

PLACE | ACADEMY

UN PROGETTO DI

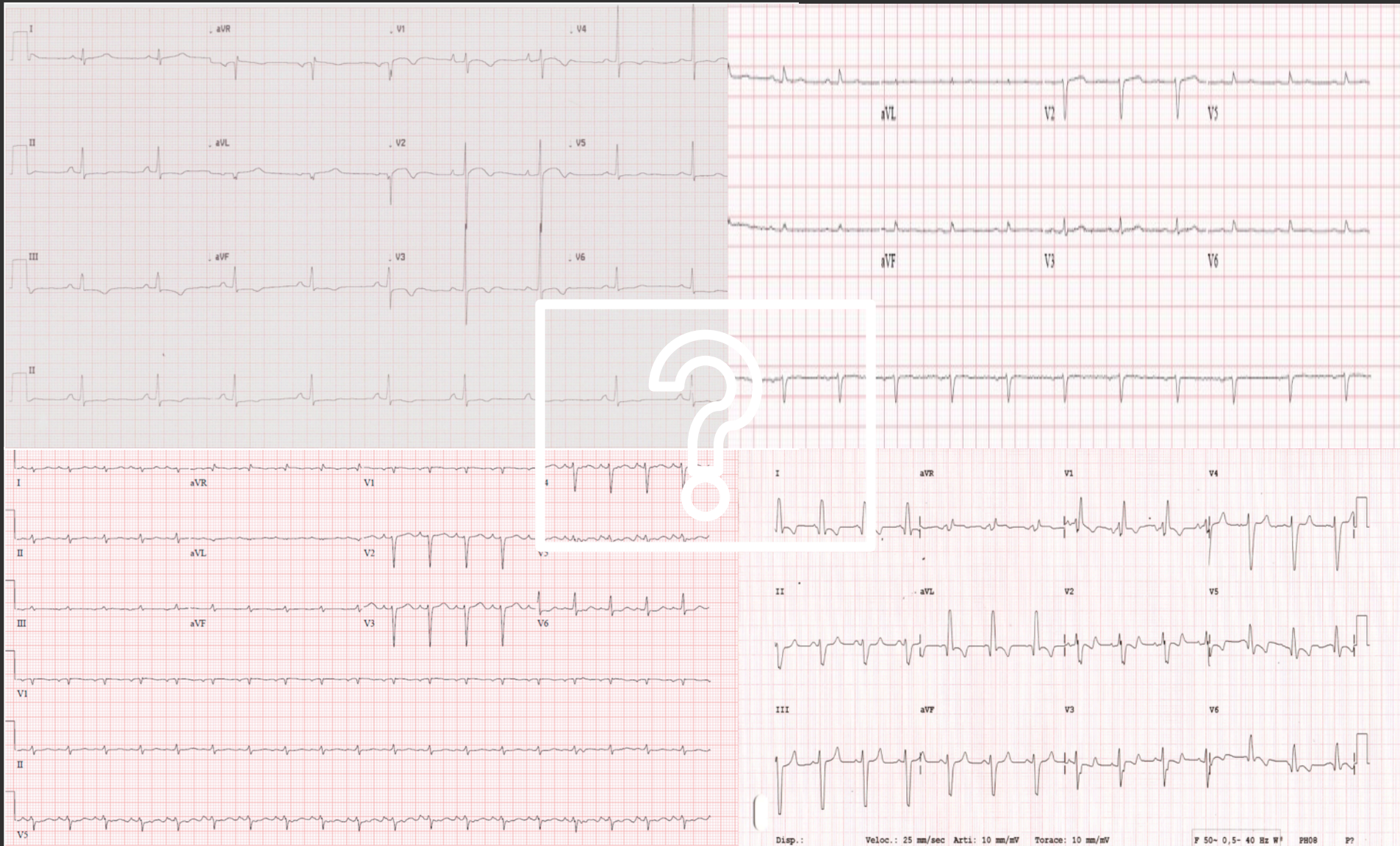


Dall'elettrocardiogramma alla genetica

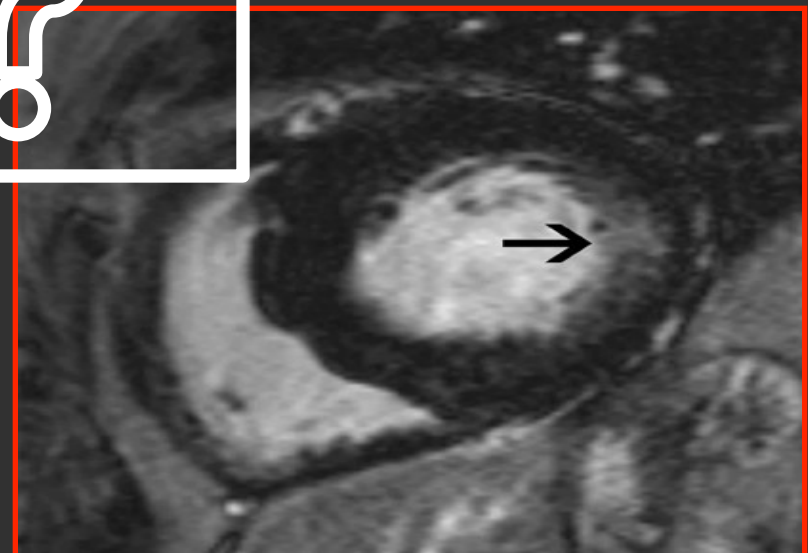
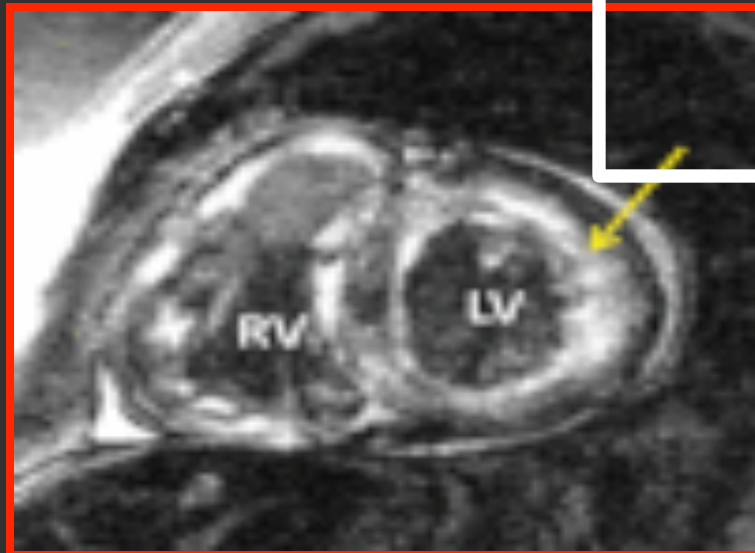
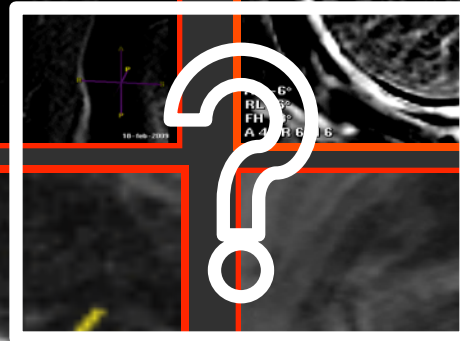
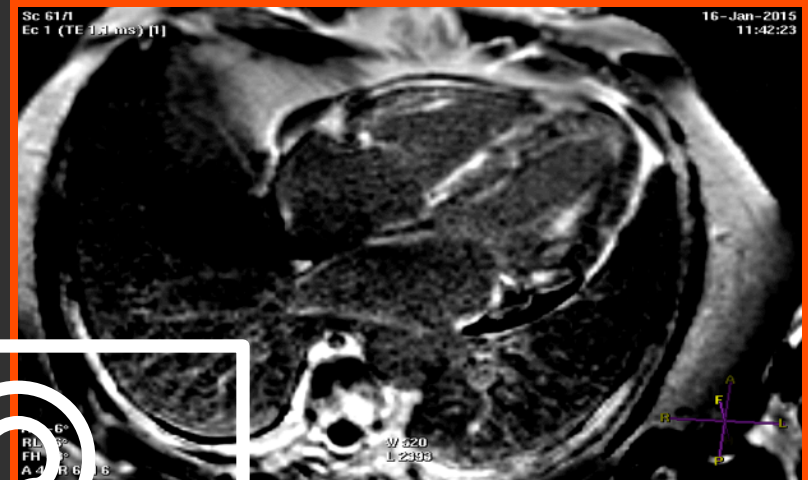
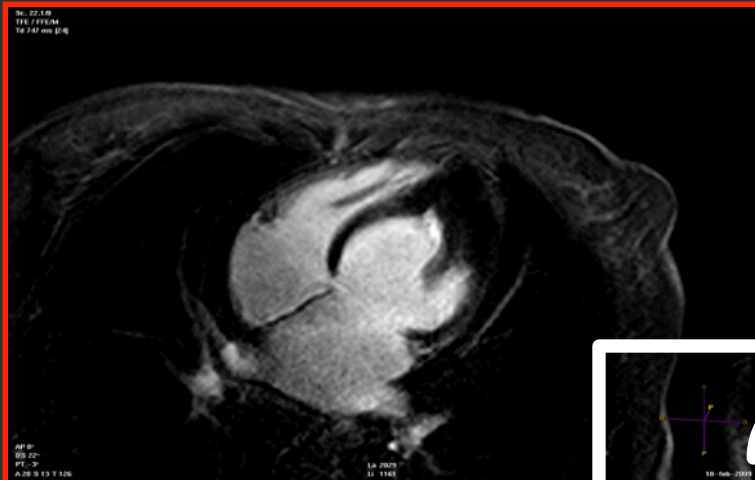
LA CARDIOPATIA A FENOTIPO IPERTROFICO 2016 UPDATE: DAL GENOTIPO AL FENOTIPO

Chiara Lanzillo MD, PhD
Cardiologia Pol. Casilino

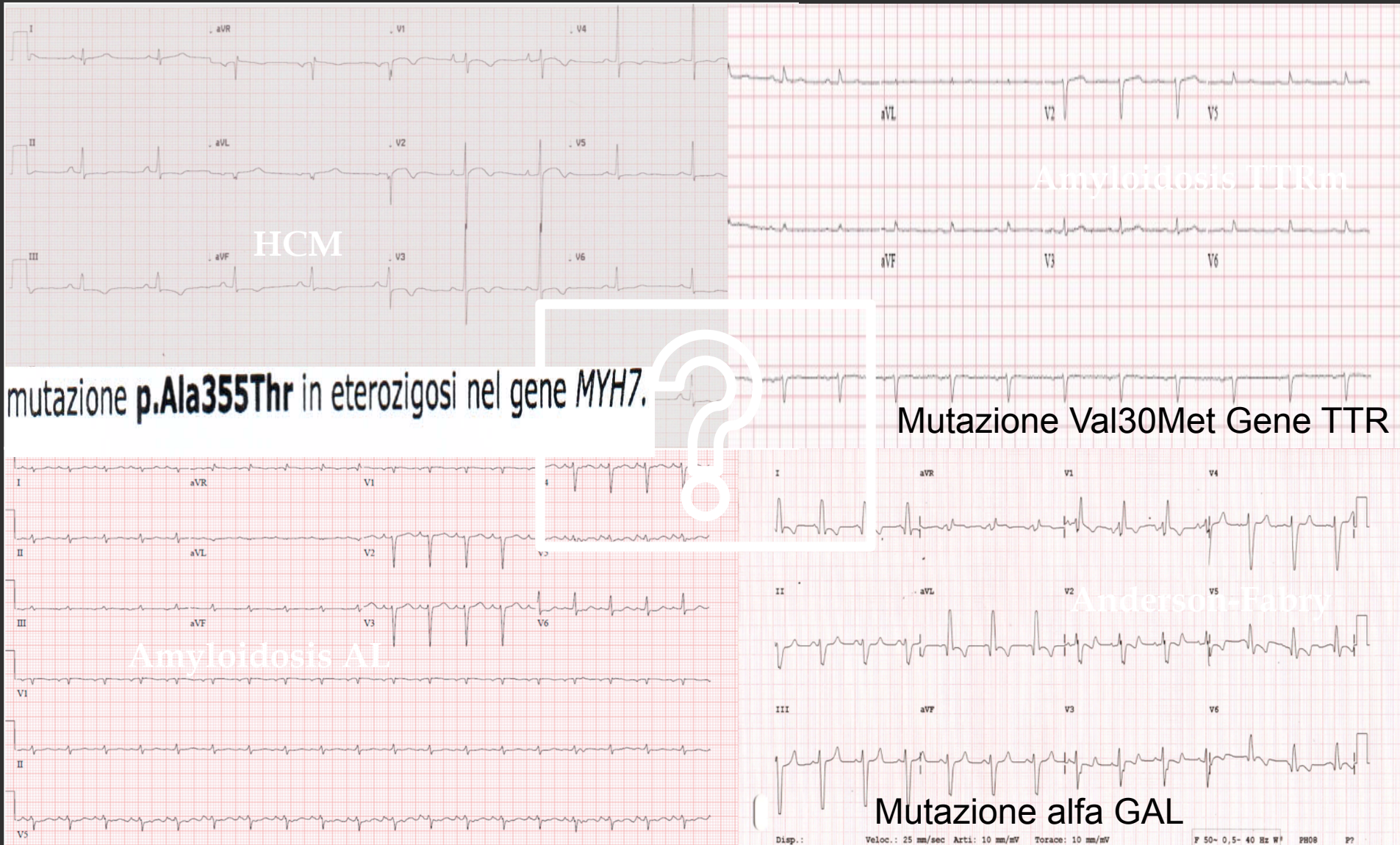
Cardiopatie a fenotipo ipertrofico

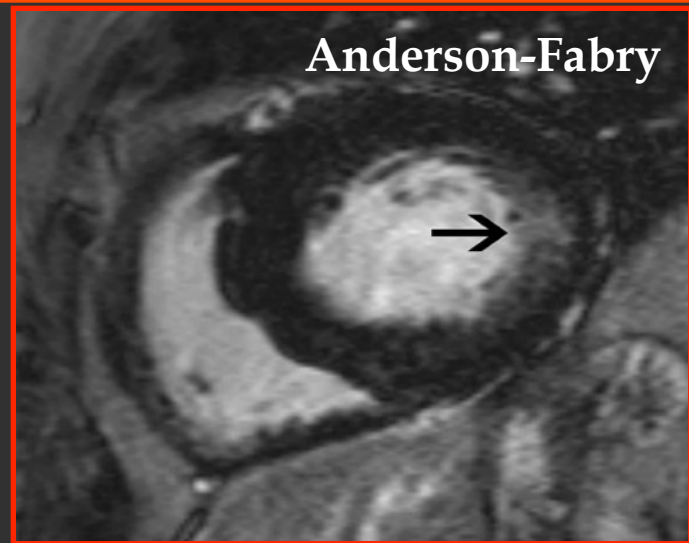
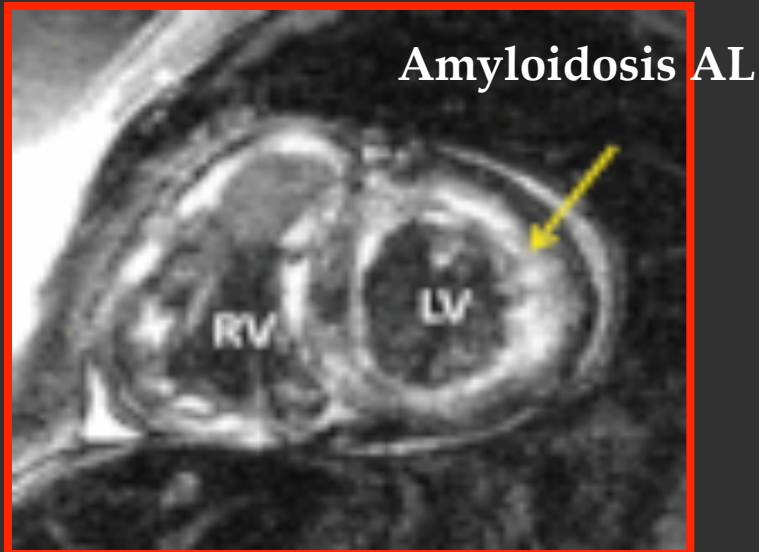
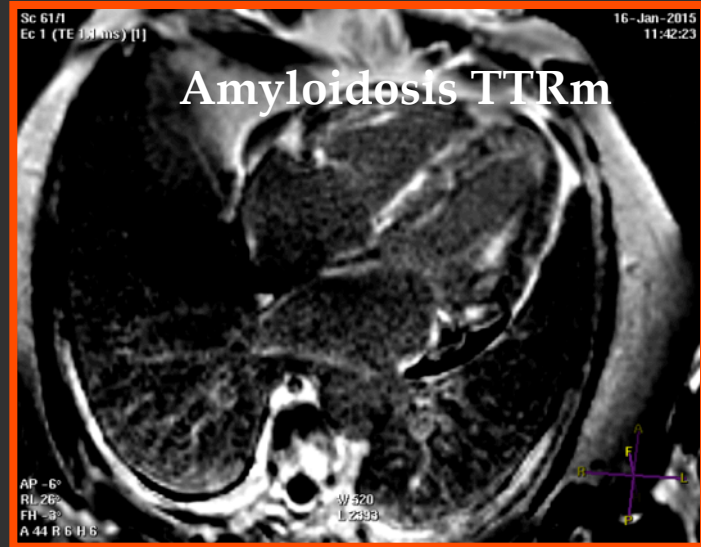
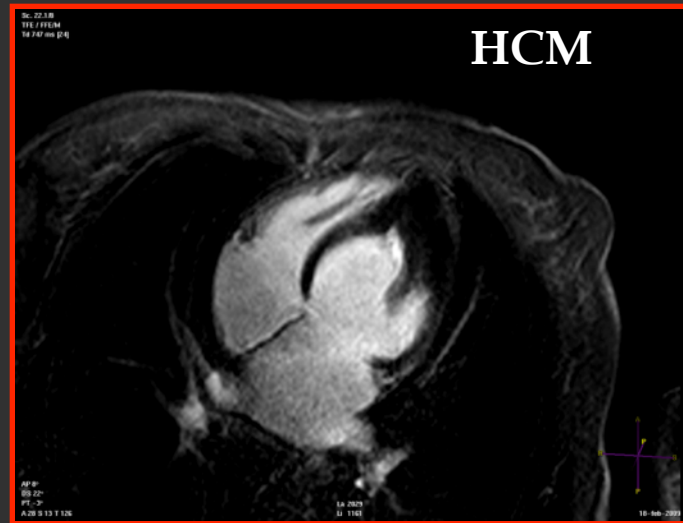


Cardiopatie a fenotipo ipertrofico



Cardiopatie a fenotipo ipertrofico



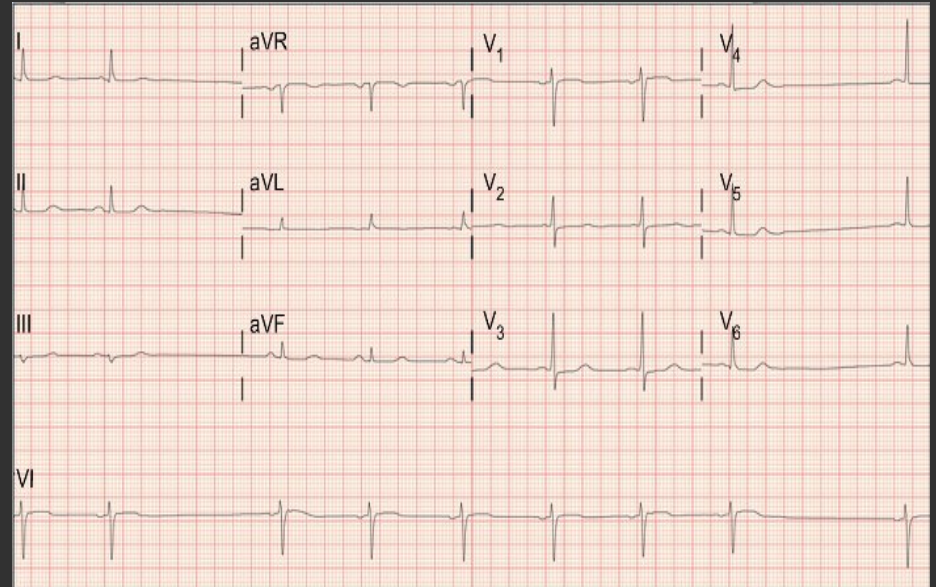


- The presence of LVH is not the end but only the beginning of the story.
- The correct diagnosis is important in therapy and prognosis.

Contribution of Electrocardiogram in the differentiation of Cardiac Amyloidosis and Nonobstructive HCM

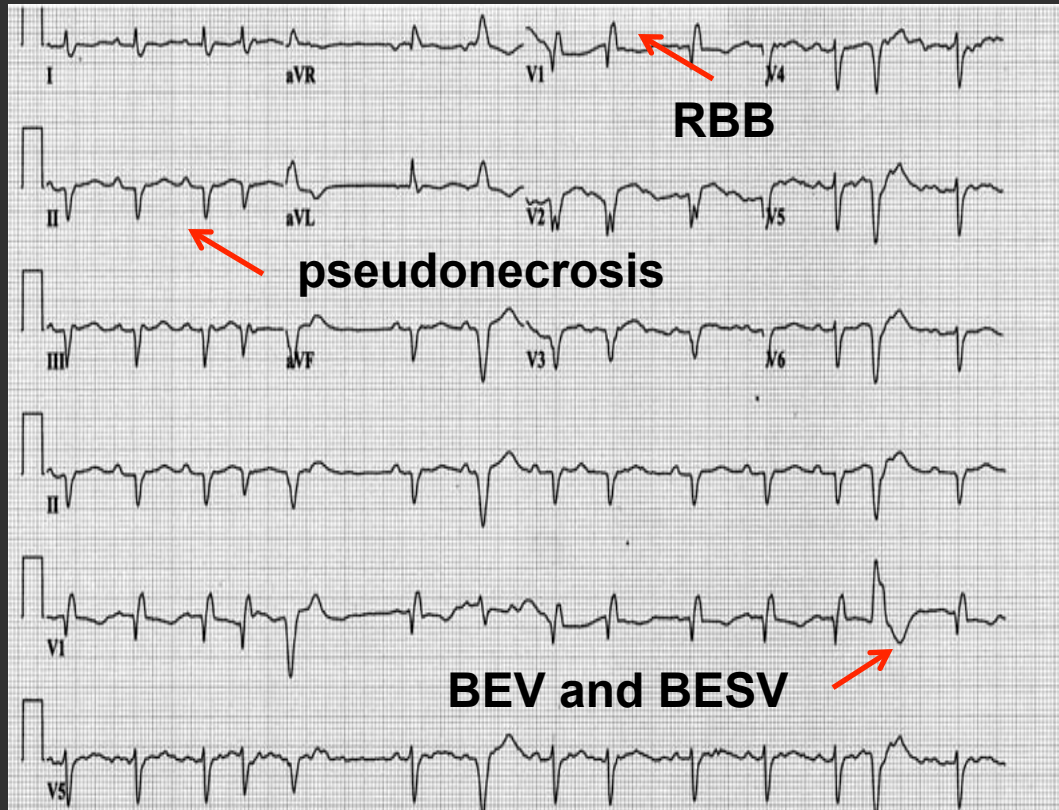
Cardiac Amyloidosis

- Low voltage on limb leads (50% vs 1,6%)
- AV block (21% vs 4%)
- Pseudoinfarct pattern (84% vs 39%)
- Longer QRS duration (104 vs 98 msec)



- R wave voltage of I and/or aVR < 0.35 mV
- QRS complex voltage of I and/or aVR < 0.45 mV

Left ventricle hypertrophy : only phenotype, what else?



- Low QRS voltage pattern
- Pseudoinfarction pattern
- AF
- Atrioventricular and interventricular block
- Unusual axis deviation

Electrocardiographic findings in primary systemic amyloidosis and biopsy-proven cardiac involvement.

Grogan M. Am J Cardiol. 2005

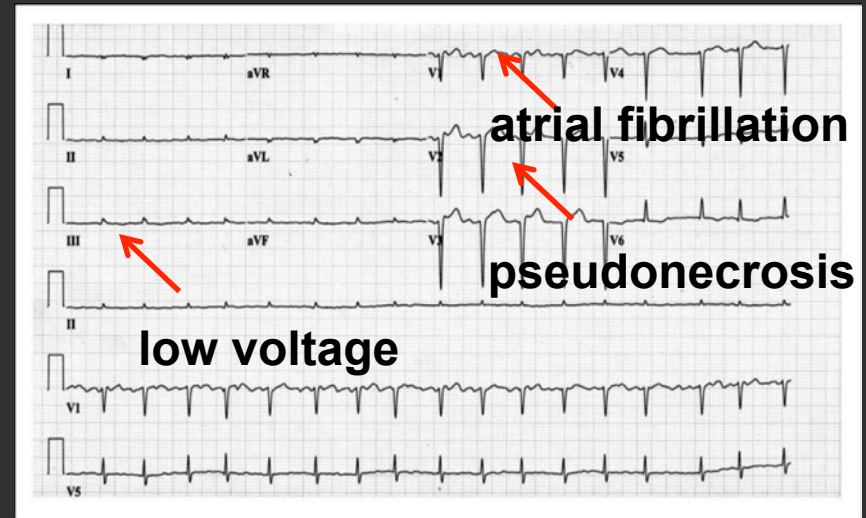
Noninvasive diagnosis of biopsy-proven cardiac amyloidosis.

Joseph E Rahman, JACC 2004

Left ventricle hypertrophy : only phenotype, what else?

- **Low voltage (46%) and a pseudoinfarct pattern (47%) were the most common findings. Criteria for left ventricular hypertrophy were present in 16% of patients**

Criteria for left ventricular hypertrophy were present in 16% of patients

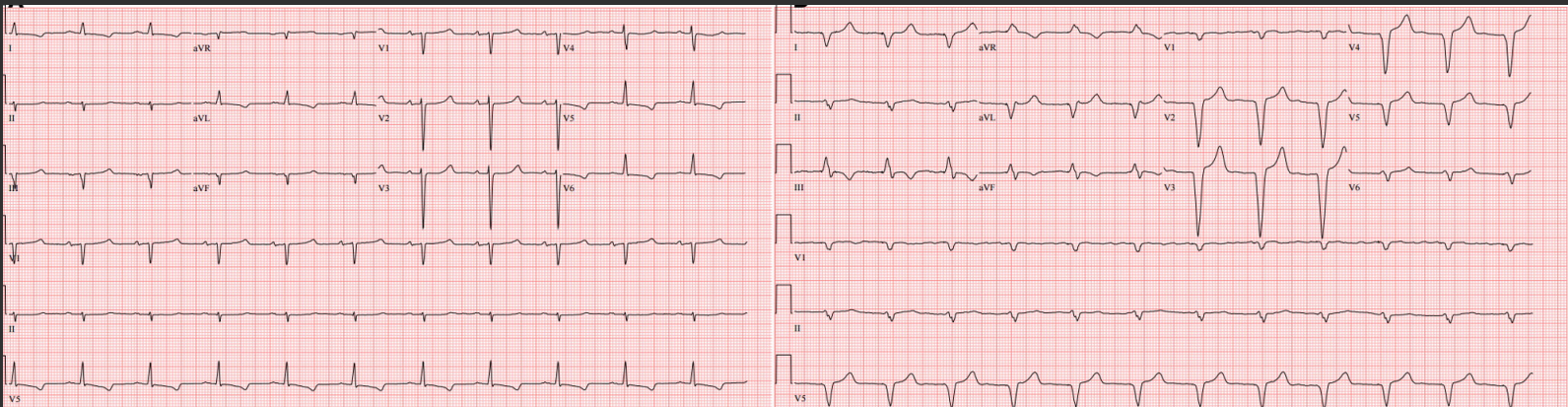


Electrocardiographic findings in primary systemic amyloidosis and biopsy-proven cardiac involvement.

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Left ventricle hypertrophy : only phenotype, what else?

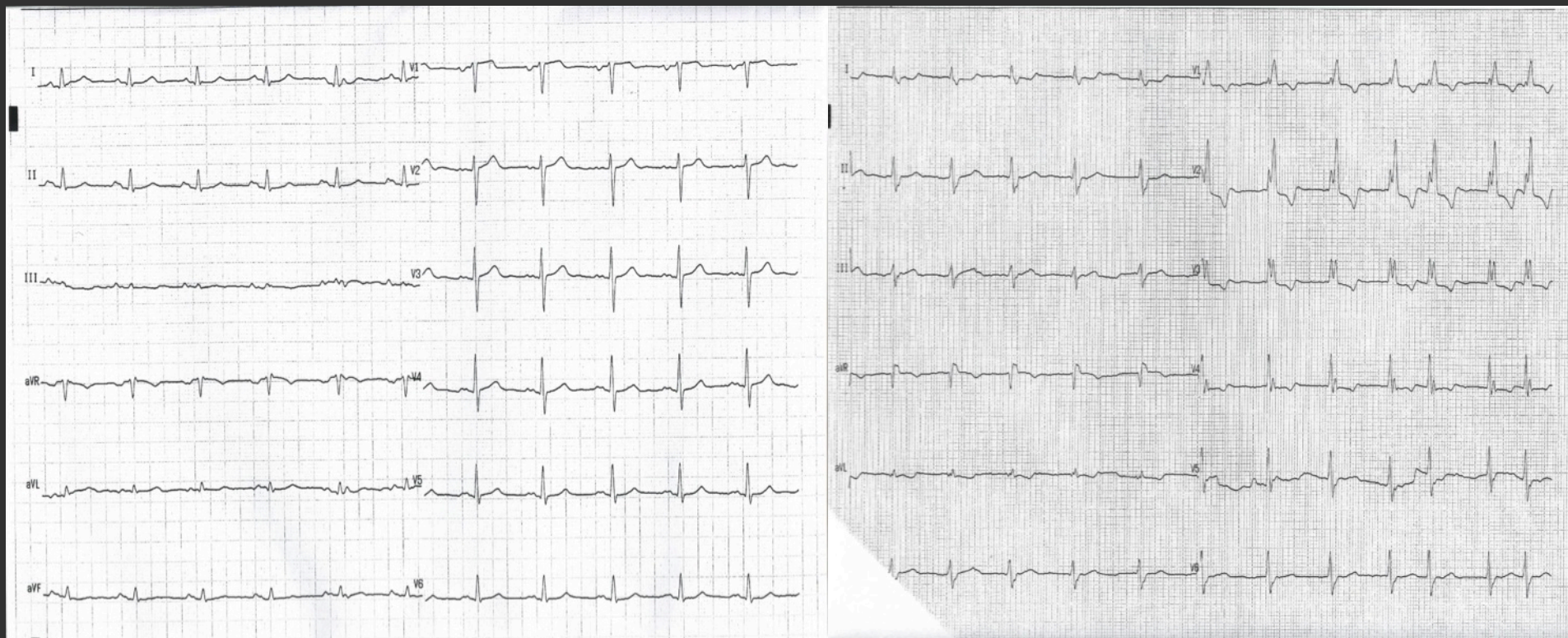
Low voltage in narrow QRS and ventricularly paced rhythm



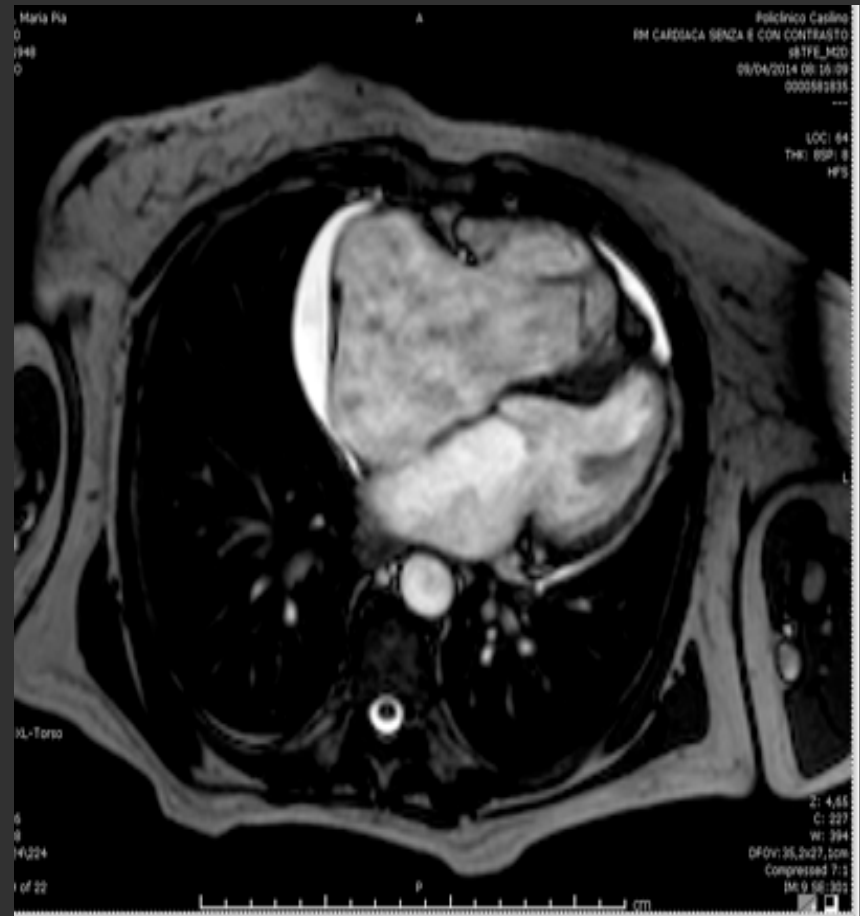
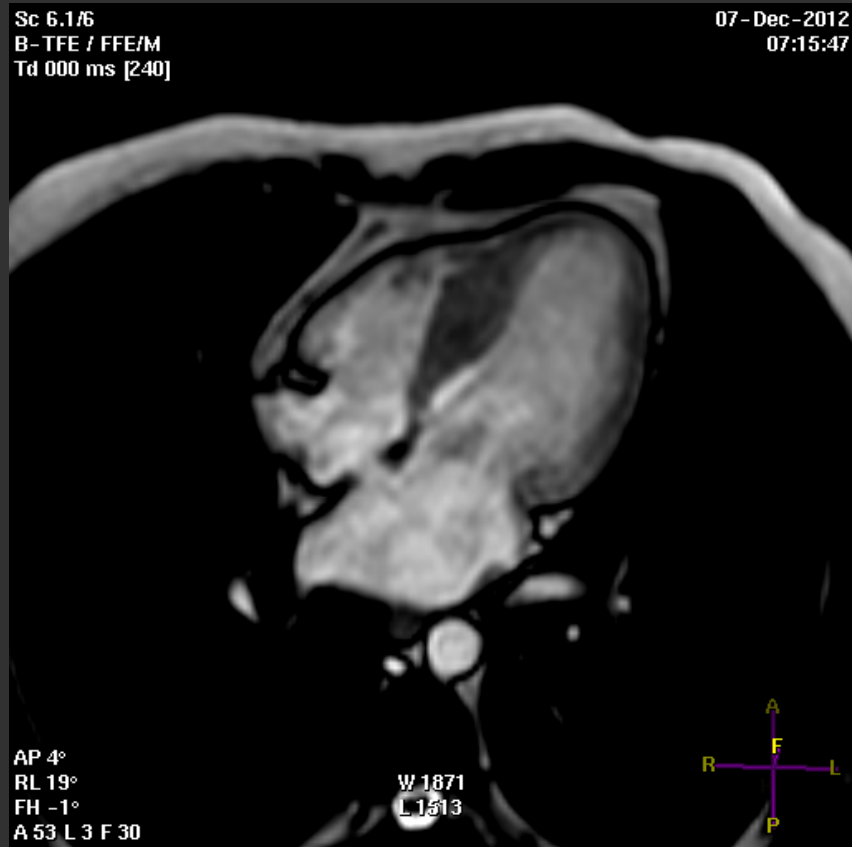
- Low voltage electrocardiography coupled with increased ventricular wall thickness is the hall- mark of cardiac amyloidosis. 30% had conduction delay (QRS N 120 ms). In those with narrow QRS, 68% met low limb, 72% low Sokolow and 57% both criteria, with lower voltages found in AL vs ATTR. Voltage was significantly associated with mortality (p b 0.001 for both criteria) after multivariable adjustment.

Are classic predictors of voltage valid in cardiac amyloidosis?
A contemporary analysis of electrocardiographic findings Brett
W. Sperry Int J Cardiology 2016

Genetica e eterogeneità del fenotipo: MYBC3

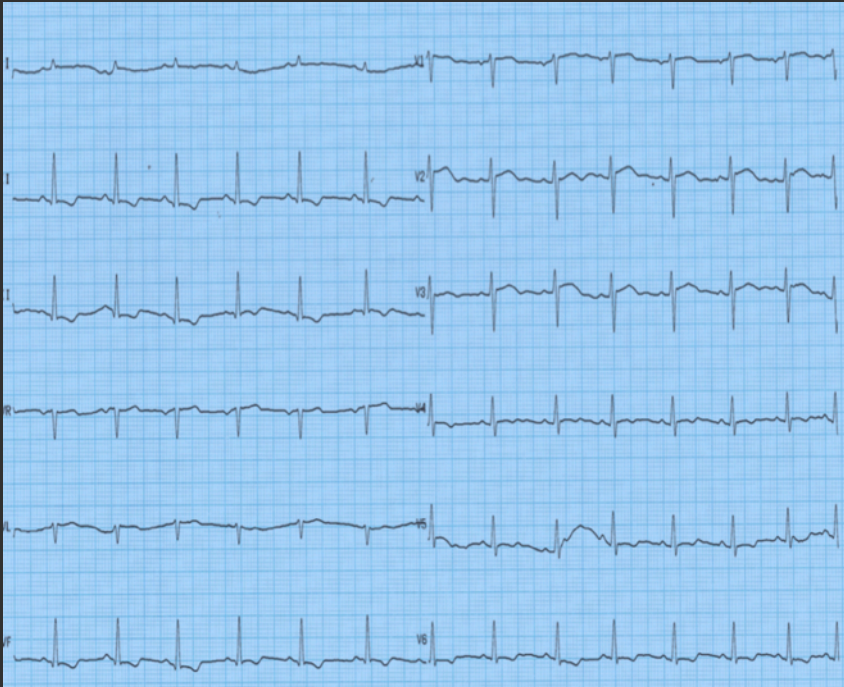


Genetica e eterogeneità del fenotipo: MYBC3



Atassia di Friedreich

ECG



Analisi genetica molecolare

Il test per la Malattia di Friedreich (espansione di triplette GAA del gene X25) è stato eseguito sul suo DNA secondo il metodo di Filla et al. 1996
L'analisi ha fornito i seguenti risultati :

alleli con numero di triplette normale (<34 GAA) : assenti

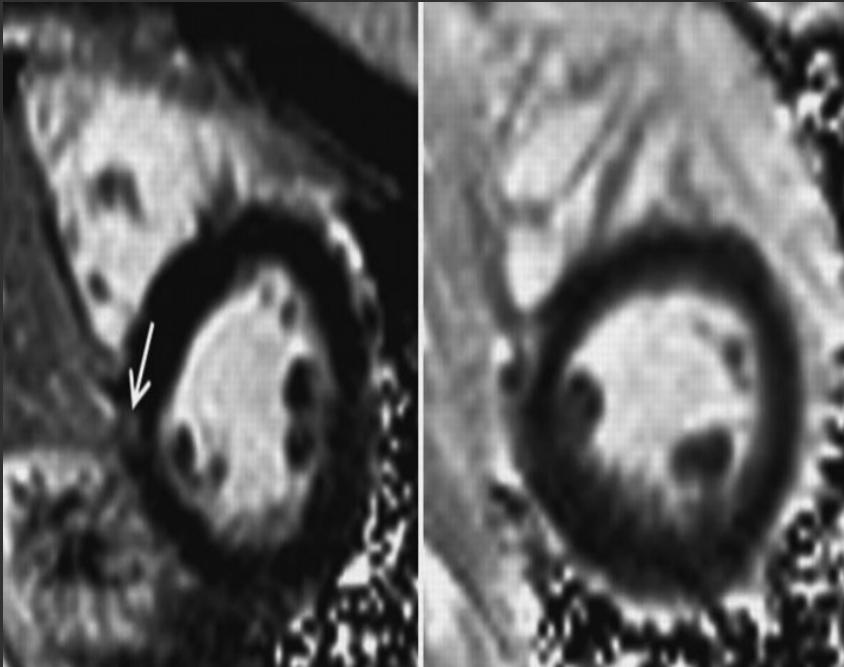
alleli con numero di triplette espanso (>90 GAA) : 2 (con numero stimato di triplette tra 900 e 1000).

Tale risultato indica che la paziente è affetta da Malattia di Friedreich. Si tratta di una malattia a trasmissione autosomica recessiva. Essa cioè viene trasmessa ad un figlio/a da genitori entrambi portatori sani della espansione. La malattia può essere trasmessa da un individuo affetto figli solo se anche il coniuge è portatore sano della stessa malattia.

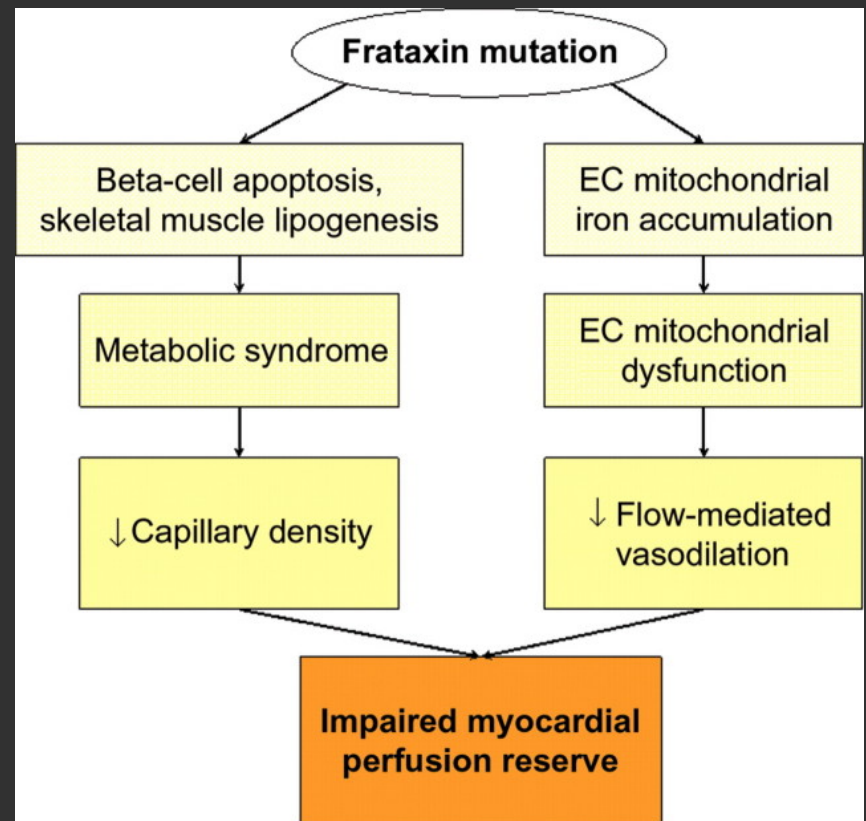
Poiché altri parenti potrebbero essere portatori sani di M. di Friedreich è consigliabile che essi richiedano una consulenza genetica.

Atassia di Friedreich

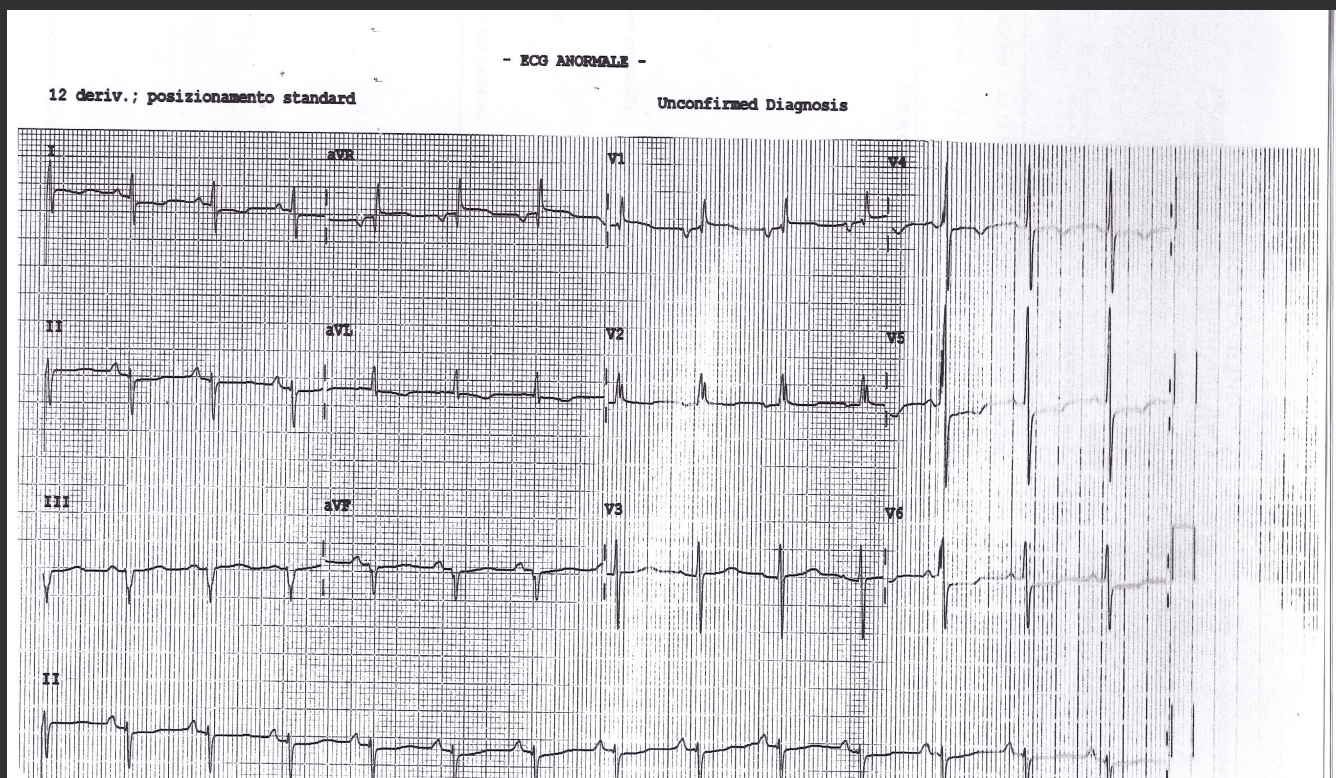
LVH



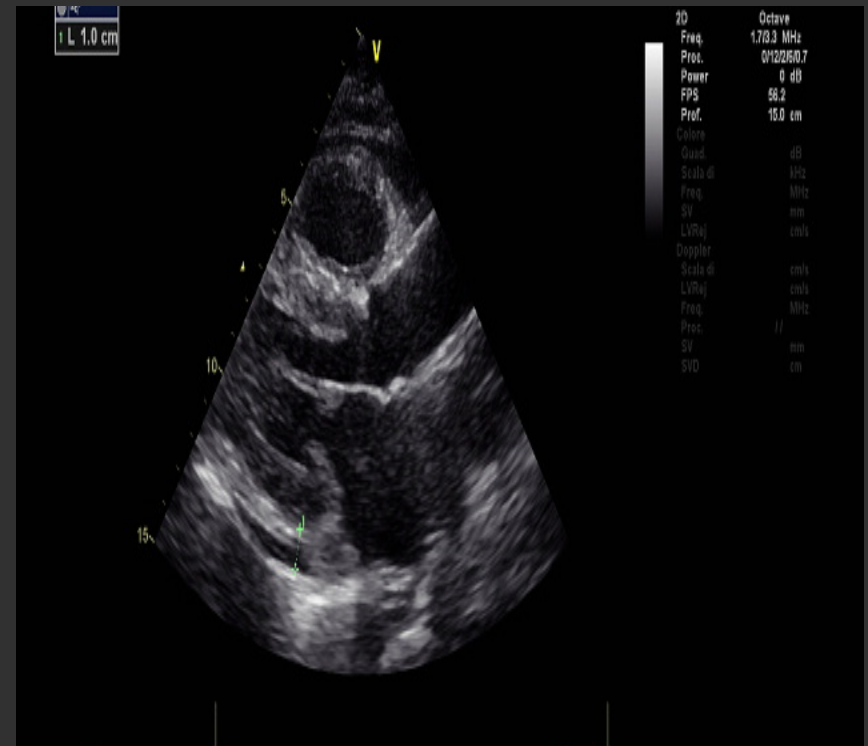
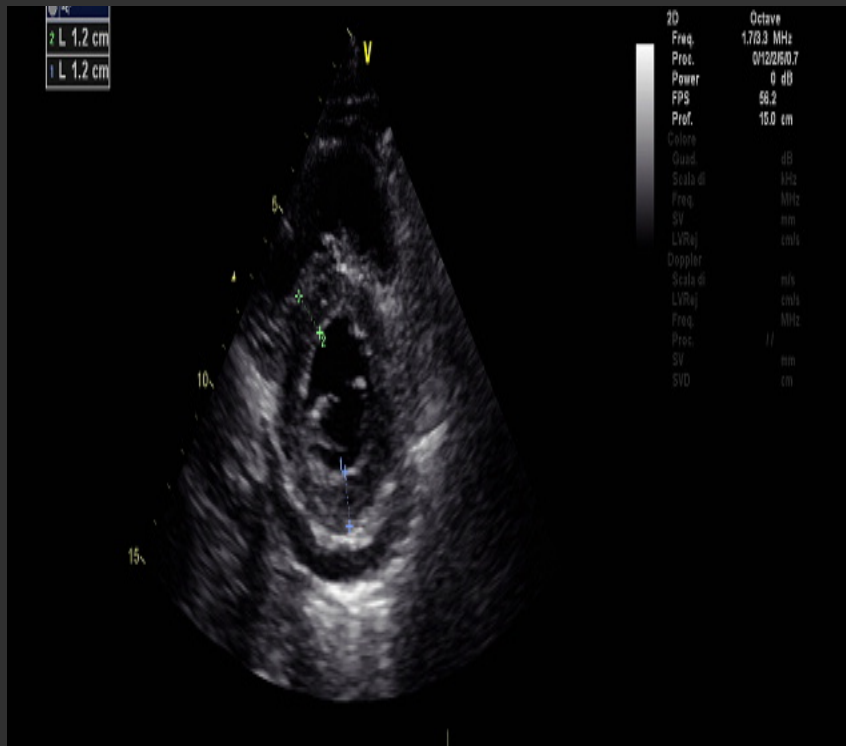
Impaired myocardial perfusion



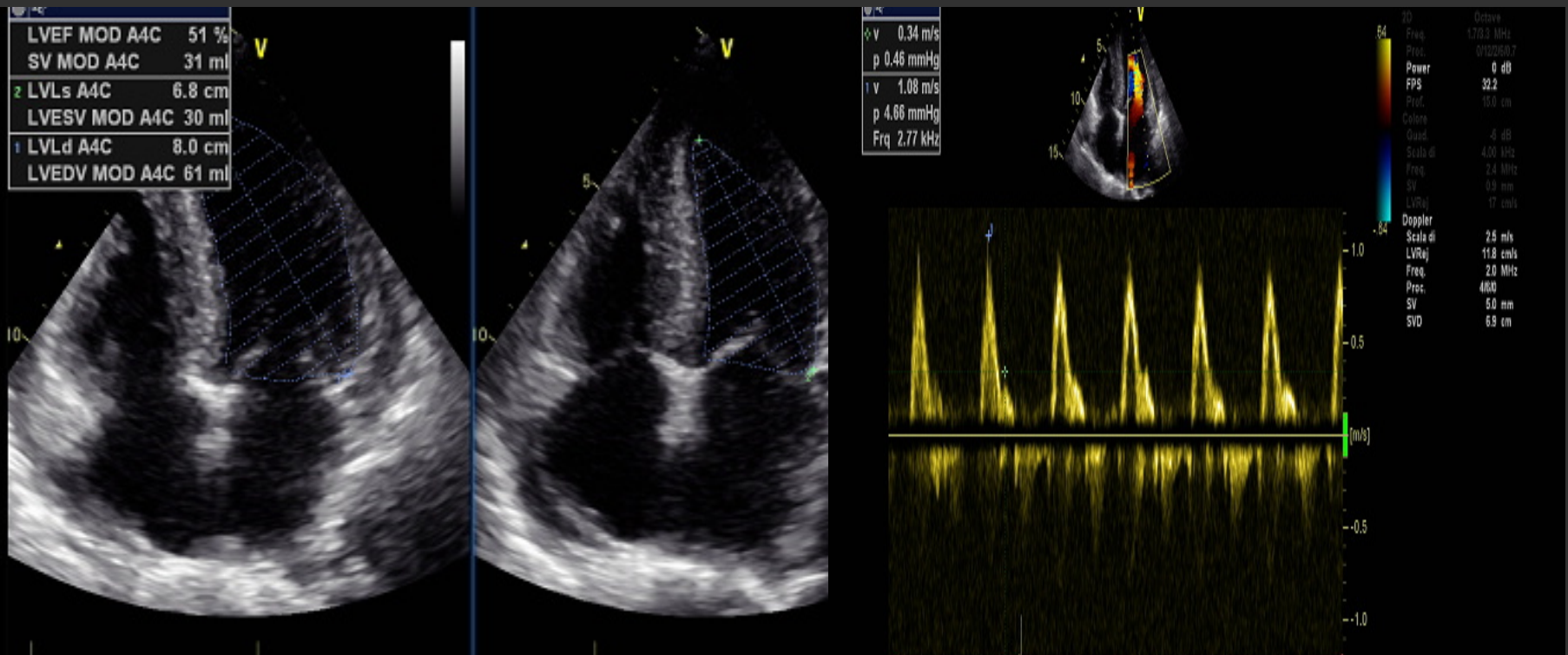
Una giovane donna con ipertrofia ventricolare sinistra e scompenso cardiaco a funzione sistolica ventricolare normale



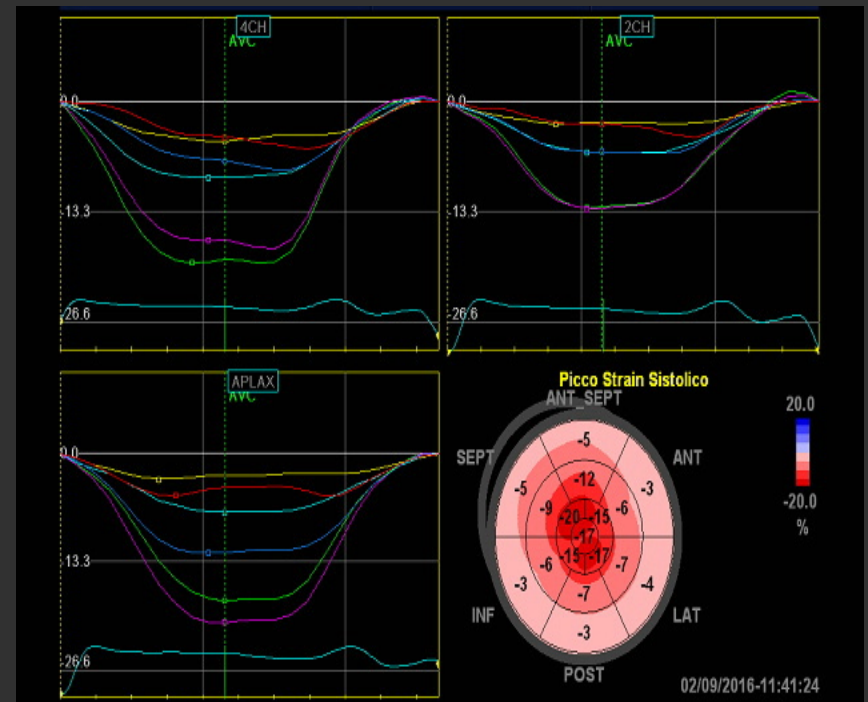
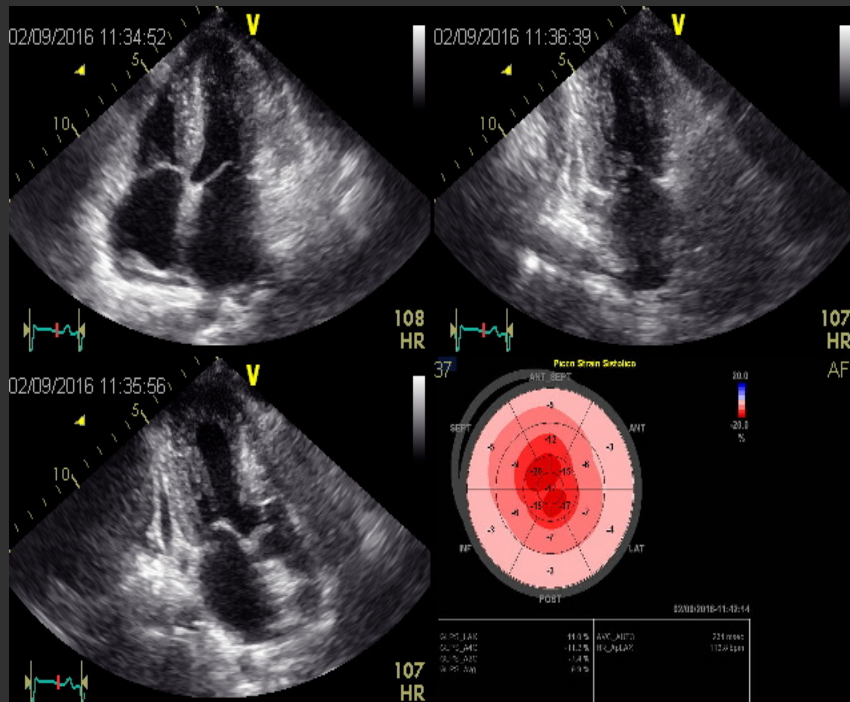
Una giovane donna con ipertrofia ventricolare sinistra e scompenso cardiaco a funzione sistolica ventricolare normale



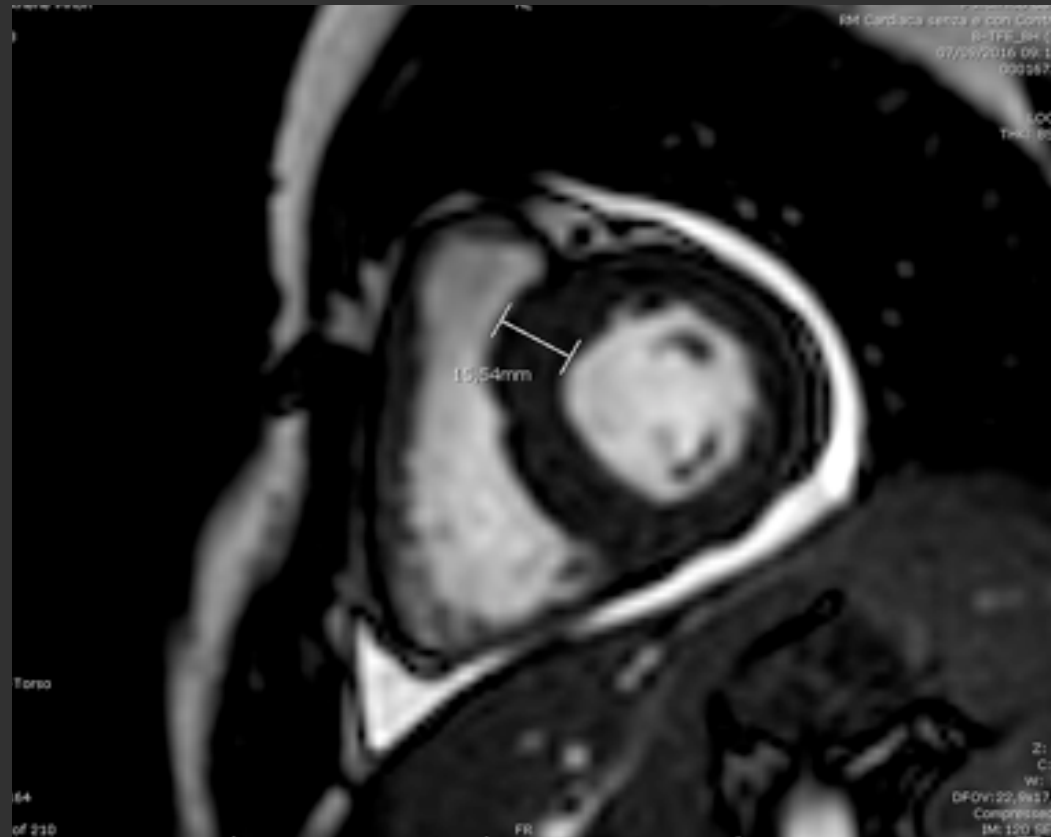
Una giovane donna con ipertrofia ventricolare sinistra e scompenso cardiaco a funzione sistolica ventricolare normale



Una giovane donna con ipertrofia ventricolare sinistra e scompenso cardiaco a funzione sistolica ventricolare normale



Una giovane donna con ipertrofia ventricolare sinistra e scompenso cardiaco a funzione sistolica ventricolare sinistra normale



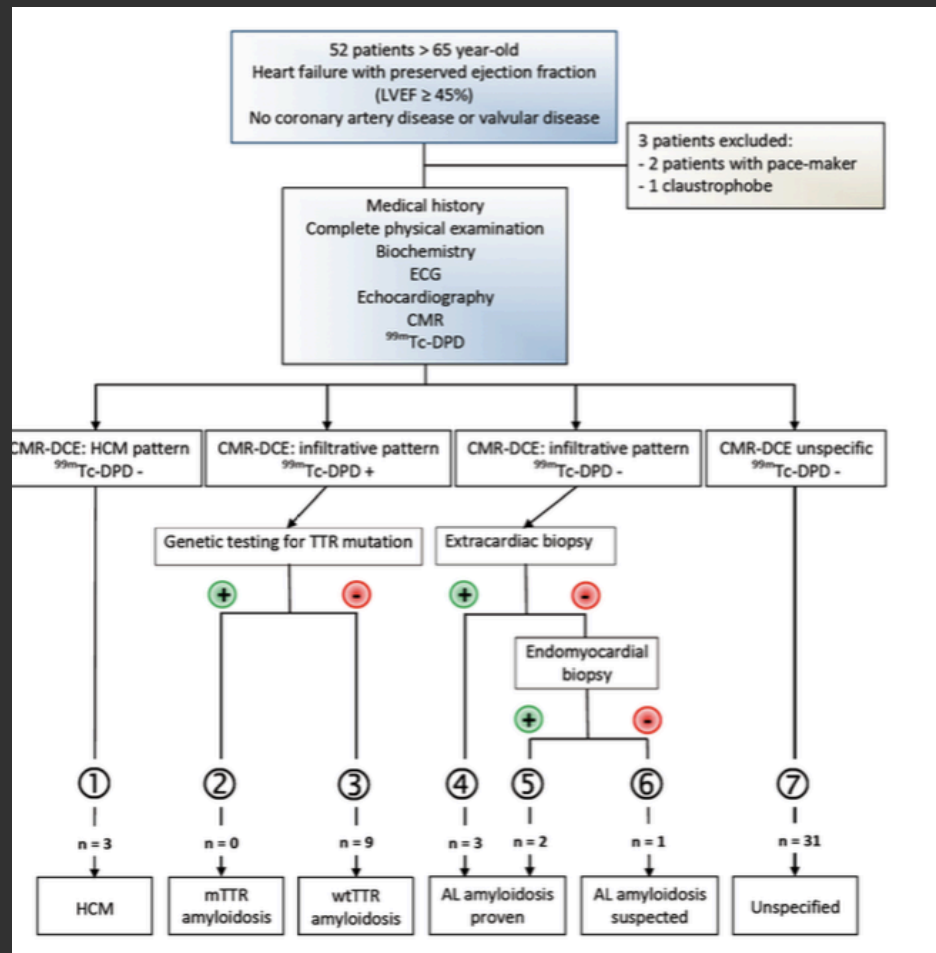
Una giovane donna con ipertrofia ventricolare sinistra e scompenso cardiaco a funzione sistolica ventricolare sinistra normale



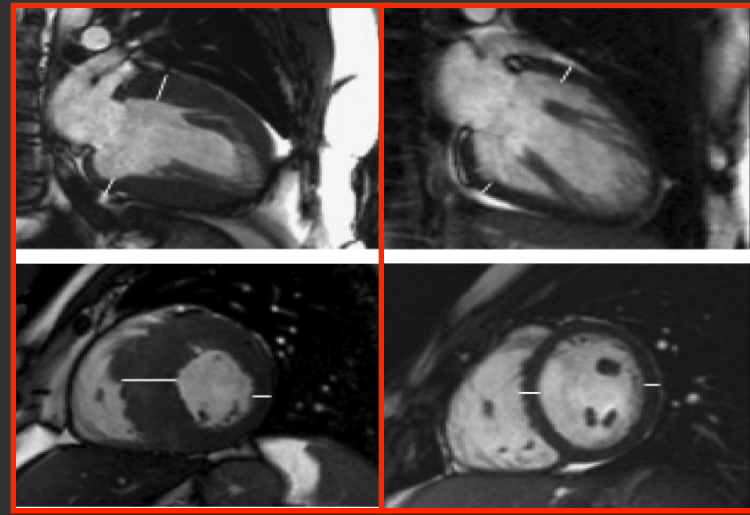
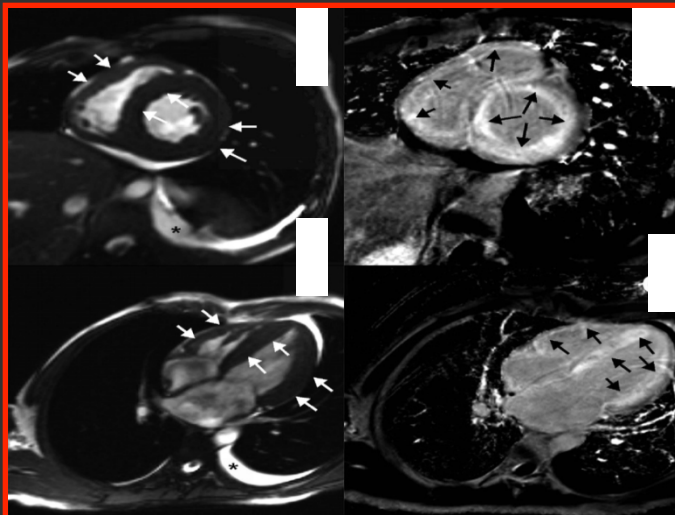
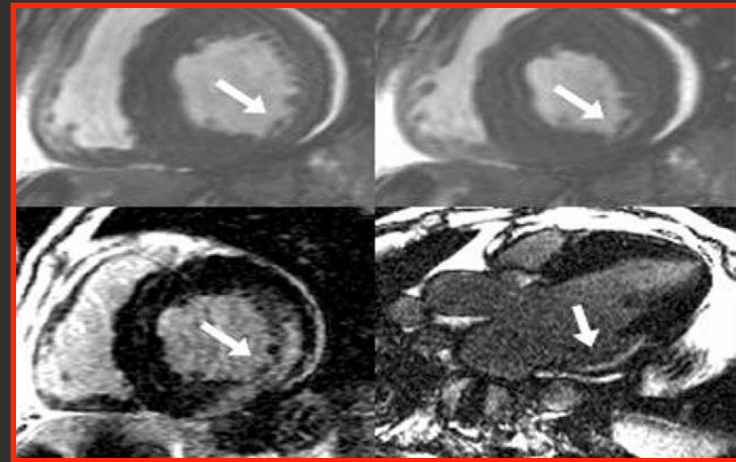
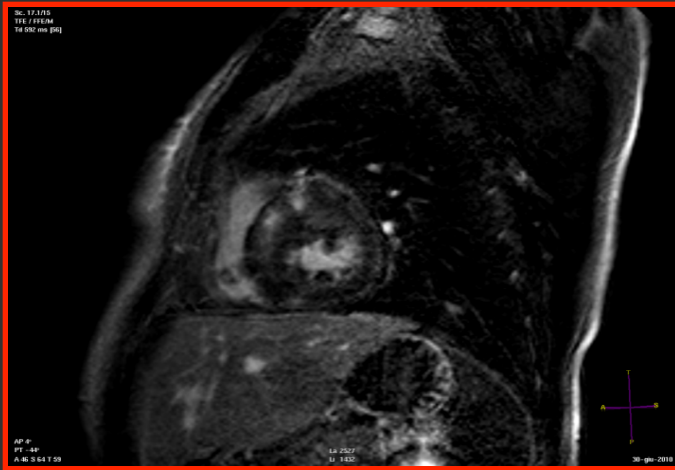
Una giovane donna con ipertrofia ventricolare sinistra e scompenso cardiaco a funzione sistolica ventricolare sinistra normale

Descrizione Esame / Test	Risultato	U.M.	Valori di Riferimento	Metodica
SIEROLOGIA E IMMUNOLOGIA				
IMMUNOFISSAZIONE S. <i>IMMUNOFISSAZIONE S.</i>	LA PRESENZA DI UNA COMPONENTE MONOCLONALE DI TIPO IG"G-LAMBDA"			
Ab anti membrana citoplasmatica	1:20			<i>I.F.A.</i>
PROTEINE DI BENGE JONES	ASSENZA DELLA PROTEINA DI BENGE JONE			<i>ELETTROFORESI</i>
CATENA KAPPA SIERICHE	136	<i>mg/dl</i>	* <i>170÷370</i>	<i>NEFELOMETRIA</i>
CATENE LAMBDA SIERICHE	152	<i>mg/dl</i>	<i>90÷210</i>	<i>NEFELOMETRIA</i>
IMMUNOFISSAZIONE URINARIA	ASSENZA DELLA PROTEINA DI BENGE JONES			<i>ELETTROFORESI</i>
C3	121	<i>mg/dl</i>	<i>90÷180</i>	<i>NEFELOMETRIA</i>
ENA (Anticorpi anti-antigeni nucleari estraibili)				
<i>SmB</i>	NEGATIVO		<i>Negativo</i>	<i>ELISA</i>
<i>SmD</i>	NEGATIVO		<i>Negativo</i>	<i>ELISA</i>
<i>RNP-70k</i>	NEGATIVO		<i>Negativo</i>	<i>ELISA</i>
<i>RNP-A</i>	NEGATIVO		<i>Negativo</i>	<i>ELISA</i>
<i>RNP-C</i>	NEGATIVO		<i>Negativo</i>	<i>ELISA</i>
<i>SSA/Ro52</i>	NEGATIVO		<i>Negativo</i>	<i>ELISA</i>
<i>SSA/Ro60</i>	NEGATIVO		<i>NEGATIVO</i>	<i>IMMUNOBLOT</i>
<i>SSB/La</i>	NEGATIVO		<i>NEGATIVO</i>	<i>IMMUNOBLOT</i>
<i>Cenp-b</i>	NEGATIVO		<i>NEGATIVO</i>	<i>IMMUNOBLOT</i>
<i>Topo-I / Scl-70</i>	NEGATIVO			
<i>Jo-1 HRS</i>	NEGATIVO			
<i>ribosomal P</i>	NEGATIVO			
<i>Histoni</i>	NEGATIVO			
Ab ANTIMITOCONDRIO (AMA)	ASSENTI		<i>ASSENTI</i>	<i>IMMUNOFLOURESCENZA INDIRECTA</i>
Ab ANTIMUSCOLO LISCIO(ASMA)	ASSENTI		<i>NEGATIVO</i>	<i>IMMUNOFLOURESCENZA INDIRECTA</i>
Ab ANTI DNA NATIVO	Assente		<i>Assenti. Titolo signif. = o > 1:</i>	<i>I.F.A.</i>
Ab ANTI-CITOPLASMA NEUTROFILI (ANCA)				
<i>c-ANCA (PR3)</i>	0,4	<i>U/ml</i>	<i>Minore di 2 Valori significativi: maggiore di 3</i>	<i>ELIA</i>
<i>p-ANCA (MPO)</i>	0,5	<i>U/ml</i>	<i>Minore di 3,5 Valori significativi: maggiore di 5</i>	<i>ELIA</i>
IMMUNOCIMICA				
Referto del 11/09/2016 ore 10:59 stampato il 11/09/2016 alle ore 10:59				

A pilot study for LV imaging phenotype of ptz over 65 years with heart failure and preserved LVEF : the high prevalence of amyloid cardiomyopathy

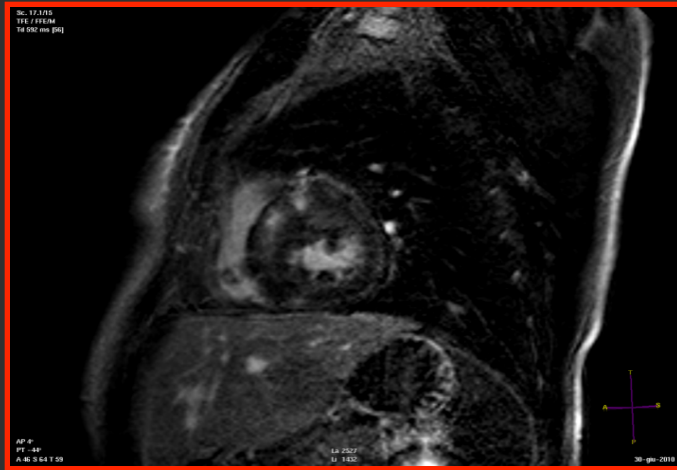


Cardiac magnetic resonance imaging in differentiating underlying causes of LVH

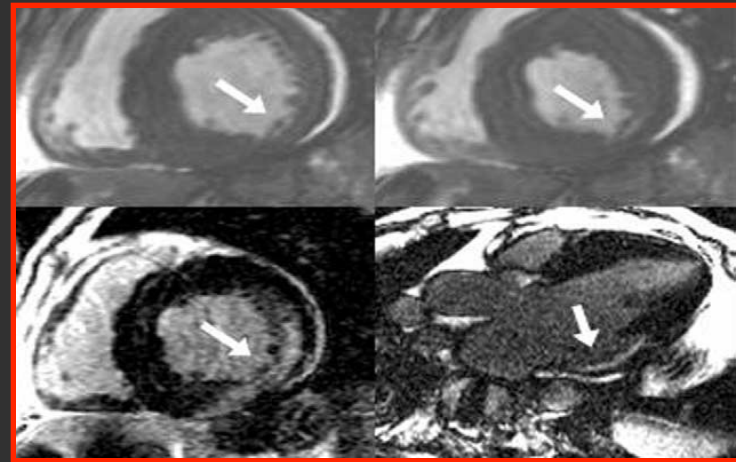


Cardiac magnetic resonance imaging in differentiating underlying causes of LVH

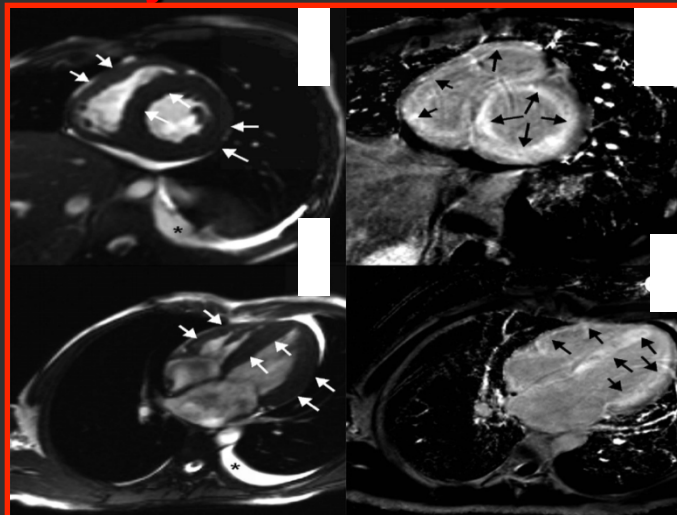
Familiar HCM



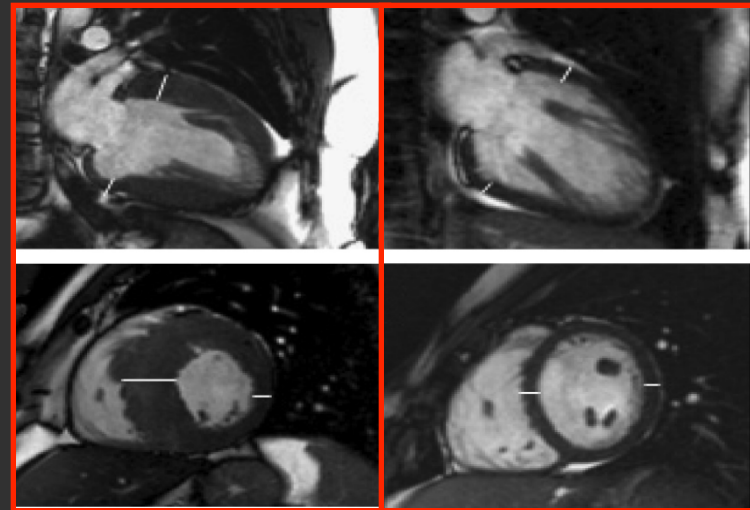
Anderson-Fabry



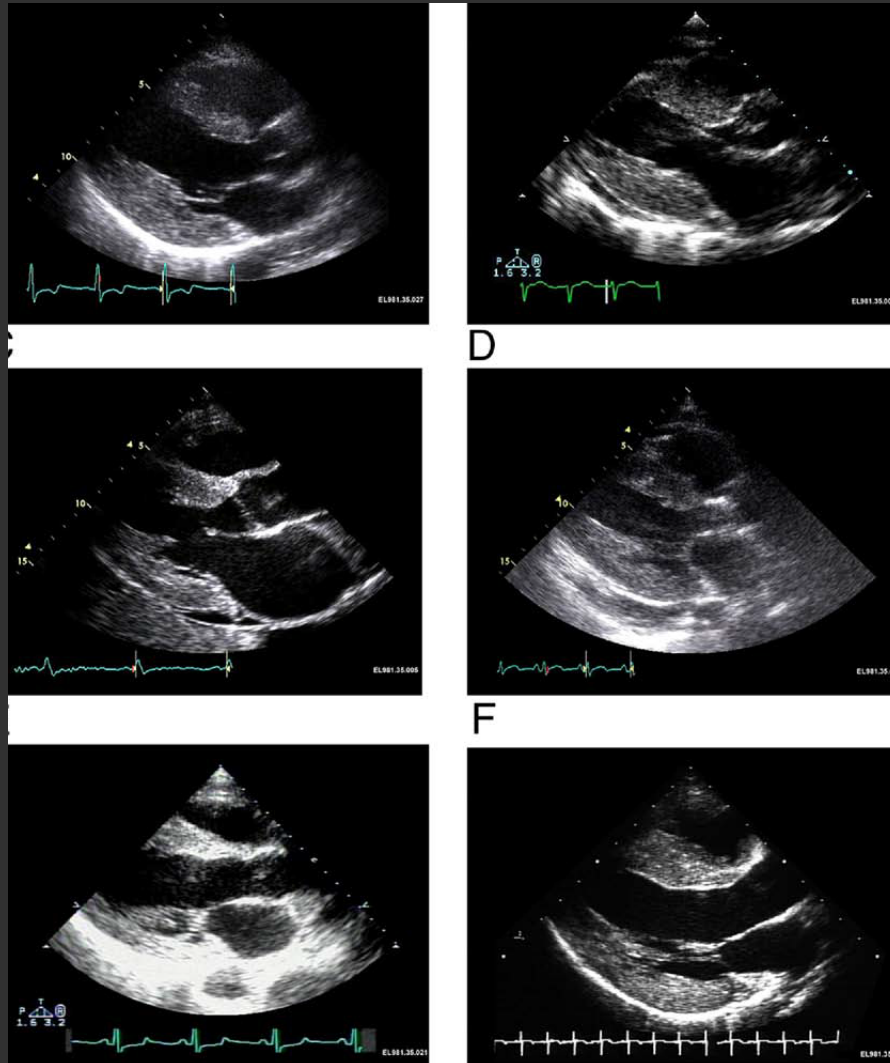
Amyloidosis



Athlete's heart

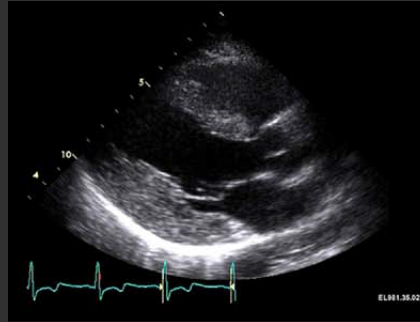


Mimicking the phenotype

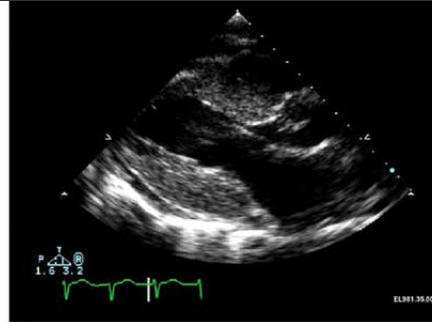


Mimicking the phenotype

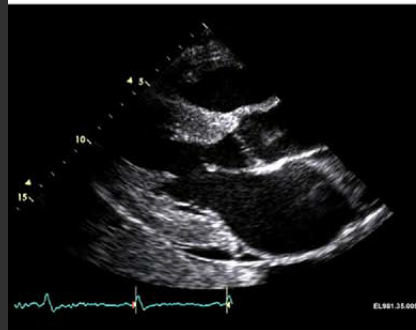
HCM



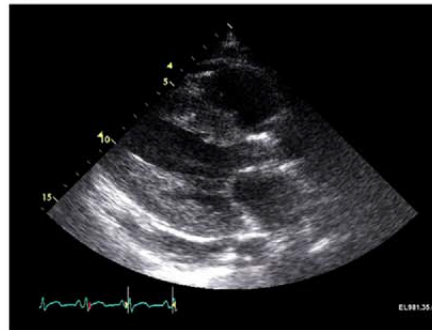
Hypertensive
Heart disease



Amyloid



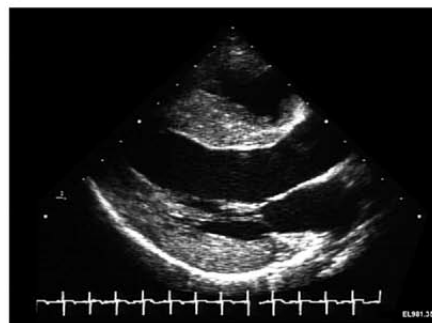
Mucopolisaccaridosis



Cardiac oxalosis

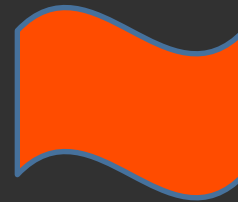


Friedreich ataxia

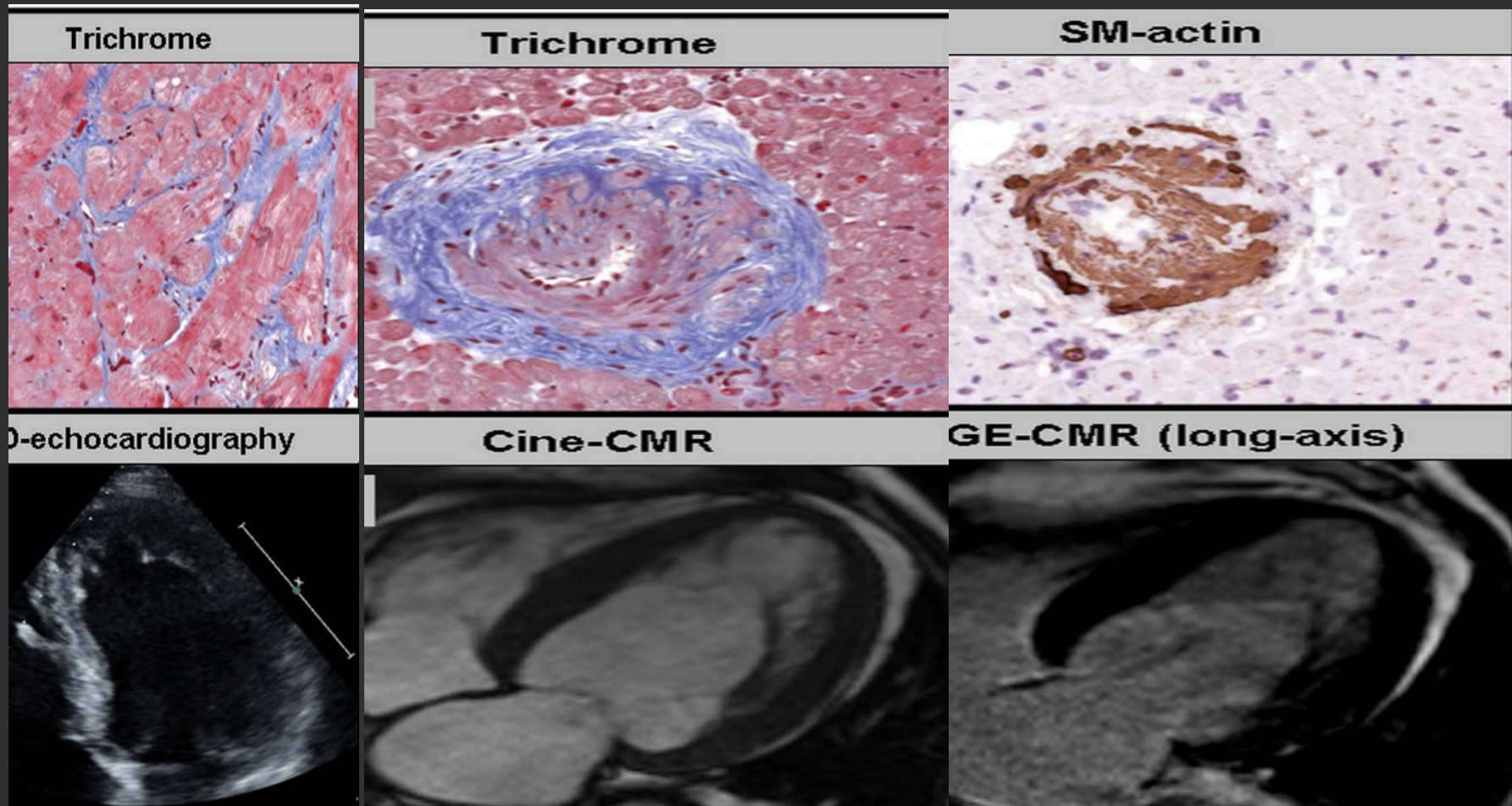


Take Home Message

- LVH is only the beginning
 - EKG and laboratory analysis
 - Use DNA analysis
 - Use BEM
- But do not forget

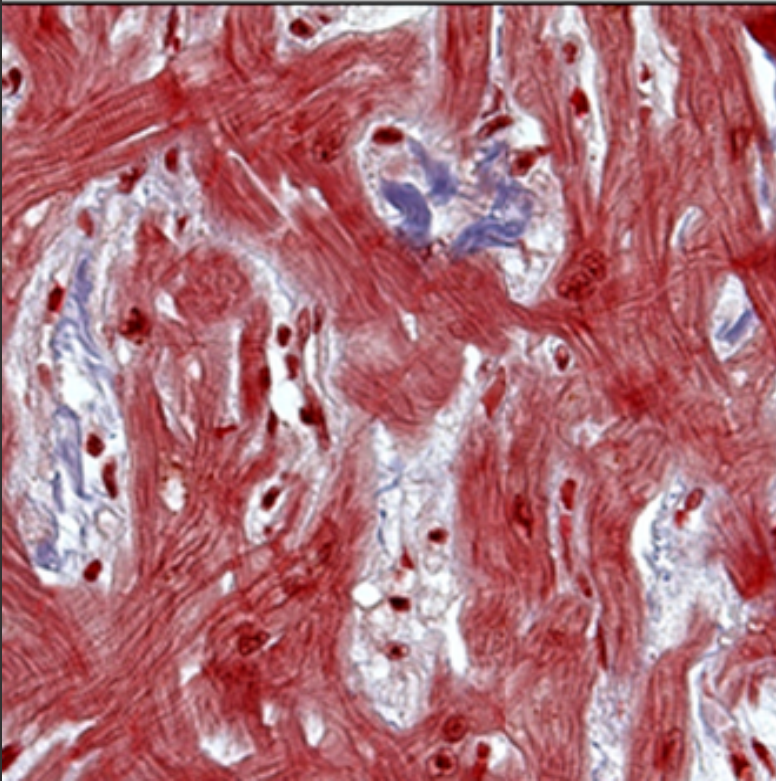


A multiparametric diagnosis for a complex disease using multimodality imaging....

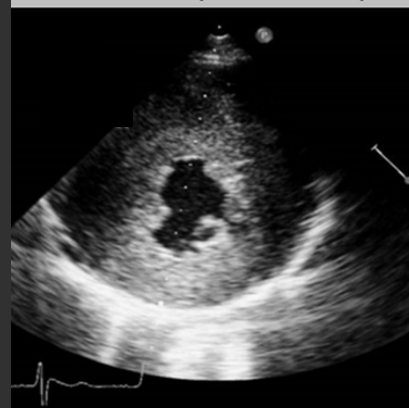


A multiparametric diagnosis for a complex disease using multimodality imaging....

Trichrome



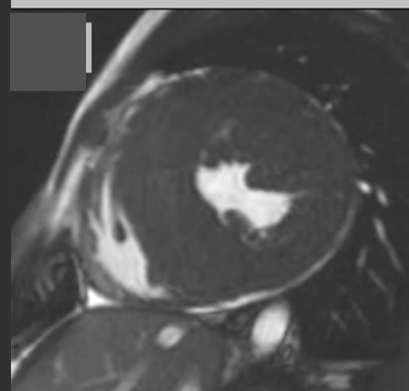
2D-echo (short-axis)



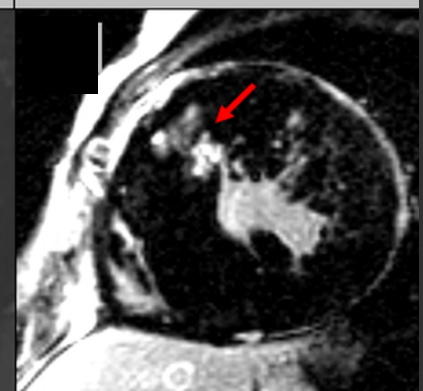
2D-echo (long-axis)



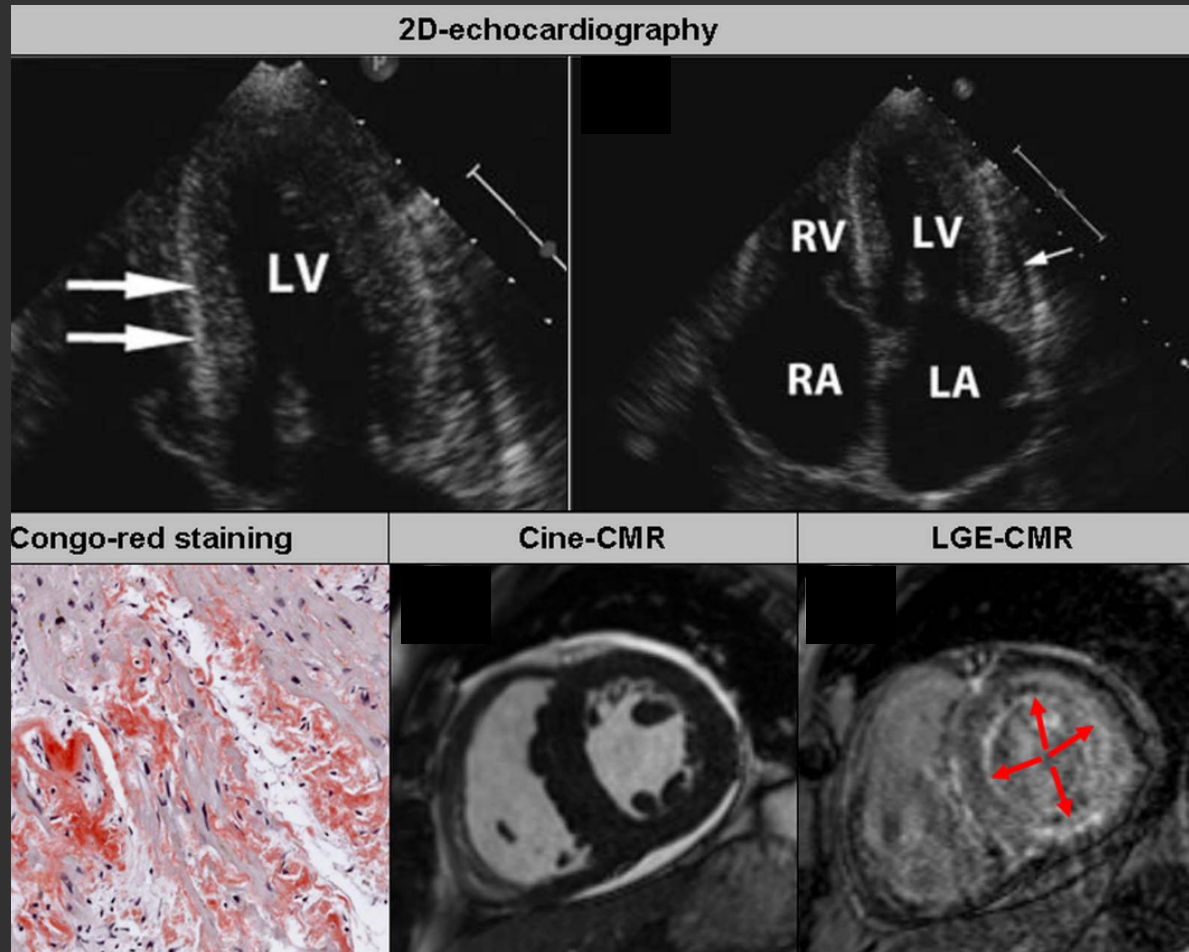
Cine-CMR



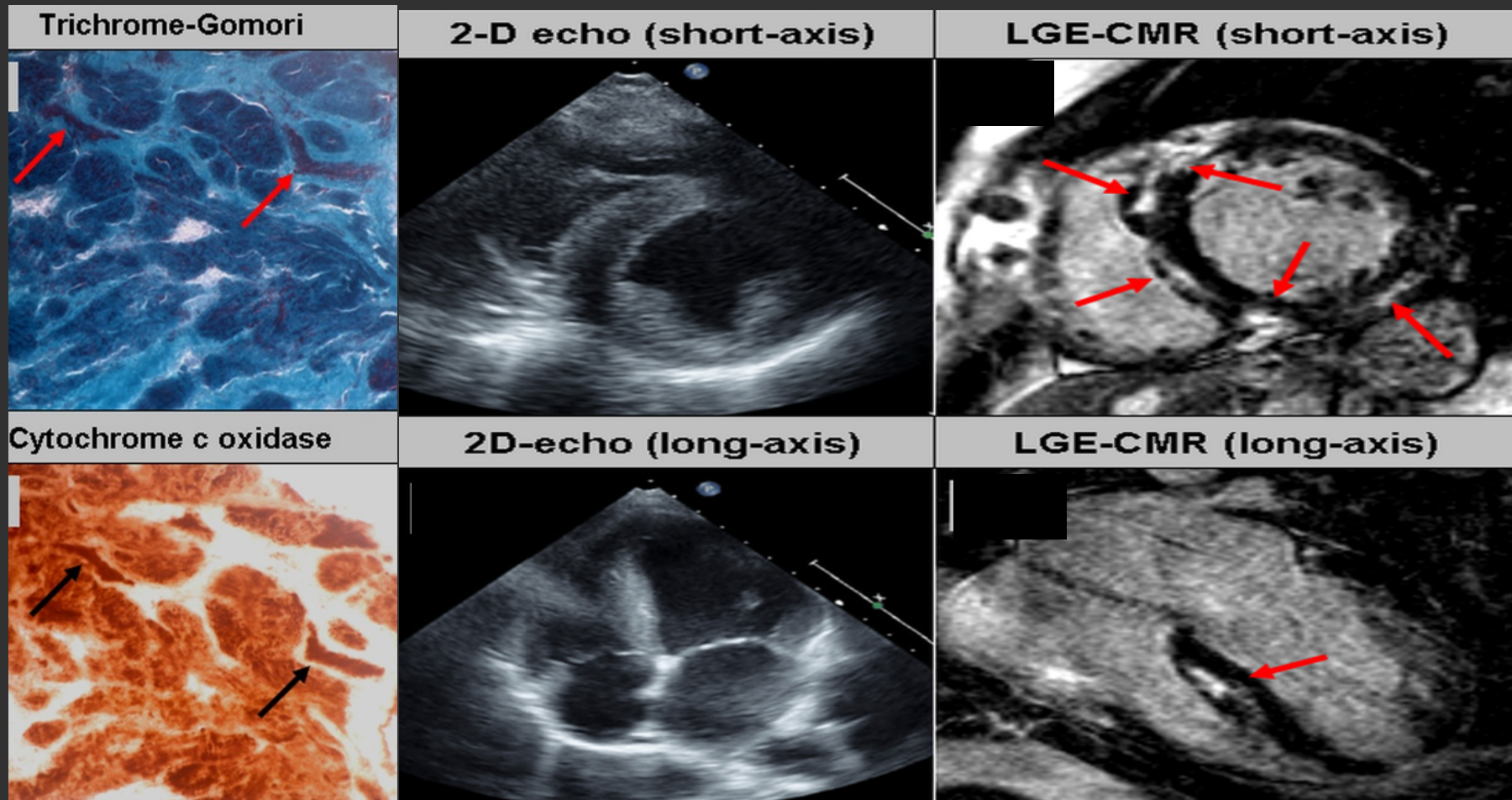
LGE-CMR



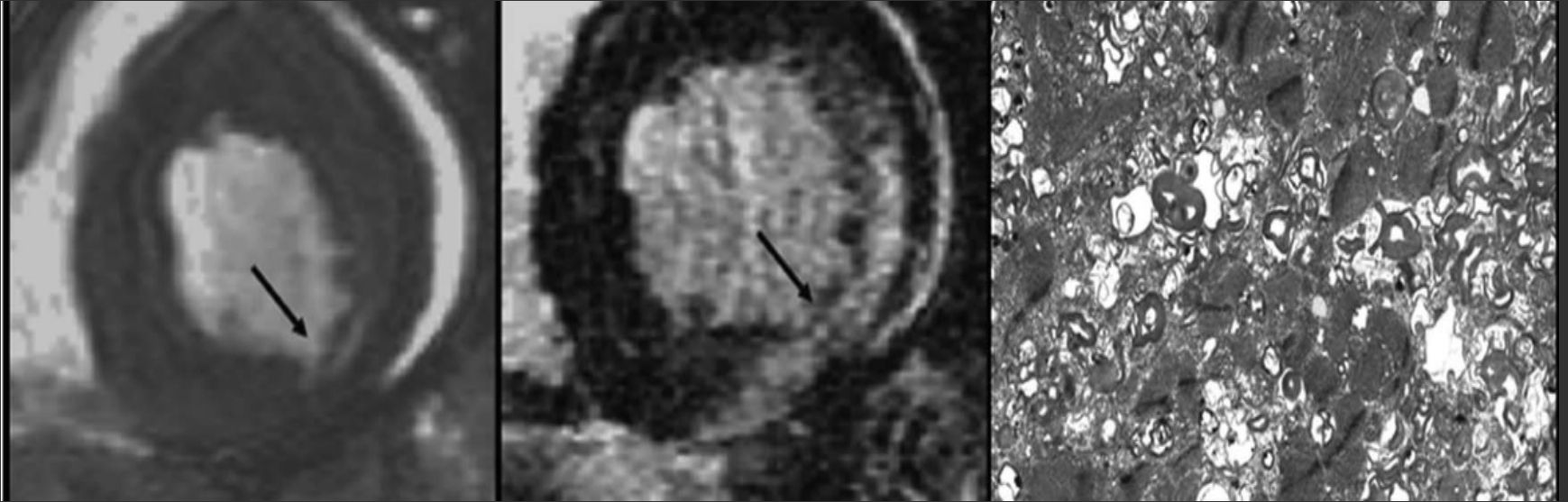
A multiparametric diagnosis for a complex disease using multimodality imaging....



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A multiparametric diagnosis for a complex disease using multimodality imaging....



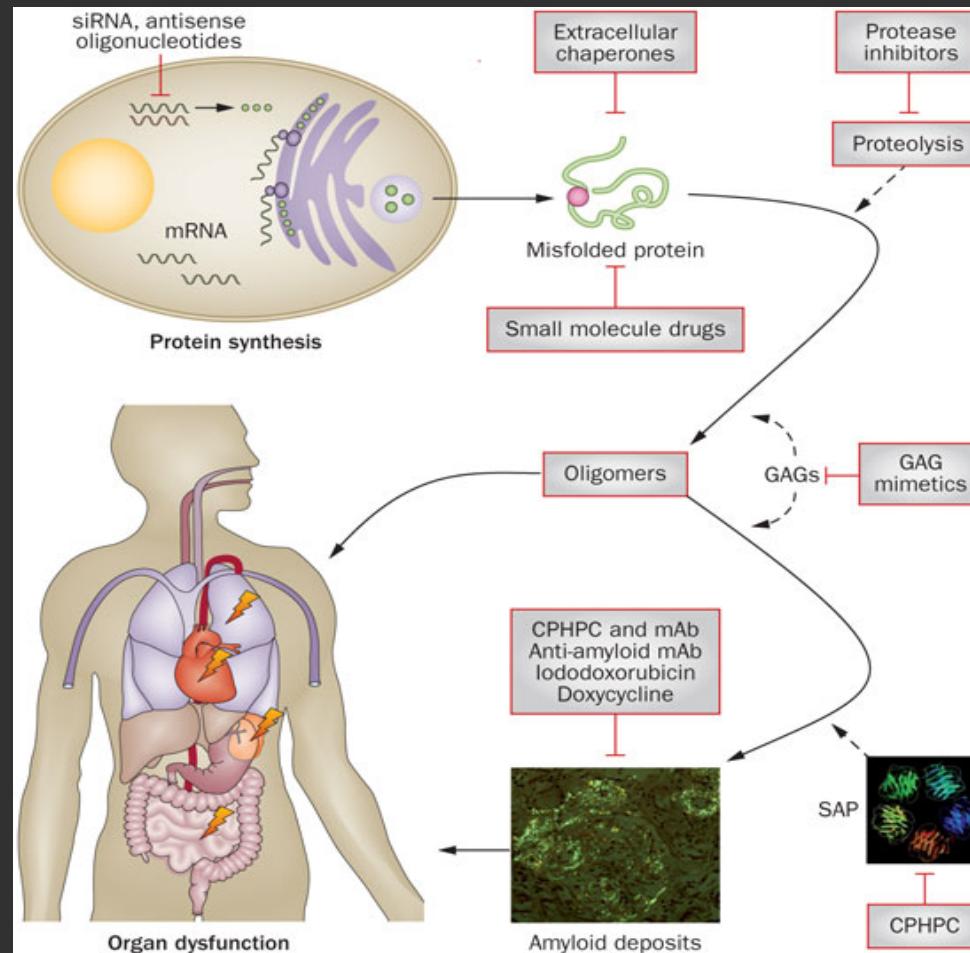
.....to get a specific therapy

CHT and autologous
stem cell
transplantation in AL
amyloidosis

Tafamidis in TTR

Small RNA
interfering
molecules

Diflunisal in TTR



Doxiciclina