University of Torino
Department of Medical Sciences
Division of Internal Medicine
Director: Prof. Franco Veglio

La cardiomiopatia ipertrofica

Simona Votta

GIOVANNI, 80 ANNI

In visita per scarso controllo pressorio

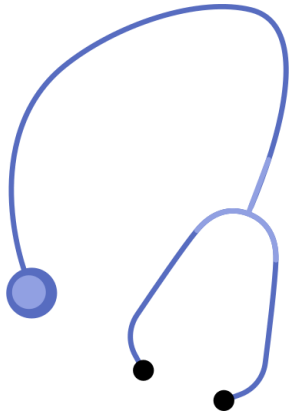
Anamnesi patologica remota: ipertensione arteriosa nota da qualche mese, artrite psoriasica trattata in passato con metotrexate, ipertrofia vescicale

Vita attiva, pratica ciclismo.

Allergie: non note

Terapia domiciliare: ramipril 5 mg

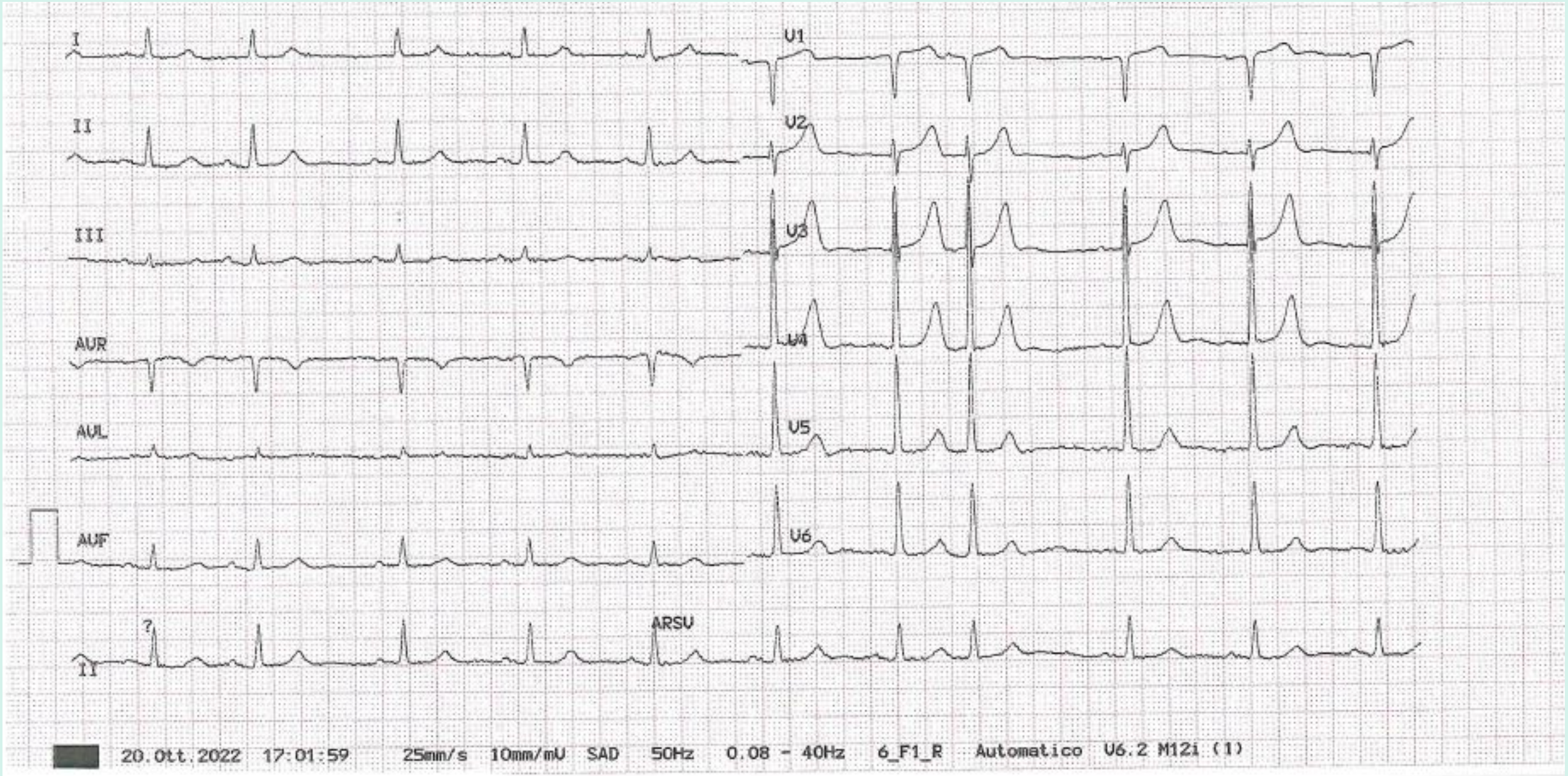
GIOVANNI, 80 ANNI



PAO 170/90 mmHg (grado 2), FC 70 bpm ritmici,

SpO2 96% in aria ambiente.

Esame obiettivo di norma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V2
V3
V4
V5
V6
?
ARSU
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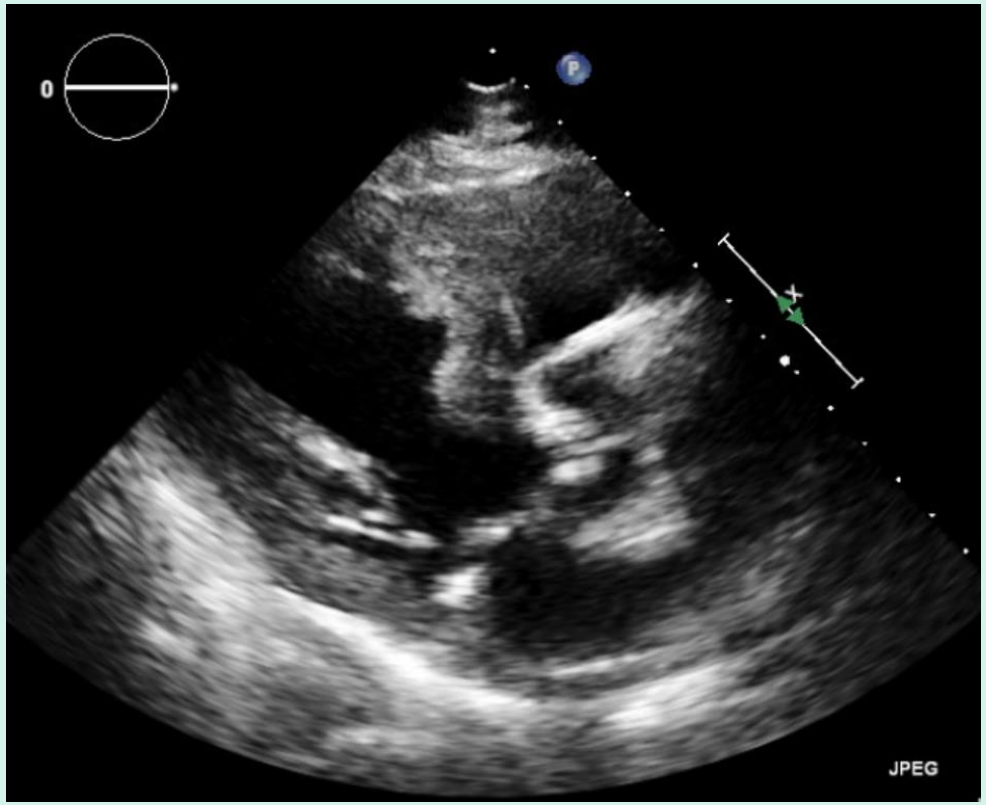
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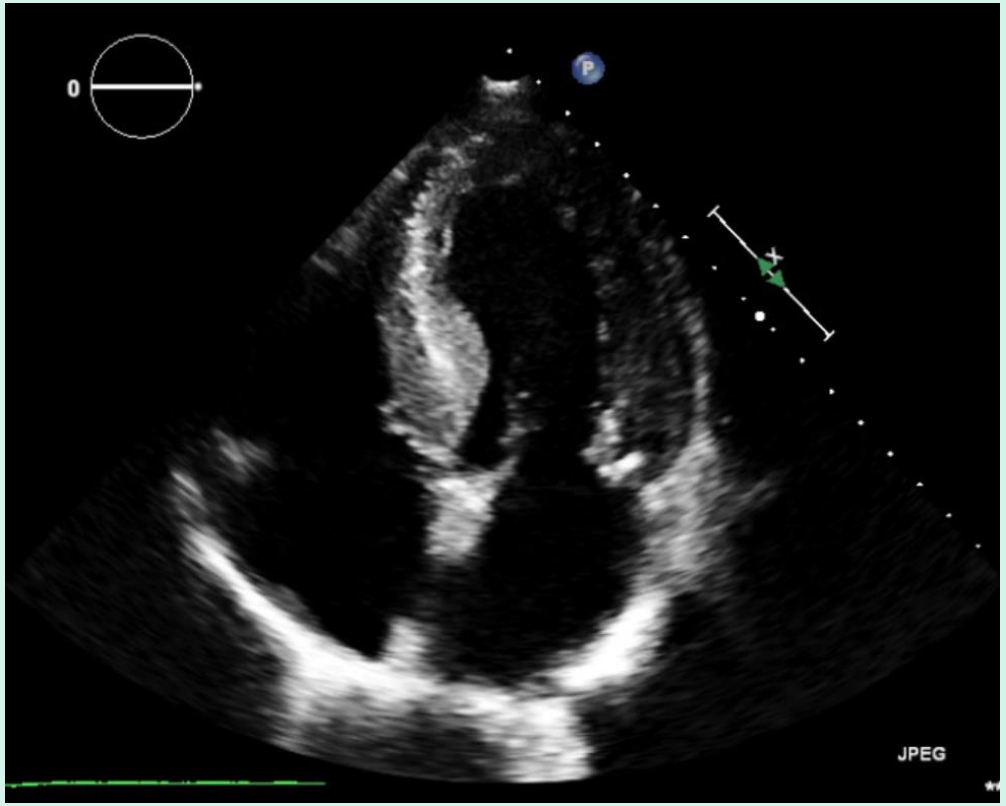
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Conclusioni ecocardiografia:

Ventricolo sinistro: ipertrofia concentrica (**LVMi 173 g/m2**). Normale funzione sistolica.(Ef=69 %). edvol=74 ml, esvol=23 ml, mv e/e'(avg)=13,7, mv e/e' (lat)=17,6.

Ventricolo destro: normali dimensioni, morfologia e funzione. S'=12,5 cm/s, TAPSE=22 mm

Setto IV: 17 mm.

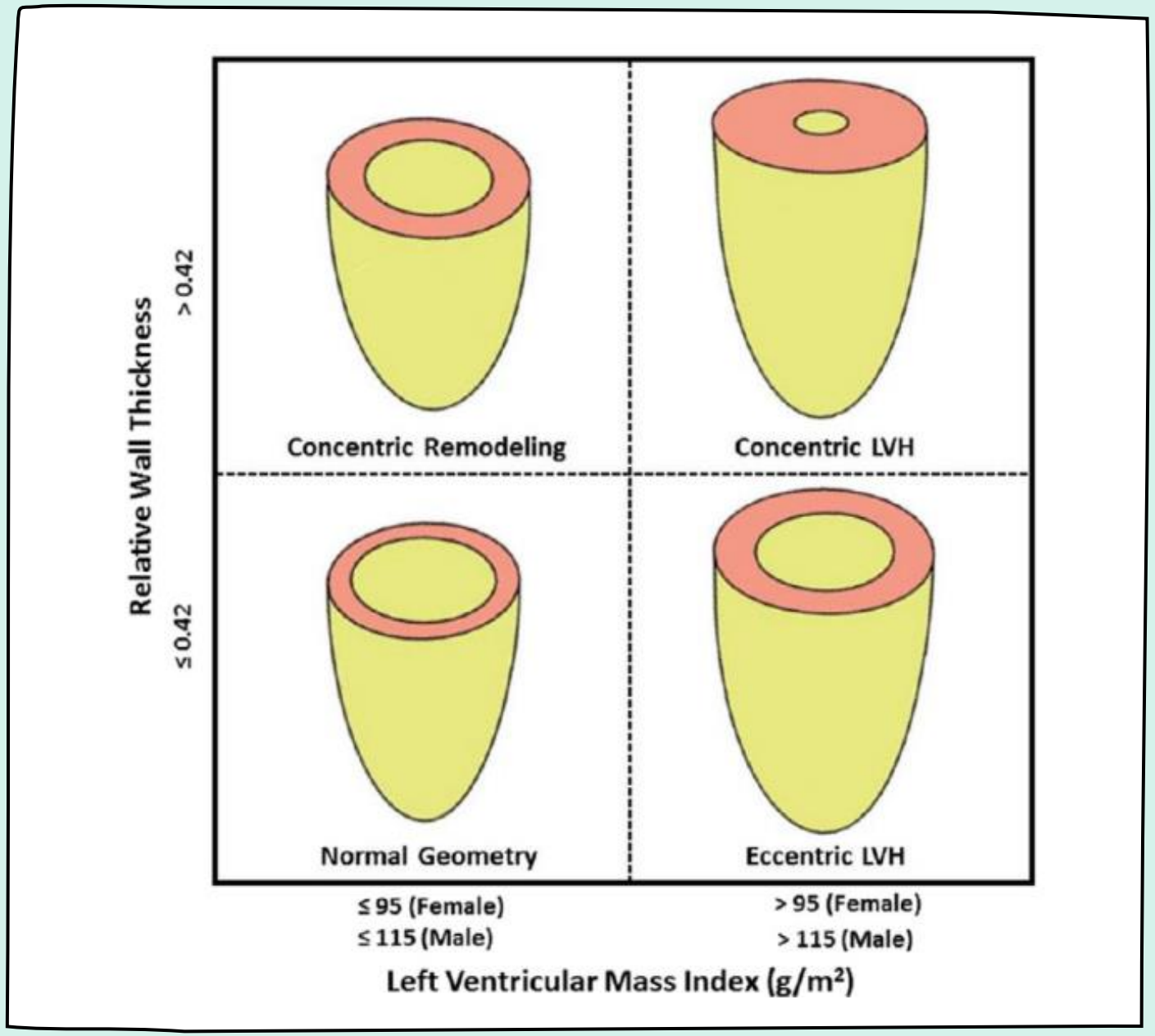
Atrio sinistro: moderatamente dilatato. Atrio destro: normali dimensioni e morfologia.

Valvola mitrale: lembi sottili che presentano normale escursione sisto-diastolica.

Valvola aortica: diffusamente ispessita, stenosi lieve e insufficienza lieve pgrad=28,0 mm hg.

Valvola tricuspide: insufficienza lieve paps=36 mm hg.

Pericardio: assenza di ispessimento o versamento pericardico.



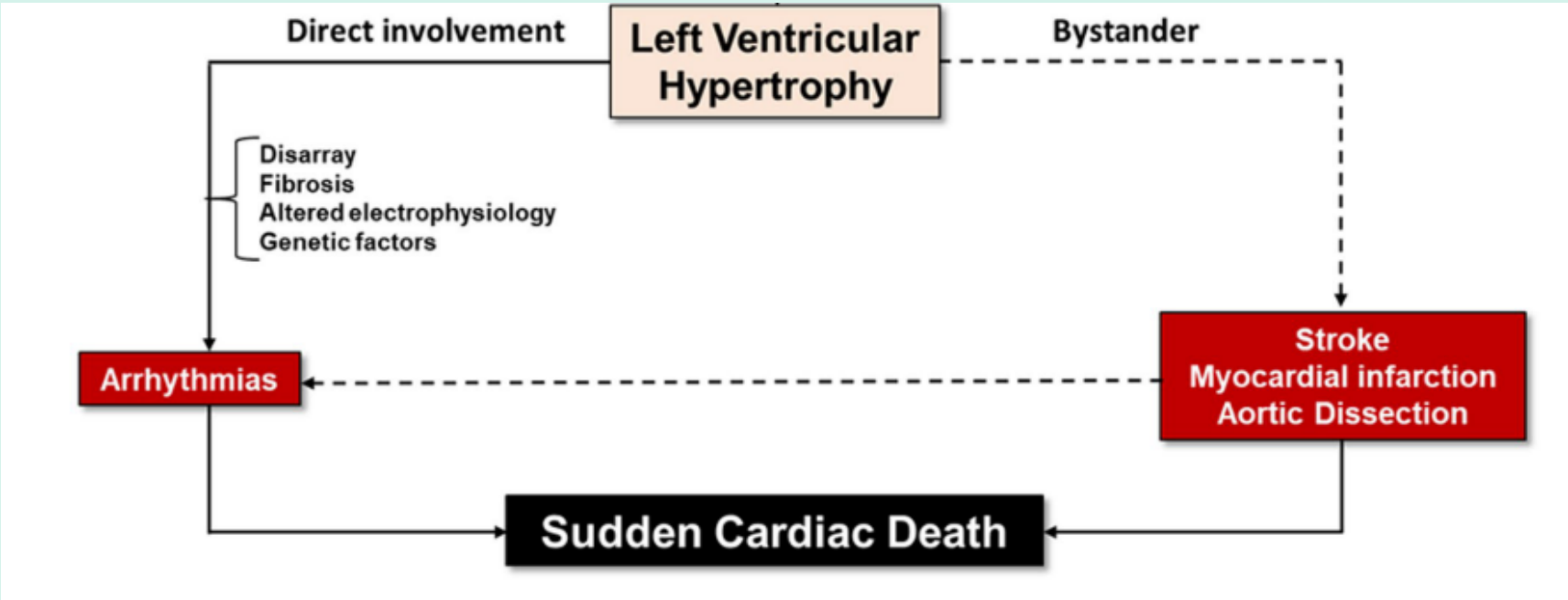
Ipertrofia ventricolare sinistra:

massa del ventricolo sinistro (indicizzato per BSA)

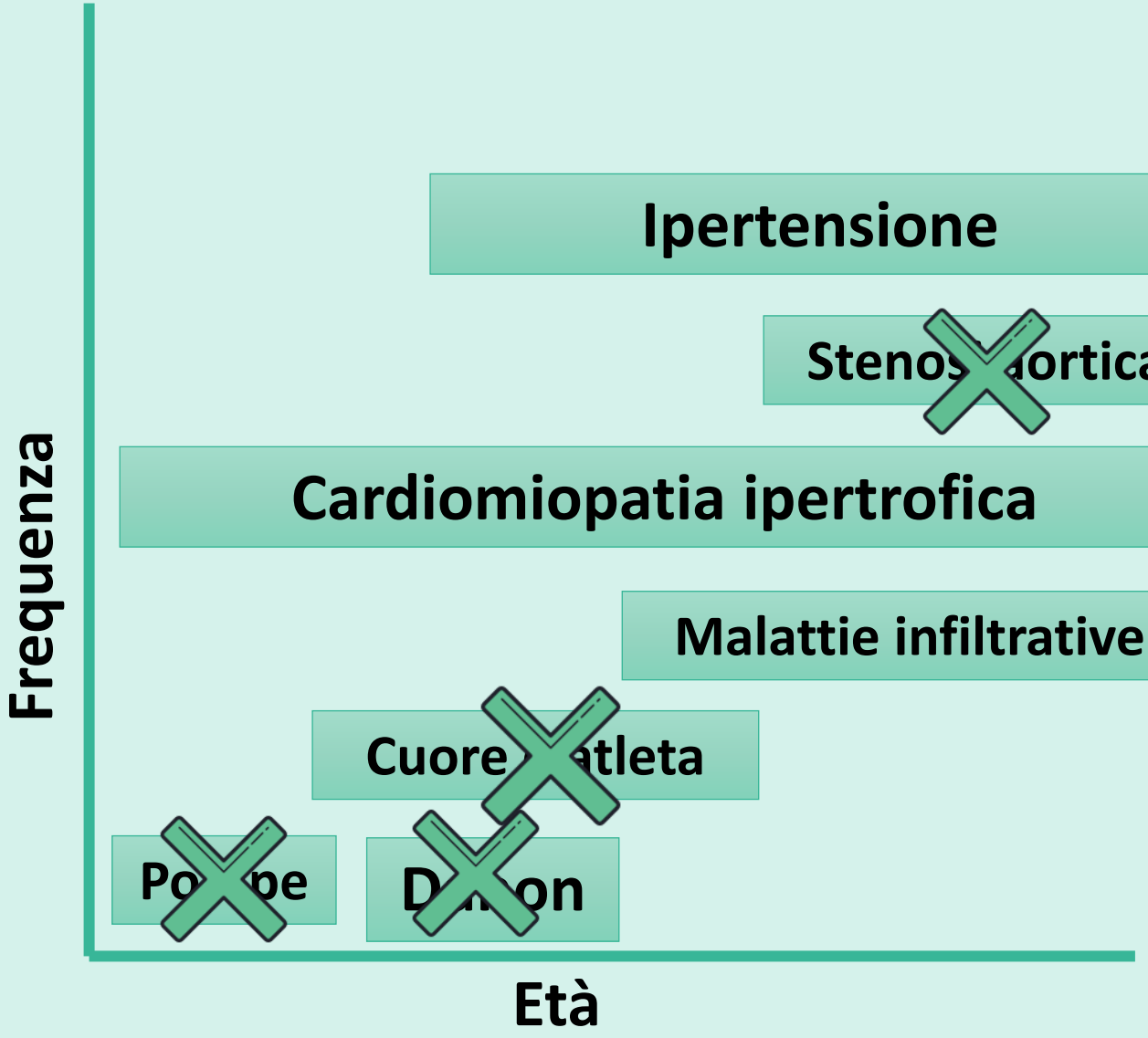
> 115 g/m2 nel genere maschile

> 95 g/m2 in quello femminile

Mehmet Yildiz a et al. 2020



Giamouzis et al, 2022



L'ipertensione frequentemente si riscontra nei pazienti adulti con HCM, con una prevalenza tra il 35 e il 50%, fino al 70% nei soggetti con età > 75 anni.

J. Geske et al., 2018
Alashi et al, 2023

ECG

-SV1 + RV5 or RV6 (Sokolow-Lyon) >35 mm,

- R wave in aVL ≥11 mm,

-SV3 + RaVL (Cornell voltage) > 28 mm for men and > 20 mm for women

ECOCARDIOGRAFIA

RISONANZA MAGNETICA

DIAGNOSI LVH

ECG

ECOCARDIOGRAFIA TRANSTORACICA

- Funzionalità sistolica e diastolica
- GLS (global longitudinal strain)
- Valutazione apparati valvolari

RISONANZA MAGNETICA

DIAGNOSI LVH

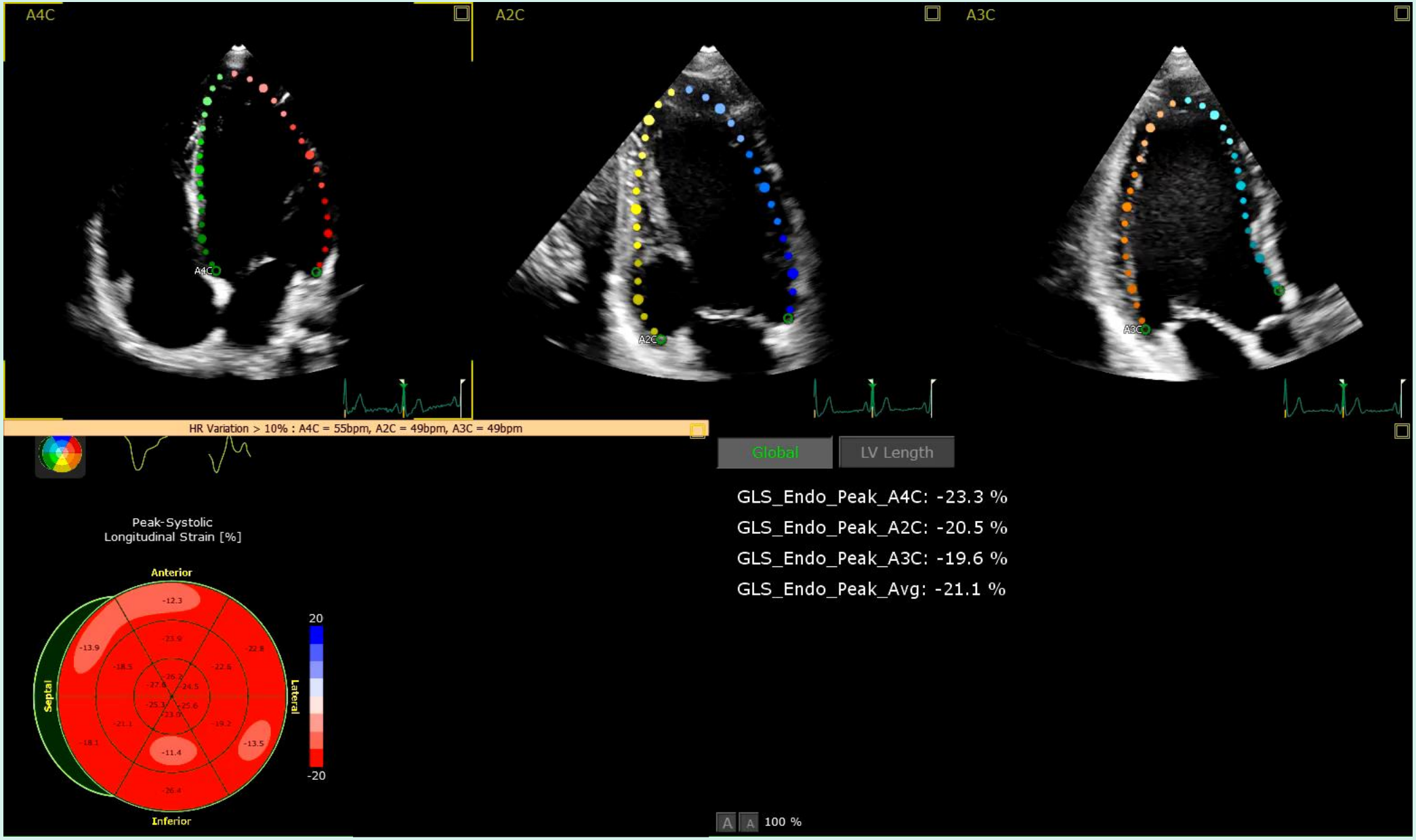
ECG

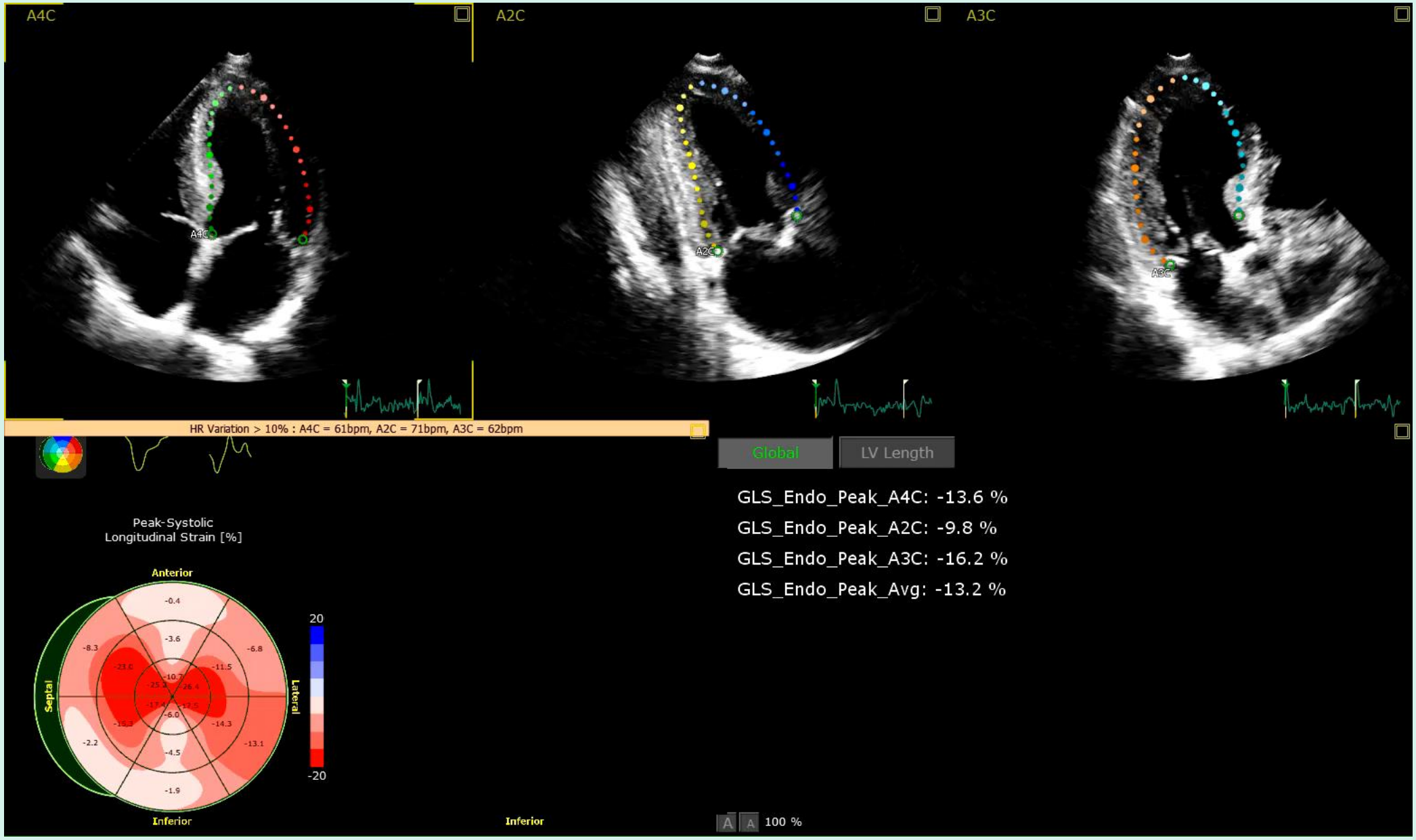
ECOCARDIOGRAFIA TRANSTORACICA

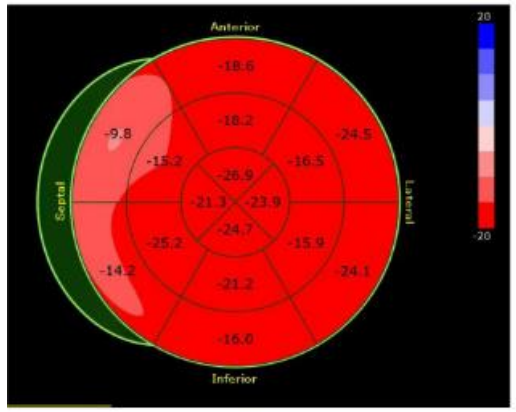
- Funzionalità sistolica e diastolica
- **GLS (global longitudinal strain)**
- Valutazione apparati valvolari

RISONANZA MAGNETICA

DIAGNOSI LVH

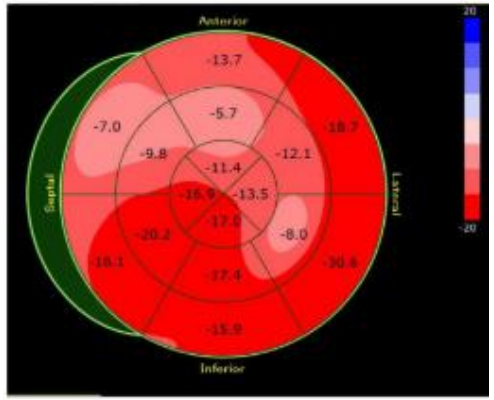

A4C
A2C
A3C
HR Variation > 10% : A4C = 55bpm, A2C = 49bpm, A3C = 49bpm
Global
LV Length
GLS_Endo_Peak_A4C: -23.3 %
GLS_Endo_Peak_A2C: -20.5 %
GLS_Endo_Peak_A3C: -19.6 %
GLS_Endo_Peak_Avg: -21.1 %
Peak-Systolic
Longitudinal Strain [%]
Anterior
Septal
Lateral
Inferior
20
-20
100 %


A4C
A2C
A3C
HR Variation > 10% : A4C = 61bpm, A2C = 71bpm, A3C = 62bpm
Global
LV Length
GLS_Endo_Peak_A4C: -13.6 %
GLS_Endo_Peak_A2C: -9.8 %
GLS_Endo_Peak_A3C: -16.2 %
GLS_Endo_Peak_Avg: -13.2 %
Peak-Systolic
Longitudinal Strain [%]
Anterior
Septal
Lateral
Inferior
20
-20
-0.4
-3.6
-8.3
-6.8
-11.5
-14.3
-2.2
-13.1
-4.5
-1.9
-6.0
Inferior
100 %



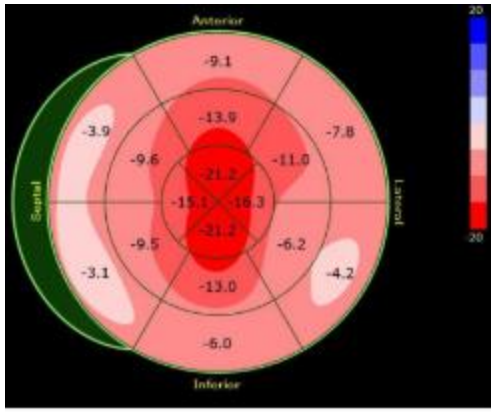
- GLS=19.8%

Ipertensione arteriosa: GLS normale o ridotto con interessamento del setto basale



- GLS=14.7%

Cardiomiopatia ipertrofica: GLS ridotto.
Ipertrofia specialmente nel setto interventricolare



- GLS=14.2%
- Relative apical longitudinal strain=1.21
- Septal apical-to-basal longitudinal strain=3.87

Amiloidosi cardiaca: GLS ridotto.
Apical sparing (variazione del GLS dalla base all'apice).

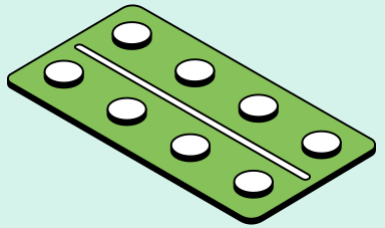
Type of LVH	GLS value	Typical impairment of longitudinal strain
Arterial hypertension	Normal or reduced	Basal septum
Hypertrophic cardiomyopathy	Reduced	The site of hypertrophy, especially in the region of the interventricular septum
Cardiac amyloidosis	Reduced	Regional variations in longitudinal strain from base-to-apex (Apical sparing)
Fabry disease	Reduced	Basal posterior-lateral wall
Aortic stenosis	Reduced	Basal LV segments
Athlete's heart	Almost normal	None
Friedreich's ataxia	Mildly reduced	None

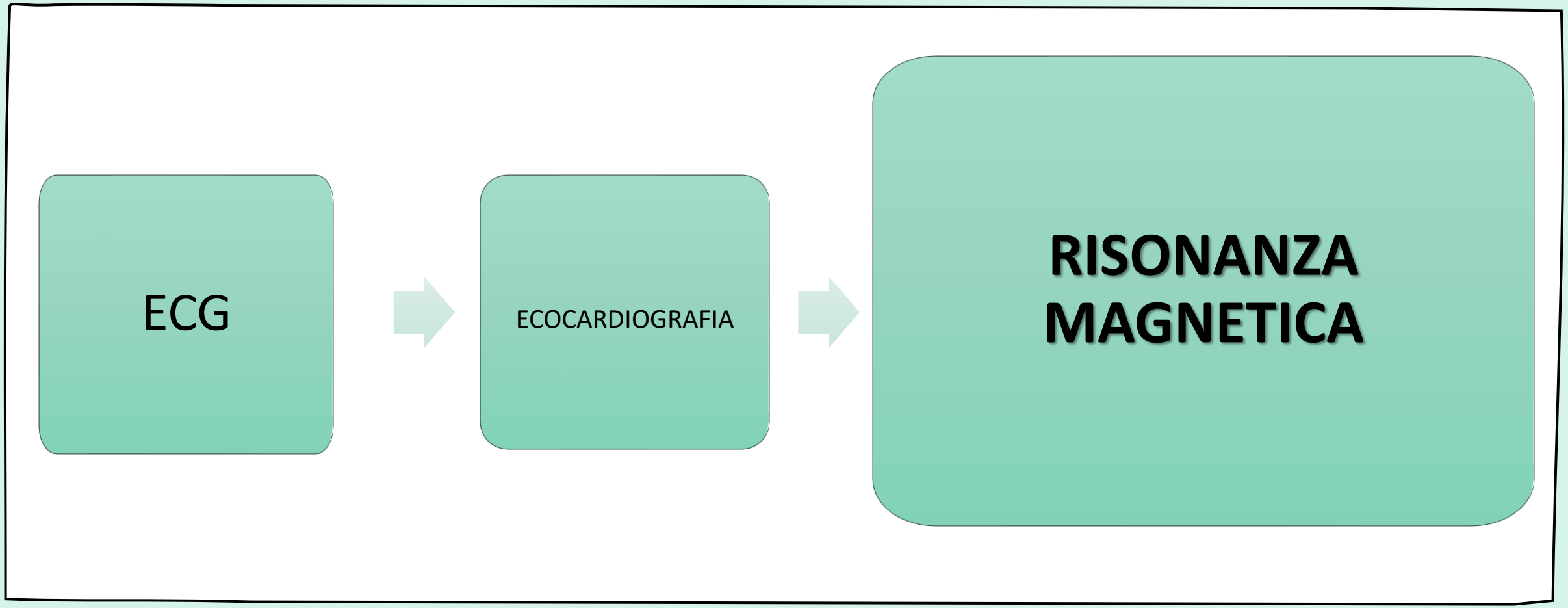
Tadic et al, 2022
Tanaka et al, 2021

Avviata disamina per ipertensione secondaria

Richiesta RM cardiaca

Terapia consigliata: telmisartan 40 mg e lercanidipina 10 mg





DIAGNOSI LVH

Torna in visita dopo 1 anno, valori pressori controllati con la terapia in atto

RM CARDIACA CON MDC: il ventricolo sinistro è ispessito a livello del setto basale e medio, ma normale frazione di eiezione, segni RM qualiquantitativi compatibili con cardiomiopatia ipertrofica non ostruttiva

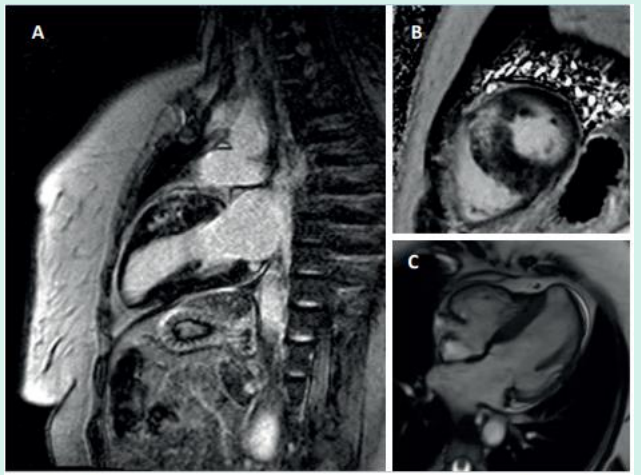


Figura 6. Cardiomiopatia ipertrofica. (*A,B*) Immagini tardive post-contrasto 2 camere e asse corto medio-ventricolare con evidenza di diffuso disomogeneo "late gadolinium enhancement" a carico dei segmenti maggiormente ipertrofici. (*C*) Immagine cine 4 camere di paziente con aneurisma apicale.

IN CONCLUSIONE: IPERTENSIONE ARTERIOSA ESSENZIALE, CARDIOMIOPATIA IPERTROFICA INDIRIZZATO IN UN CENTRO PER LE CARDIOMIOPATIE

LA CARDIOMIOPATIA IPERTROFICA

La cardiomiopatia ipertrfoica è una malattia autosomica dominante con penetranza variabile, associate a mutazioni nei geni che codificano per le proteine dei sarcomeri cardiaci

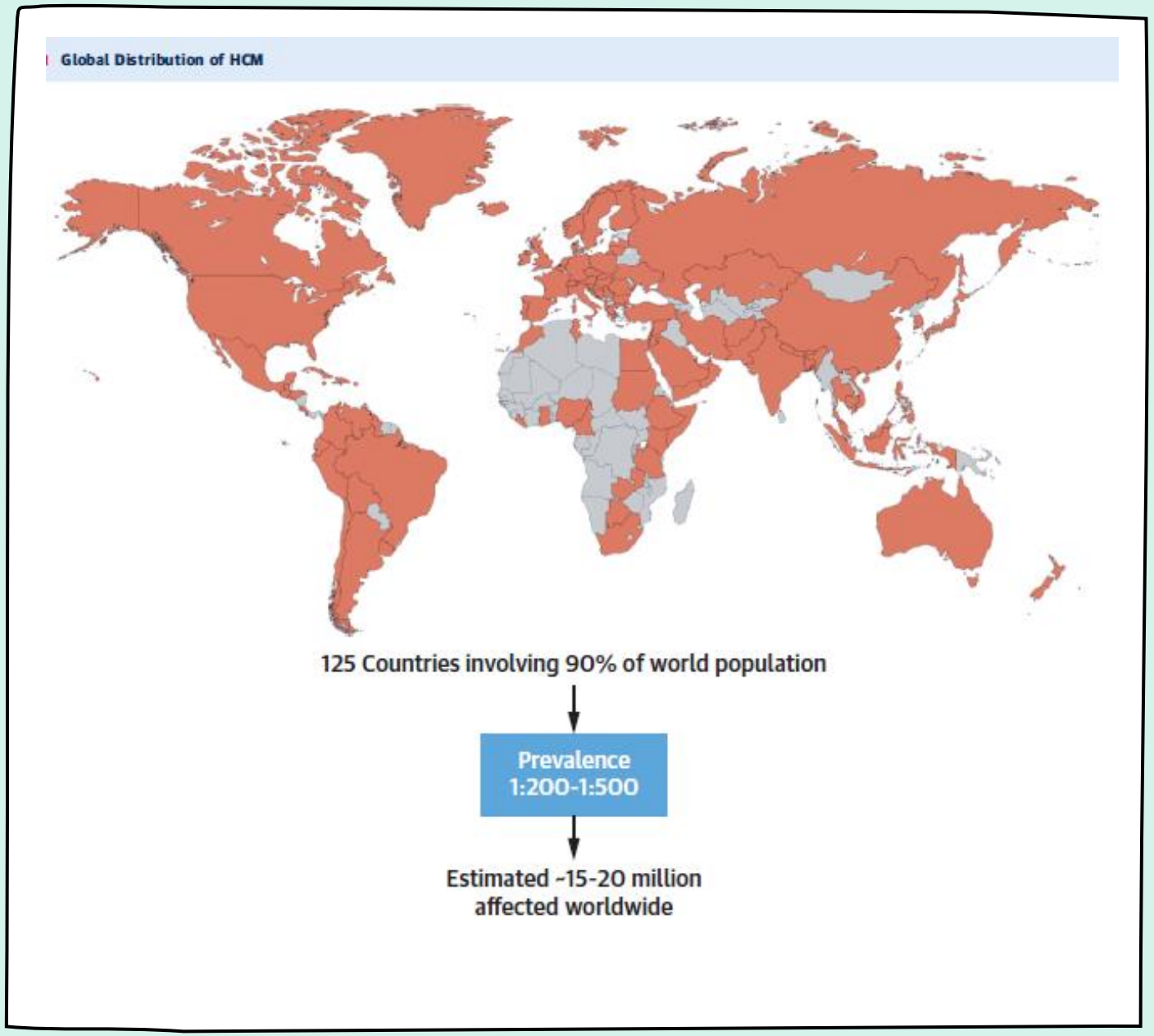


TABLE 1 Genetic and Molecular Basis of Disease for HCM and Phenocopies

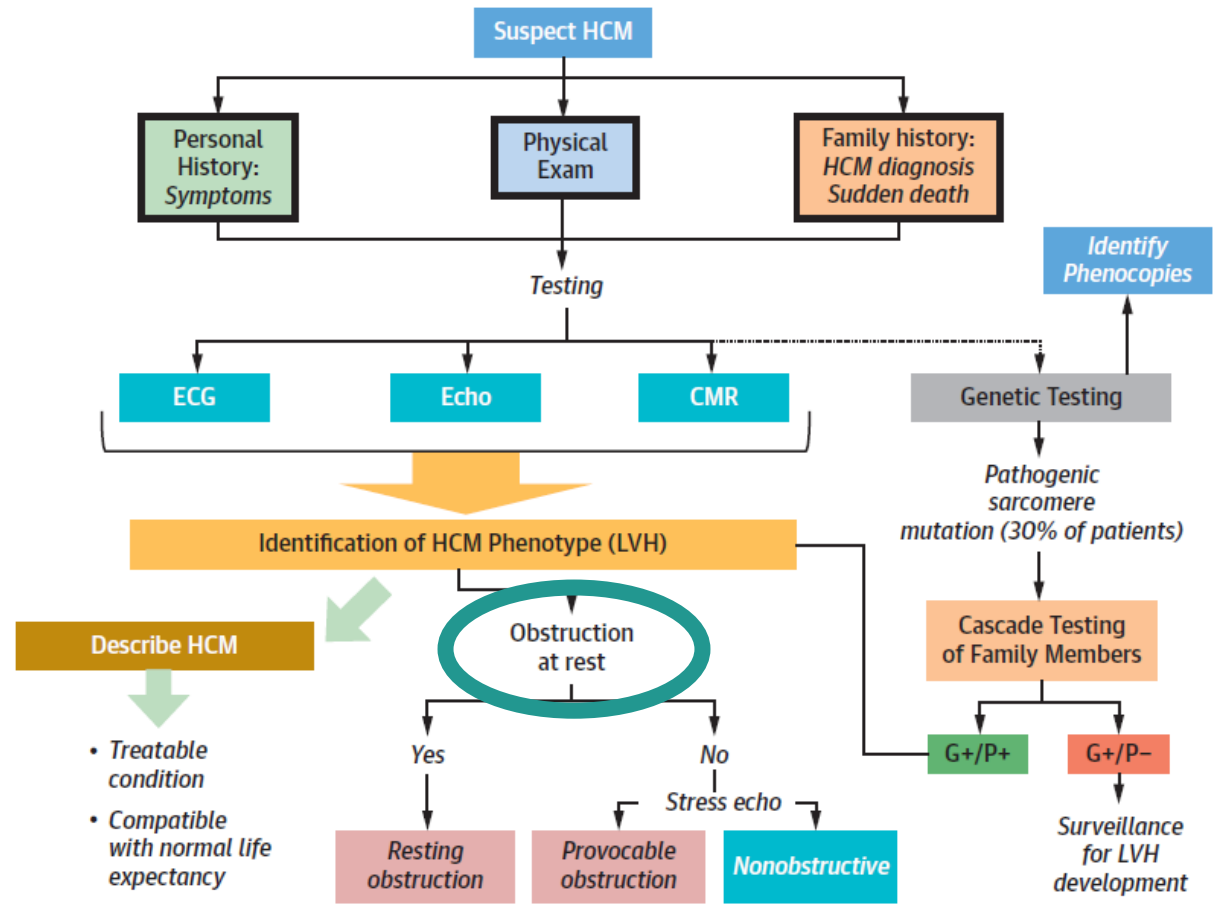
Disease	Gene	Locus	Protein	Frequency (%)
Hypertrophic cardiomyopathy				
HCM: myofilament mutation	*MYBPC3*	11p11.2	Cardiac myosin-binding protein C	15–25
	MYH7	14q11.2–q12	β-Myosin heavy chain	15–25
	TNNI3	19p13.4	Cardiac troponin I	<5
	TNNT2	1q32	Cardiac troponin T	<5
	TPM1	15q22.1	α-Tropomyosin	<5
	MYL2	12q23–q24.3	Ventricular regulatory myosin light chain	<2
	ACTC	15q14	α-Cardiac actin	<1
	MYH6	14q11.2–q12	α-Myosin heavy chain	<1
	MYL3	3p21.2–p21.3	Ventricular essential myosin light chain	<1
	TNNC1	3p21.3–p14.3	Cardiac troponin C	<1
	TTN	2q24.3	Titin	<1
HCM: Z-disc mutation	*LBD3*	10q22.2–q23.3	LIM binding domain 3 (alias: ZASP)	1–5
	ACTN2	1q42–q43	α-Actinin 2	<1
	ANKRD1	10q23.33	Ankyrin repeat domain 1 (alias: CARP)	<1
	CSRP3	11p15.1	Muscle LIM protein	<1
	MYOZ2	4q26–q27	Myozenin 2	<1
	TCAP	17q12–q21.1	Telethonin	<1
	VCL	10q22.1–q23	Vinculin/metavinculin	<1
HCM: calcium-handling	*JPH2*	20q12	Junctophilin-2	<1
	PLN	6q22.1	Phospholamban	<1
Hypertrophic cardiomyopathy phenocopies				
Barth syndrome/left ventricular noncompaction	*DTNA*	18q12	α-Dystrobrevin	–
	TAZ	Xq28	Tafazzin (G4.5)	–
Danon disease/Wolff-Parkinson-White syndrome	*LAMP2*	Xq24	Lysosome-associated membrane protein 2	–
Fabry's disease	*GLA*	Xq22	α-Galactosidase A	–
Forbes disease	*AGL*	1p21	Amylo-1,6-glucosidase	–
Friedreich's ataxia	*FXN*	9q13	Frataxin	–
Noonan syndrome	*KRAS*	12p12.1	v-Ki-ras2 Kirsten rat sarcoma viral oncogene homolog	–
	SOS1	2p22–p21	Son of sevenless homolog 1	–
Noonan syndrome, LEOPARD syndrome	*PTPN11*	12q24.1	Protein tyrosine phosphatase, non-receptor type 11, SHP-2	–
	RAF1	3p25	V-RAF-1 murine leukemia viral oncogene homolog 1	–
Pompe disease	*GAA*	17q25.2–q25.3	α-1,4-glucosidase deficiency	–
Wolff-Parkinson-White syndrome/HCM	*PRKAG2*	7q35–q36.36	AMP-activated protein kinase	–

Adapted with permission from Ginsburg (48).

Maron et al, 2022

T. Kubo et al., 2017

CENTRAL ILLUSTRATION Recommendations for Initial Clinical Evaluation and Testing Algorithm for Patients With or Suspected of Having Hypertrophic Cardiomyopathy



Maron, B.J. et al. J Am Coll Cardiol. 2022;79(4):372-389.

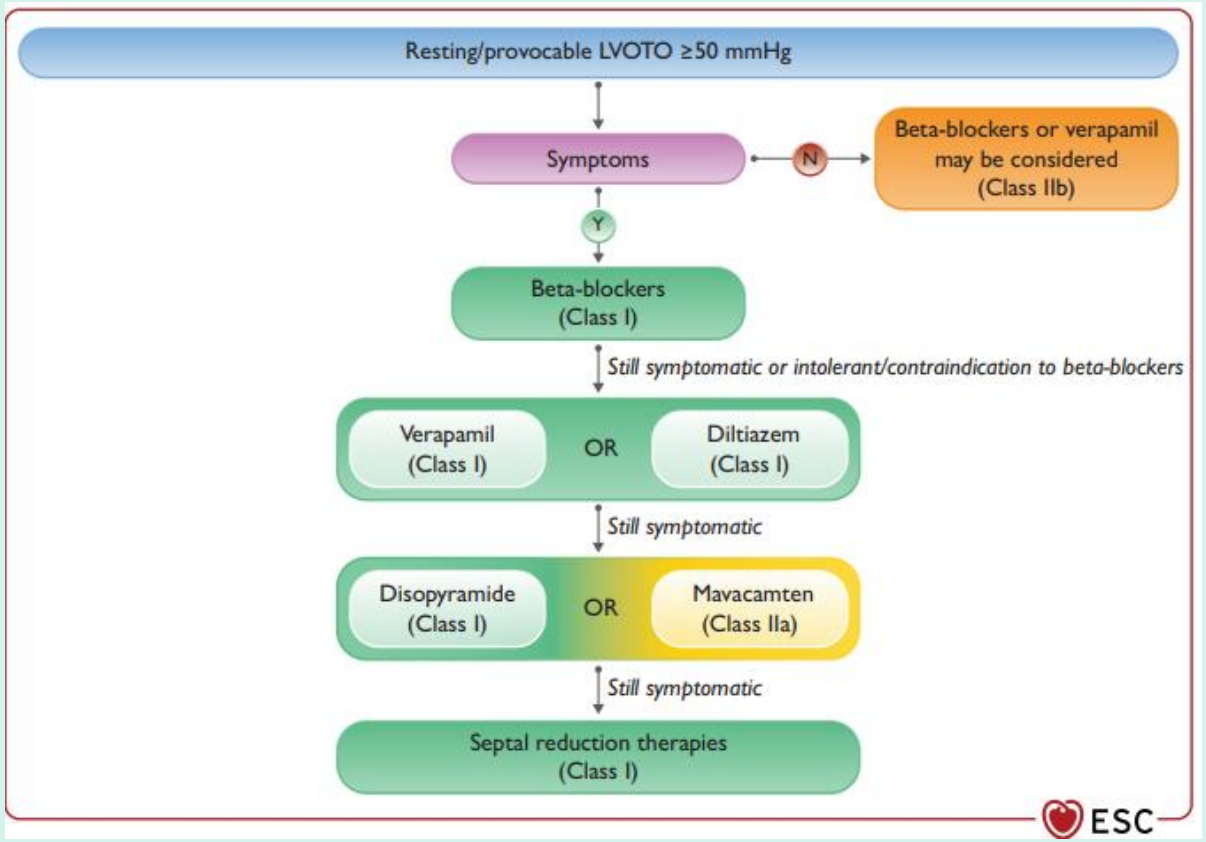
Evaluation of the HCM patient requires, in addition to targeted personal and family history and physical examination, noninvasive testing to define hemodynamic and clinical subgroups, as well as the identification of gene carriers. CMR = cardiac magnetic resonance; G = genotype; ECG = electrocardiography; echo = echocardiography; HCM = hypertrophic cardiomyopathy; LVH = left ventricular hypertrophy; P = phenotype; SAM = systolic anterior motion.

Maron et al, 2022

ESC European Society of Cardiology
European Heart Journal (2023) **44**, 3503–3626
https://doi.org/10.1093/eurheartj/ehad194

2023 ESC Guidelines for the management of cardiomyopathies

Beta-blockers and calcium antagonists (verapamil or diltiazem) should be considered to improve symptoms in patients with angina-like chest pain even in the absence of LVOTO or obstructive CAD.[740–744]	IIa	C
Oral nitrates may be considered to improve symptoms in patients with angina-like chest pain, even in the absence of obstructive CAD, if there is no LVOTO.	IIb	C
Ranolazine may be considered to improve symptoms in patients with angina-like chest pain even in the absence of LVOTO or obstructive CAD.[738,739]	IIb	C

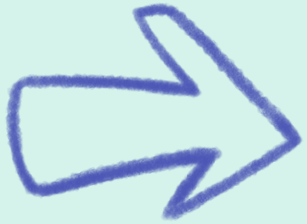


Recommendation Table 31 — Exercise recommendations for patients with cardiomyopathy

Recommendations	Class[a]	Level[b]
All cardiomyopathies		
Regular low- to moderate-intensity exercise is recommended in all able individuals with cardiomyopathy.	I	C
An individualized risk assessment for exercise prescription is recommended in all patients with cardiomyopathy.	I	C
HCM		
High-intensity exercise and competitive sport should be considered in genotype-positive/ phenotype-negative individuals who seek to do so.[1124]	IIa	C
High-intensity exercise and competitive sport may be considered in asymptomatic low-risk[c] individuals with morphologically mild hypertrophic cardiomyopathy in the absence of resting or inducible left ventricular outflow obstruction and exercise-induced complex ventricular arrhythmias.[1107,1113,1125,1126]	IIb	B
High-intensity exercise, including competitive sport, is not recommended in high-risk individuals and in individuals with left ventricular outflow tract obstruction and exercise-induced complex ventricular arrhythmias.	III	C

2023 ESC Giudelines for the management of cardiomyopathies

TAKE HOME MESSAGE

L’ipertrofia ventricolare sinistra  **elevata prevalenza**

In base al grado considerare cause rare

ECHOLAB

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