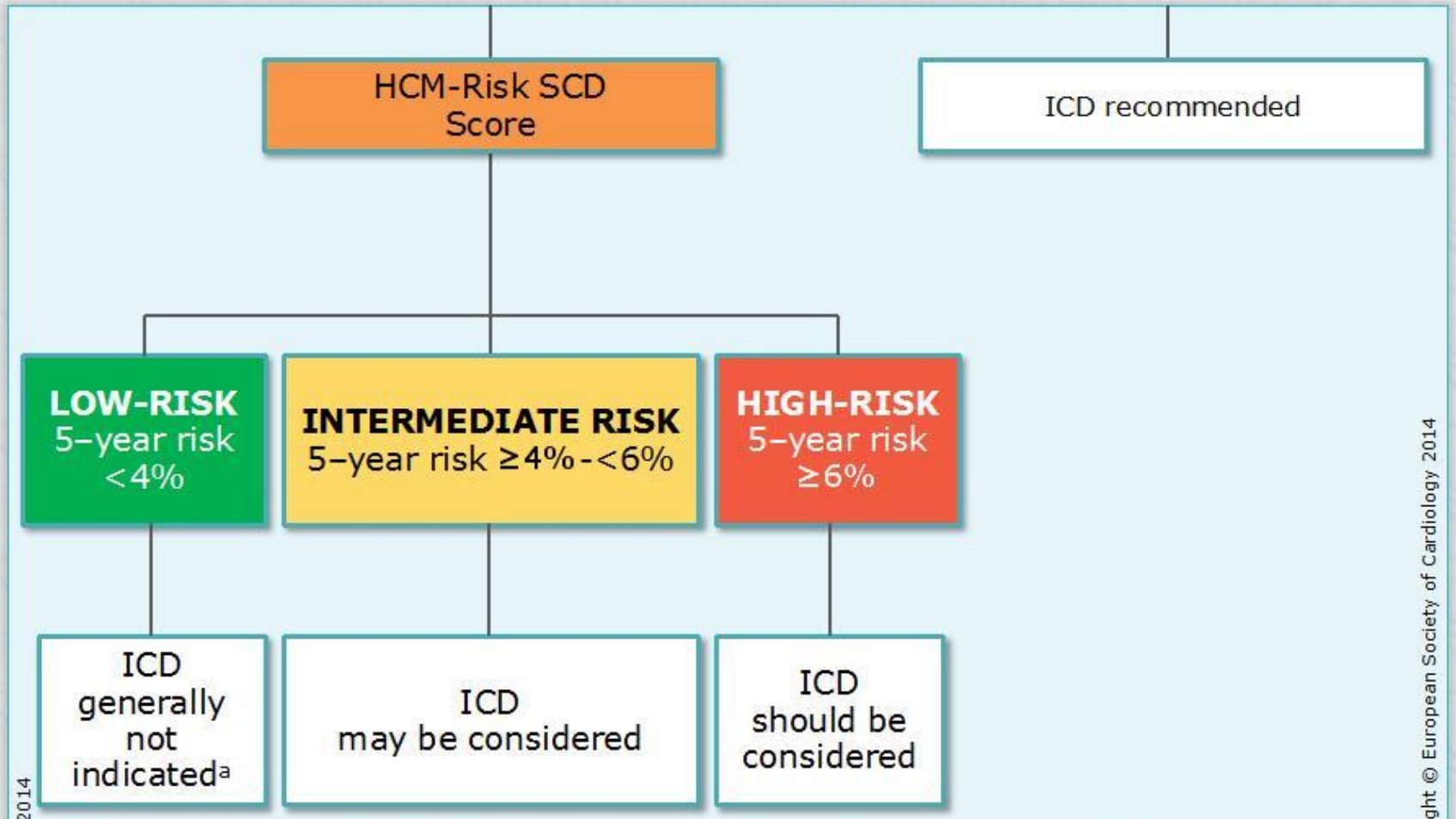
Flow chart for ICD implantation



[a]ICD not recommended unless there other clinical features that are of potential prognostic importance and when the likely benefit is greater thant the lifelong lrisk of complication s and the impact of an ICD on lifestyle, socioeconomic status and psychological health.

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ICD implantation in children

Recommendations	Class	Level
ICD implantation is recommended in children who have survived a cardiac arrest or experienced documented sustained ventricular tachycardia.	I	B
ICD implantation should be considered in children with two or more major paediatric risk factors* after appropriate counselling and when an assessment of the lifelong risk of complications and the impact of an ICD on lifestyle and psychological health suggests a net benefit from ICD therapy.	IIa	C
ICD implantation may be considered in children with a single major paediatric risk factor* after appropriate counselling and when an assessment of the lifelong risk of complications and the impact of an ICD on lifestyle and psychological health suggests a net benefit from ICD therapy.	IIb	C

Major paediatric risk factors: Maximum left ventricular wall thickness ≥ 30 mm or a Z-score ≥ 6, unexplained syncope, non-sustained ventricular tachycardia (≥3 consecutive ventricular beats at ≥ 120 BPM lasting,30 seconds), family history of SCD (one or more first-degree relatives with SCD aged, 40 years with or without the diagnosis of HCM, or SCD in a first-degree relative at any age with an established diagnosis of HCM).

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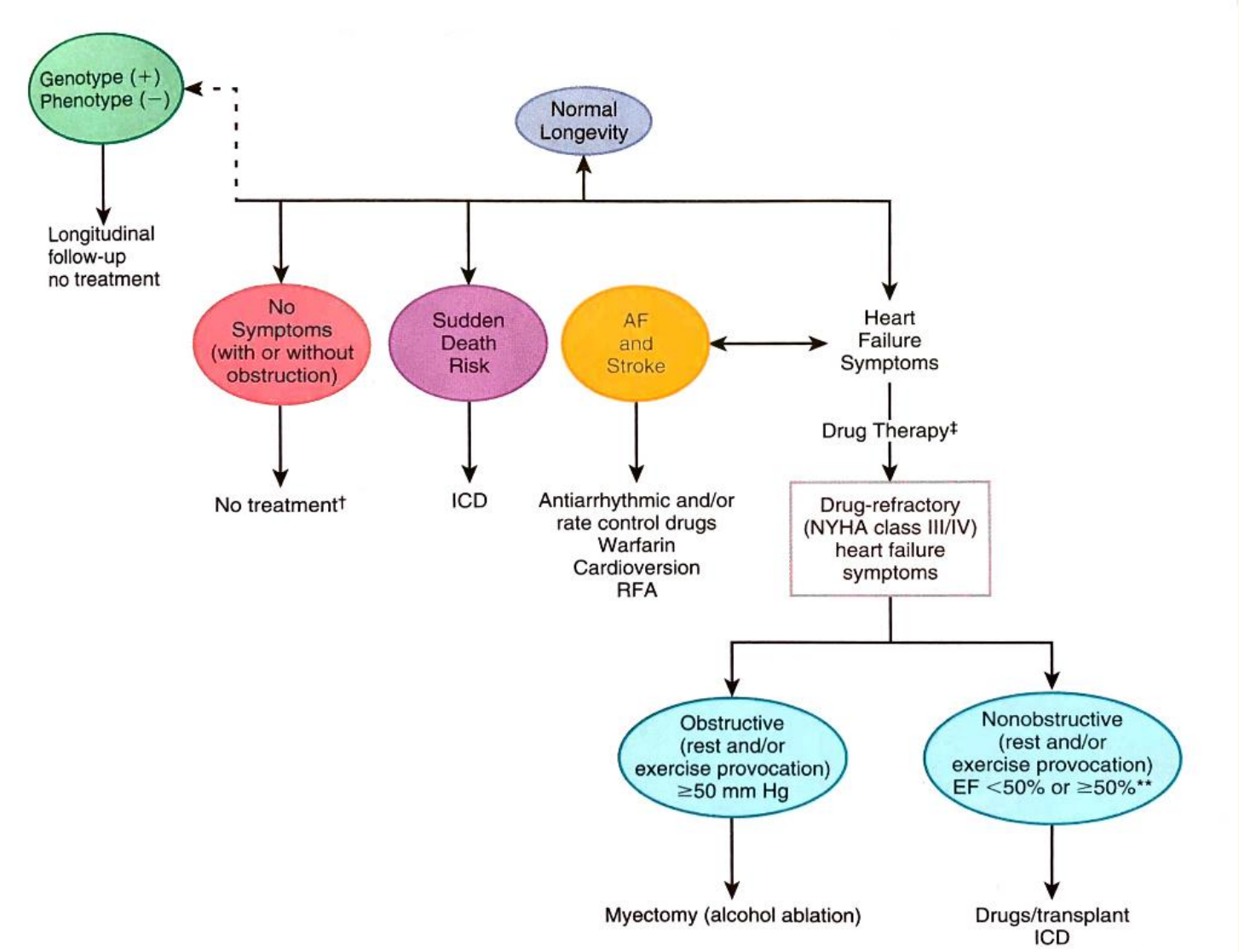
Chiến lược điều trị và tiên lượng bệnh nhân BCTPĐ



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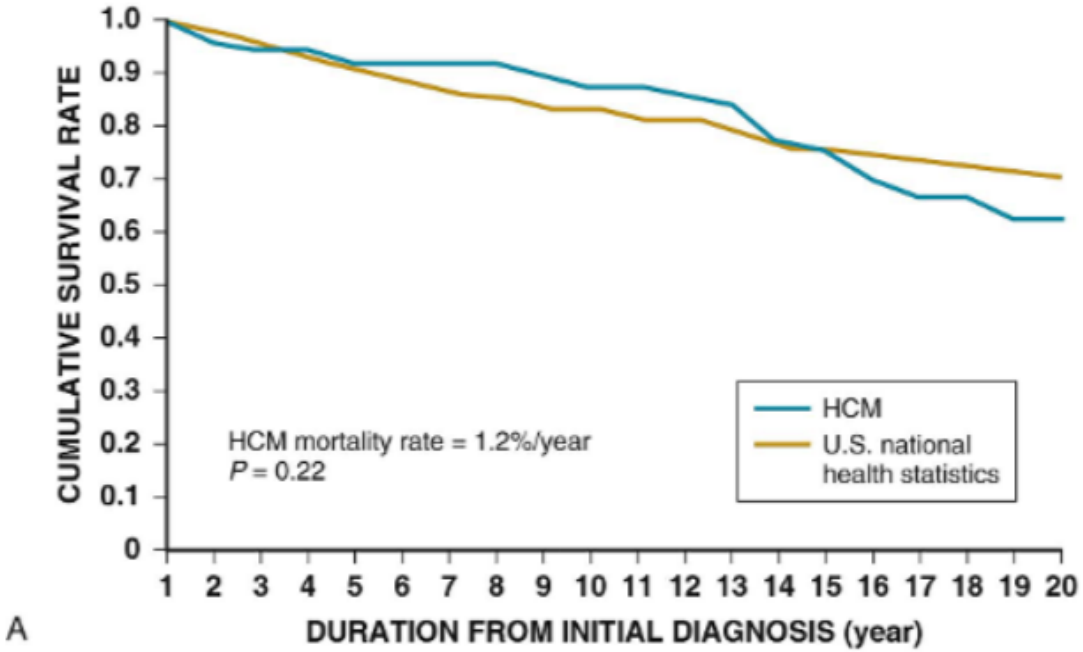
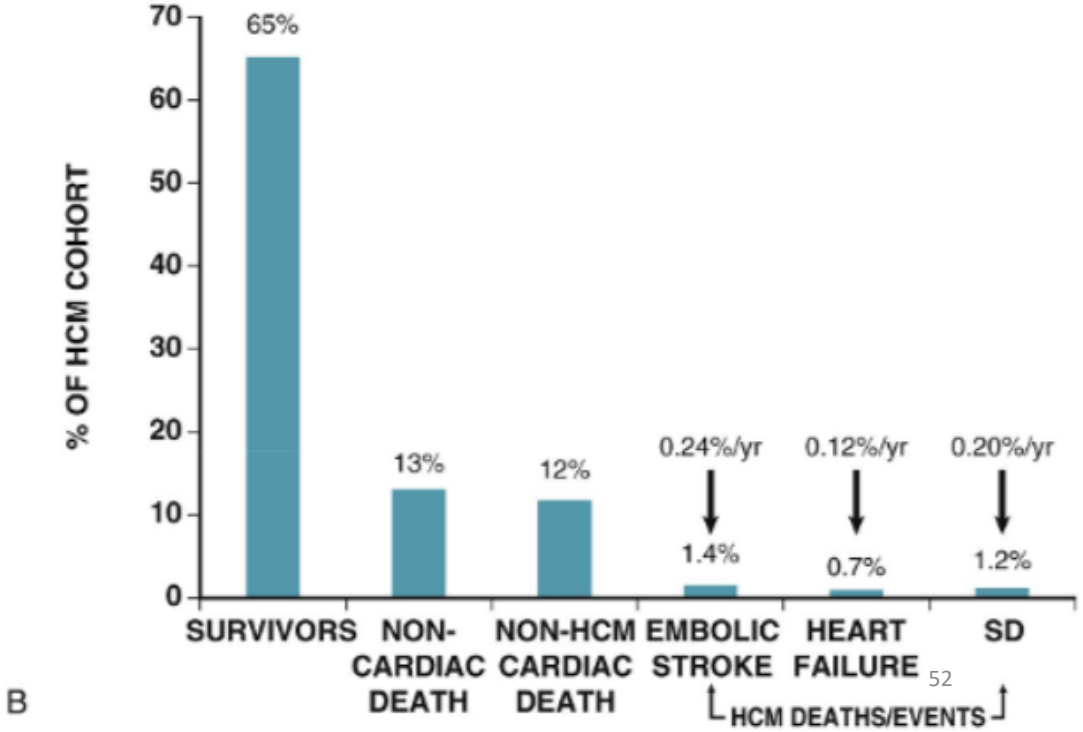
TL: Maron BJ et al. Braunwald's Heart Disease 11th ed, 2018 Elsevier Saunders, p. 1602-1615

Longterm outcome in HCM





TL: Maron BJ et al. JAMA 281: 650,199

Pham Nguyen Vinh

Kết luận

- Tần suất: 1/500 (USA)
- Di truyền:
  - 1500 đột biến gen đã xác nhận
  - Cần khảo sát di truyền: B/n, thế hệ 1
- Chẩn đoán: LS, ECG, siêu âm tim, siêu âm Dobutamin, MRI, thông tim, khảo sát di truyền
- Điều trị:
  - Thuốc, myomectomie, alcohol ablation, PM
  - Điều trị đúng: sống còn như bình thường

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