



# Childhood cardiomyopathies

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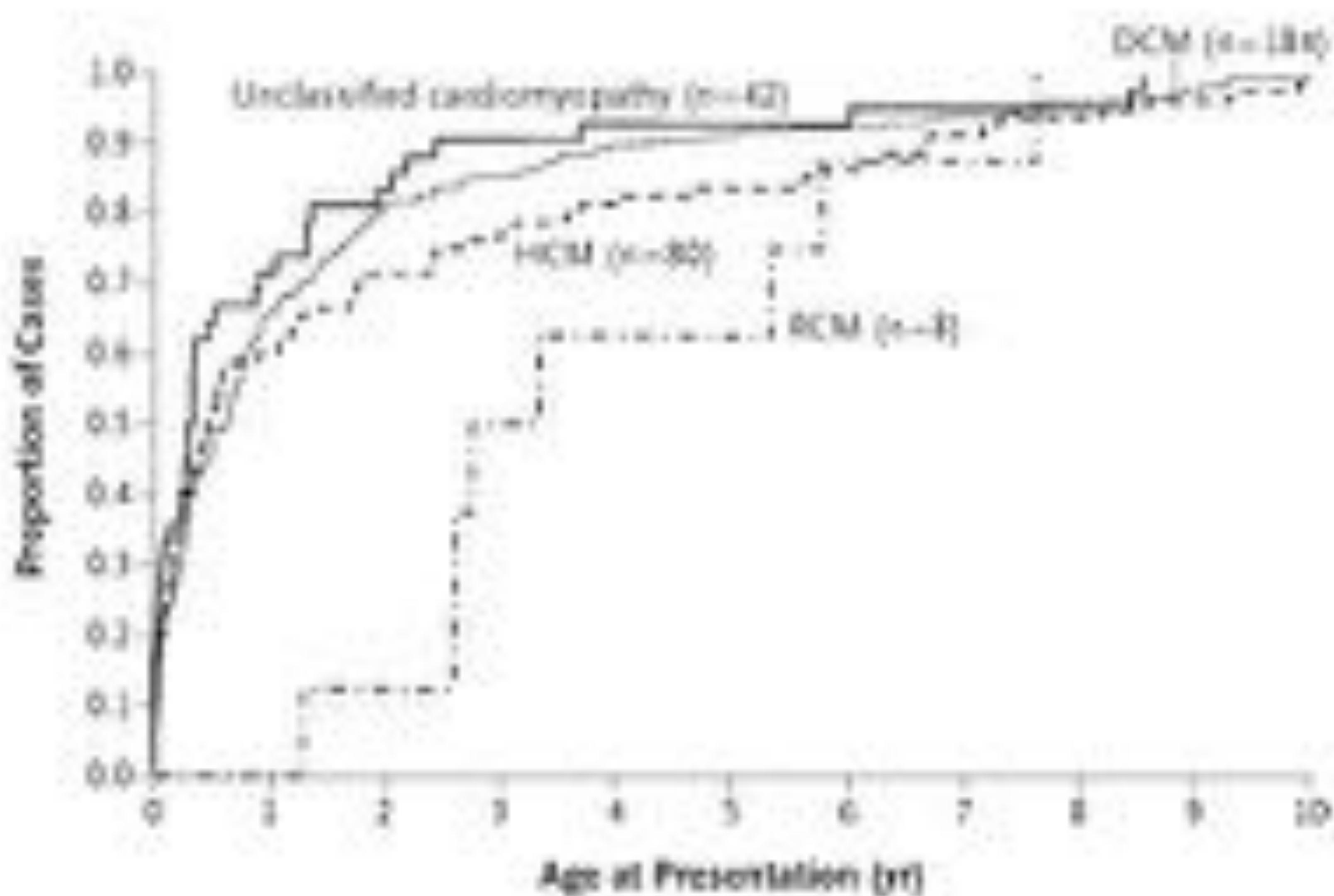
Centre de Référence Maladies Rares  
Malformations Cardiaques Congénitales Complexes-M3C  
Centre de Référence Maladies Rares  
Maladies Cardiaques Héréditaires- CARDIOGEN



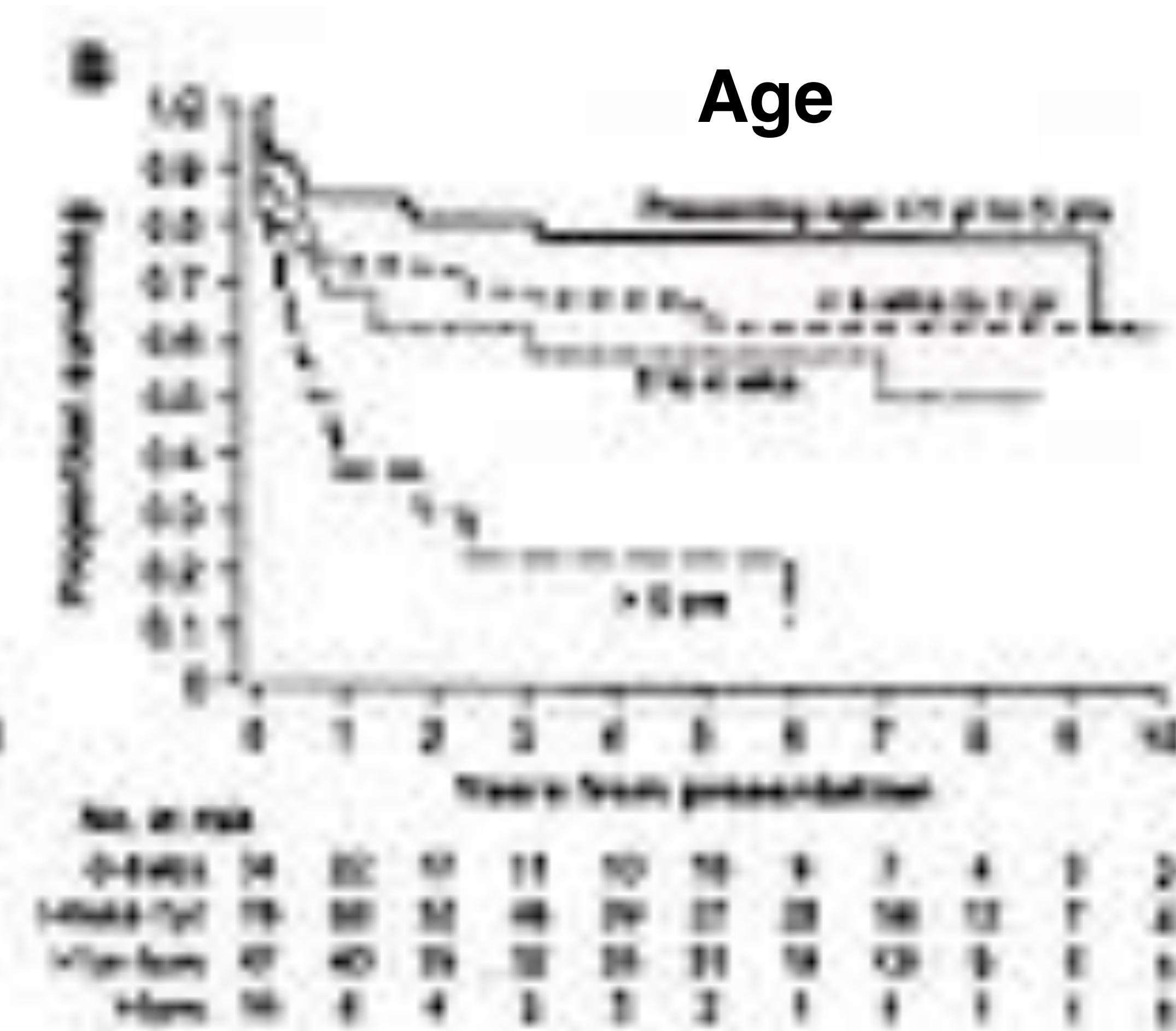
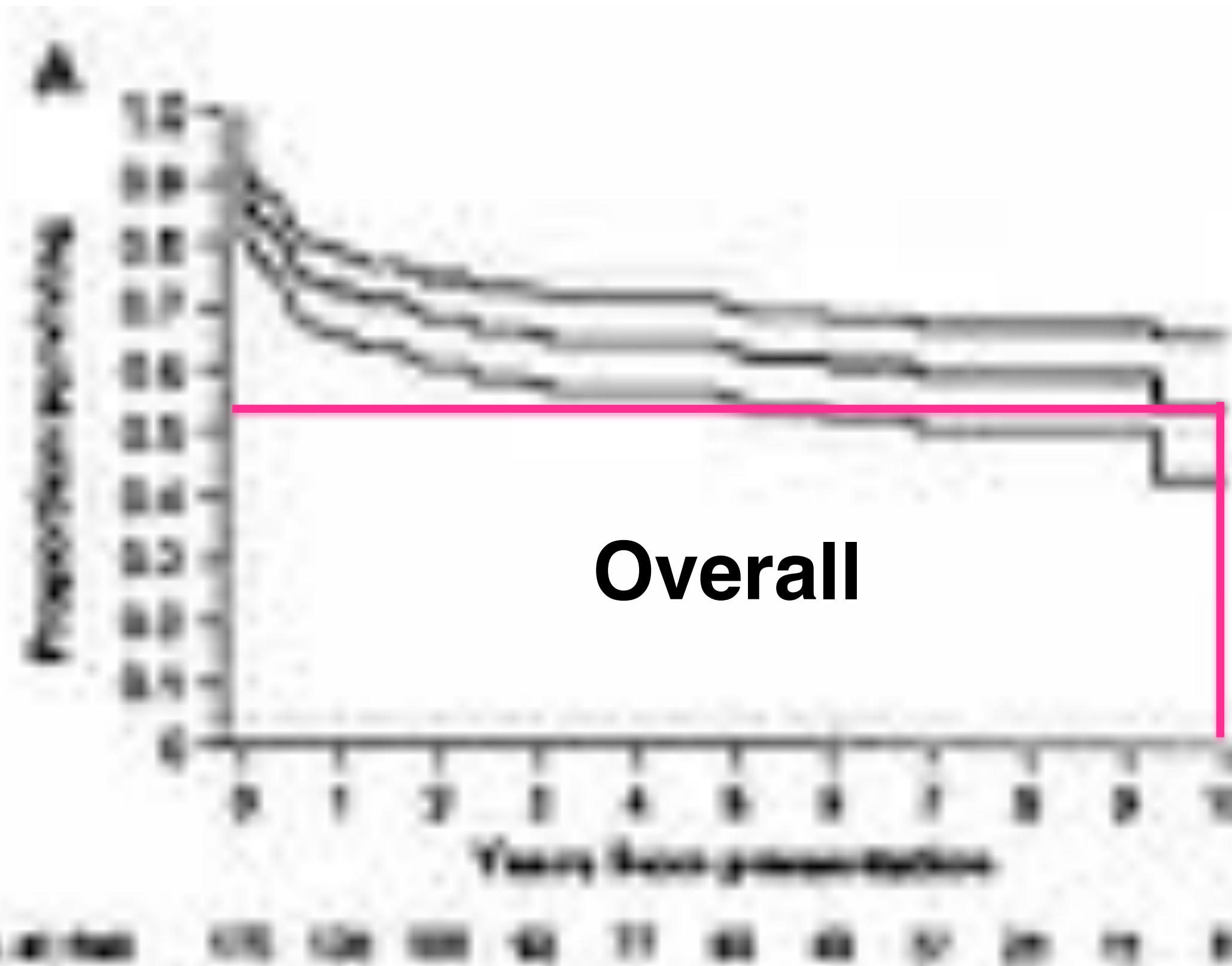
# Epidemiology

- Annual incidence of childhood cardiomyopathies : 1.13 per 100,000
- Incidence higher among children <1 year :  
8.34 vs. 0.70 per 100,000
- Categorized according to type :
  - Hypertrophic 42 %
  - Dilated 51%
  - Restrictive 2.5%
  - Non compaction 9.2%
- Sudden death as presenting symptom 3.5%

# Cumulative frequency distribution of age at presentation



# Survival to death or transplantation from time to presentation in pediatric cardiomyopathies



Daubeney PEF et al. Circulation 2006  
Alexander PMA et al. Circulation 2013

Wittlieb-Weber CA et al. J Card Failure 2015;21:76-82  
Rossano JW et al. J Card Fail 2012;18:459-70.

# **Evidence-based data for the treatment of chronic heart failure in children is scarce**

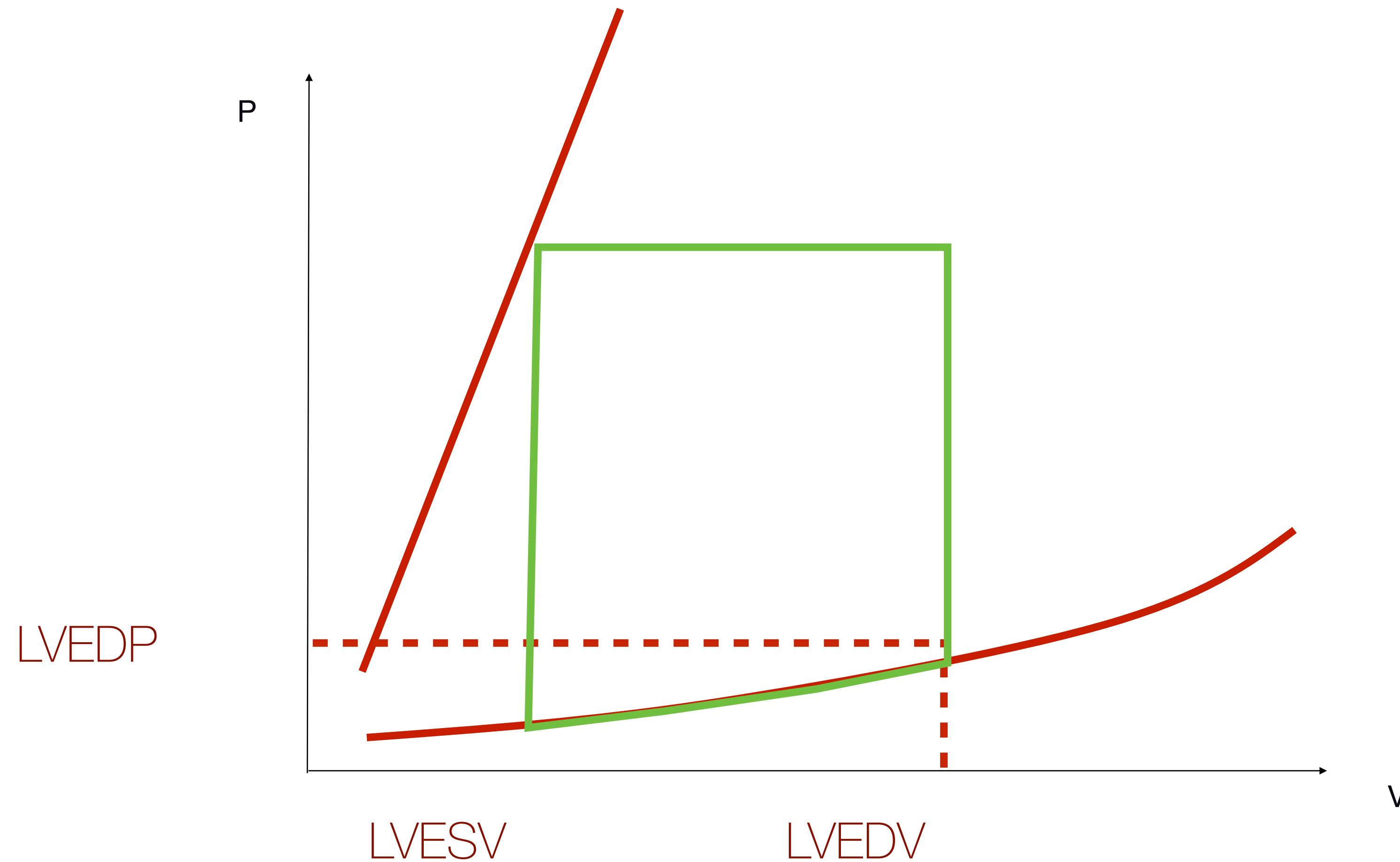
## **Informations from registries**

- 1-Chronic heart failure due to cardiomyopathies has a high mortality
- 2-*Survival depends on age at presentation*
- 3-Cause is a major predictor of outcome
- 4-*Recovery of left ventricular function is possible*
- 5-A child with HF admitted to the hospital has an over 20-fold increase in the risk of death compared to a child without HF
- 6-*Children with cardiomyopathy hospitalized with HF have significantly increased morbidity, mortality, and resource utilization compared to adults.*

## **Informations from clinical trials and guidelines**

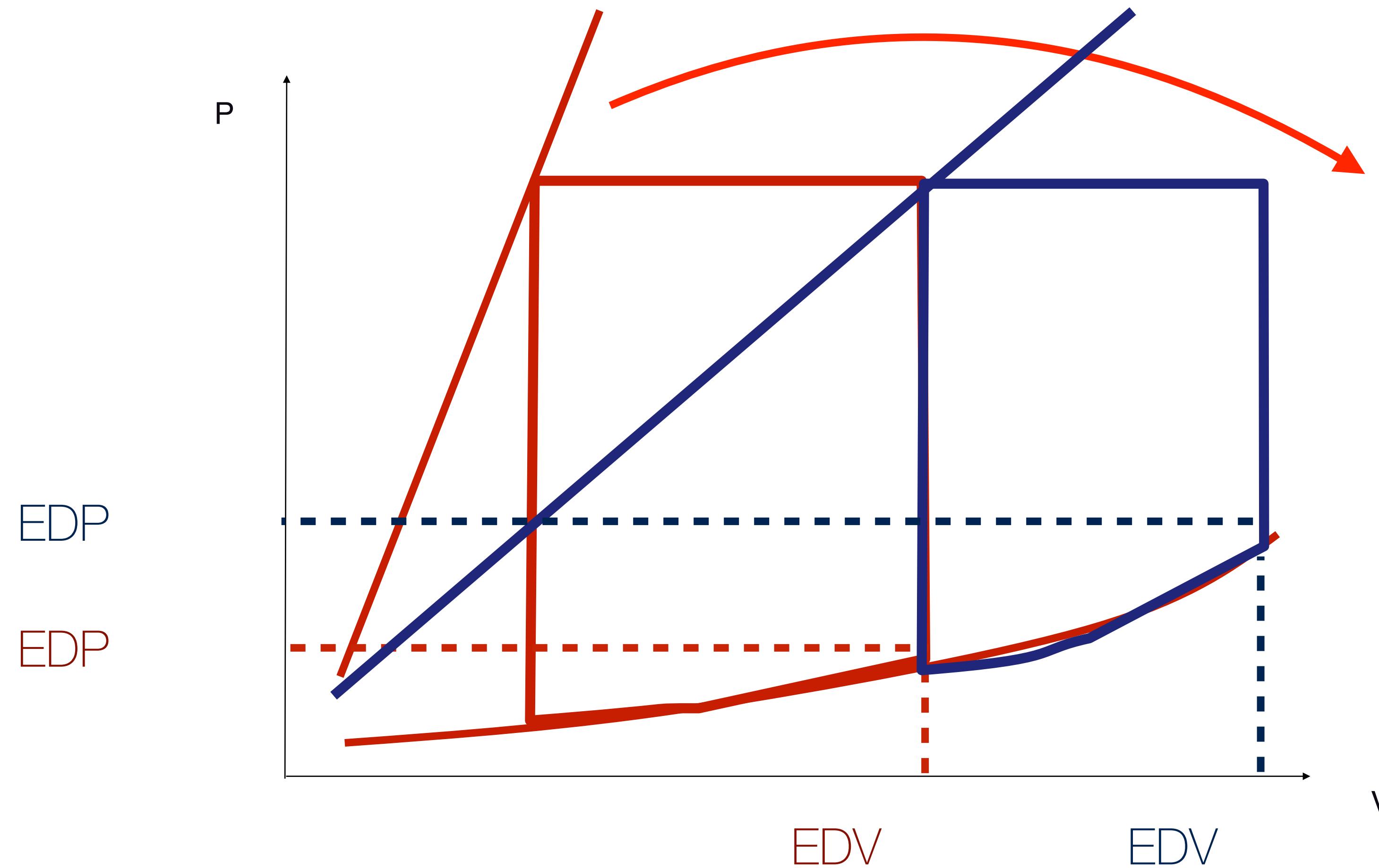
- 1-No RCT has ever proven efficacy of any drug on mortality in pediatric heart failure
- 2-*Guidelines level of evidence are Level C*

# Pressure volume curve



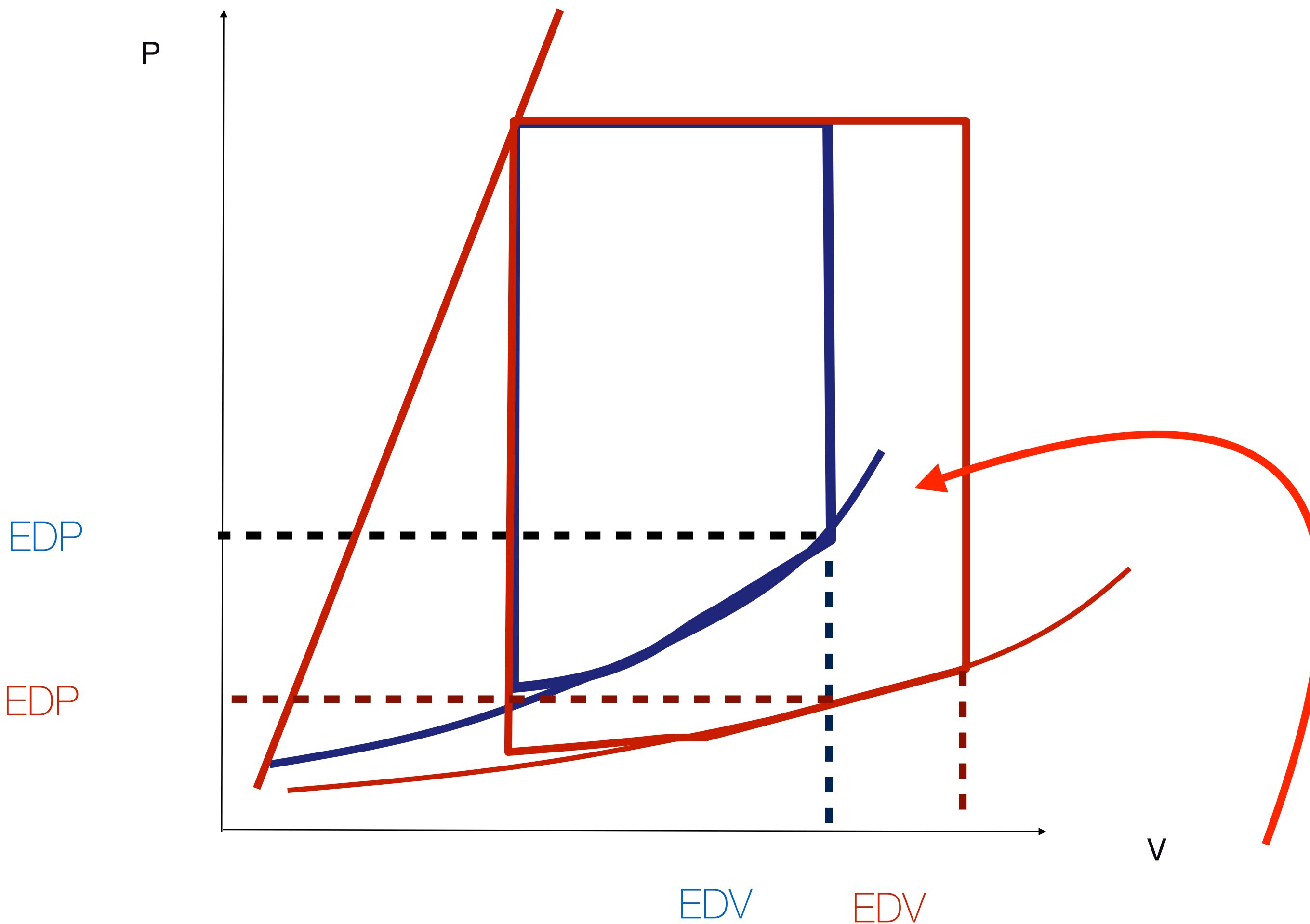
# Systolic dysfunction

## Low contractility

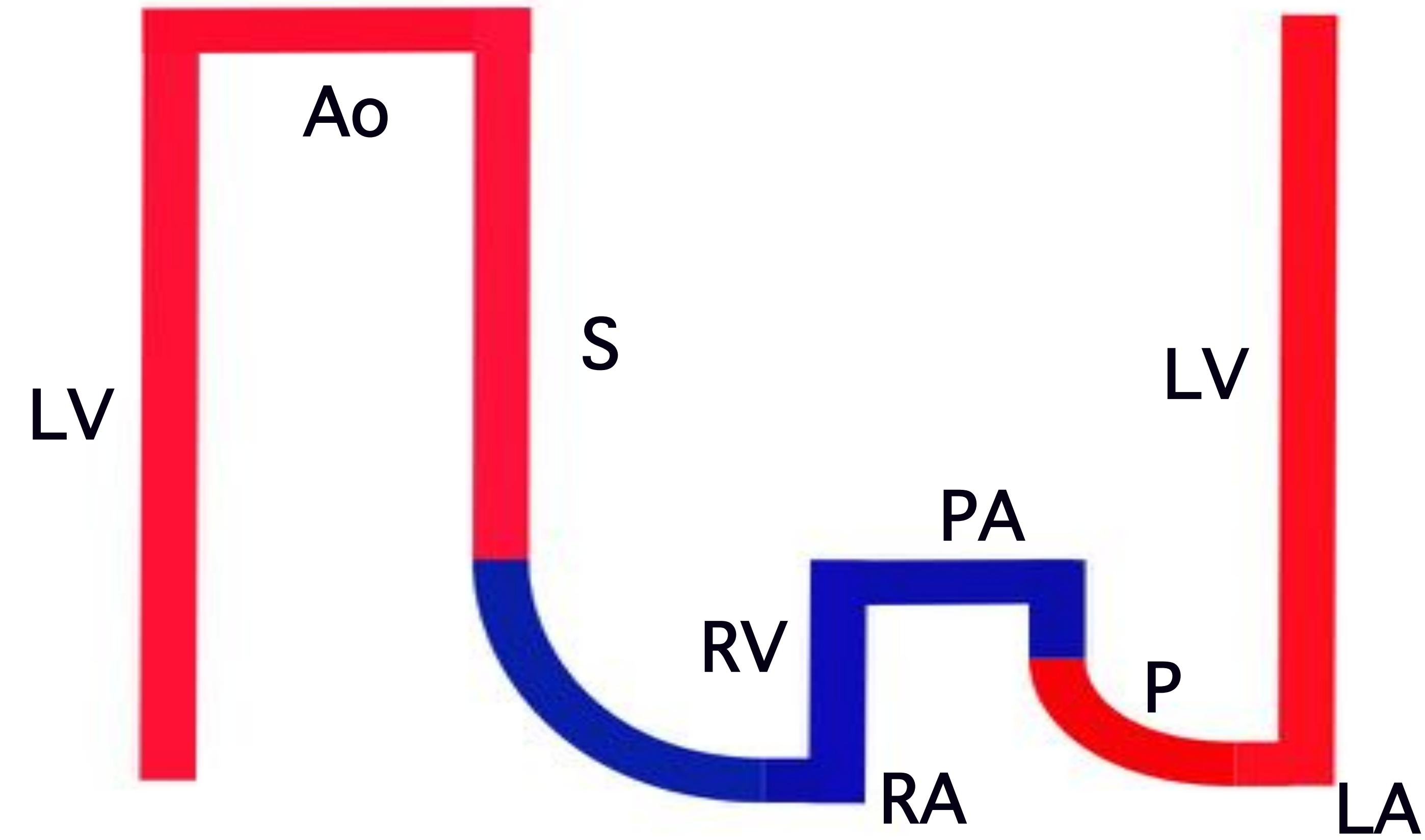


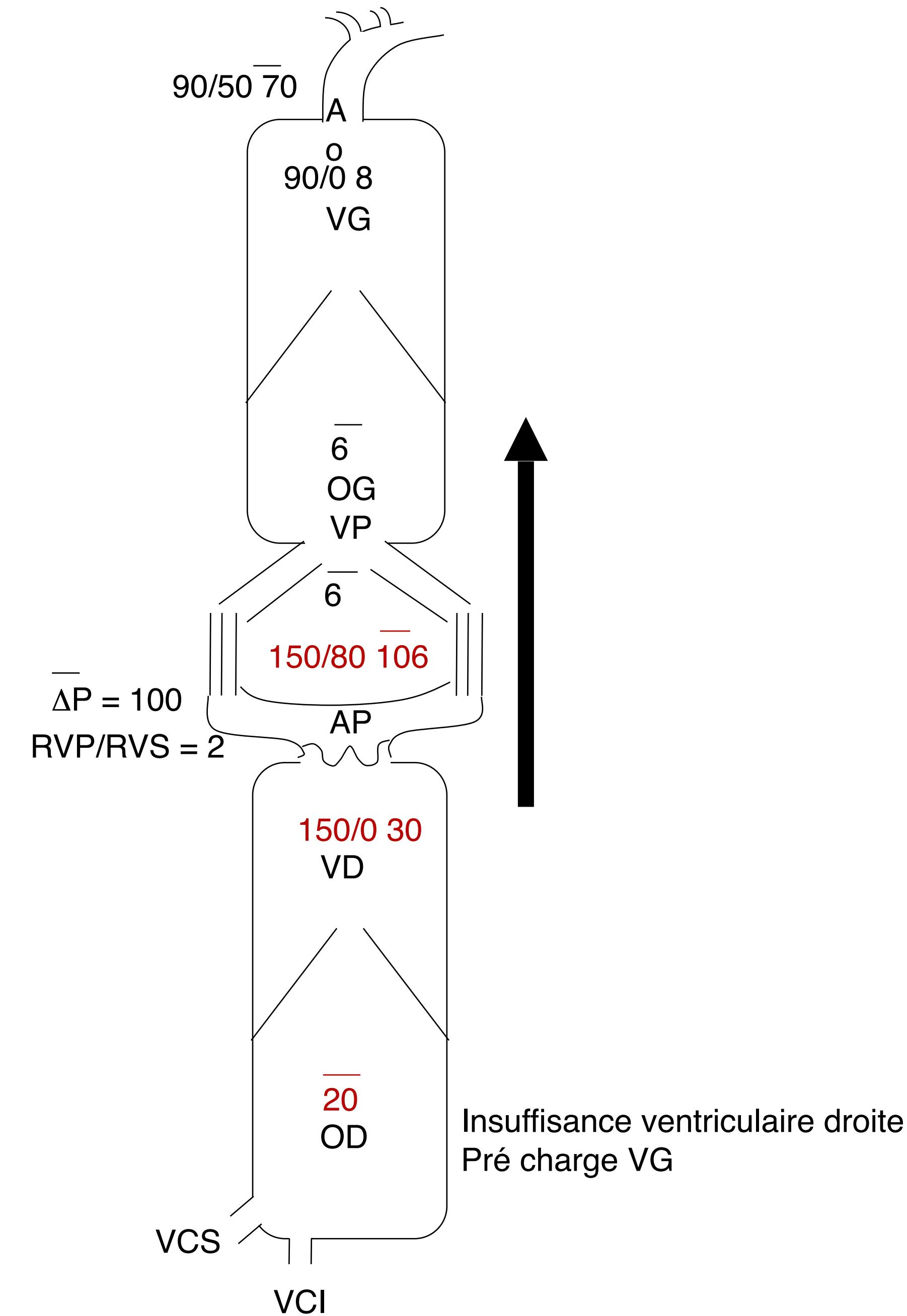
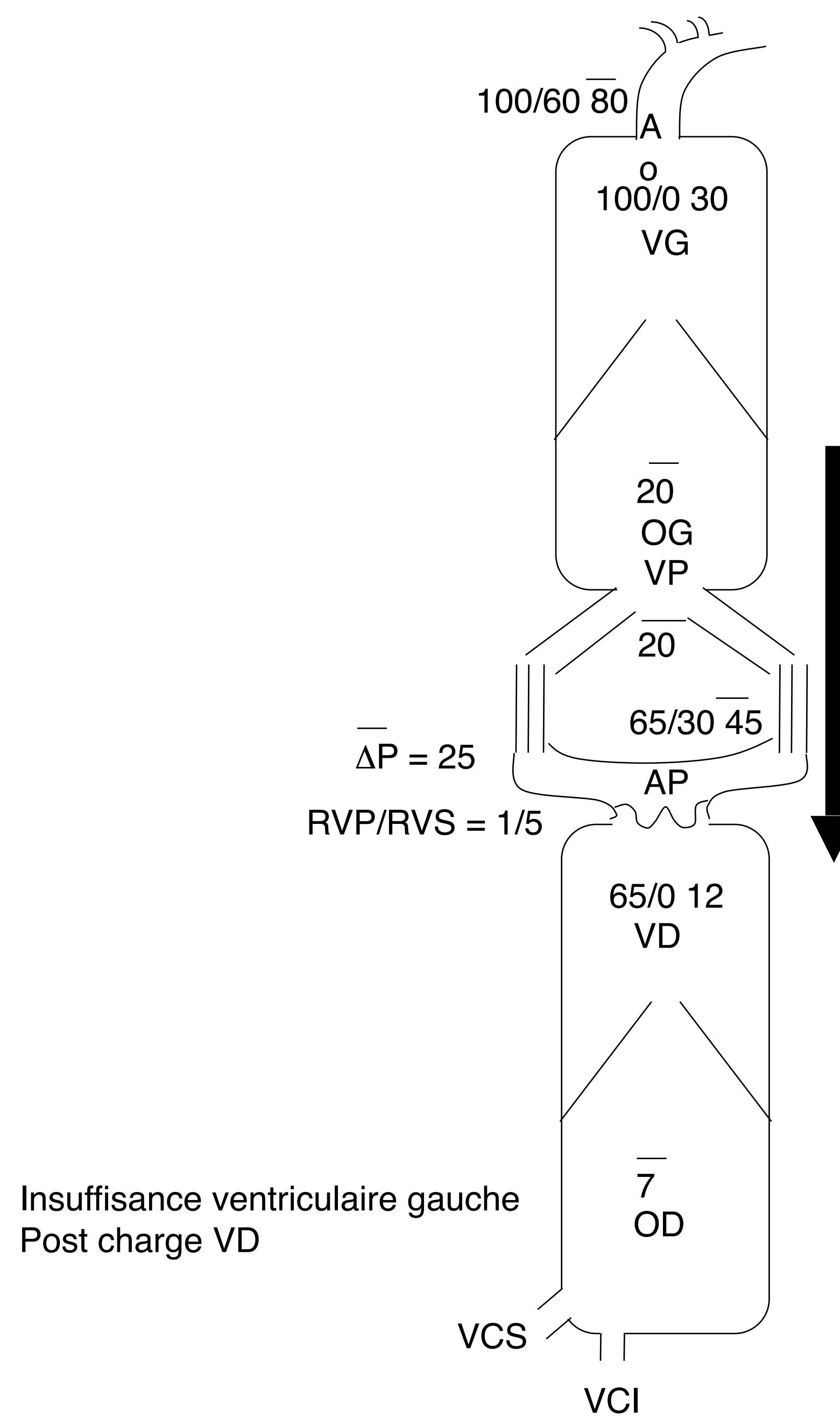
# Preserved EF heart failure

## Low compliance



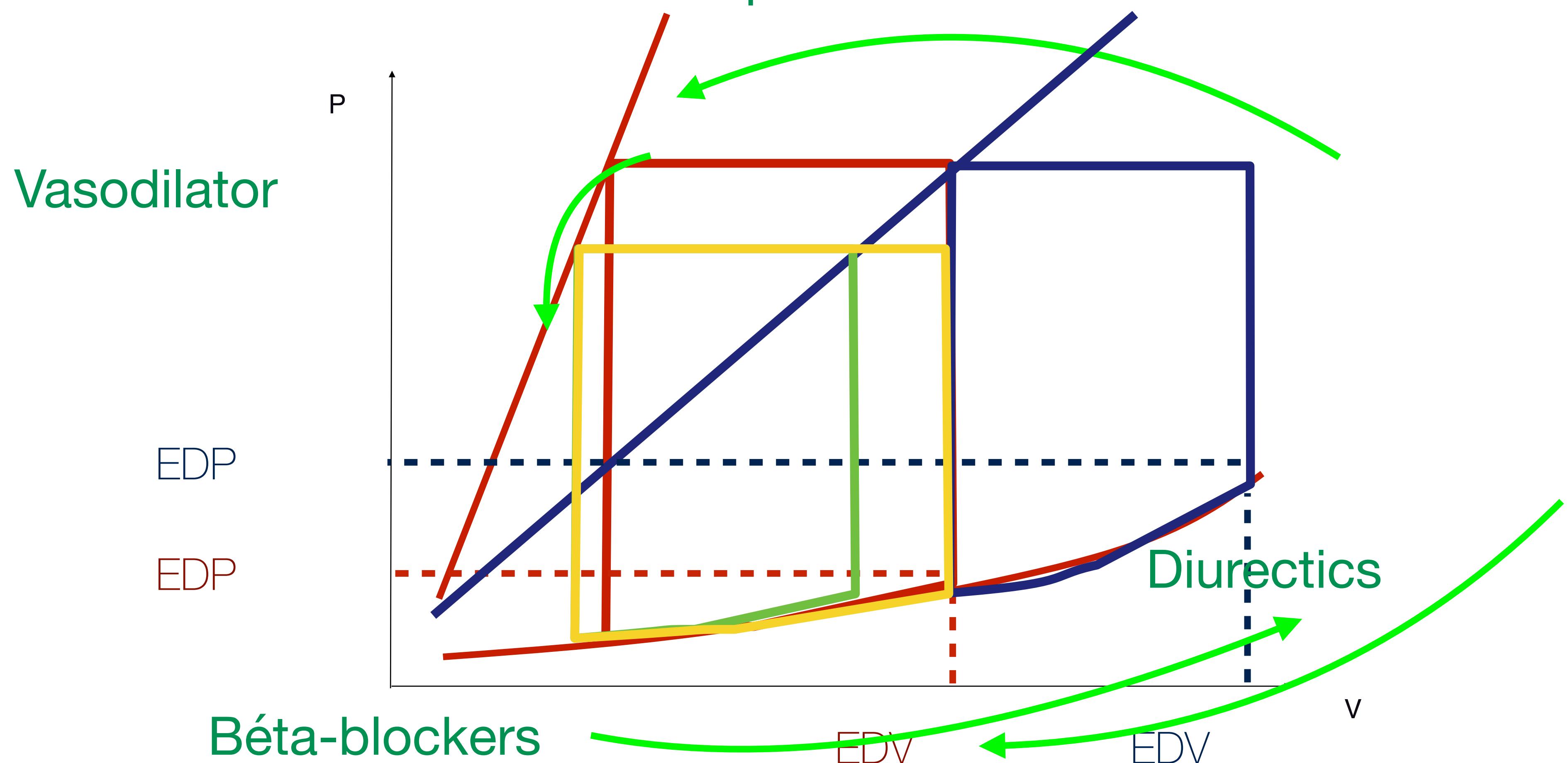
# Schéma hémodynamique du Cœur en Série



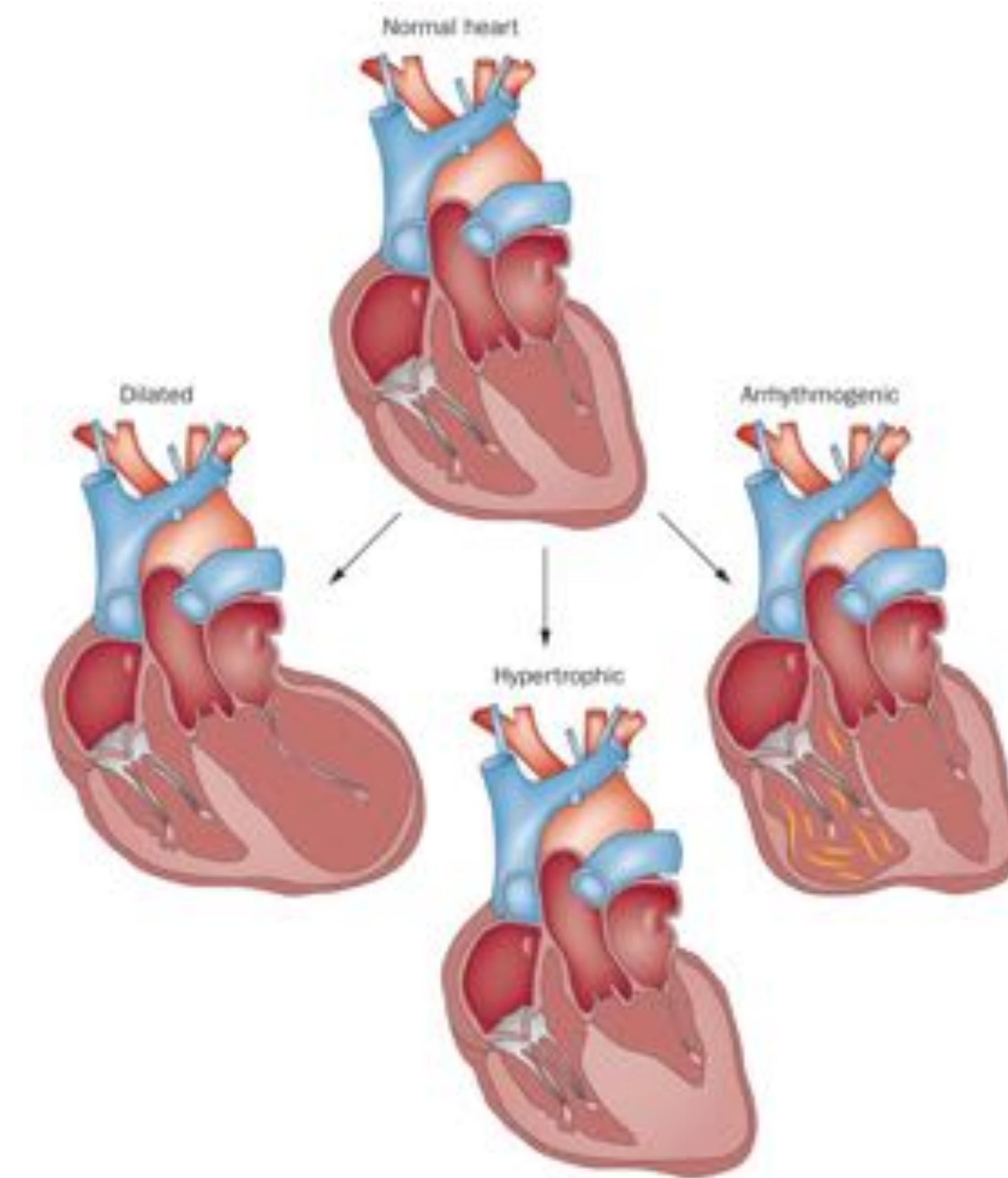


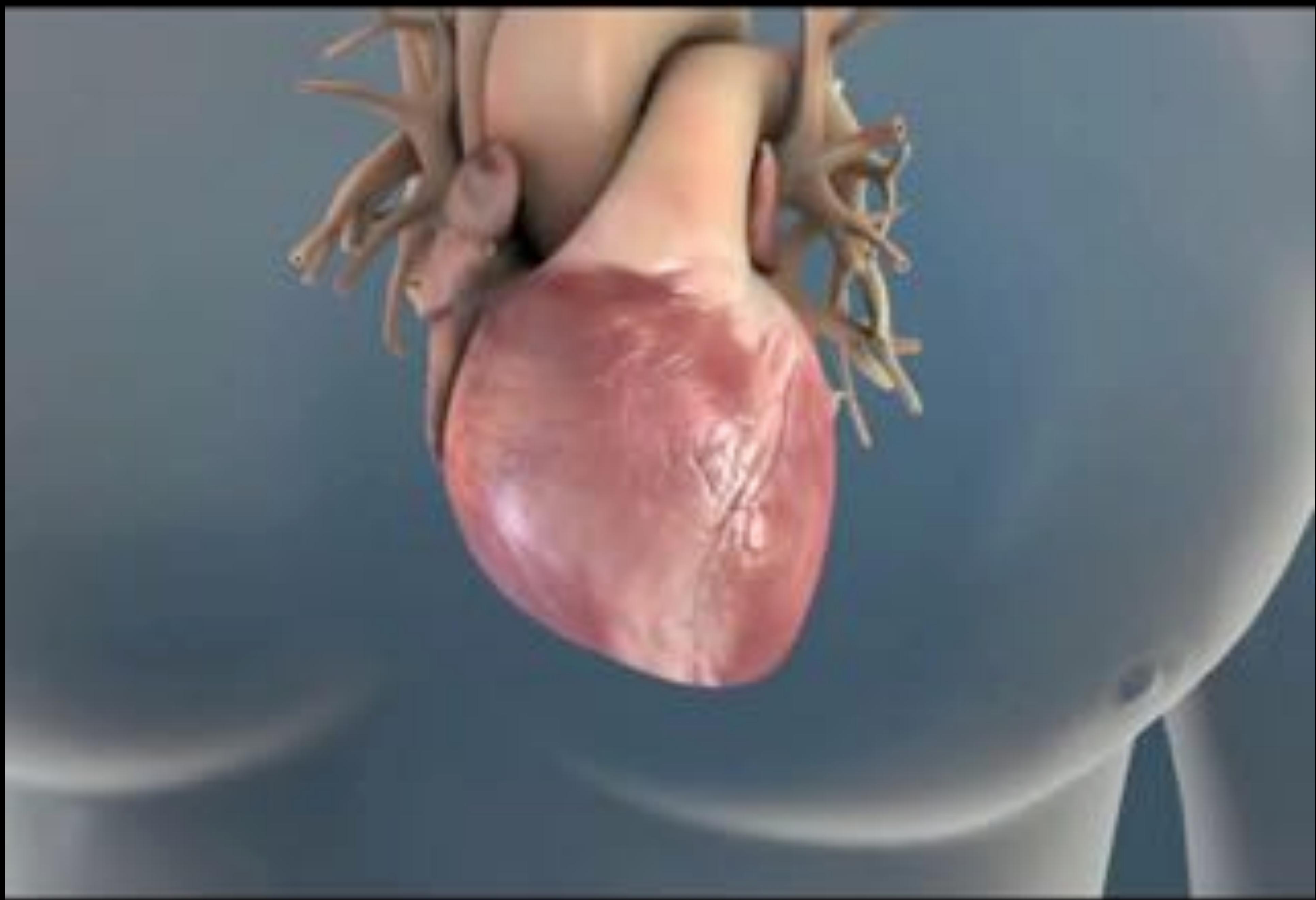
# Systolic dysfunction

## Low contractility/treatment

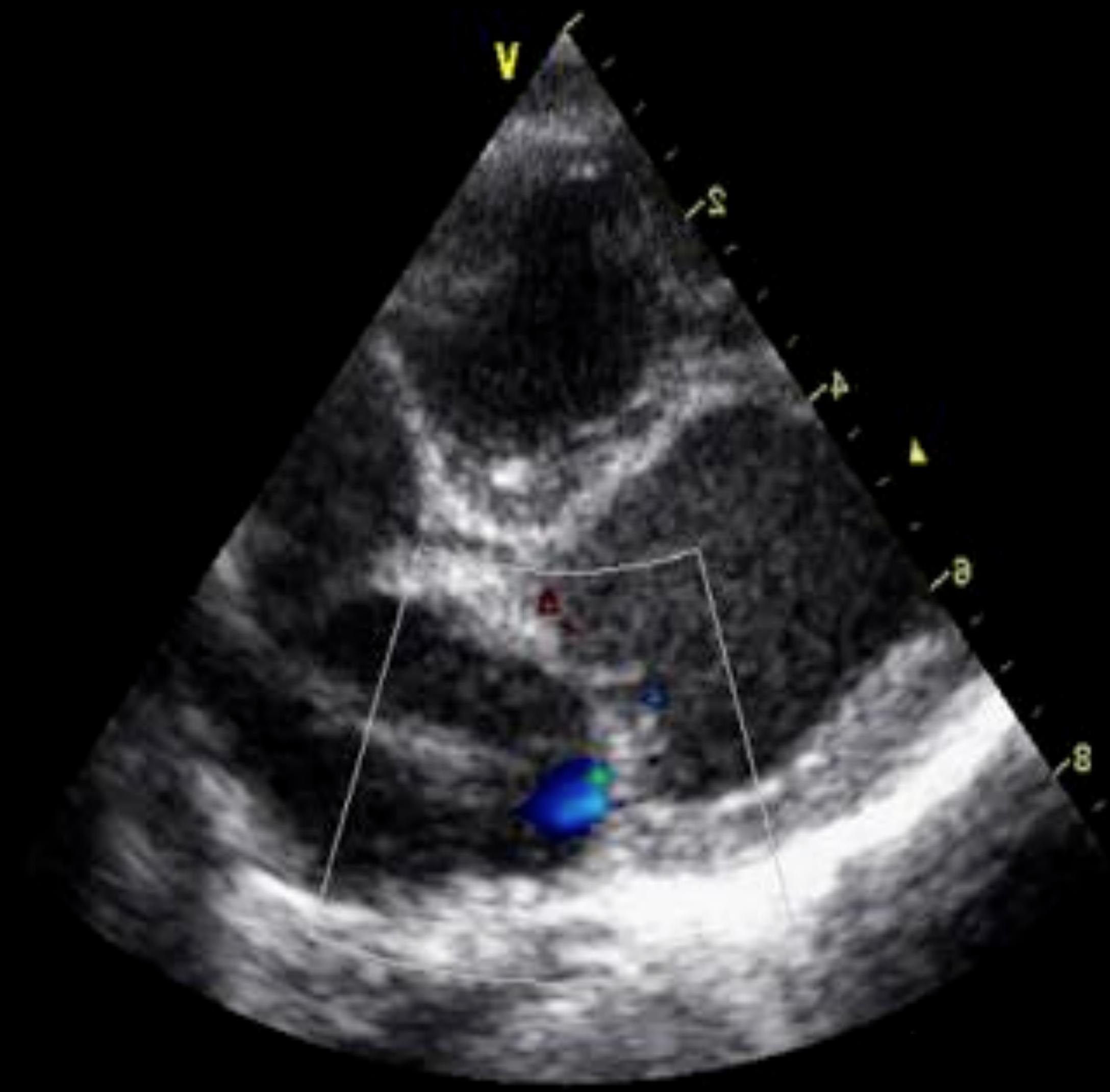
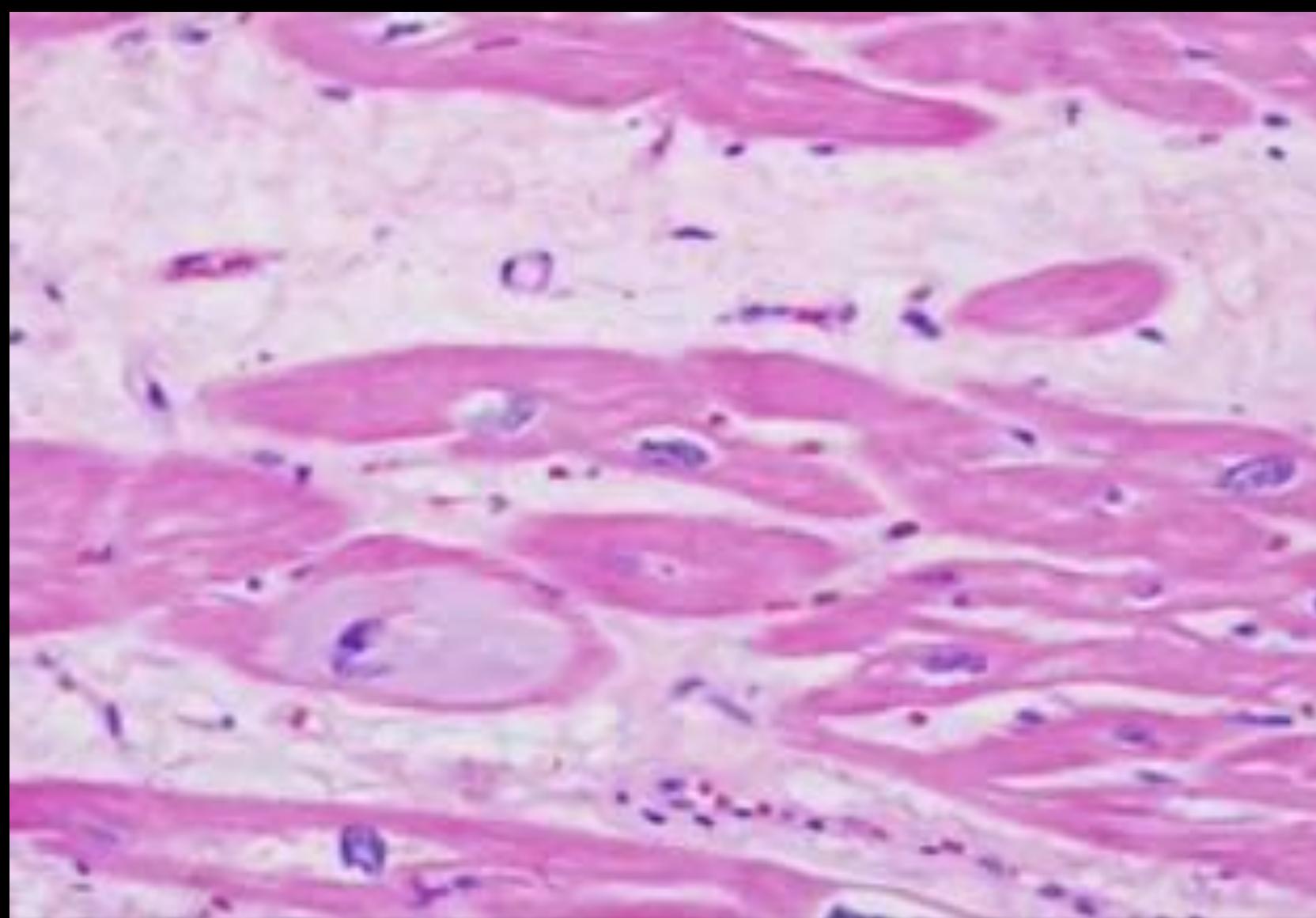
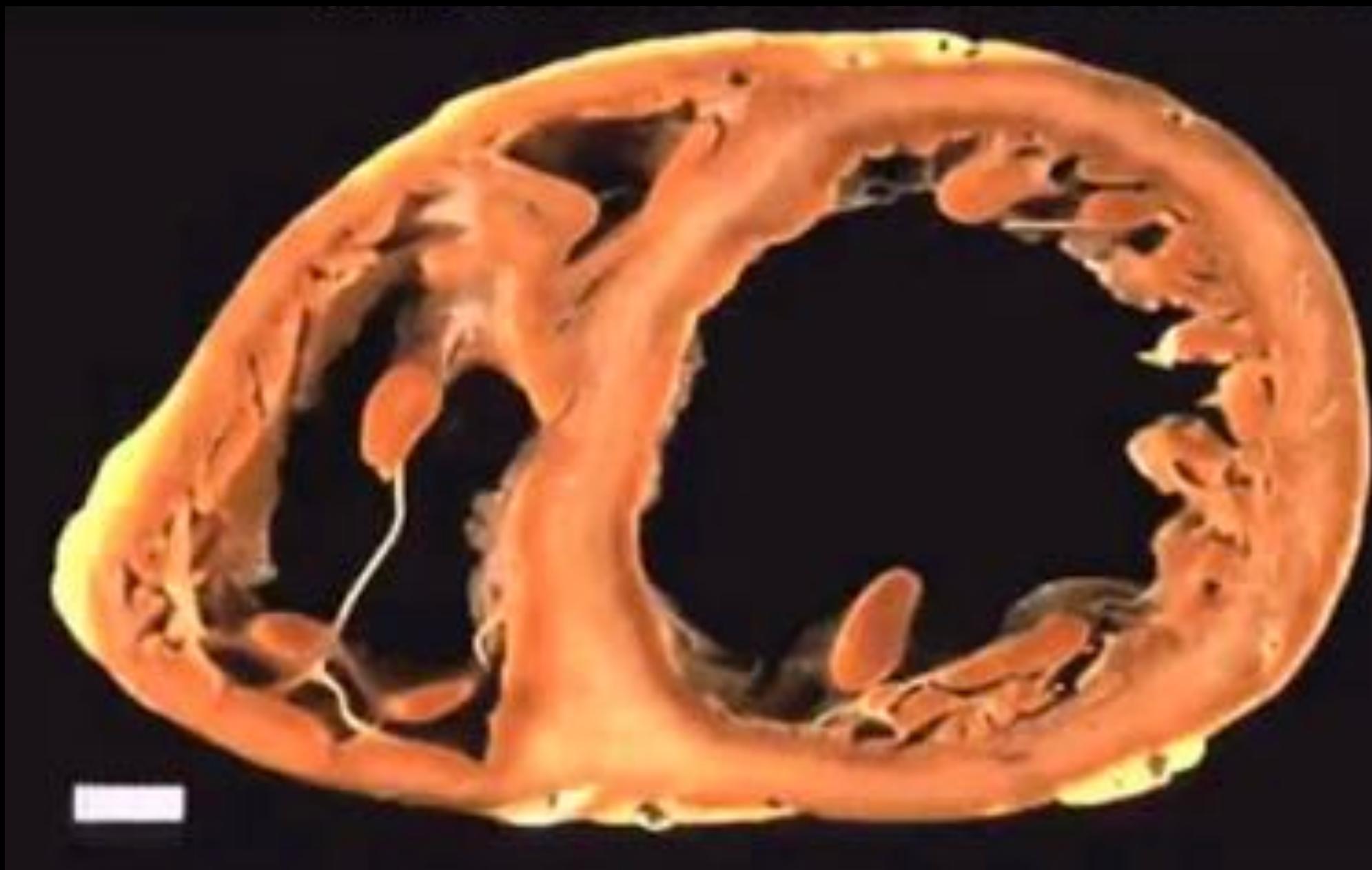


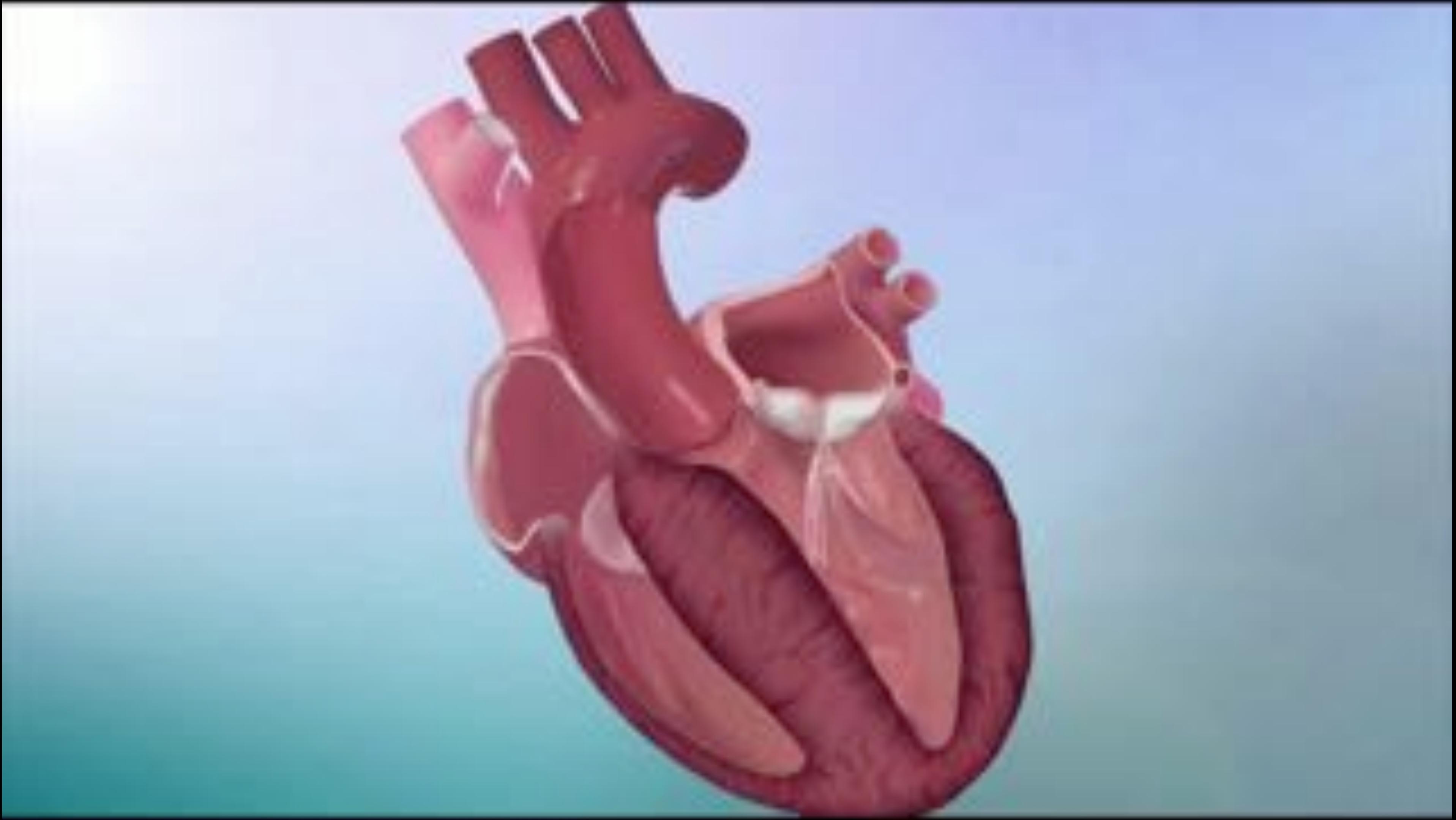
# Cardiac phenotypes



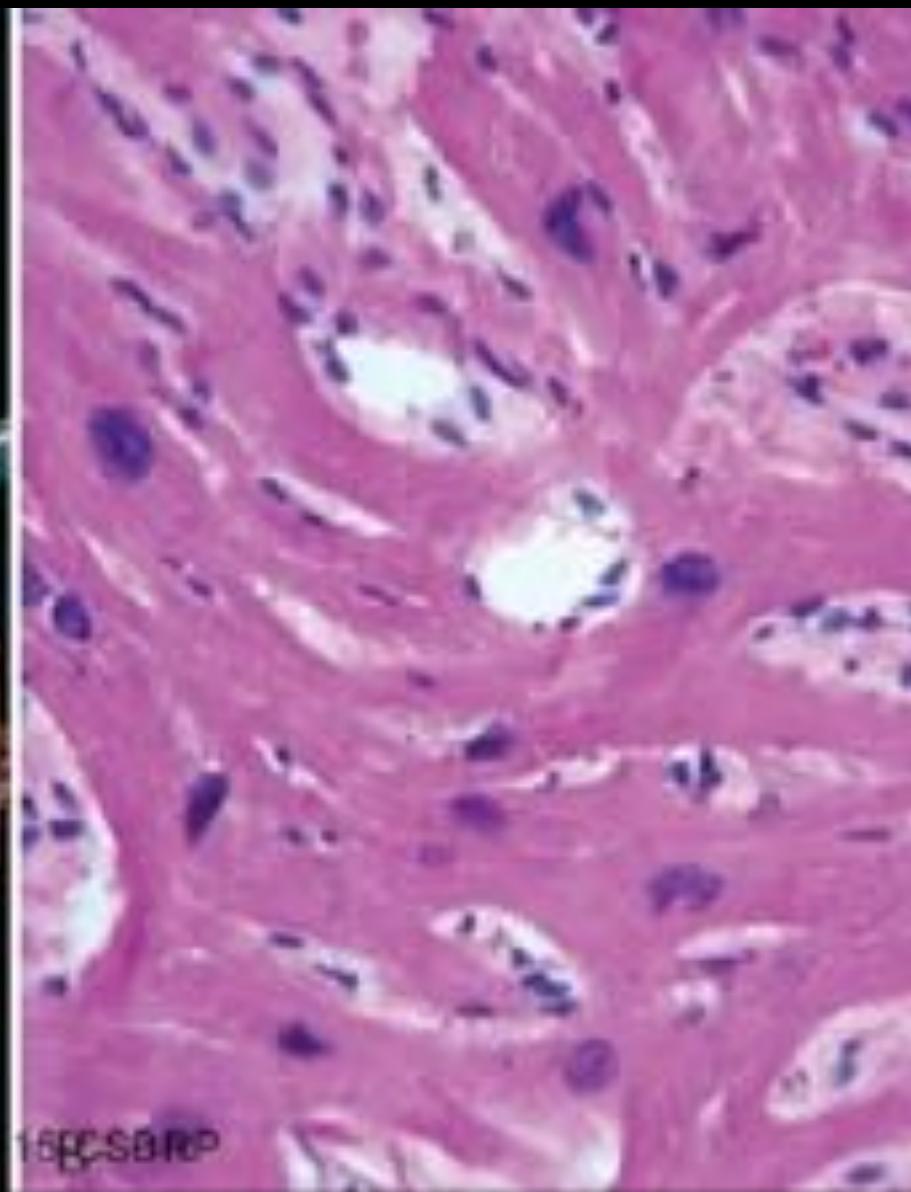
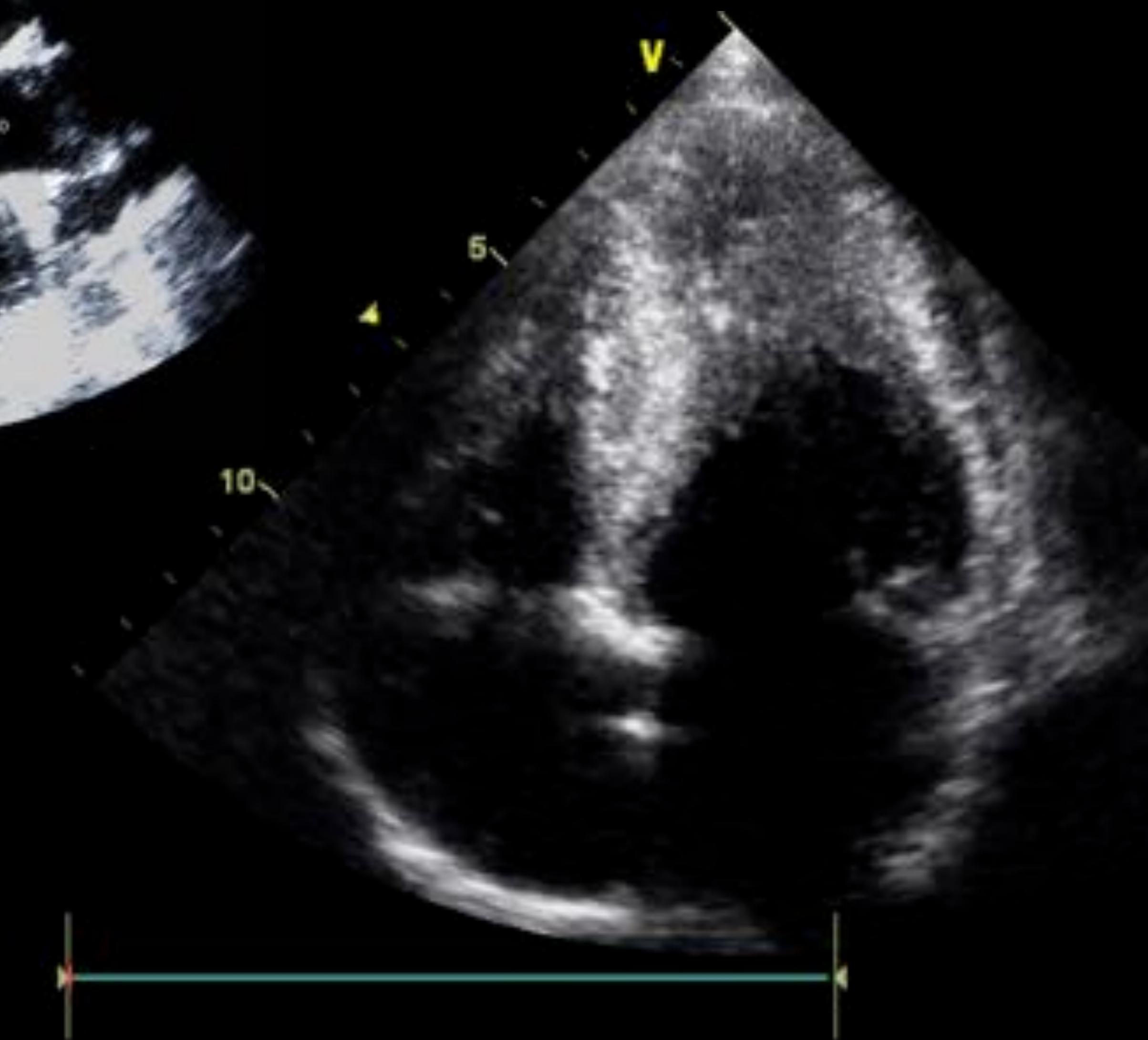
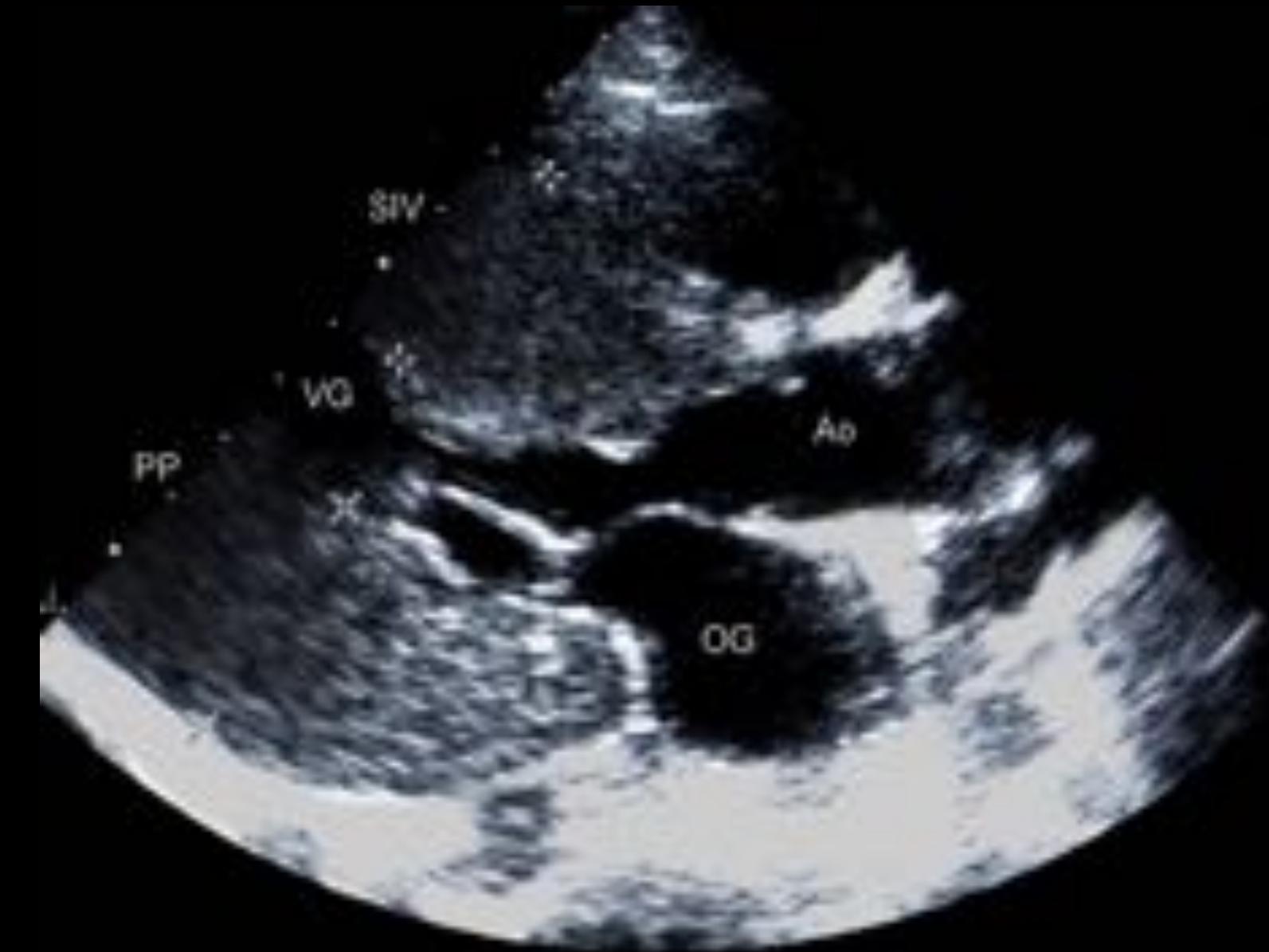


# Dilated cardiomyopathy

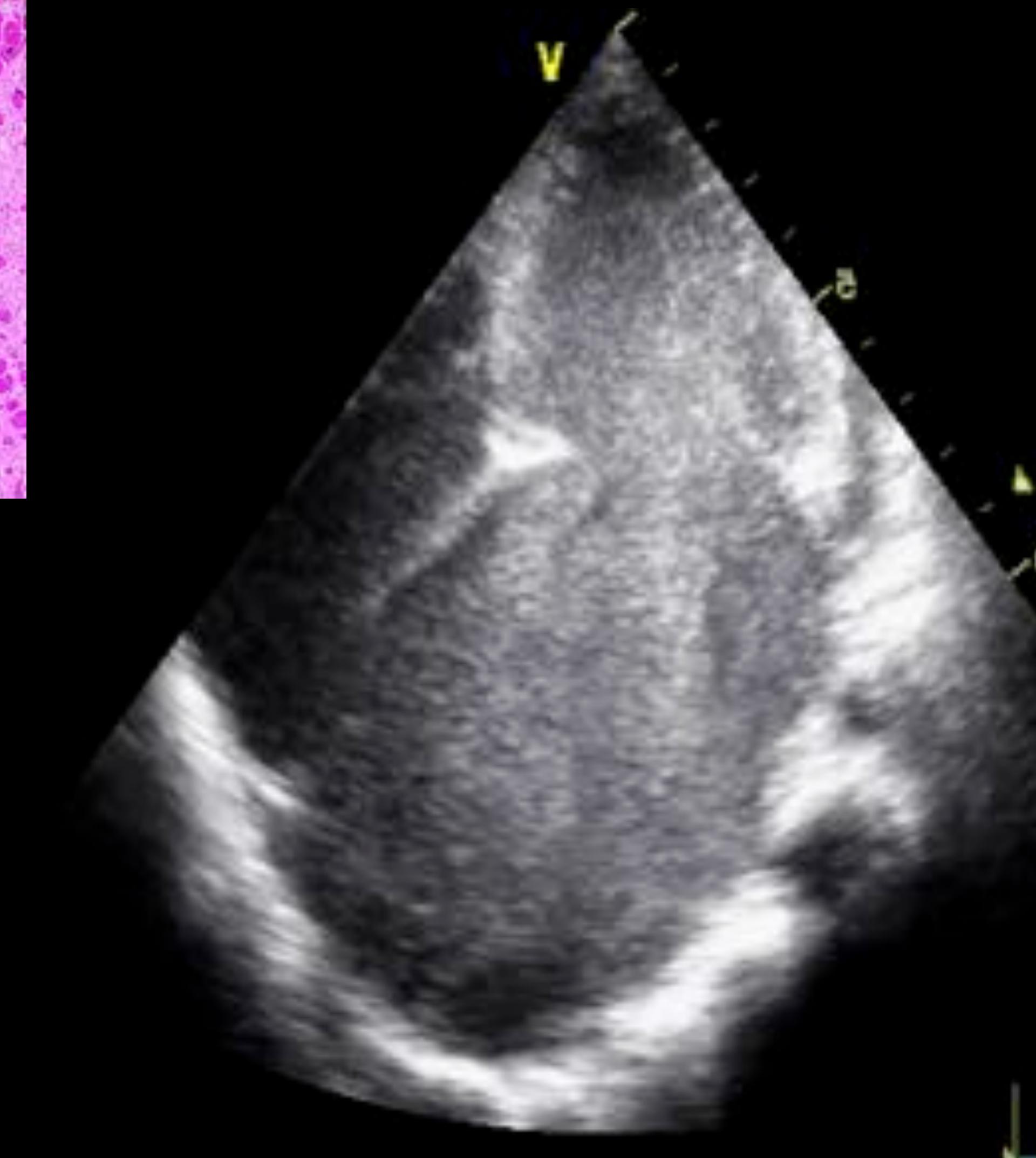
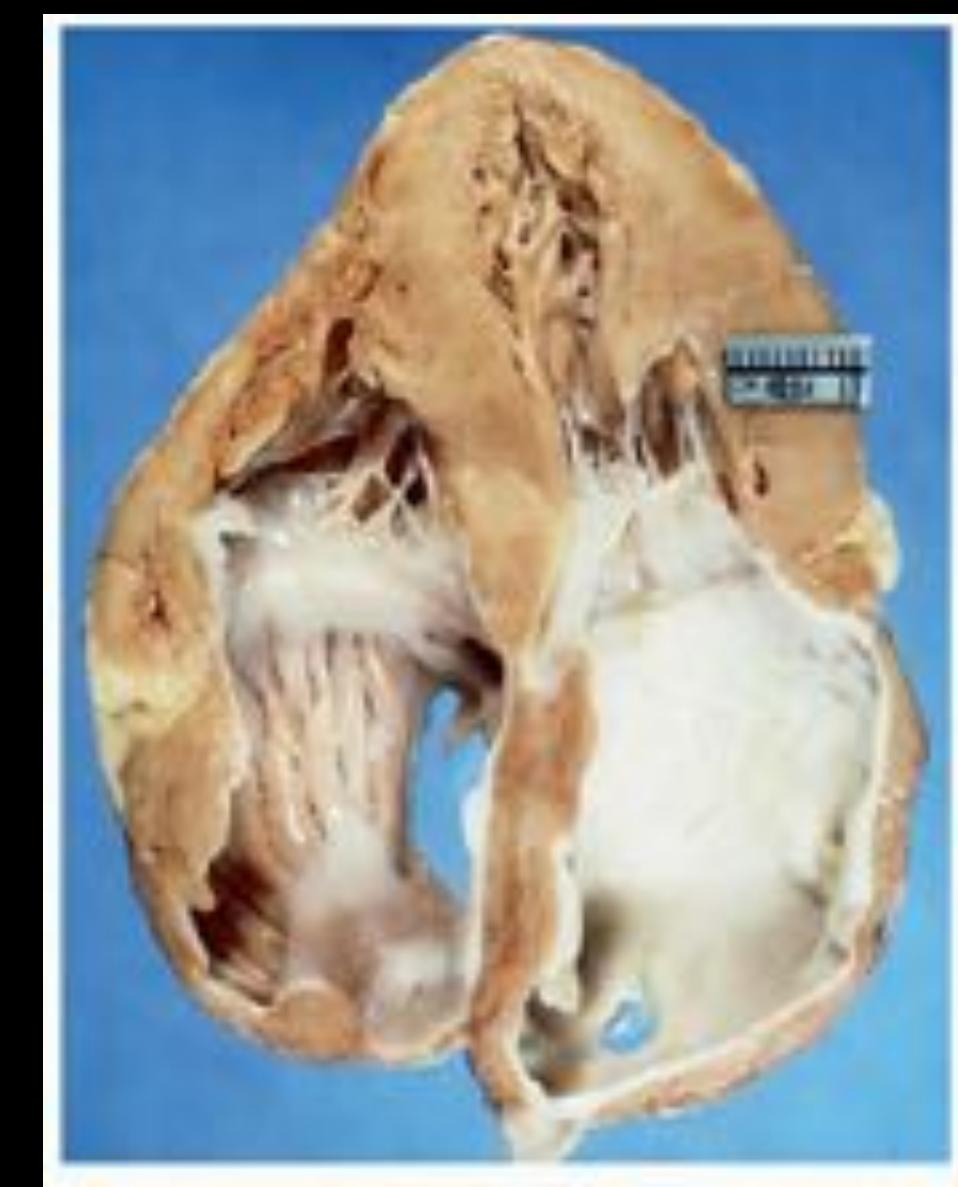
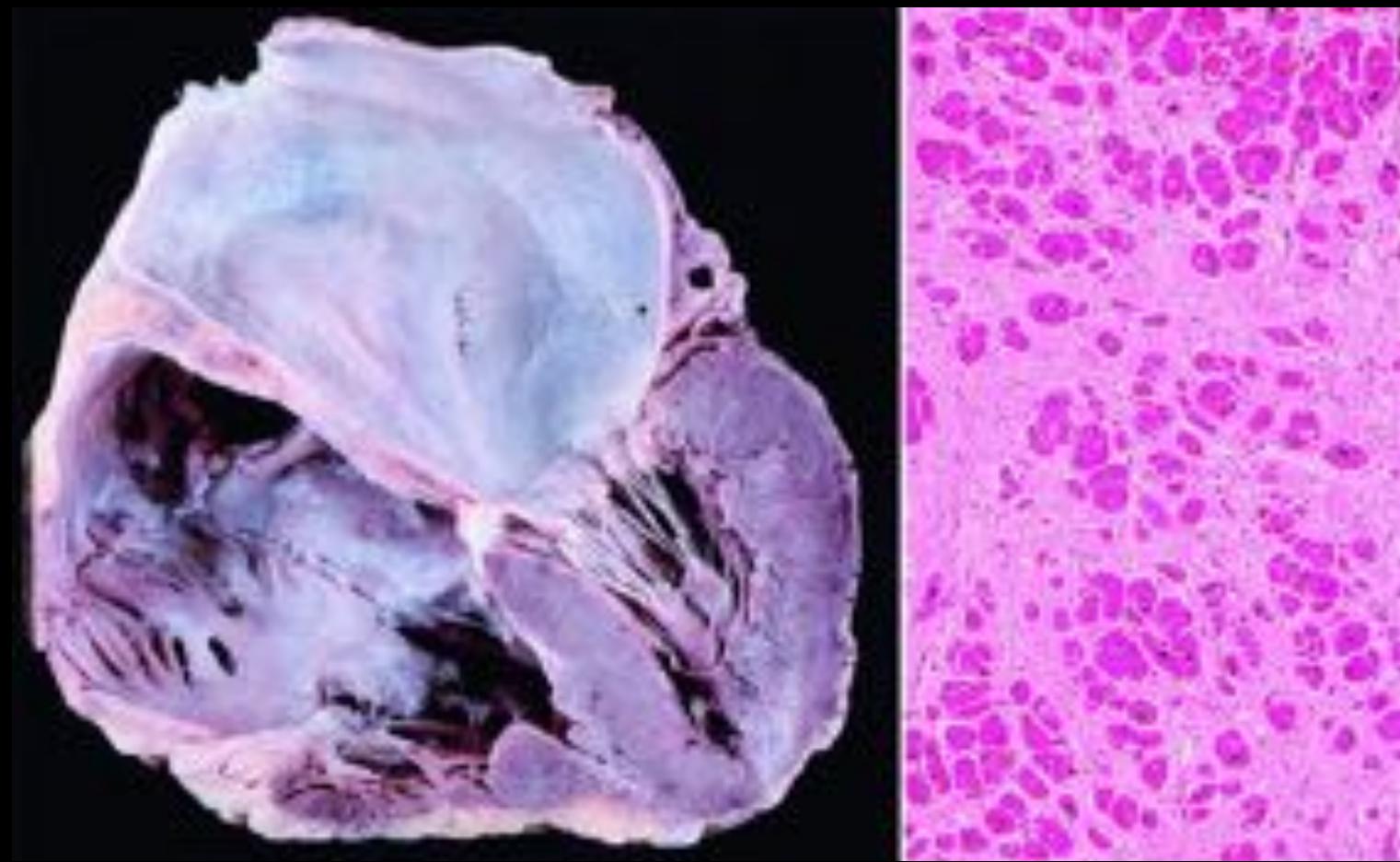




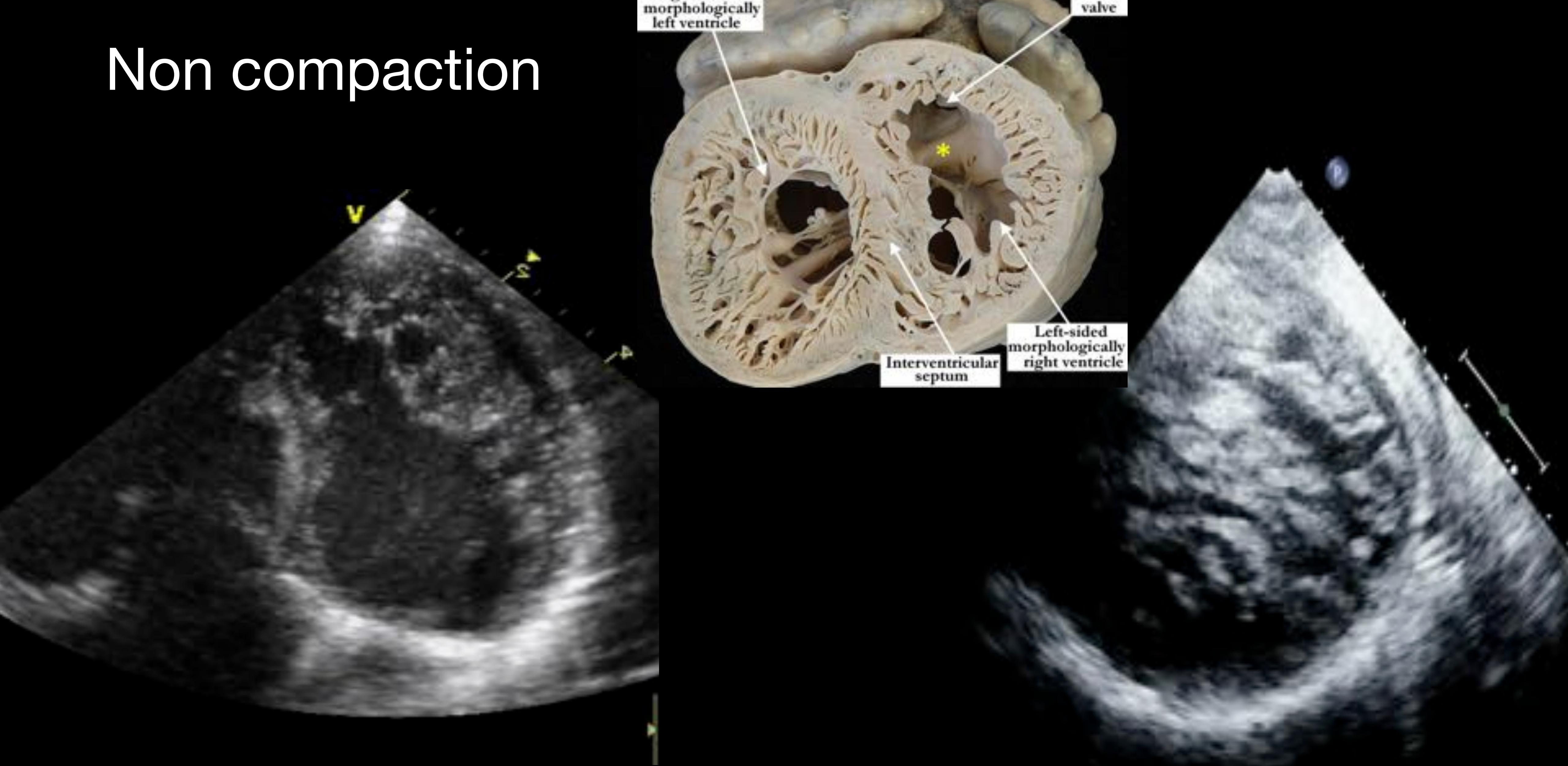
# Hypertrophic cardiomyopathy



# Restrictive cardiomyopathy



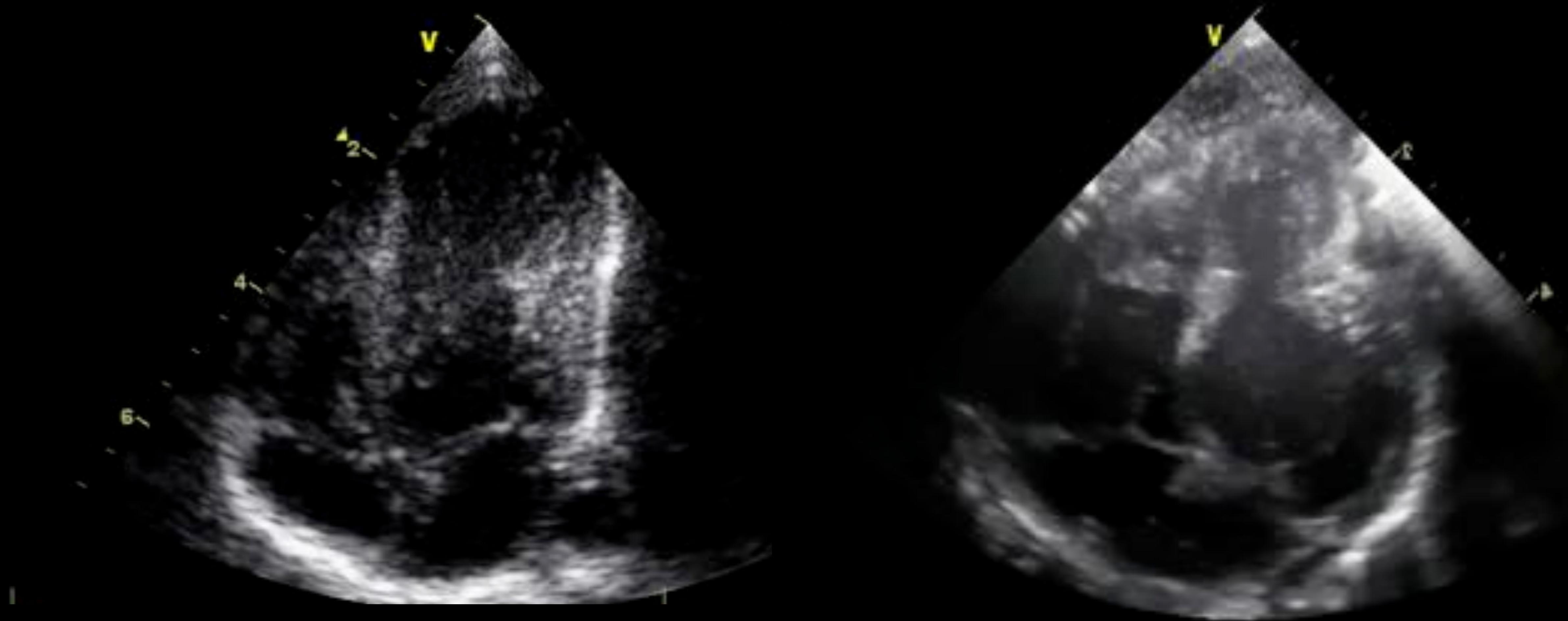
# Non compaction



# Ischemic



# Aneurysm and diverticulum



## Heart failure without myocardial disease



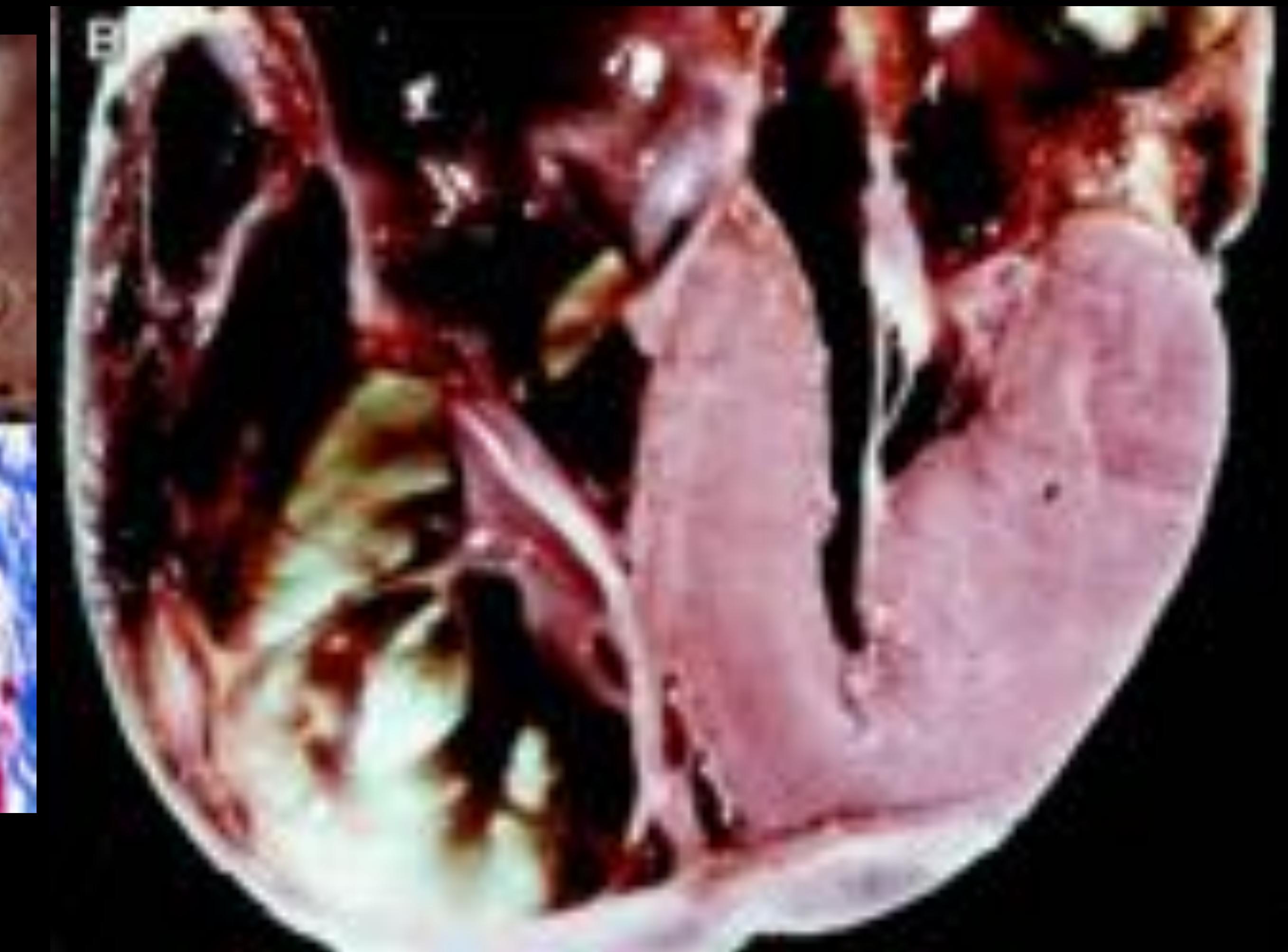
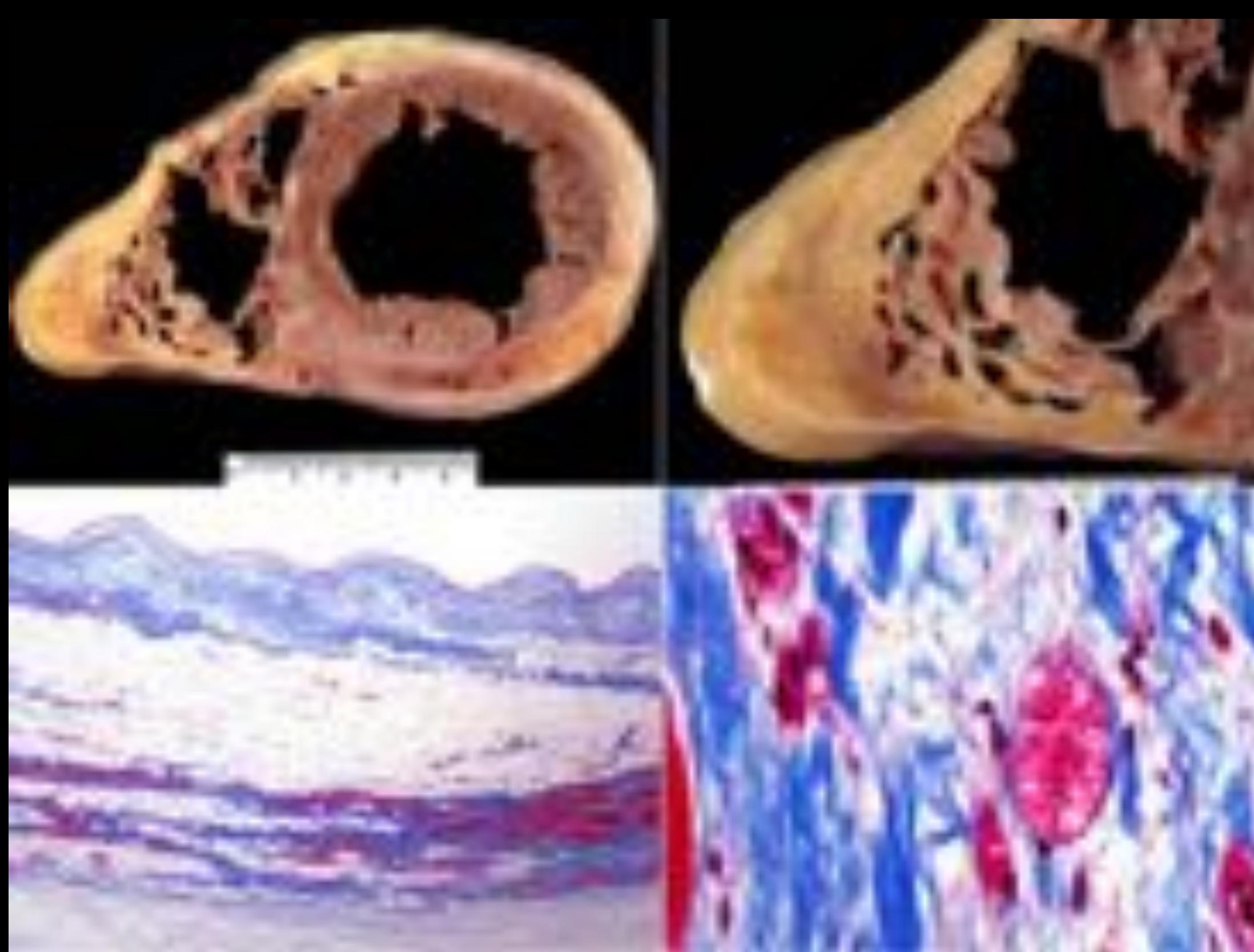
# Chronic constrictive pericarditis



# Right ventricular cardiomyopathies



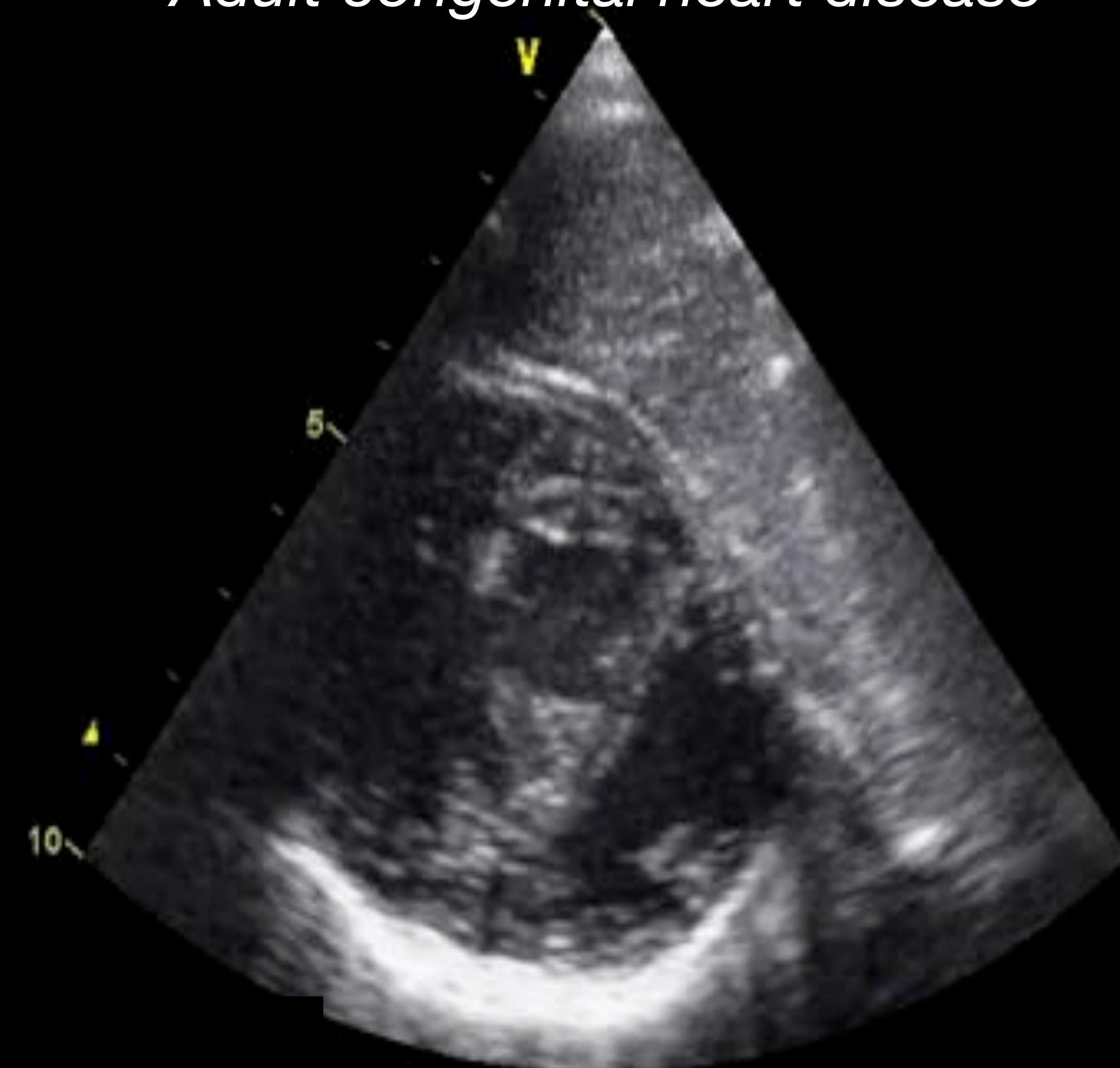
A.R.V.D



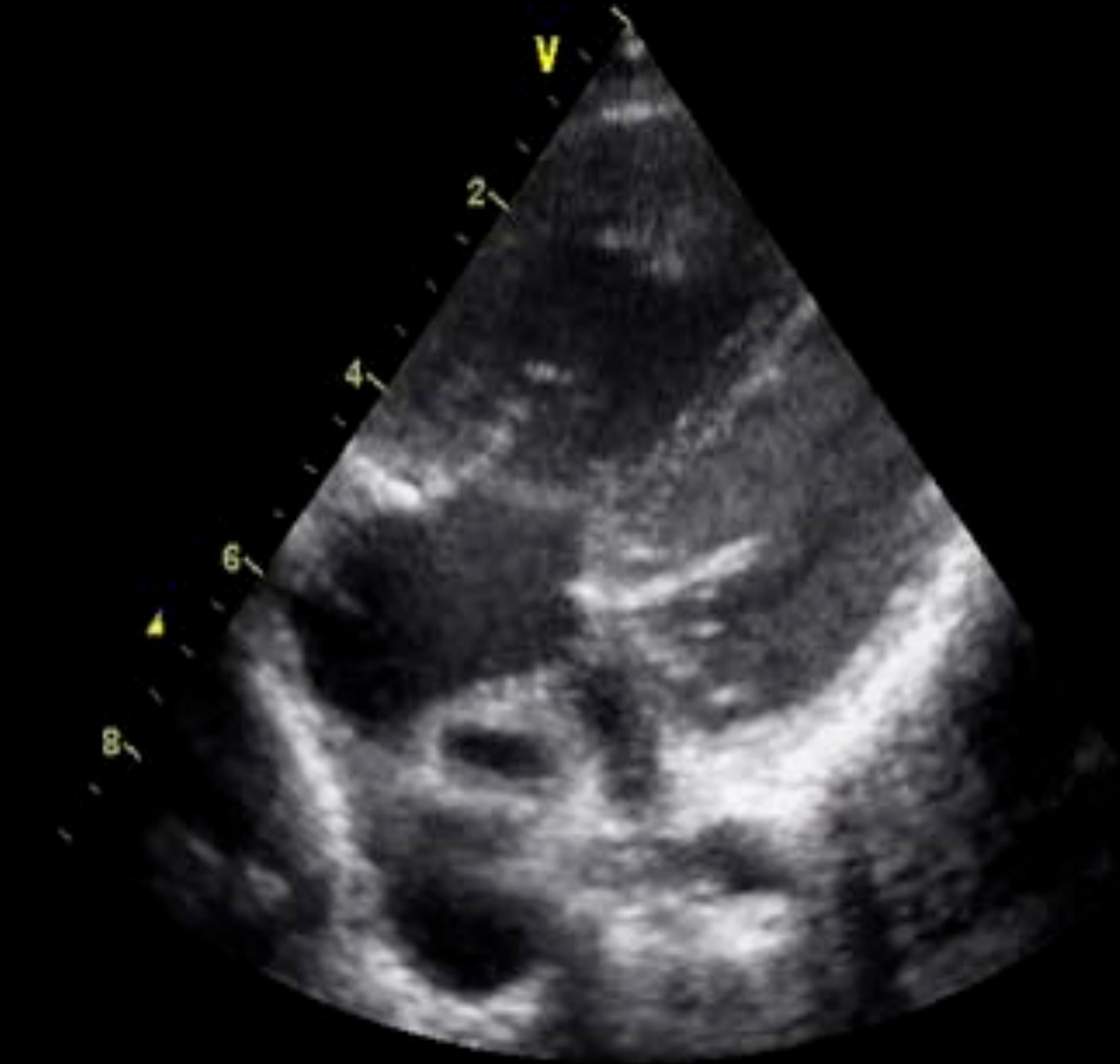
A.R.V.D



Booby-traps  
*Adult congenital heart disease*



Booby-traps  
*Adult congenital heart disease*



## Right ventricular dysfunction in PAH



# Difficulties in phenotyping

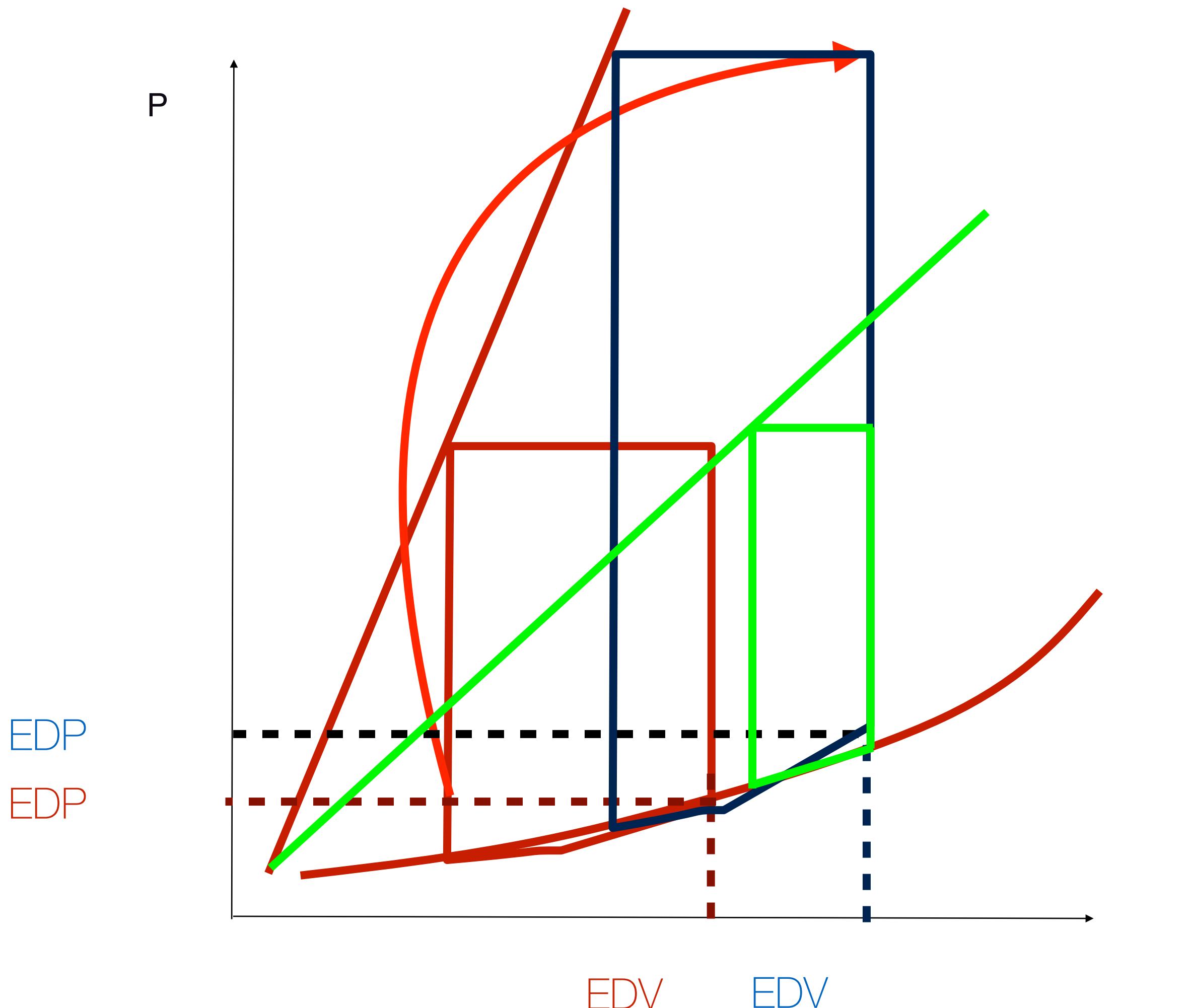
- Unusual phenotypes
  - Dilated with hypertrophic walls and restrictive physiology
- Changing phenotype
  - From hypertrophic to dilated
- Uncertain phenotype
  - Penetrance increasing with age

# **Cardiomyopathies are rarely familial and a known cause of ventricular dilatation and/or hypertrophy should be extensively searched**

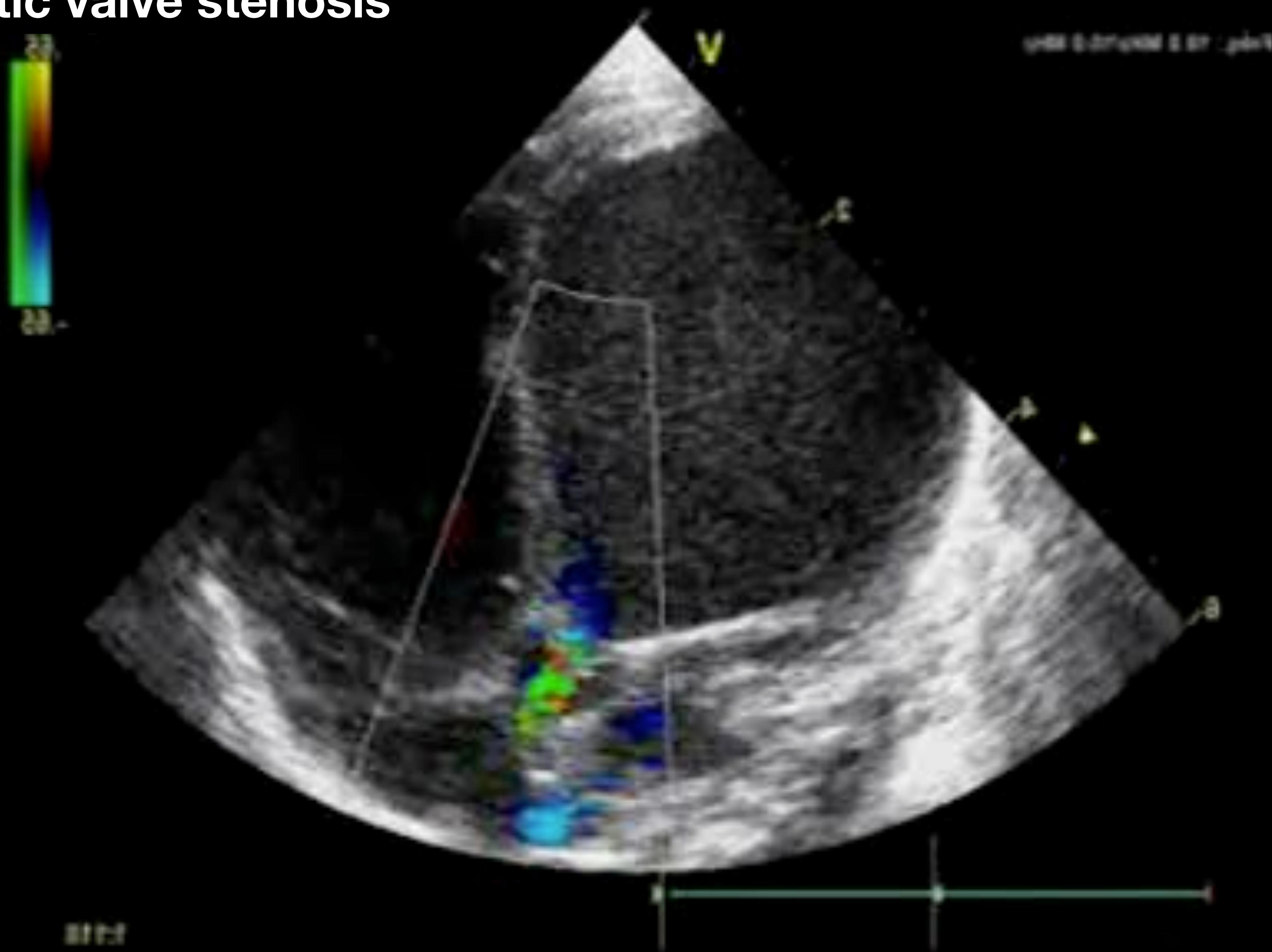
- Volume and pressure overload
- Myocardial ischemia
- Sustained arrhythmias
- Infective myocarditis
- Toxic
- Neuromuscular disorders
- Syndromic cardiomyopathies
- Metabolic diseases
- Inherited cardiomyopathies

# Heart failure due to increased afterload

## Normal contractility and compliance

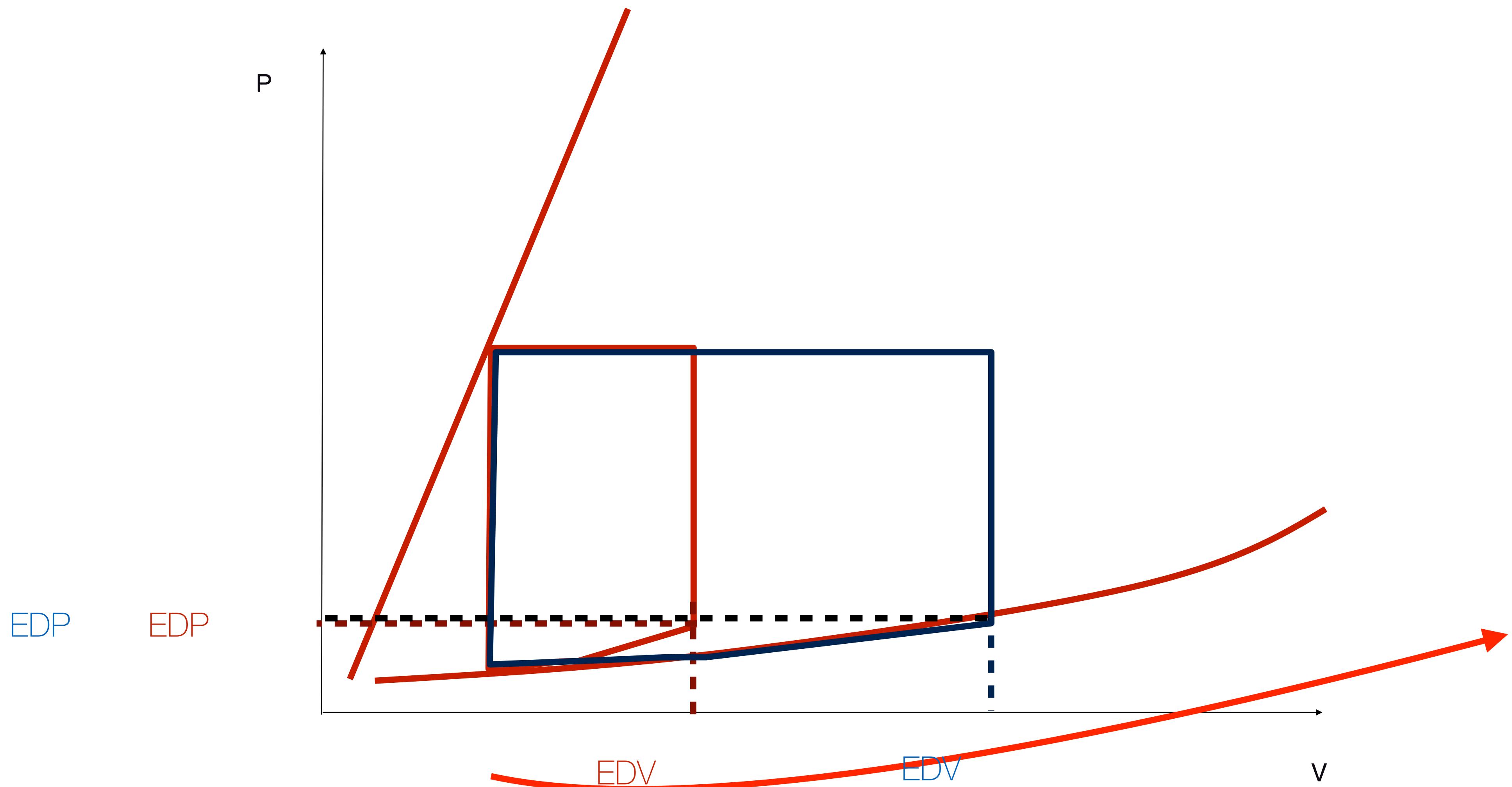


# Critical aortic valve stenosis



# Heart failure due to increased preload

## Normal contractility and compliance



# Severe mitral valve regurgitation

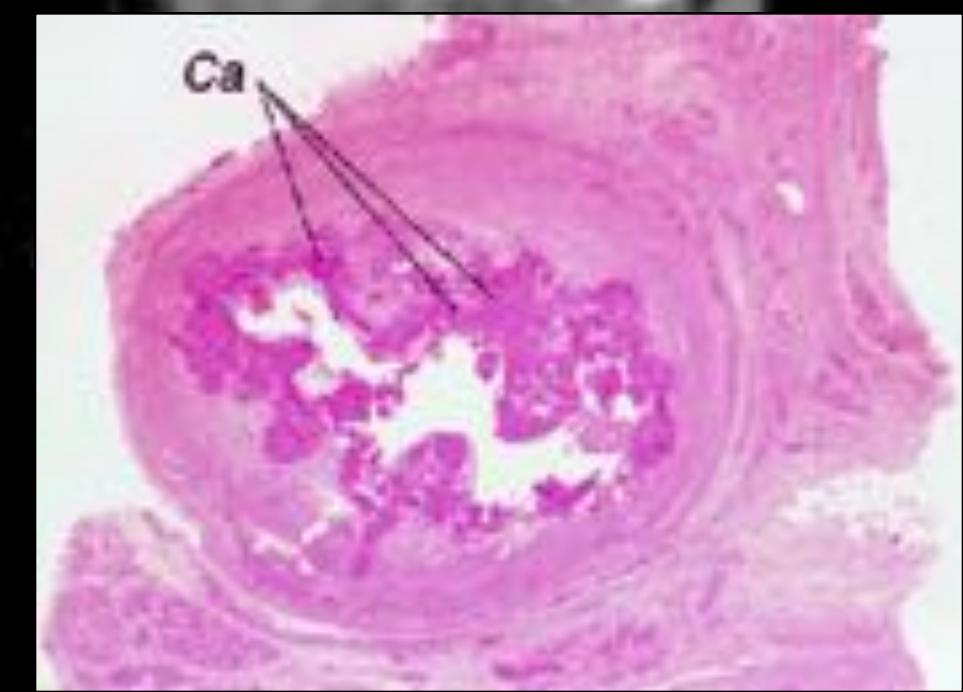
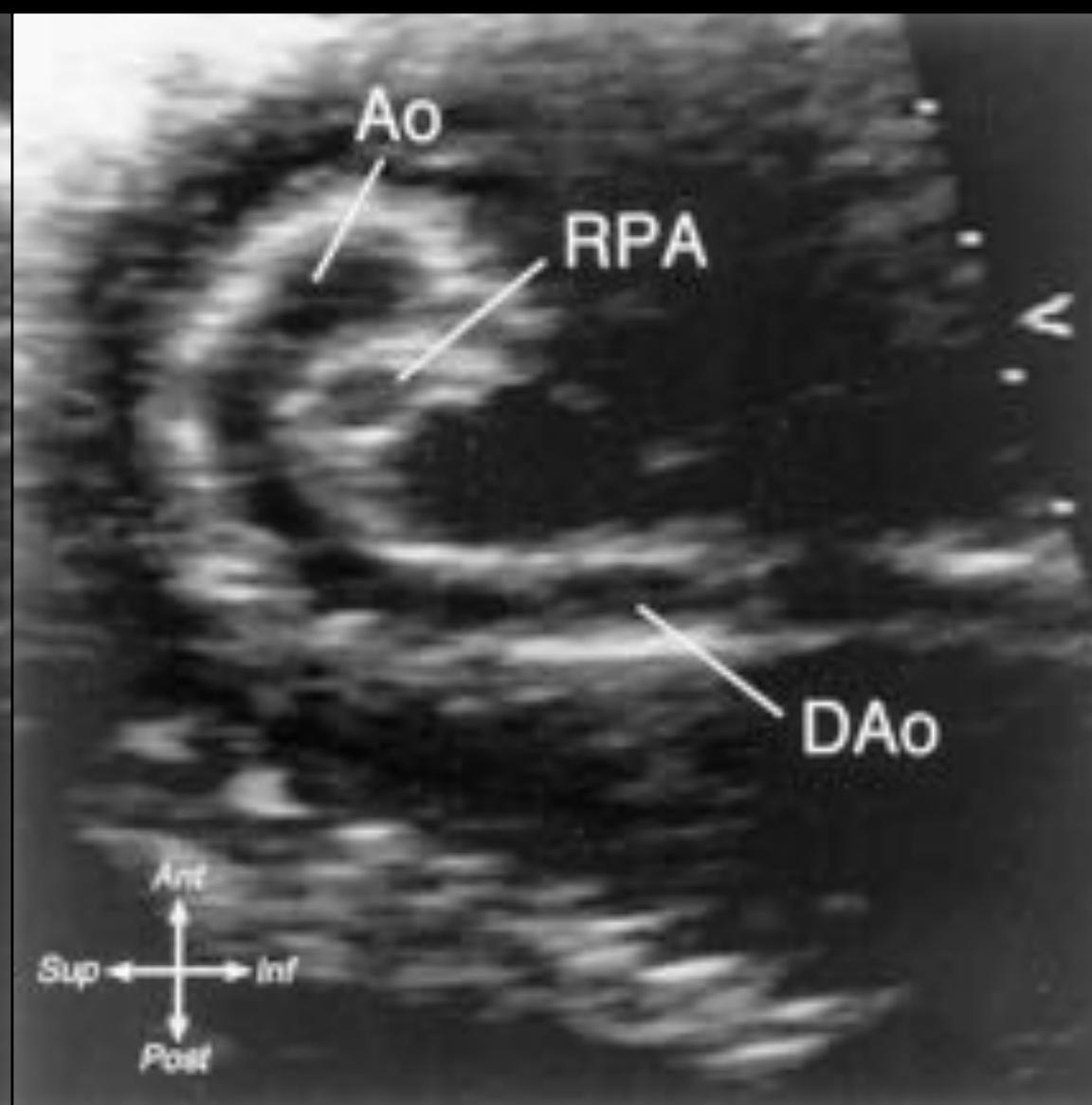


# Ischemic cardiomyopathies



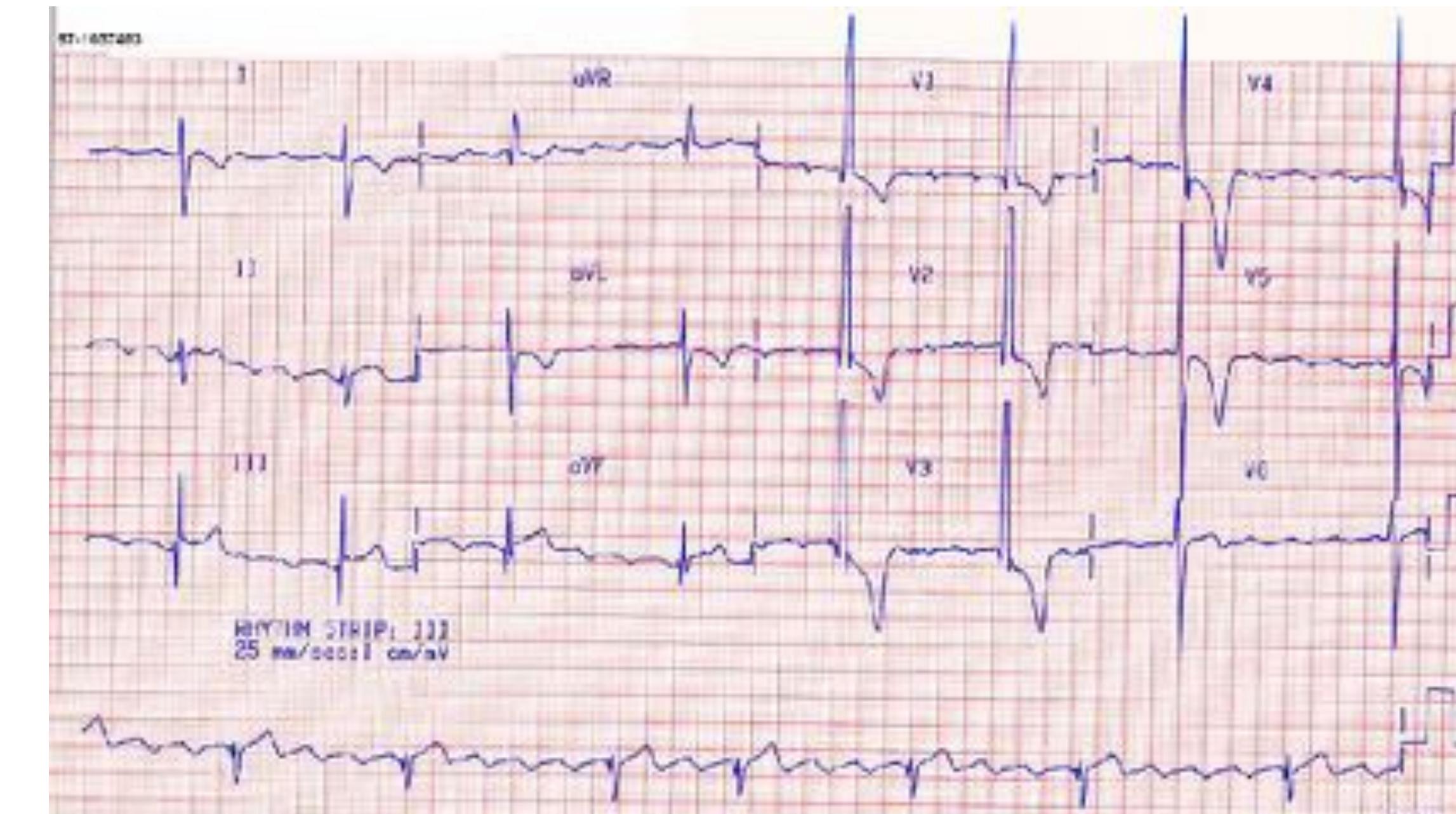
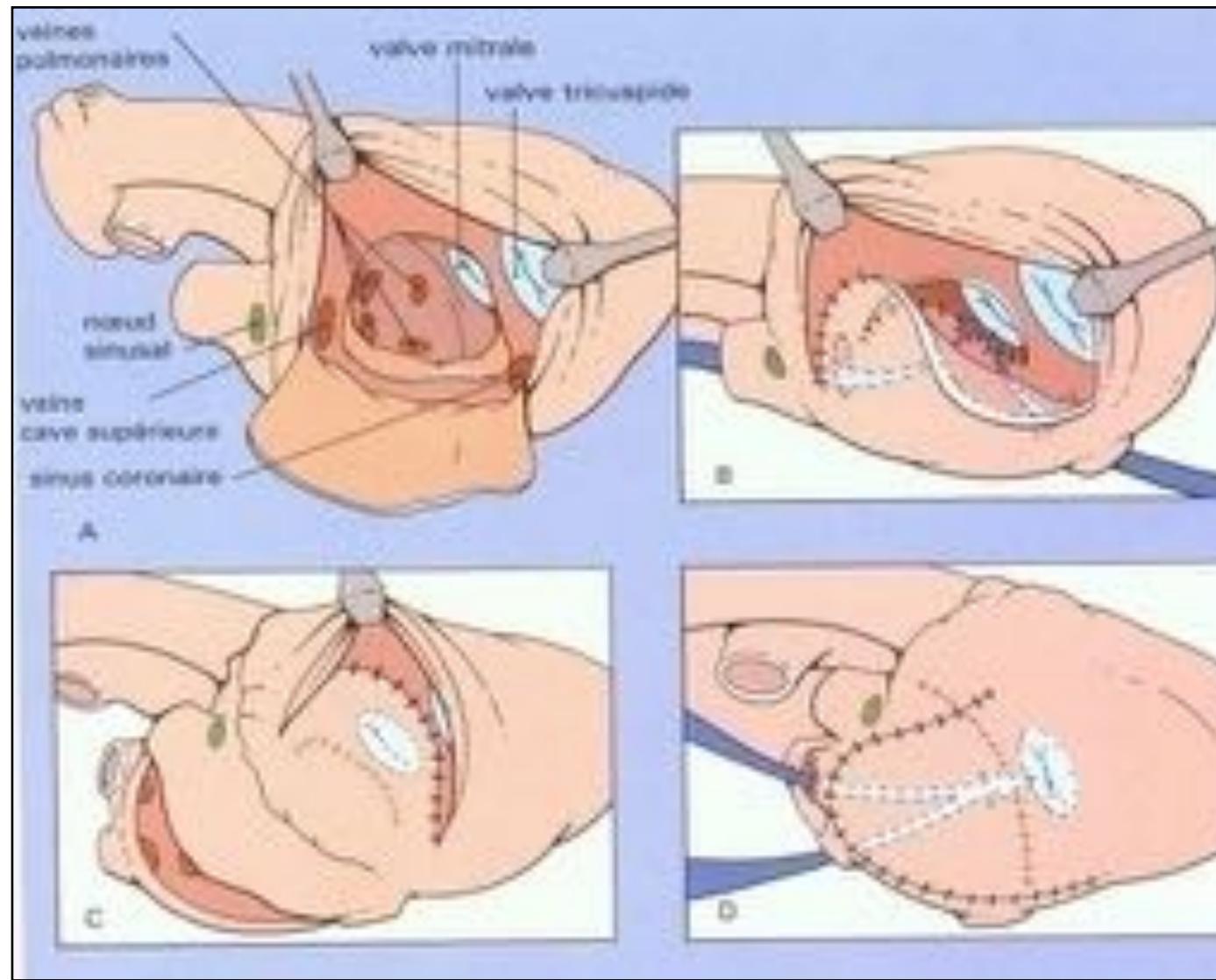
ALCAPA-Main stem atresia  
Post-operative  
TGA  
Kawasaki disease  
Hypercholesterolemia  
GACI

GACI



# Arrhythmic cardiomyopathy

- Supraventricular tachycardia of the newborn
- Booby-traps
  - Atrial arrhythmias after atrial correction of TGA
  - Arrhythmias after TCPC



# Arrhythmic cardiomyopathy fetal

JT/AVB



# Infectious cardiomyopathies

Viral myocarditis

Lyme disease

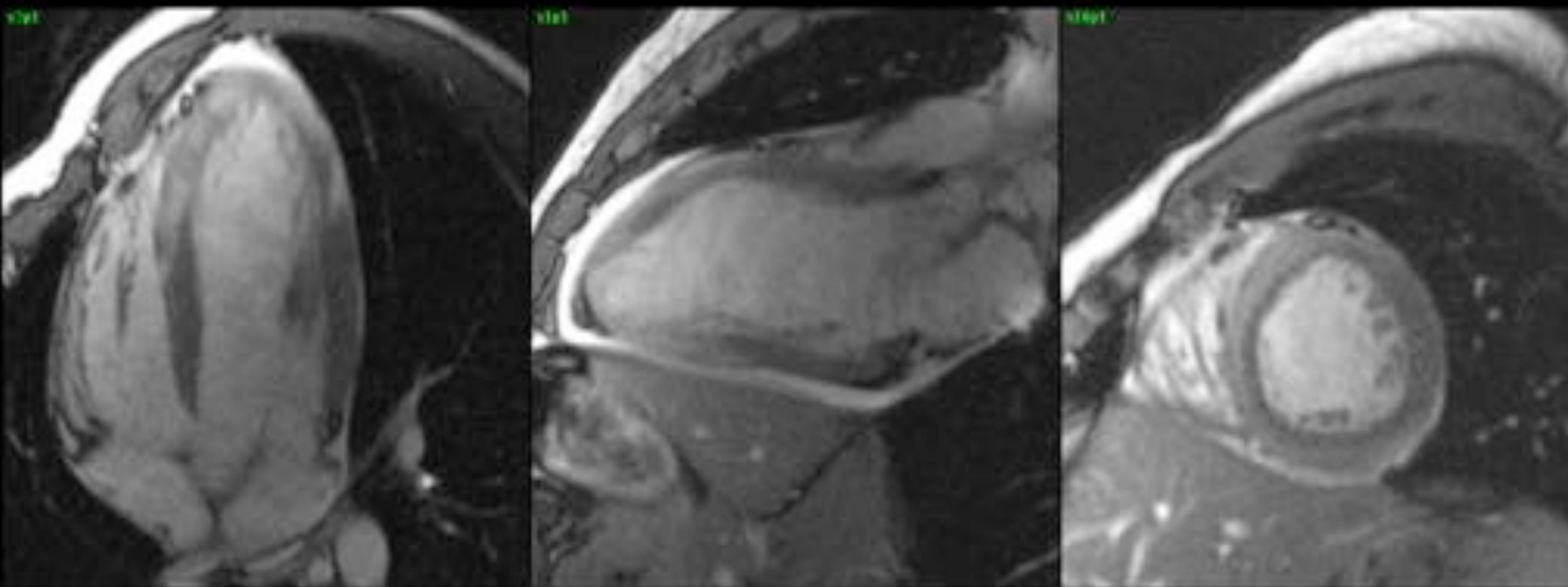
Chagas disease

HIV

Toxoplasmosis

Rheumatic

# Myocarditis



# Toxic

- Anthracyclines
- Radiations

# Neuromuscular disorders

- Dystrophinopathies
  - Duchenne de Boulogne
  - Becker
- Emery-Dreyfus: laminopathies
- Steinert
- Friedreich

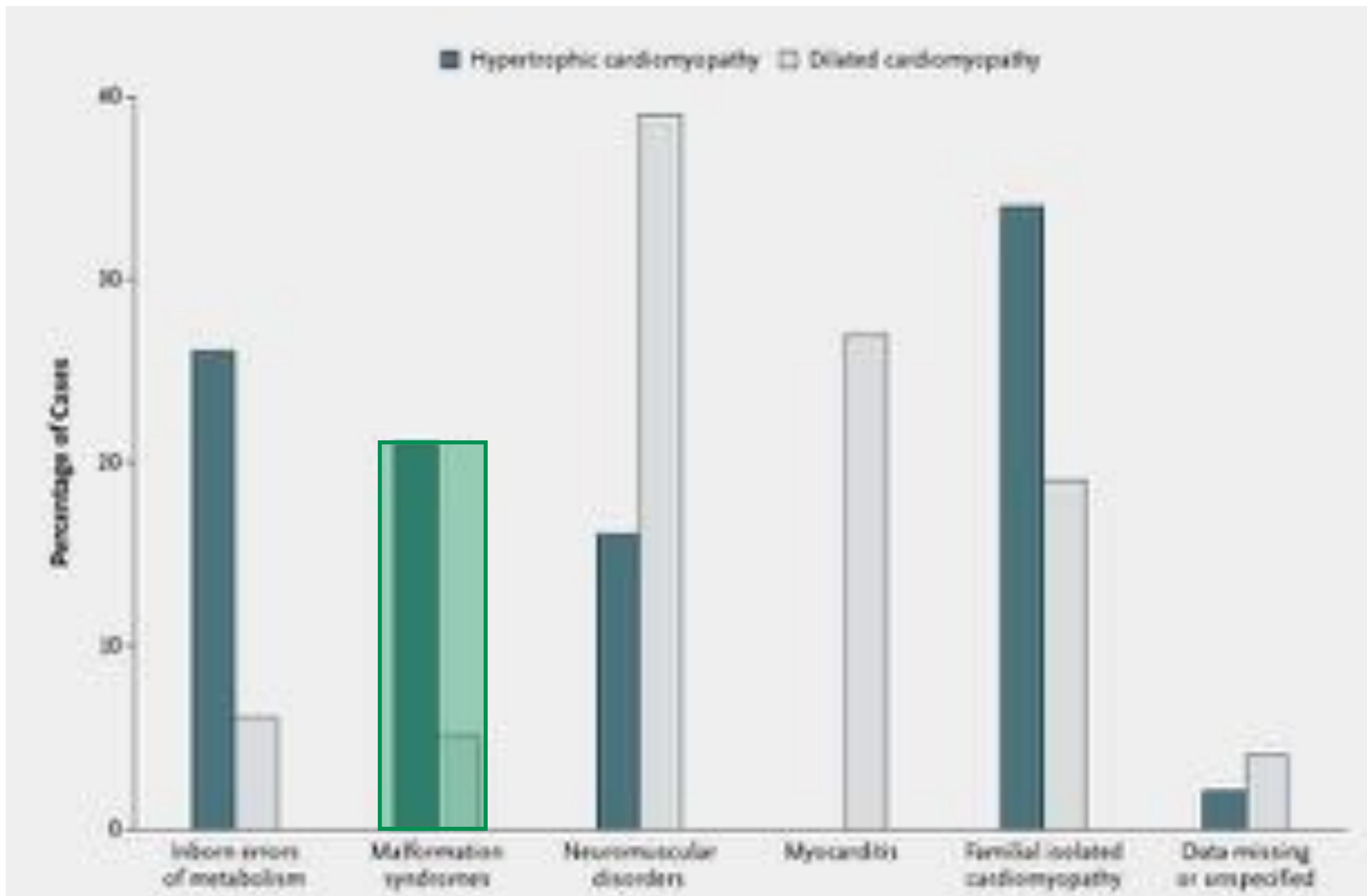


**First stop !**

# What have you done ?

- Clinical examination
- Medical history
- ECG
- Echocardiography
- Troponine
- MRI

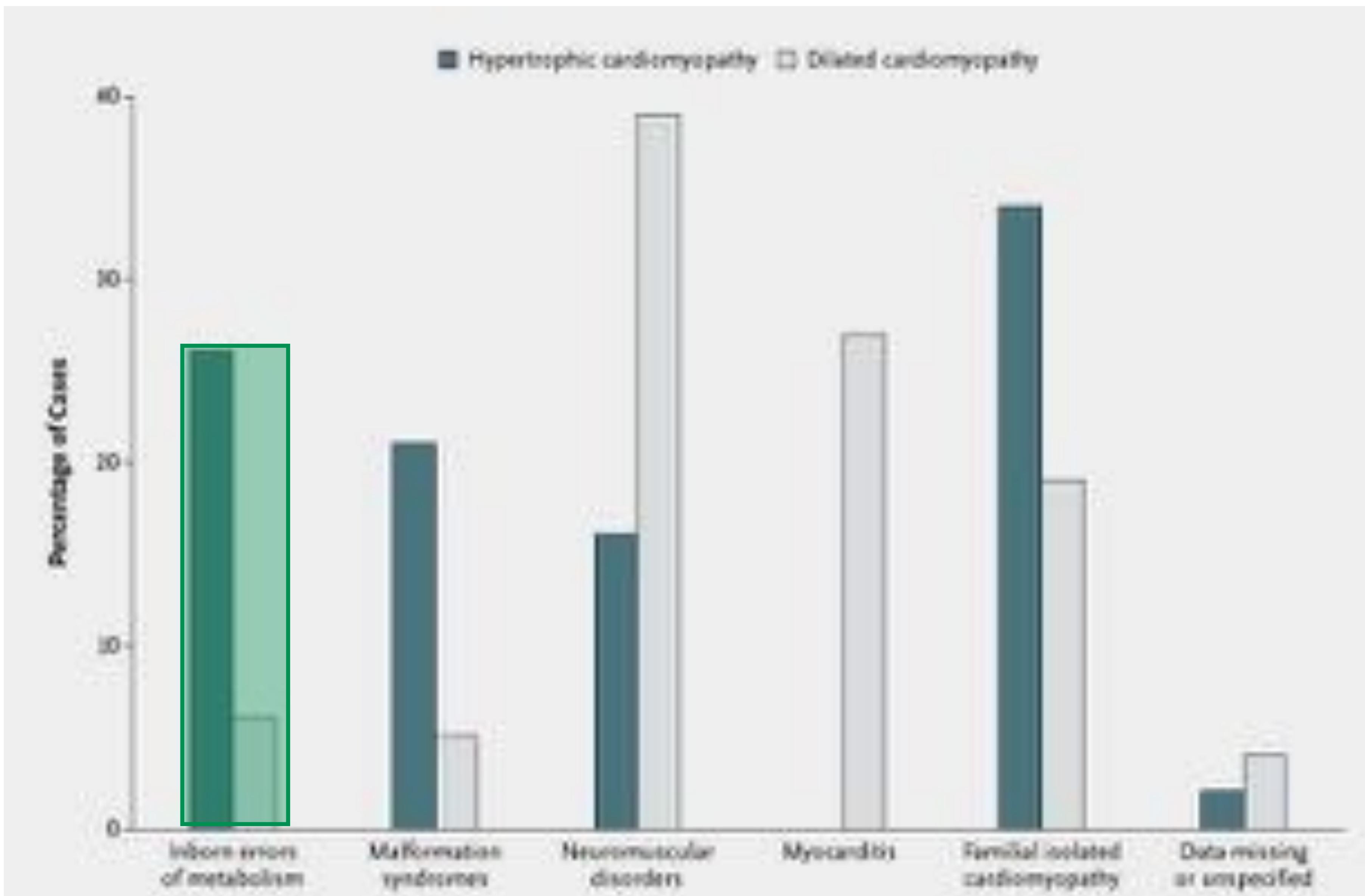
# Primary causes of CMP in children





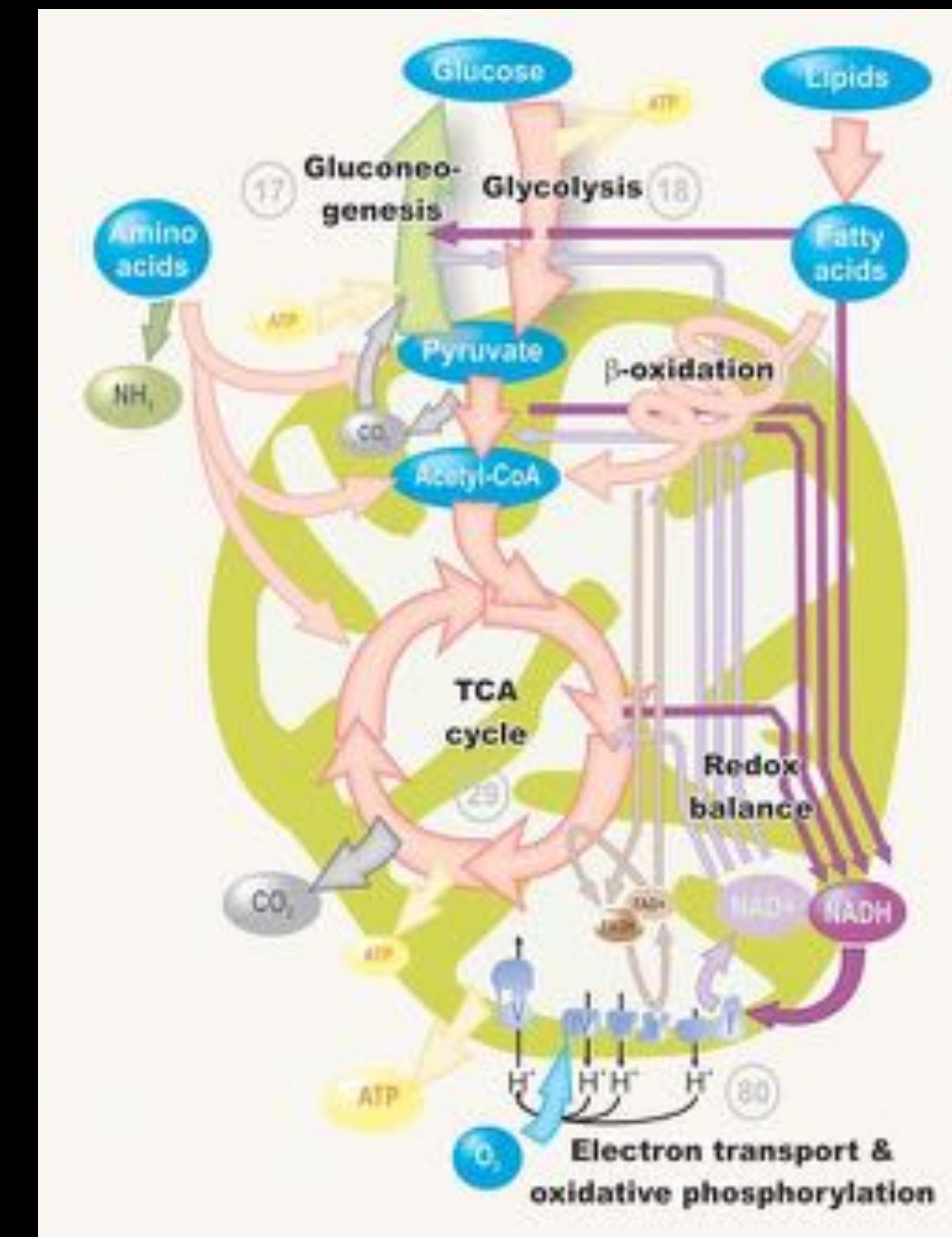
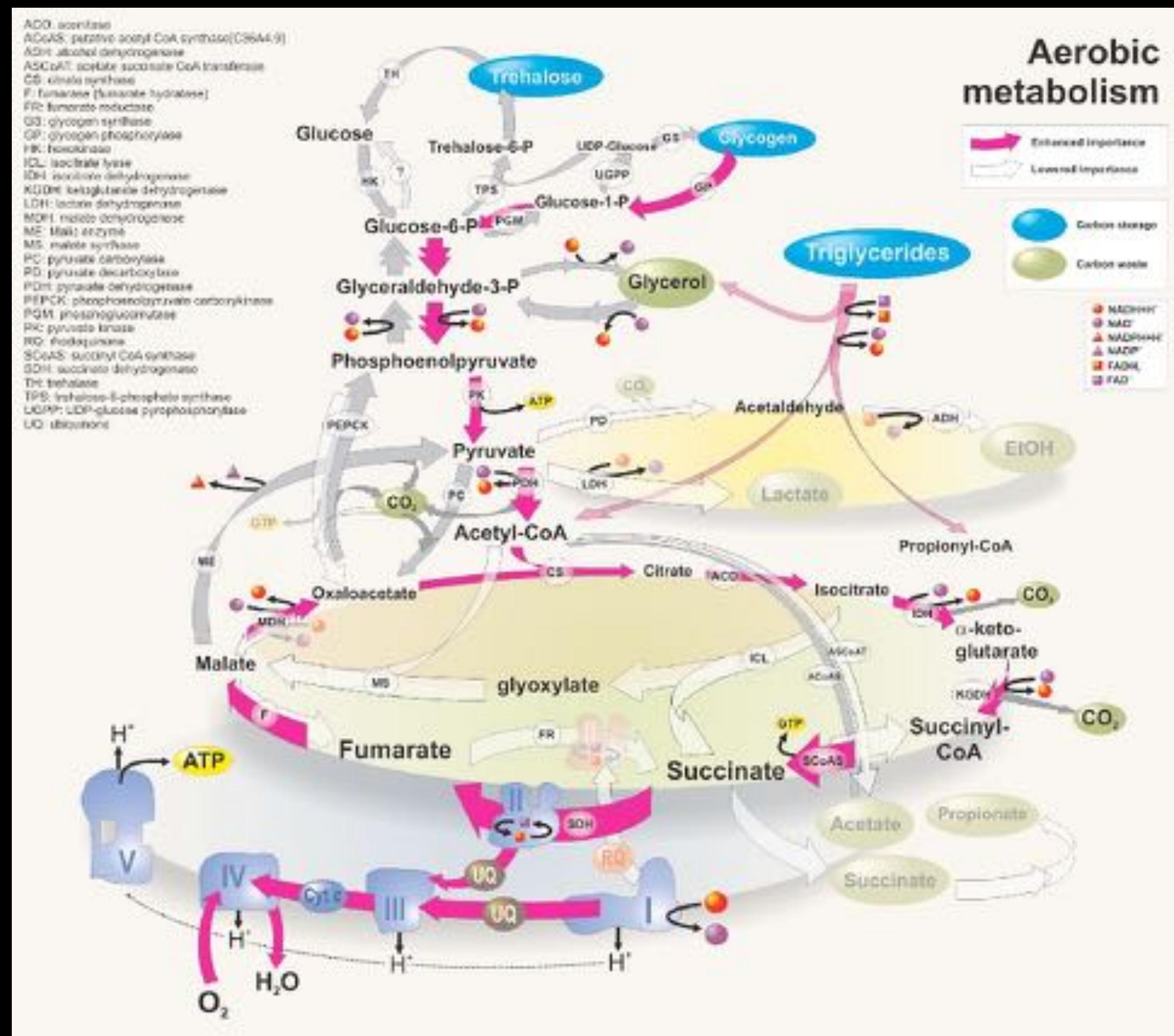
RASopathies

# Primary causes of CMP in children



# **When should you think of metabolic cardiomyopathy ?**

- Family history of sudden death or unexplained death in infancy
- Multisystemic disease
- Changing phenotype
- Severe hemodynamic compromise with mild alteration of LV function
- Atypical anomalies of ECG : left bundle branch bloc, AVB, ventricular tachycardia



# Cardiac metabolism for pediatric cardiologists

Substrate accumulation (non toxic):  
storage diseases

Lysosomal : HCM, valves  
Peroxisomal  
Reticulum: glycosylation

Substrate

Metabolism

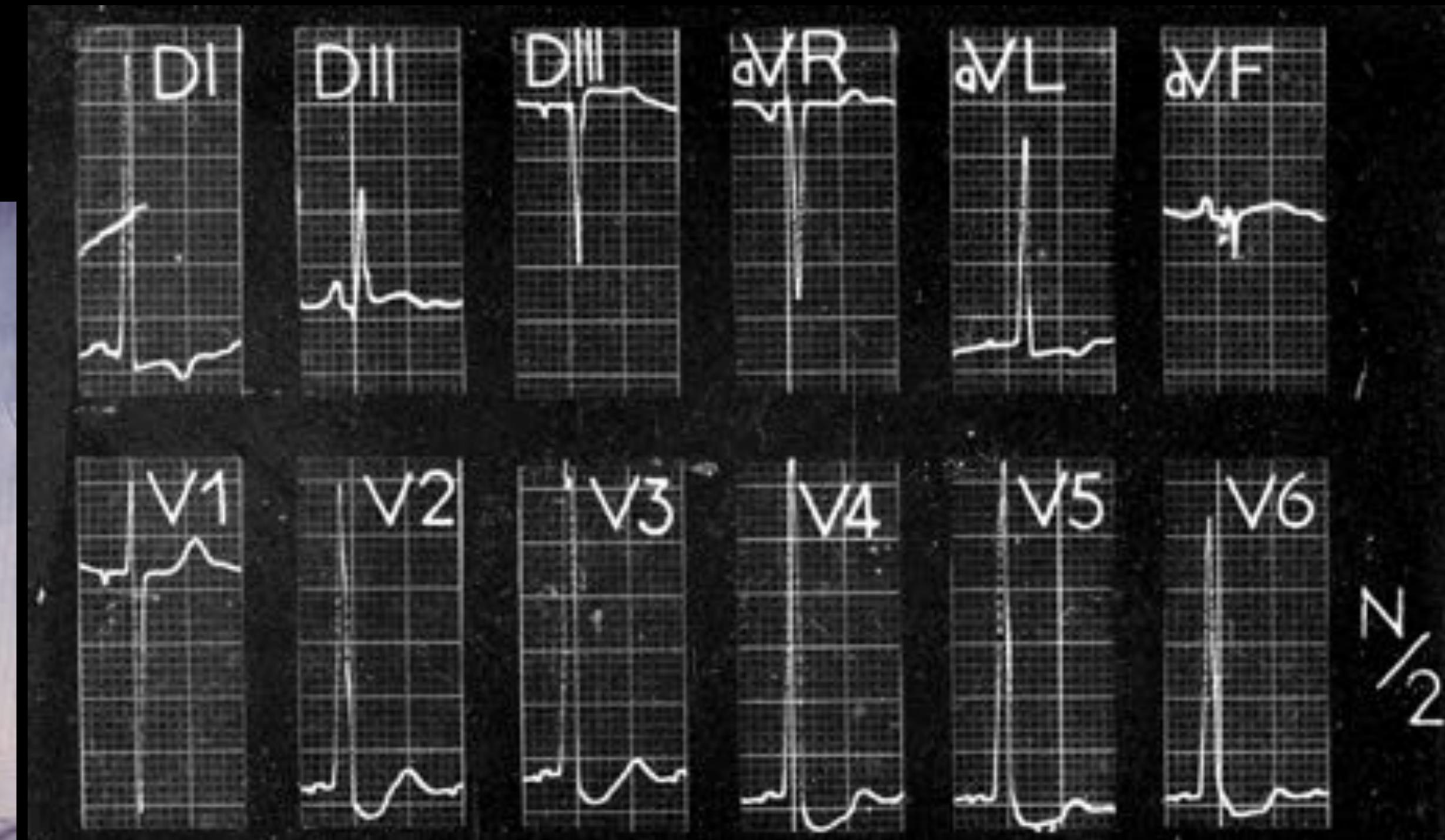
Product

Organic aciduria

Fatty-acid oxydation  
Respiratory chain  
Krebs cycle  
Glycogenoses

Substrate accumulation (toxic):  
intoxication diseases

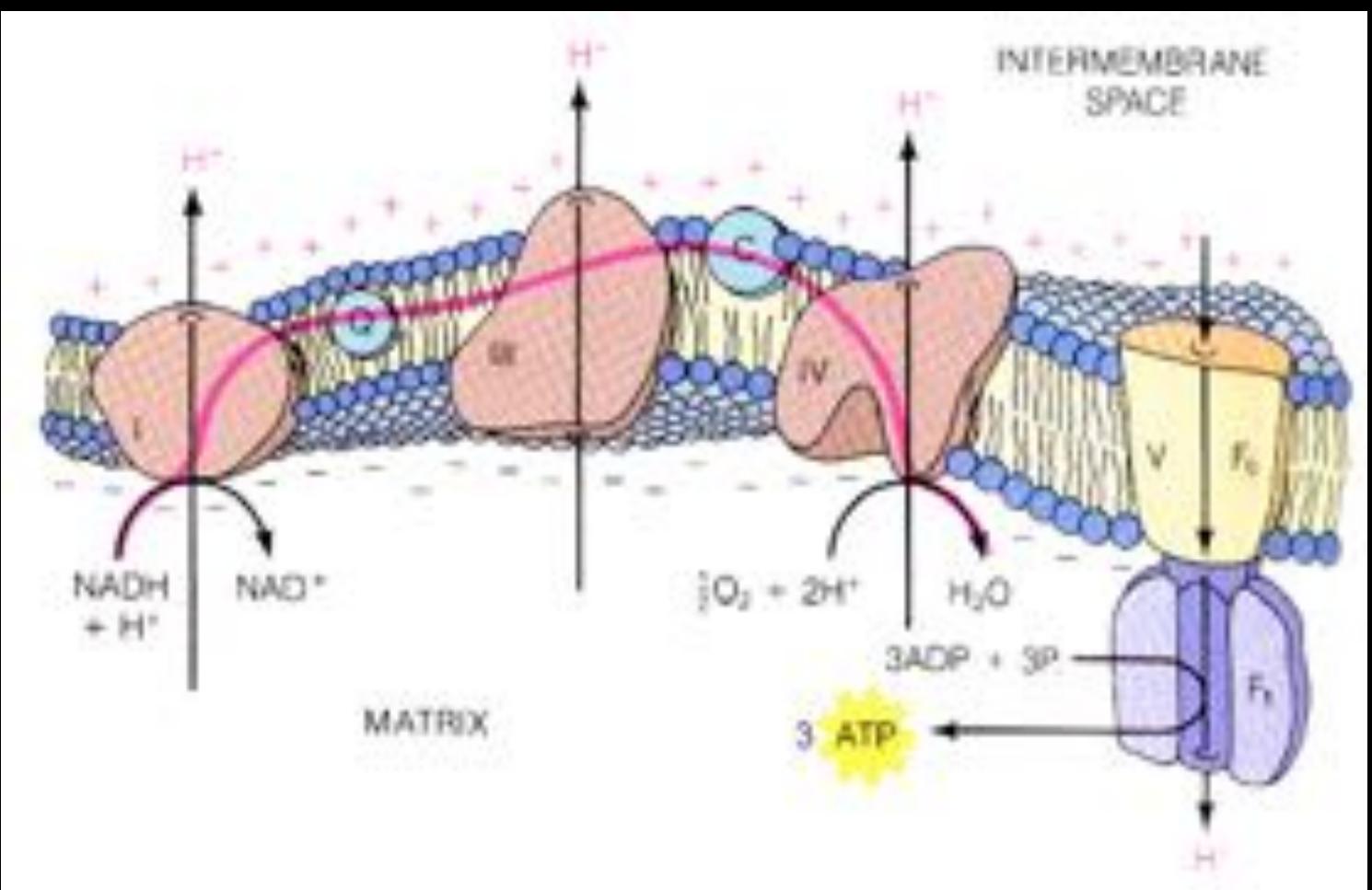
Product decrease or absent :  
energetic defects



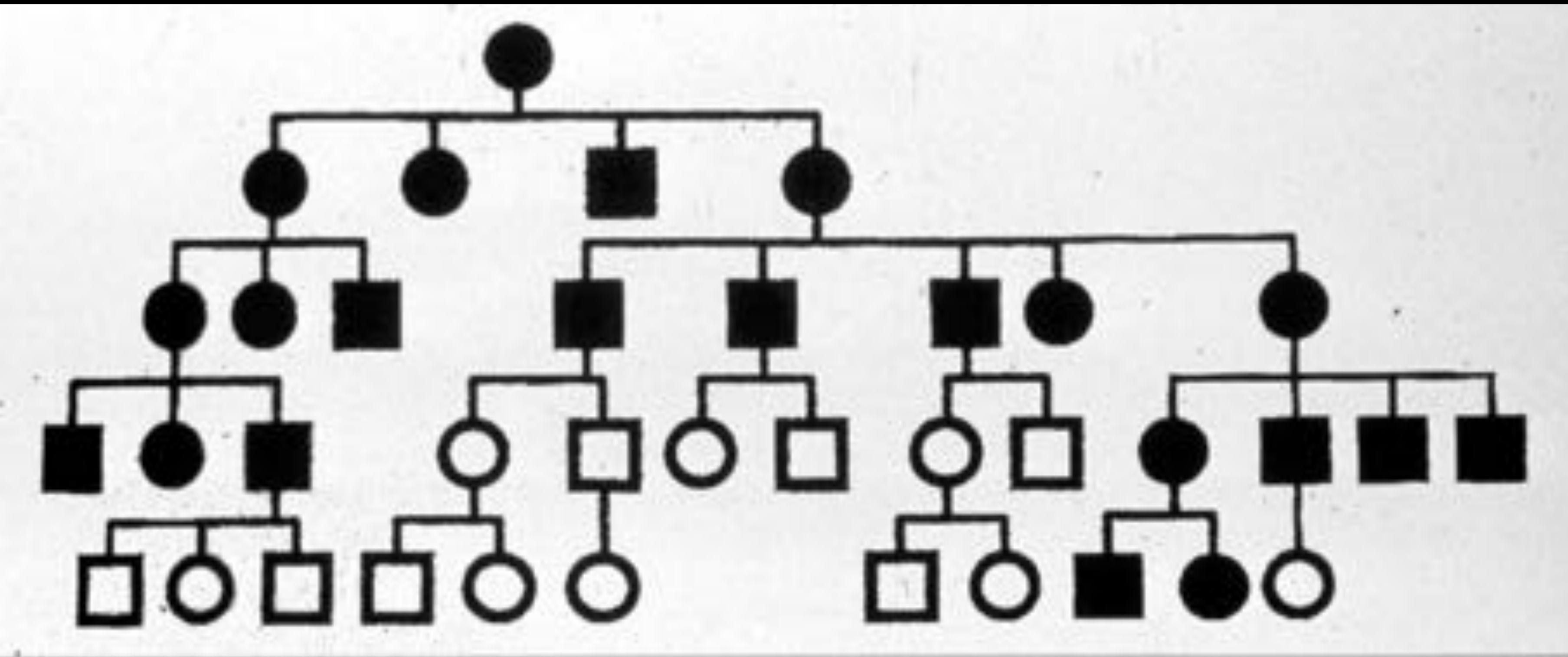
Pompe's disease



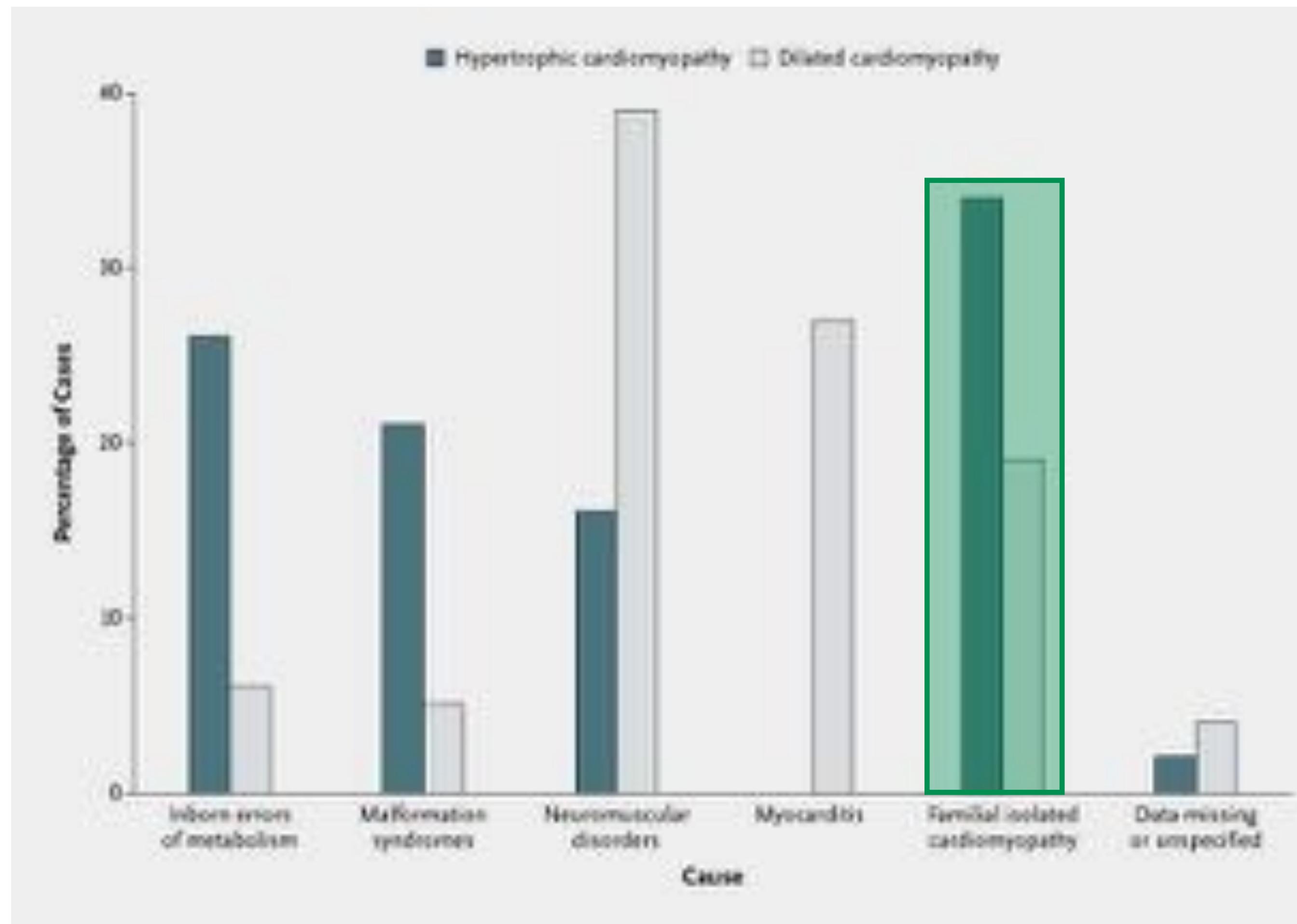
Carnitine deficiency



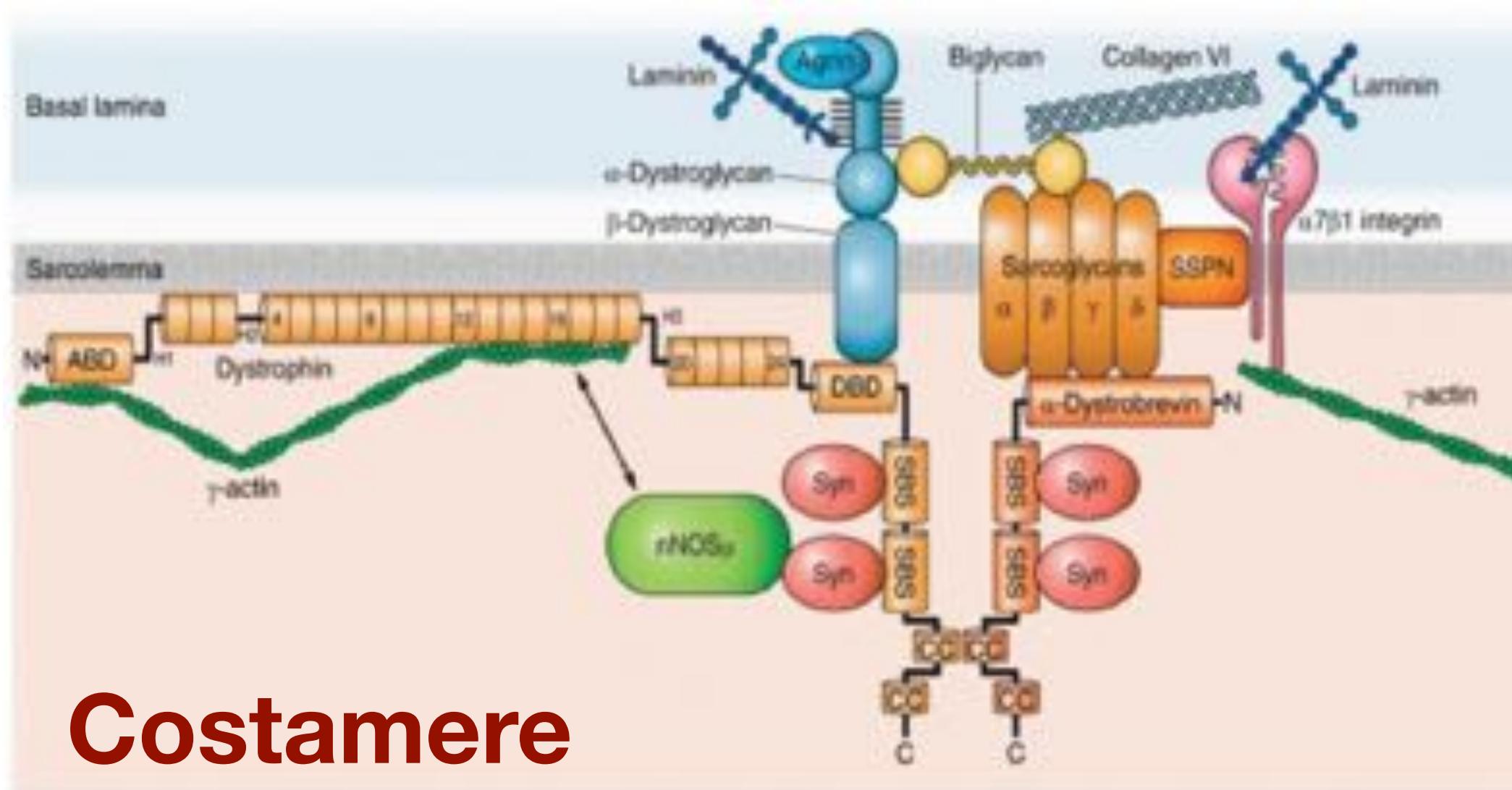
## Mitochondrial disease



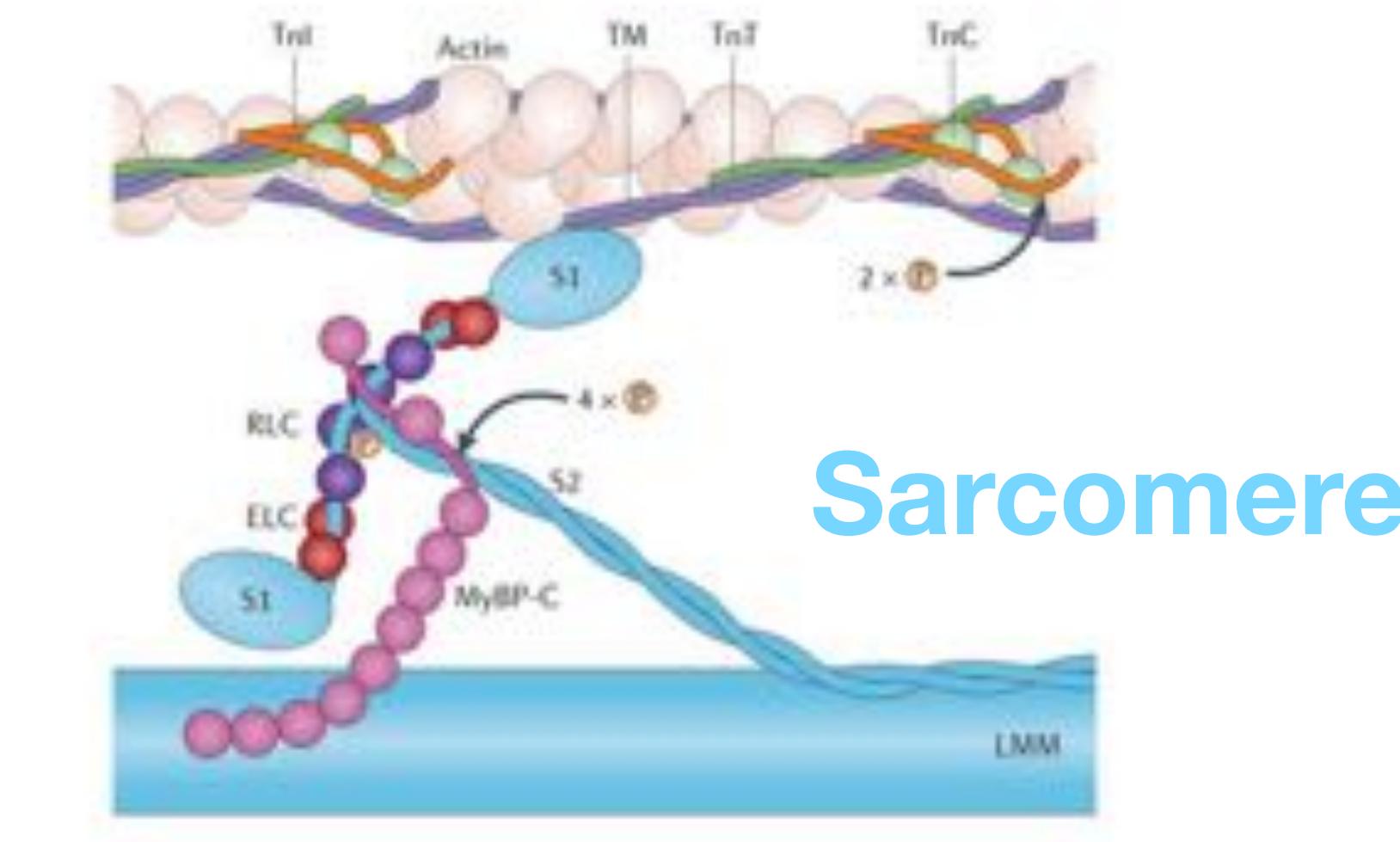
# Primary causes of CMP in children



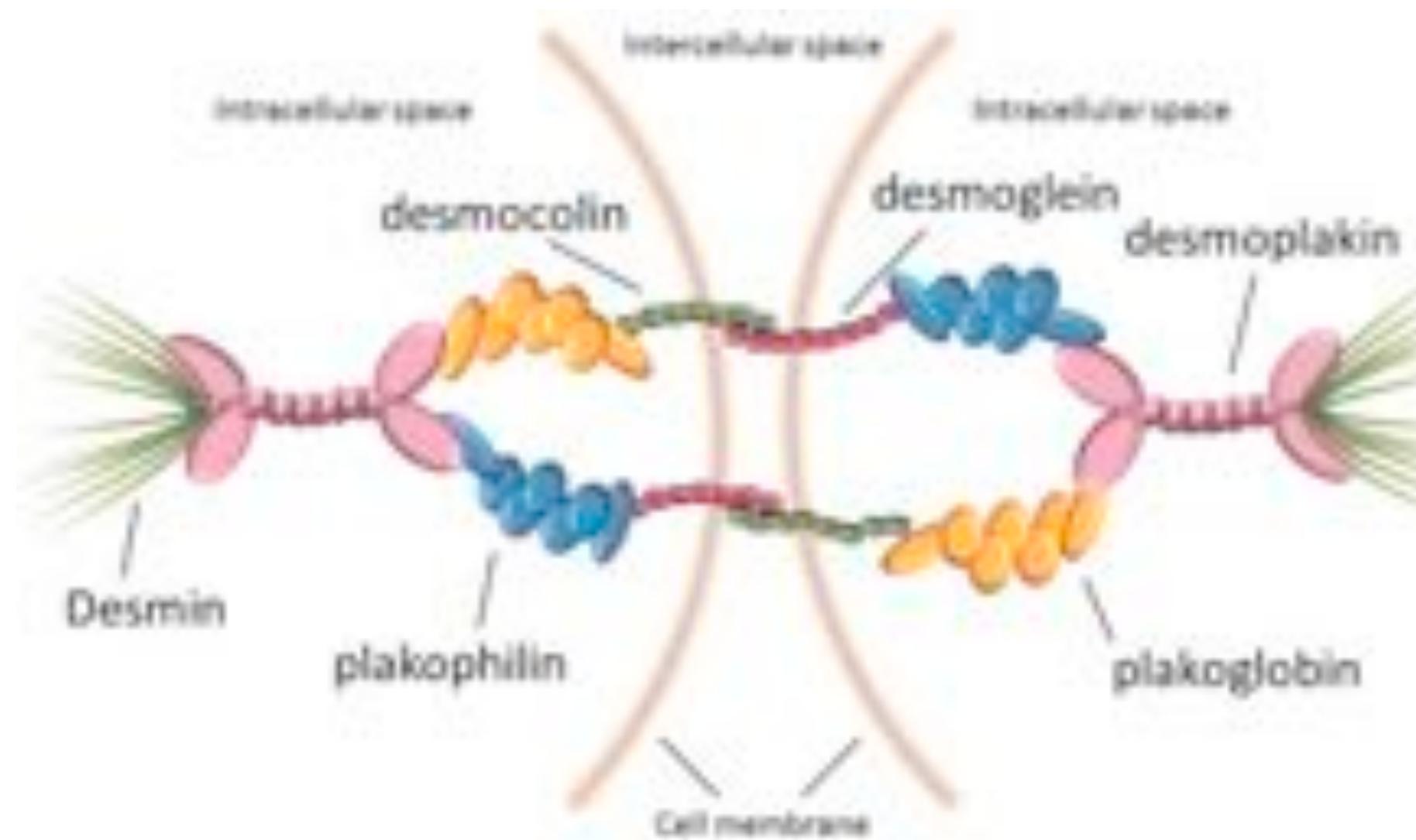
# Cardiomyopathy genes in pediatric inflammatory cardiomyopathy



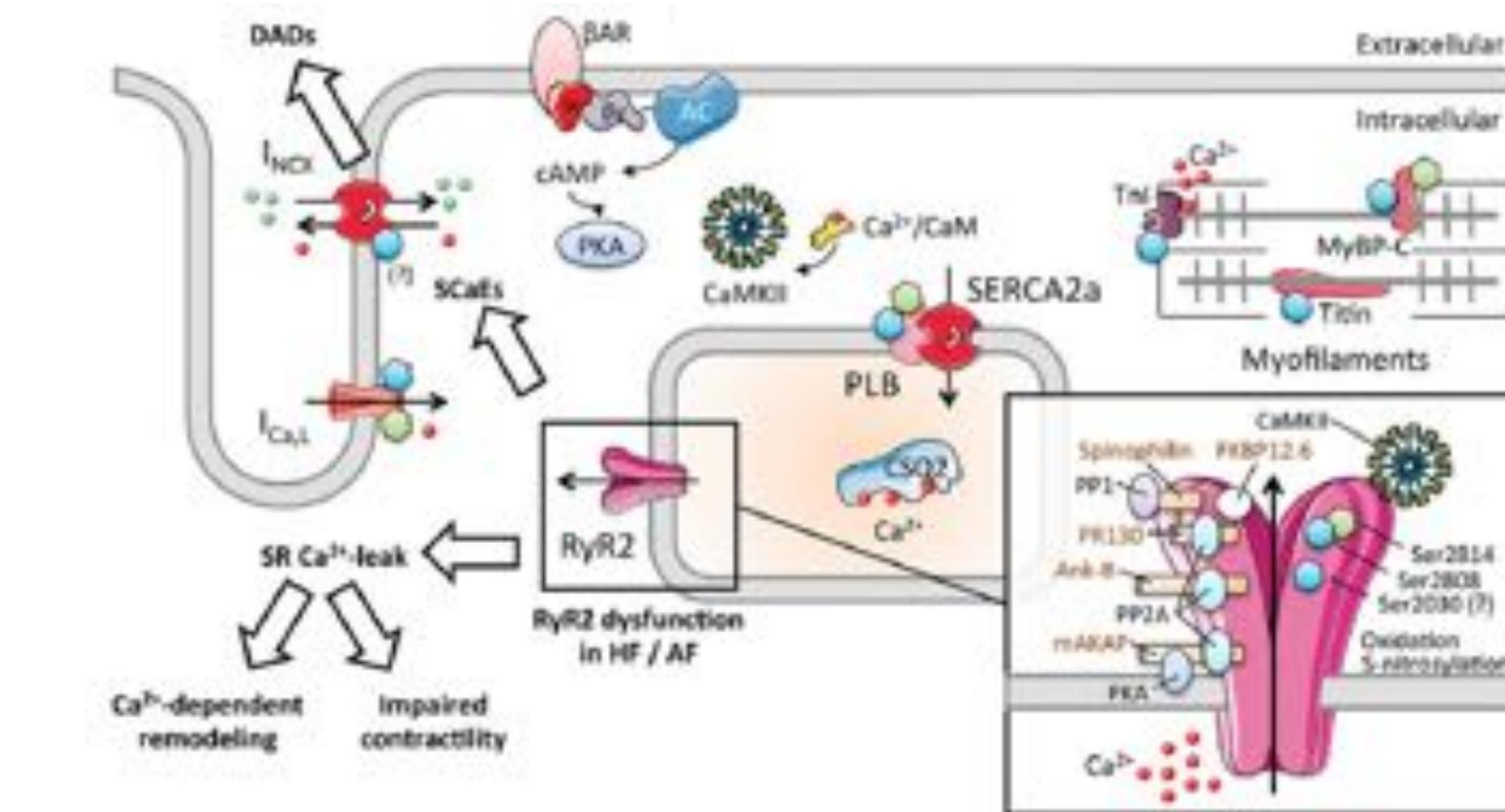
Costamere



Sarcomere



Desmosome



Ion channels

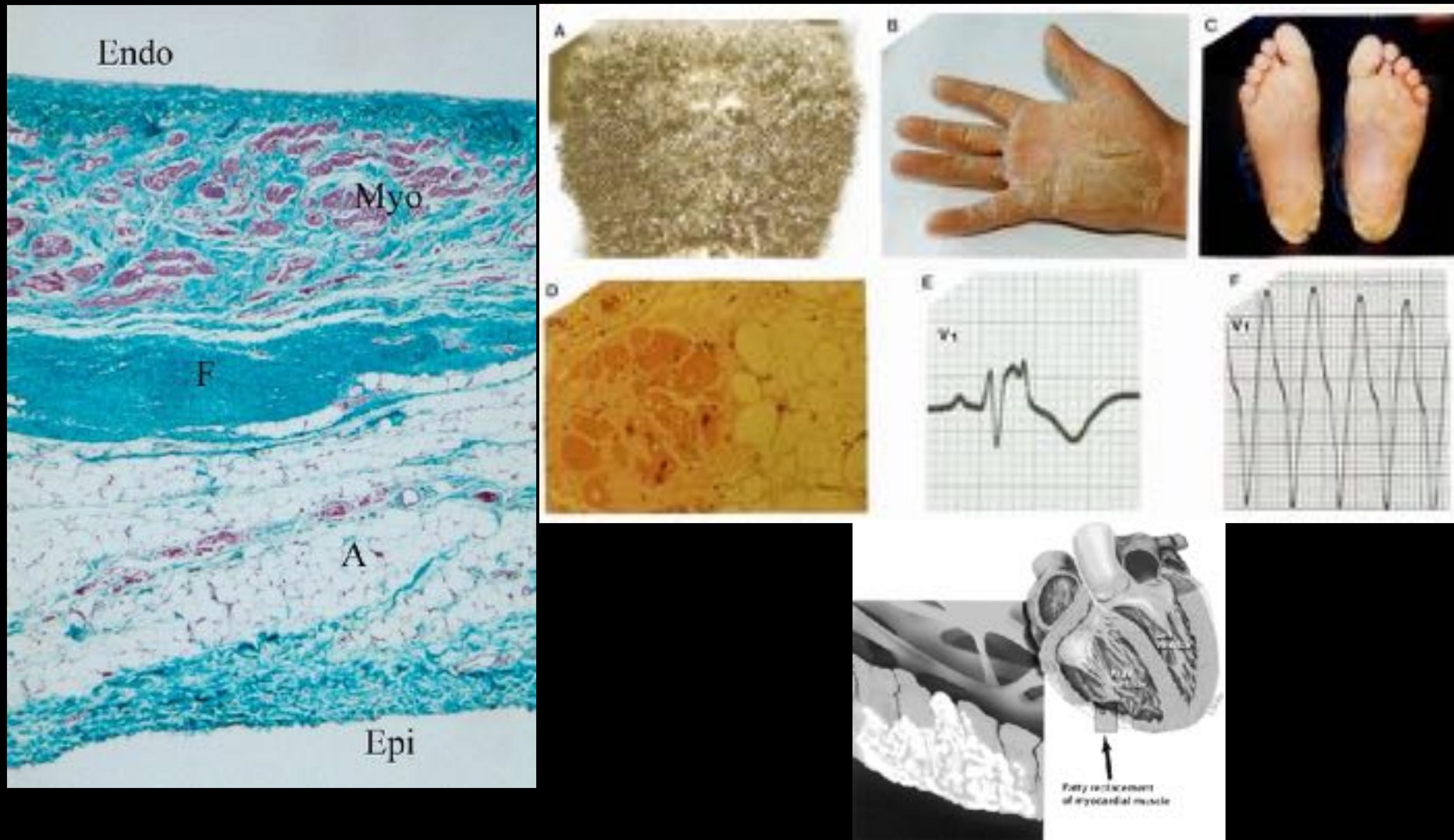
# **Formes familiales dominantes de cardiomyopathies hypertrophiques**

- 20% de formes familiales
- Objectifs de la prise en charge
  - Dépistage des facteurs de risque de MS
    - Antécédents familiaux
    - Adaptation tensionnelle à l'effort
    - TVNS
    - Epaisseur septale
    - Obstruction gauche

# **Formes familiales dominantes de cardiomopathies dilatées**

- <20% de formes familiales
- Insuffisance cardiaque progressive
- Pronostic très difficile à évaluer
  - Risque rythmique
  - Anémie
  - Nutrition
  - Réponse au traitement médical

# Dysplasie arythmogène du ventricule droit



# Enquête familiale

- Apparentés au 1er degré
- ECG
- Echocardiographie
- Enquête génétique

# Résumé de la démarche diagnostique

- Cardio standard + anamnèse
- ECG + Echocardiographie
- IRM + troponine pour myocardite
- Dysmorphologistes pour les formes syndromiques
- Métabolisme simple
  - Glycémie, corps cétoniques, lactates,
  - Chromato AO, profil acylcarnitines, carnitine T+L
  - Et c'est tout!



TATOO

