Risultati delle ricerche cliniche effettuate in questi 60 anni nella cardiomiopatia ipertrofica e quelle ancora necessarie

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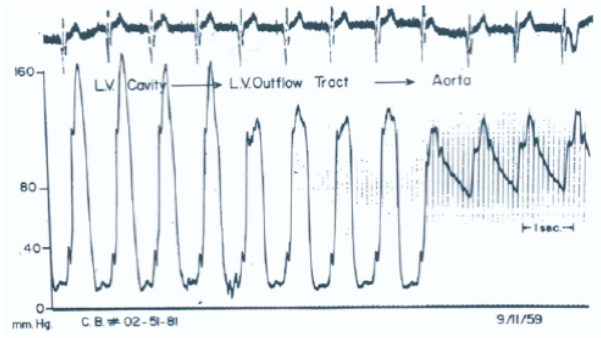

Circulation
JOURNAL OF THE AMERICAN HEART ASSOCIATION
American Heart Association®
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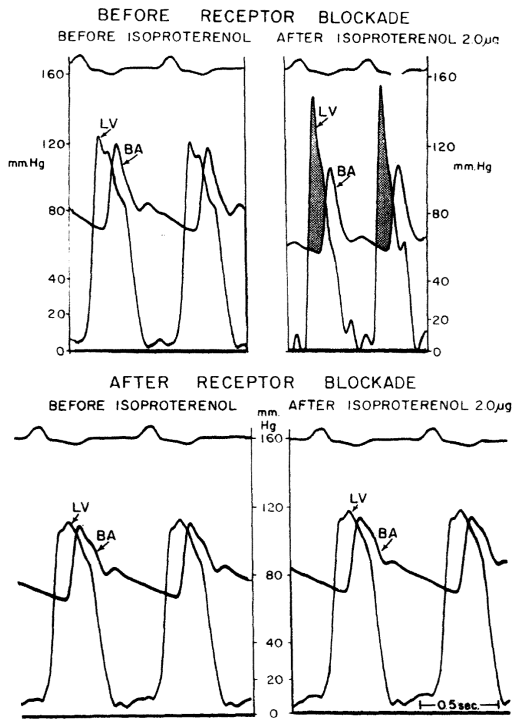
Circulation 1964, 29:84-98

Effects of Beta Adrenergic Blockade on the Circulation, with Particular Reference to Observations in Patients with Hypertrophic Subaortic Stenosis

DONALD C. HARRISON, EUGENE BRAUNWALD, GERALD GLICK, DEAN T. MASON, CHARLES A. CHIDSEY and JOHN ROSS, JR.



Il **Nethalide** (noto anche come **Pronethalolo** o con il nome commerciale **Alderlin**) è stato uno dei primi farmaci **beta-bloccanti** non selettivi ad essere sintetizzato.
Sviluppato nei primi anni '60 dal pioniere James Black, fu il primo farmaco di questa classe ad entrare in uso clinico per il trattamento di angina e aritmie
Tuttavia fu ritirato precocemente per lo scarso profilo di sicurezza, come anche successivamente il **Practololo.**

Il Propanololo dimostrò invece un profilo di sicurezza ottimale ed è ancora in uso

HYPERTROPHIC CARDIOMYOPATHY

HCM is a PHENOTYPE

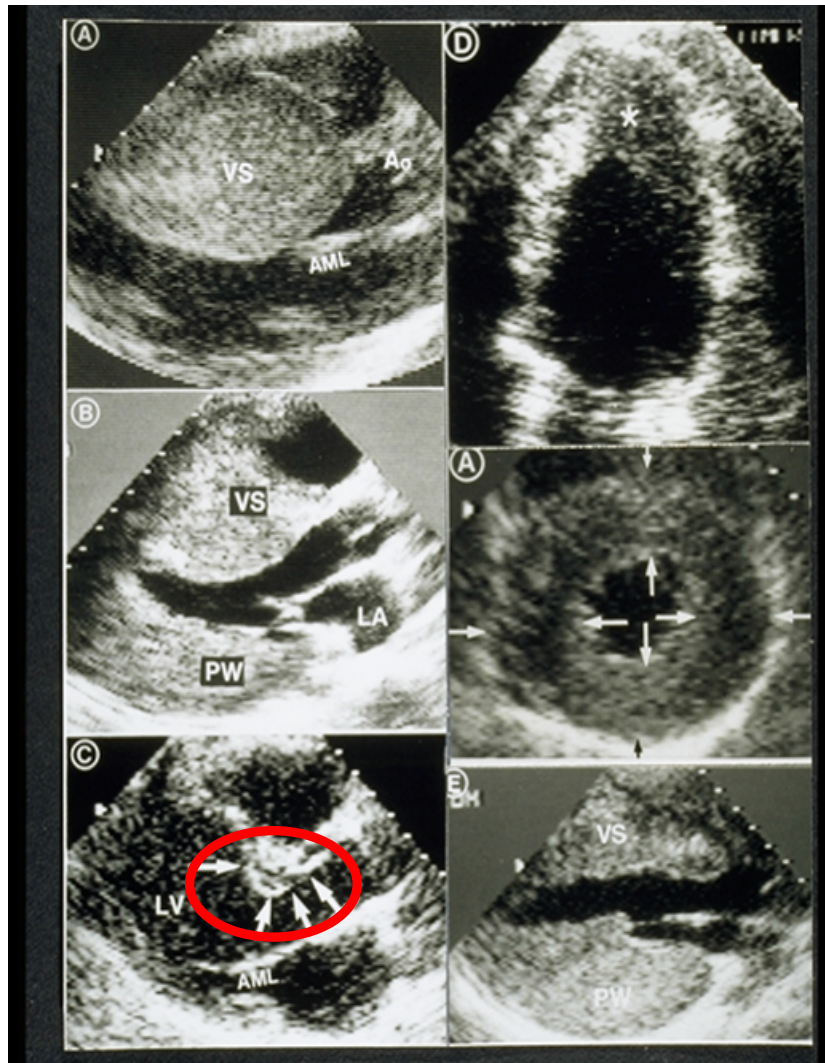
Cut off value LV wall thickness ≥ 15 mm in any LV segment that is not explained solely by loading conditions (ESC 2023)

→ Prevalence in Adults **1:500** (2 per ‰)

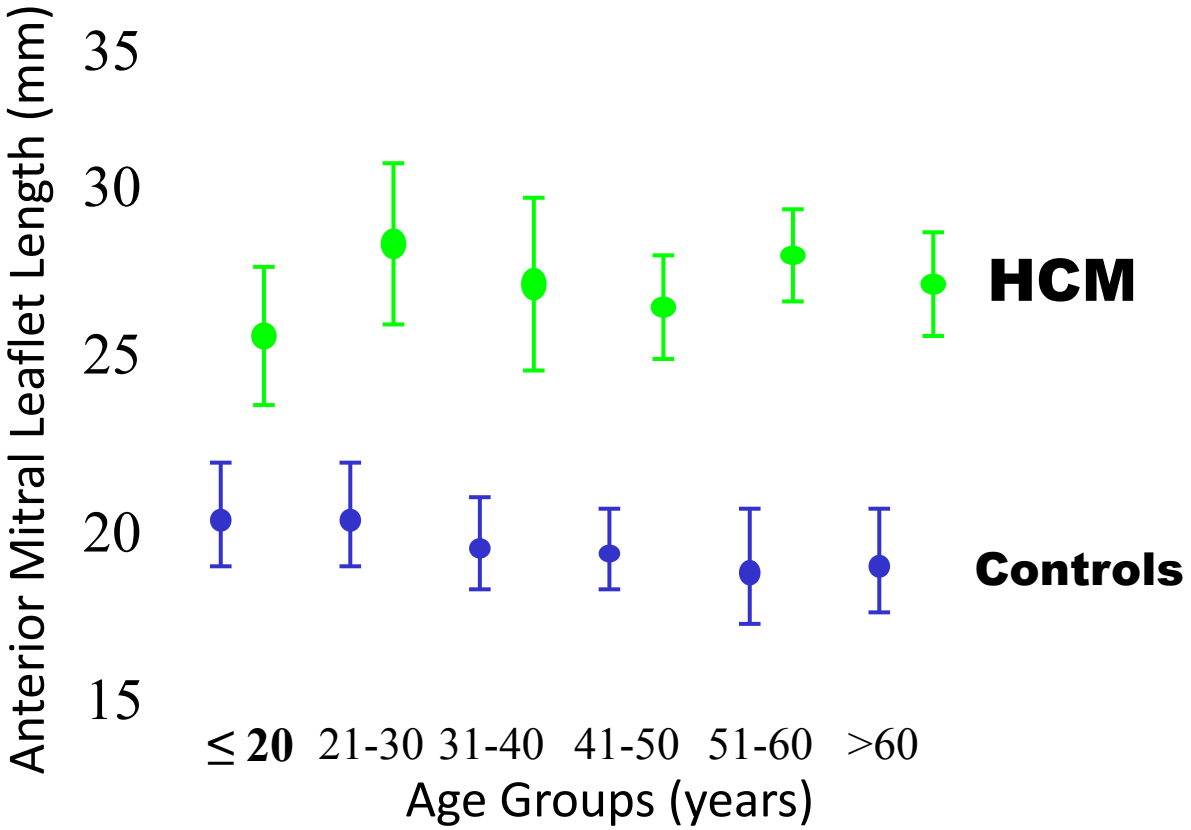
Family members ≥13 mm → Prevalence in Adults **1:200** (5 per ‰)

Heterogeneous

Variable (Dynamic)

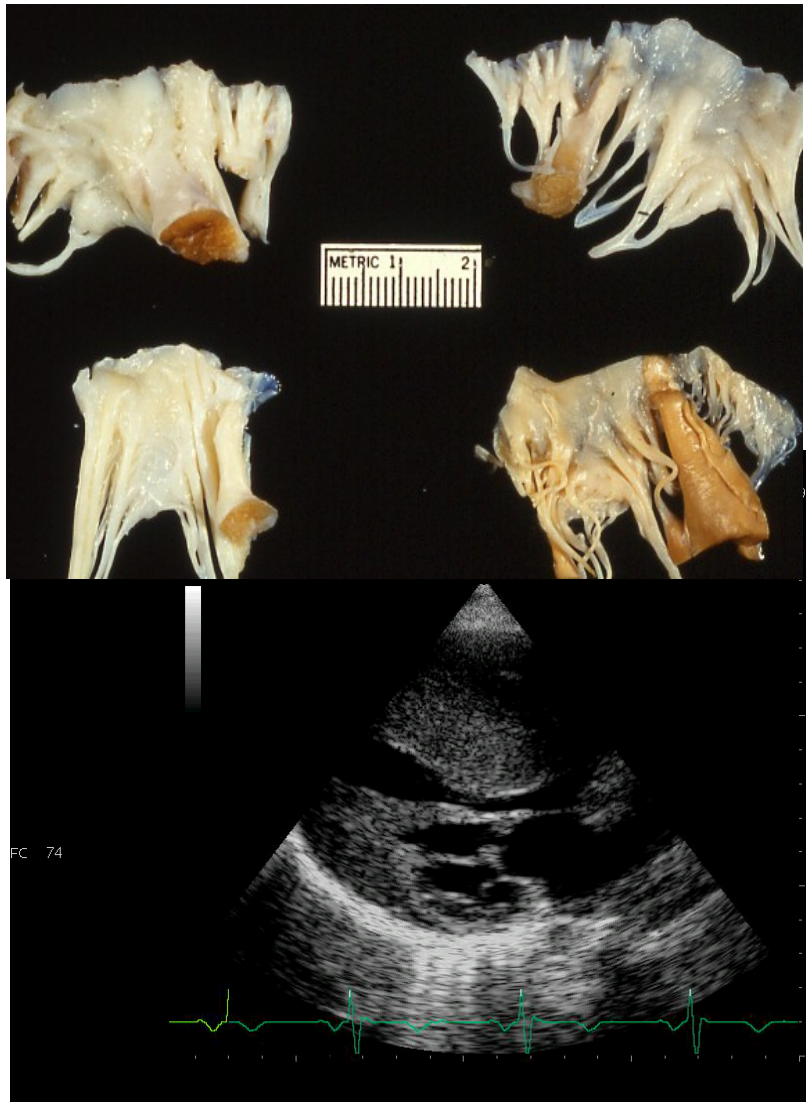


Aumentata lunghezza del lembo anteriore mitralico per classi di età



Maron MS et al. Circulation 2011

Attacco anomalo del muscolo papillare



L’ostruzione all’efflusso del ventricolo sinistro è una condizione VARIABILE , che può aggravarsi o migliorare a seconda di alcune condizioni :

- Pasti abbondanti (pasta, pizza, etc)

- Sforzi importanti
  - sollevamento pesi
  - esercizio fisico

- Farmaci: Diuretici, sartani

- **Disidratazione**

Hypertrophic Cardiomyopathy Is Predominantly a Disease of Left Ventricular Outflow Tract Obstruction

Martin S. Maron, MD; Iacopo Olivotto, MD; Andrey G. Zenovich, MSc; Mark S. Link, MD; Natesa G. Pandian, MD; Jeffery T. Kuvin, MD; Stefano Nistri, MD; Franco Cecchi, MD; James E. Udelson, MD; Barry J. Maron, MD

Circulation 2006

CMI ostruttiva provocabile (latente):

1. manovra di Valsalva
2. dopo pasto carboidrati
3. Eco da sforzo su pedana o cicloergometro

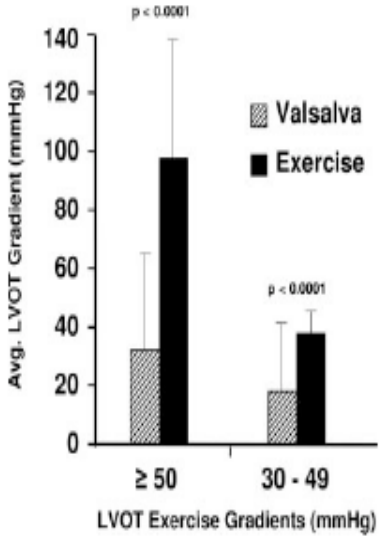
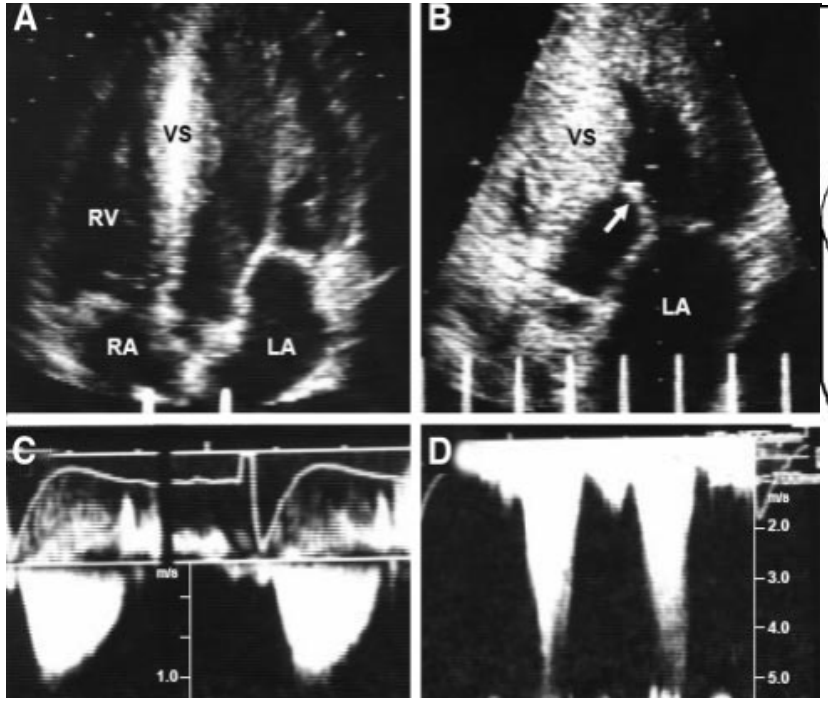
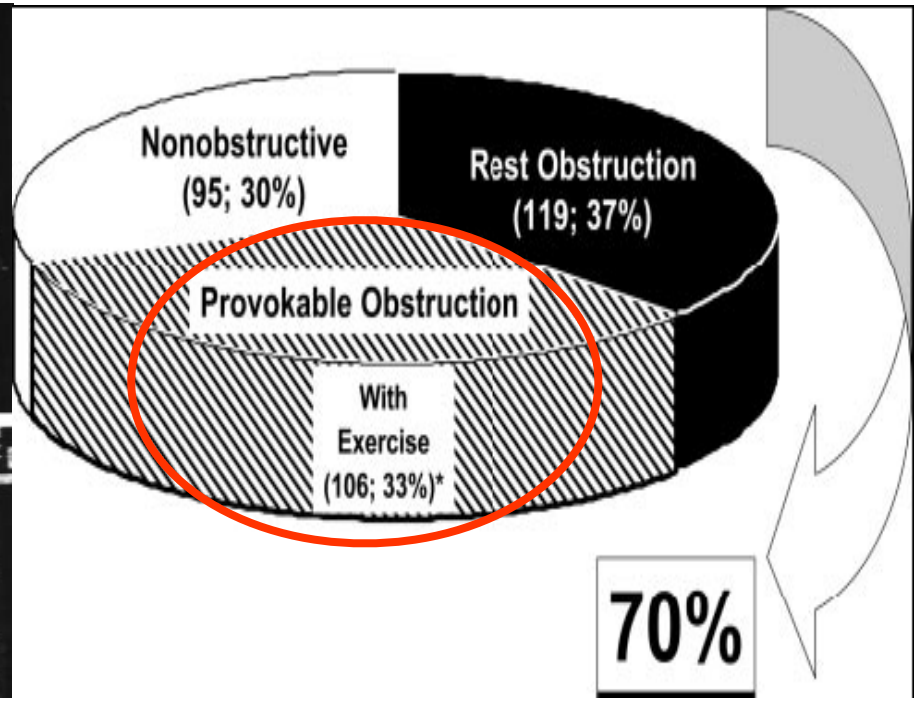


Figure 4. Comparison between LV outflow tract (LVOT) gradients (≥30 mm Hg) provoked with exercise and with Valsalva maneuver. Valsalva significantly underestimates gradients vs those provoked with exercise. Avg indicates average.





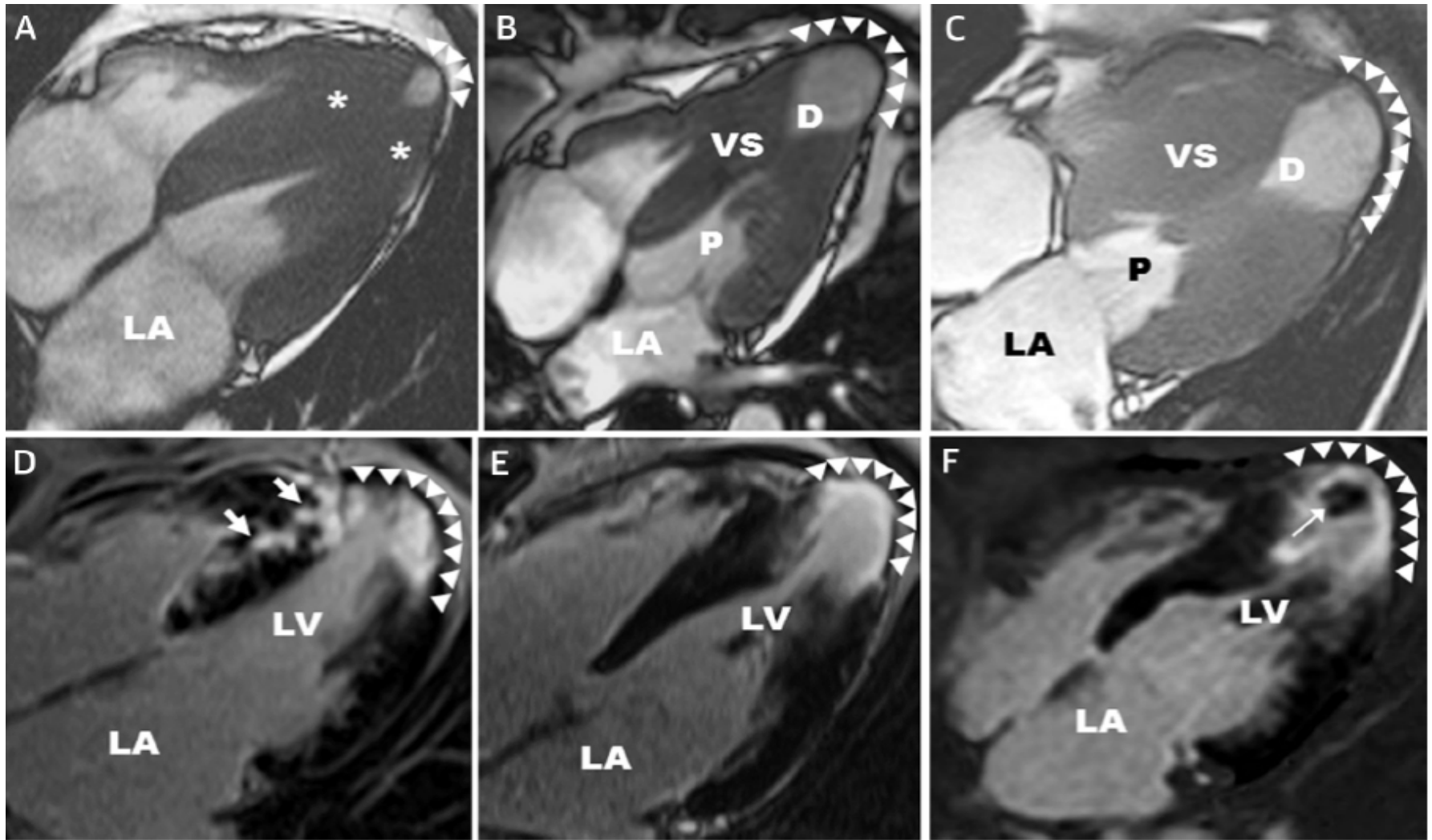
Hypertrophic Cardiomyopathy With Left Ventricular Apical Aneurysm

Implications for Risk Stratification and Management

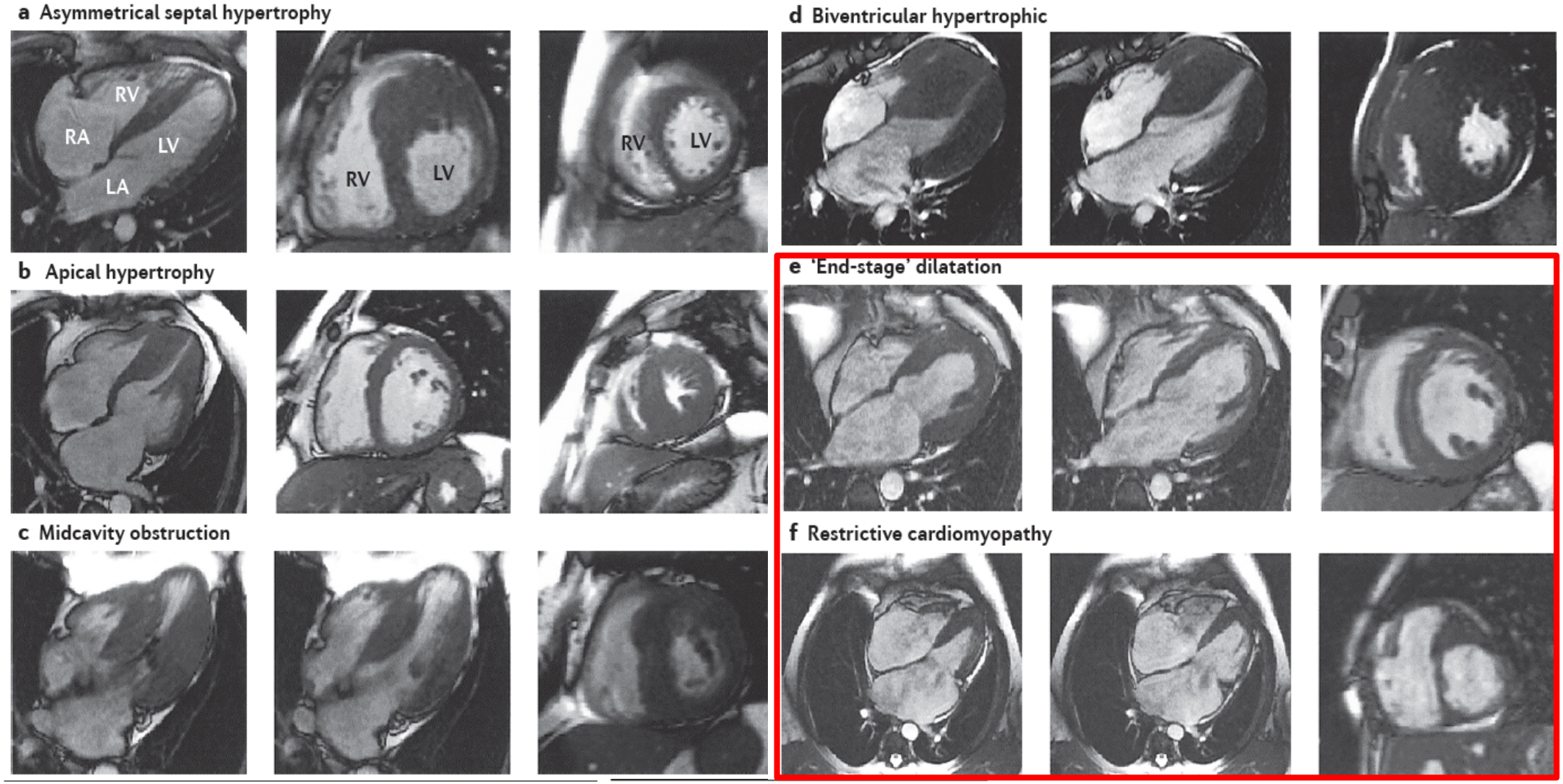
Ethan J. Rowin, MD,[a] Barry J. Maron, MD,[a] Tammy S. Haas, RN,[b] Ross F. Garberich, MS,[b] Weijia Wang, MD,[a] Mark S. Link, MD,[a] Martin S. Maron, MD[a]

JACC 2017

- **Trombosi apicale**
- **TV sostenuta monomorfa**



Heterogeneous Variable (Dynamic)



Srijita Sen-Chowdhry[1,2], Daniel Jacoby[3], James C. Moon[1,4] and William J. McKenna[5]

NATURE REVIEWS | CARDIOLOGY 2016

HCM

PATIENTS JOURNEY

Patterns of Disease Progression in Hypertrophic Cardiomyopathy

An Individualized Approach to Clinical Staging

Iacopo Olivotto, MD; Franco Cecchi, MD; Corrado Poggesi, MD; Magdi H. Yacoub, MD, FRS

Circ Heart Fail. 2012;5:535-546

STAGE II

"Classic" Phenotype

Prevalence ~75%

LVEF >65%

~20-25% Basal LVOTO± MR

~20-25% Provokable LVOTO± MR

~50% Symptoms

No or mild Fibrosis (LGE)

Stable

Development of LV Hypertrophy

STAGE III

Adverse Remodeling

Prevalence ~15%

LVEF 50%-65%

~20-25% Basal LVOTO ± MR

~20-25% Provokable LVOTO ± MR

~100% Symptoms

Moderate-severe Fibrosis (LGE)

Non-Hypertrophic

No Symptoms

ECG may be abnormal

STAGE I (gene +)

STAGE IV

Overt Dysfunction

Prevalence ~5-10%

LVEF <50%

NO LVOTO ~

100% MR

Severe symptoms

Severe Fibrosis (LGE)

HCM PHENOTYPE

Sarcomeric versus non sarcomeric disease

Red flag approach

2014 ESC Guidelines on diagnosis and management of hypertrophic cardiomyopathy

The Task Force for the Diagnosis and Management of Hypertrophic Cardiomyopathy of the European Society of Cardiology (ESC)

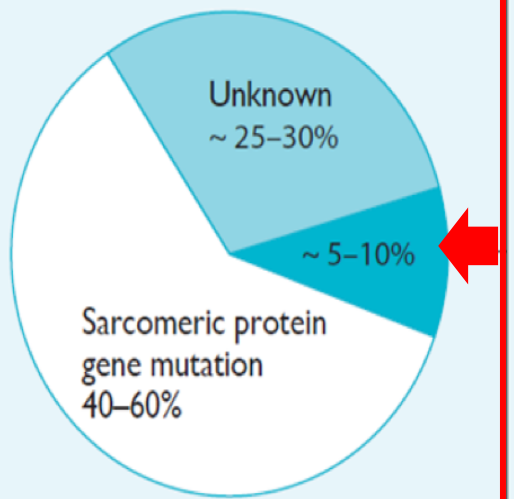
European Heart Journal
doi:10.1093/eurheartj/ehu284

Rare Variants (10%)
MYL2,ALPK3,et al

MYL3
TPM1
TNNI3
TNNT2
MYH7
MYBPC3

Poligenic + environmental

- Obesity
- Hypertension (youngs)
- Drugs
- Intense sport activity



The majority of cases in adolescents and adults are caused by mutations in sarcomere protein genes.

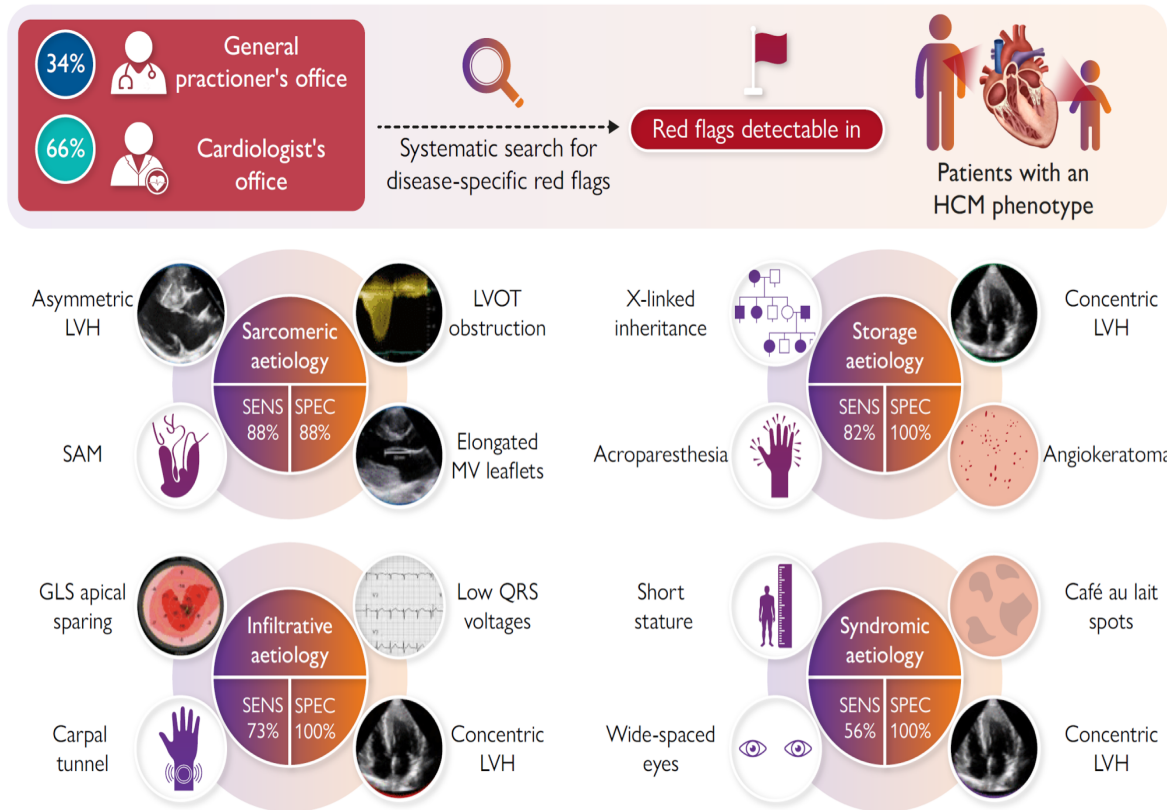
Other genetic or non genetic causes

- **Inborn errors of metabolism**
  - Glycogen storage diseases:
    - Pompe
    - Danon
  - AMP-Kinase (PRKAG2)
  - Carnitine disorders
  - Lysosomal storage diseases
    - Anderson-Fabry
- **Neuromuscular diseases**
  - Friedreich's ataxia
  - FHL1
- **Mitochondrial diseases**
  - MELAS
  - MERFF
- **Malformation Syndromes**
  - Noonan
  - LEOPARD
  - Costello
  - CFC
- **Amyloidosis**
  - Familial ATTR
  - Wild type TTR (senile)
  - AL amyloidosis
- **Newborn of diabetic mother**
- **Drug-induced**
  - Tacrolimus
  - Hydroxychloroquine
  - Steroids

Hypertrophic cardiomyopathy: prevalence of disease-specific red flags

Niccolò Maurizi[1,2,*], Emanuele Monda[3], Elena Biagini[4], Ella Field[5,6], et al.

European Heart Journal (2025)



HCM

TREATMENT OPTIONS

Pharmacological treatment of hypertrophic cardiomyopathy: current practice and novel perspectives

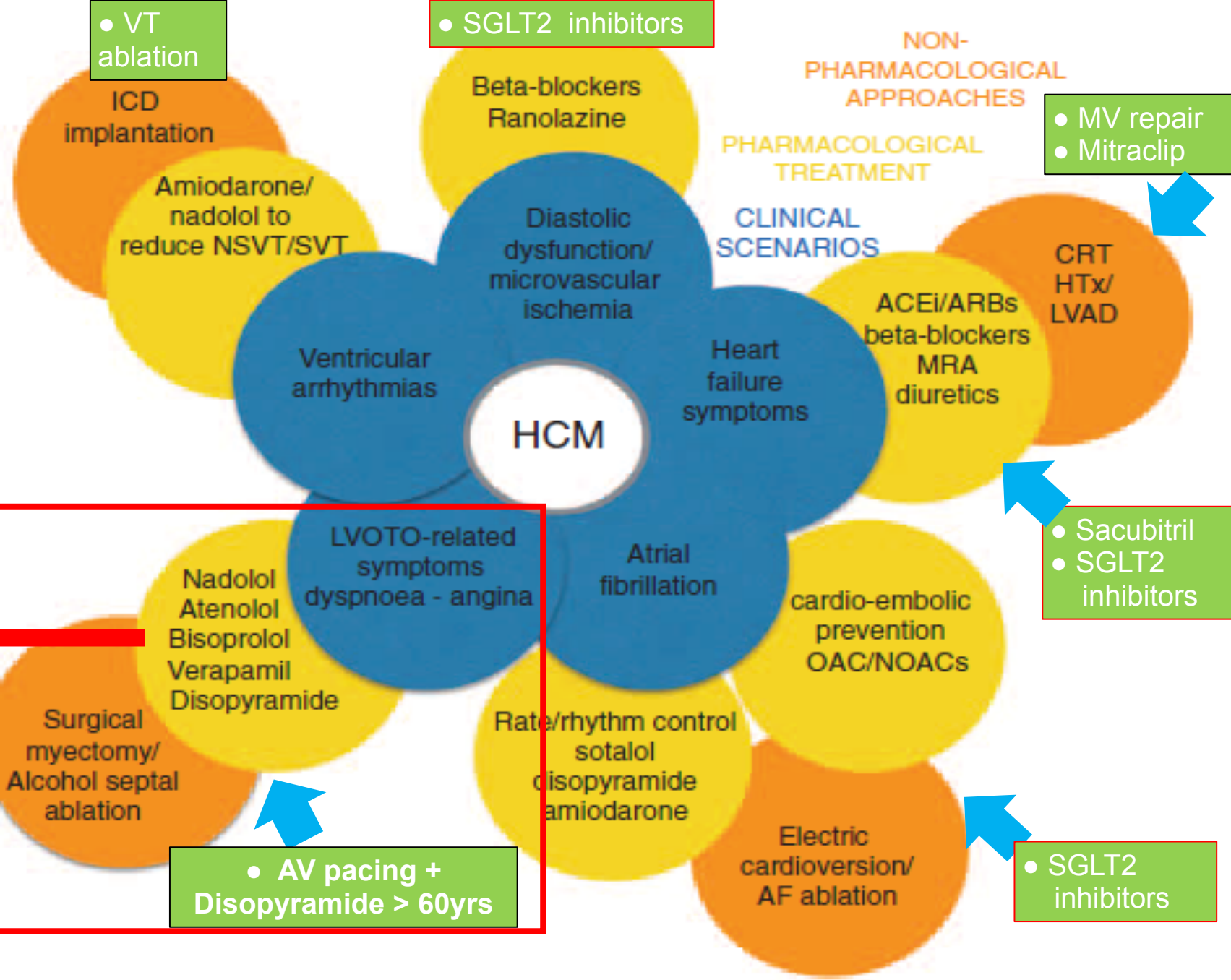
Enrico Ammirati[1]*, Rachele Contri[2], Raffaele Coppini[3], Franco Cecchi[3], Maria Frigerio[1], and Iacopo Olivotto[3]*

European Journal of Heart Failure (2016)

Therapeutic armamentarium in 2026

Myosin Inhibitors
Mavacamten
Aficamten

Nadolol
Disopyramide
or
Nadolol &
Disopyramide



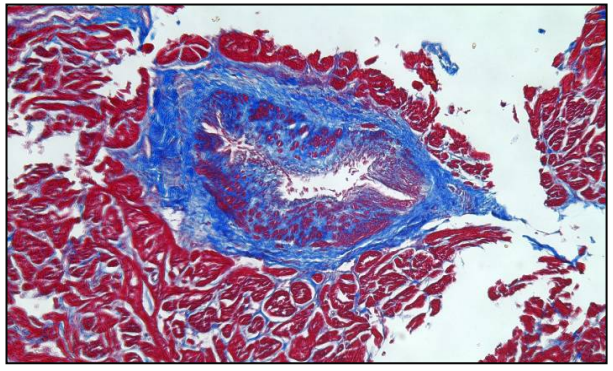
HCM: WHAT ARE WE STILL MISSING ?

HCM: WHAT ARE WE STILL MISSING ? NEXT STEPS

ORIGINAL ARTICLE

Coronary Microvascular Dysfunction and Prognosis in Hypertrophic Cardiomyopathy

Franco Cecchi, M.D., Iacopo Olivotto, M.D., Roberto Gistri, M.D., Roberto Lorenzoni, M.D., Giampaolo Chiriatti, M.D., and Paolo G. Camici, M.D.



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Relevance of Coronary Microvascular Flow Impairment to Long-Term Remodeling and Systolic Dysfunction in Hypertrophic Cardiomyopathy

Iacopo Olivotto, MD,* Franco Cecchi, MD,* Roberto Gistri, MD,* Roberto Lorenzoni, MD,† Giampaolo Chiriatti, MD,‡ Francesca Girolami, BSc,§ Francesca Torricelli, BSc,§ Paolo G. Camici, MD, FACC, FRCP‖

Florence, Lucca, Pescia, and Pisa, Italy; and London, England

- WHY CARDIAC ARREST OCCUR
- HOW TO BETTER IDENTIFY HIGH RISK PTS
- ANY OPTION BEYOND ICD ?
- HOW TO PREVENT MYOCARDIAL FIBROSIS ?
- THE ROLE OF
  - GENE VARIANTS (e.g. VOUS)
  - SNP
  - ENVIRONMENT
- THE ROLE OF GENE THERAPY

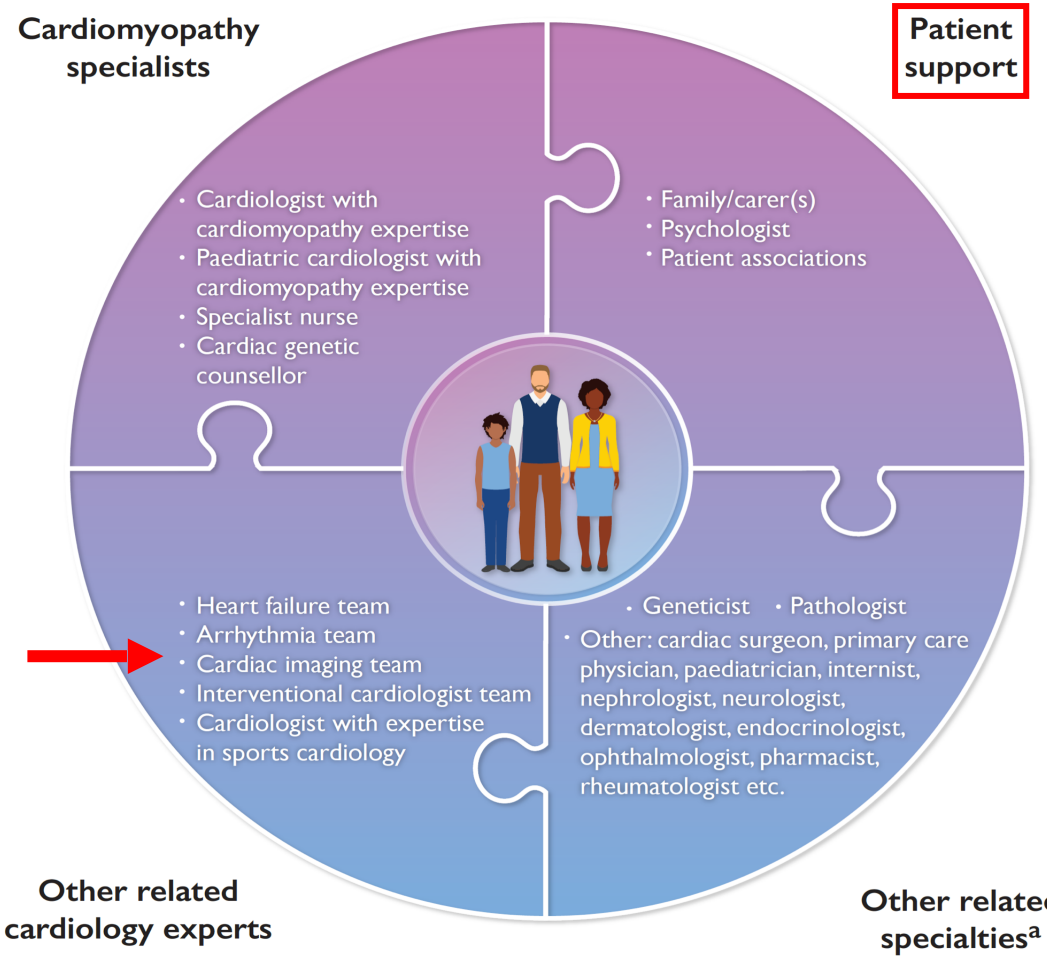
2023 ESC Guidelines for the management of cardiomyopathies

Developed by the task force on the management of cardiomyopathies of the European Society of Cardiology (ESC)

Authors/Task Force Members: Elena Arbelo [*†], (Chairperson) (Spain), Alexandros Protonotarios [†], (Task Force Co-ordinator) (United Kingdom), Juan R. Gimeno [‡], (Task Force Co-ordinator) (Spain)

European Heart Journal (2023) **00**, 1–124

MULTIDISCIPLINARY HCM CENTERS



AICARM ASSOCIAZIONE ITALIANA CARDIOMIOPATIE

Aicarm.it

AICARM APS "Servizio «Cuori in ascolto"

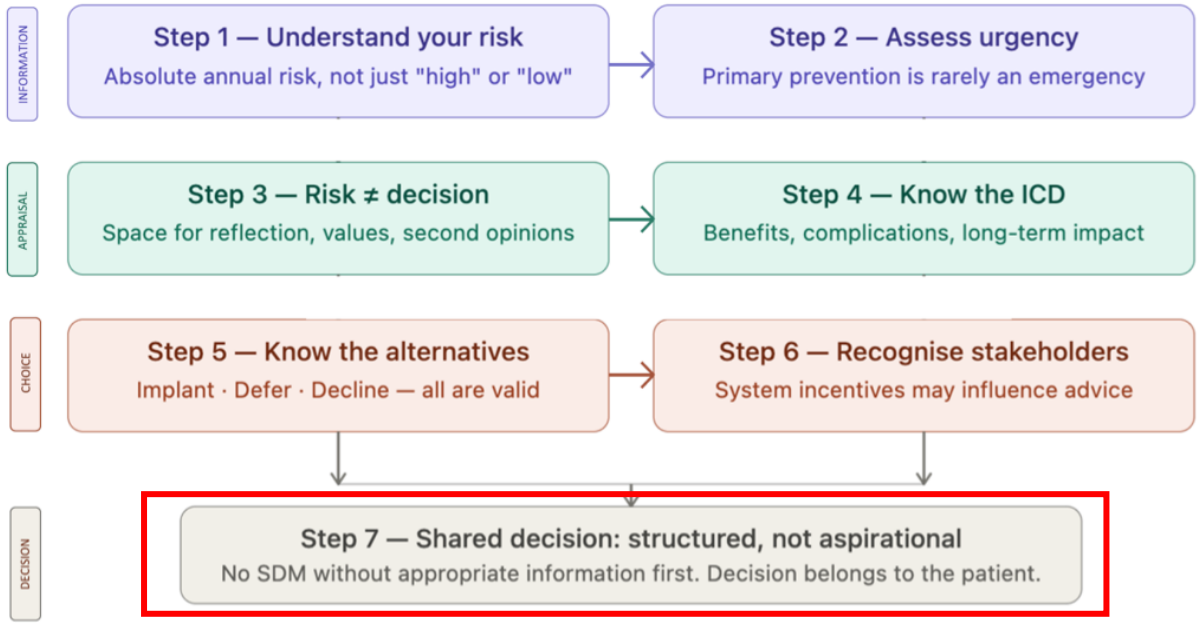
Chiama

cuori in ascolto

055 06 20 178

- **Dal 2022, oltre 650 pazienti o familiari hanno utilizzato il servizio di sostegno ai pazienti con Cardiomiopatia, prestato telefonicamente da volontari esperti**
- **Gli utenti di Cuori in Ascolto possono ottenere sostegno psicologico e confrontarsi con altri pazienti in merito alle proprie esperienze di malattia e percorso terapeutico**

A



Global Spotlights European Heart Journal (2026) **00**, 1–3
https://doi.org/10.1093/eurheartj/ehag412

Assessing the need for cardioverter defibrillator implantation in hypertrophic cardiomyopathy: patient association's perspectives

Niccolò Maurizi [1,2,3,*], Iacopo Olivotto[1,4,5], and Franco Cecchi[3]; on behalf of AICARM-HCM Patient Foundation

B

The communication gap: what physicians say, what patients hear, what should be said

Physician framing	Patient perception	How to convey it
"High / intermediate / low risk" (relative to population)	"This could happen to me at any time"	Give explicit absolute annual risk e.g. "<1% per year" + visual aid
"Life-saving therapy" (ICD benefit framing)	"Complete protection from death"	Clarify risk reduction, not risk elimination
Complications "listed as rare events"	Underestimated / not fully internalised	Discuss lived experience, cumulative long-term burden
Urgency implied / prompt action needed	"This is an emergency — I must decide now"	Distinguish urgent (2° prevention) vs elective (1° prevention)
"ICD vs no ICD" (binary framing)	Fear of choosing "no protection"	Explicitly present 3 options: implant · defer · decline
Focus on short-term prevention	Lifelong consequences not fully considered	Frame decision over decades, especially in the young
Patient values as secondary consideration	Values unclear / not elicited	Elicit preferences early; document them explicitly

aicarm

GRAZIE !

prof.francocecchi@gmail.com www.cecchifranco.it