Cardiomyopathies

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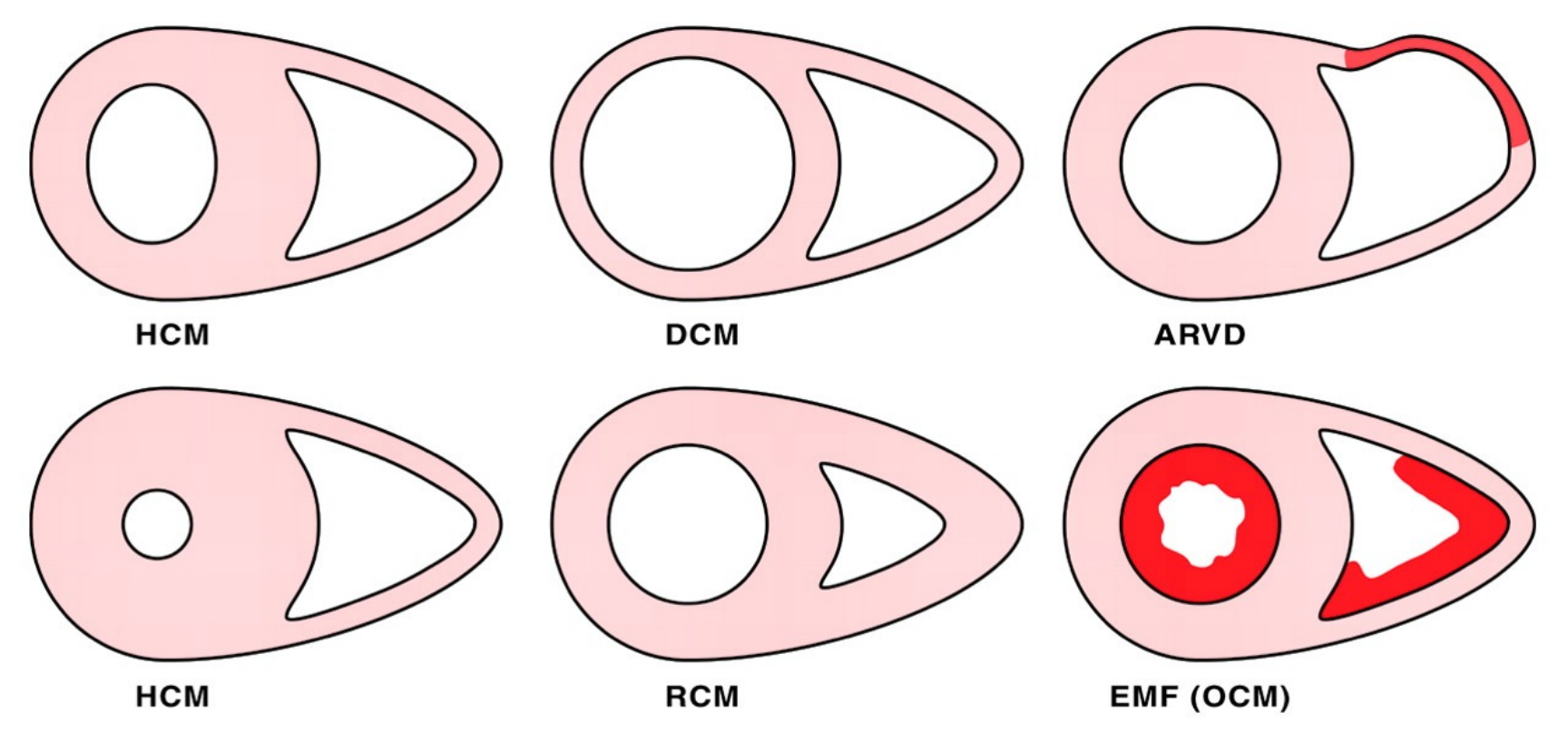
CENTRE FOR INHERITED CARDIOVASCULAR DISEASES

Definition

- **“A myocardial disorder in which the heart muscle is structurally and functionally abnormal, in the absence of coronary artery disease, hypertension, valvular disease and congenital heart disease sufficient to cause the observed myocardial abnormality.”**

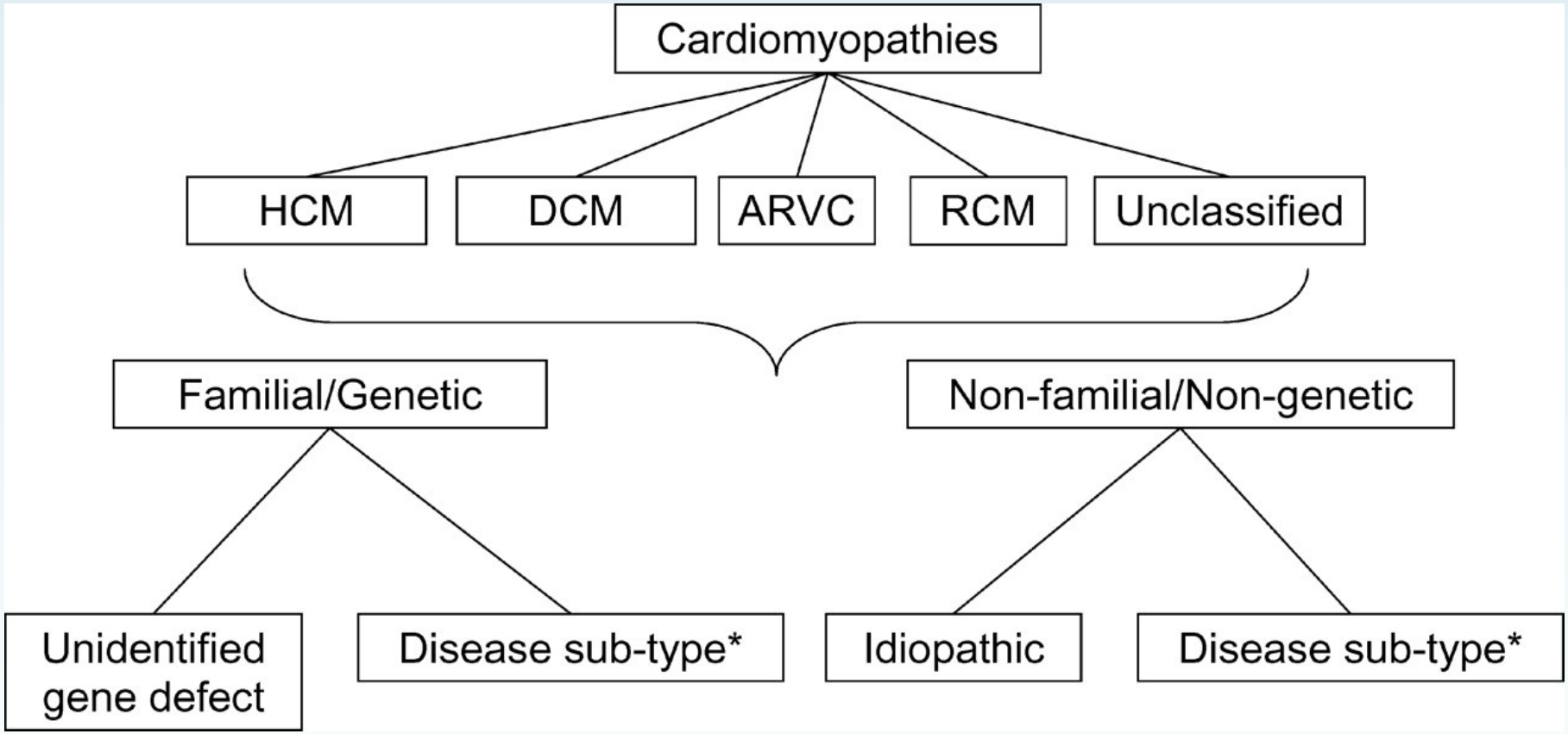
ESC Classification of Cardiomyopathies, 2008

Classification



Davies M J Heart 2000;83:469-474

Classification



Elliott P et al. Eur Heart J 2008;29:270-276

HYPERTROPHIC CARDIOMYOPATHY

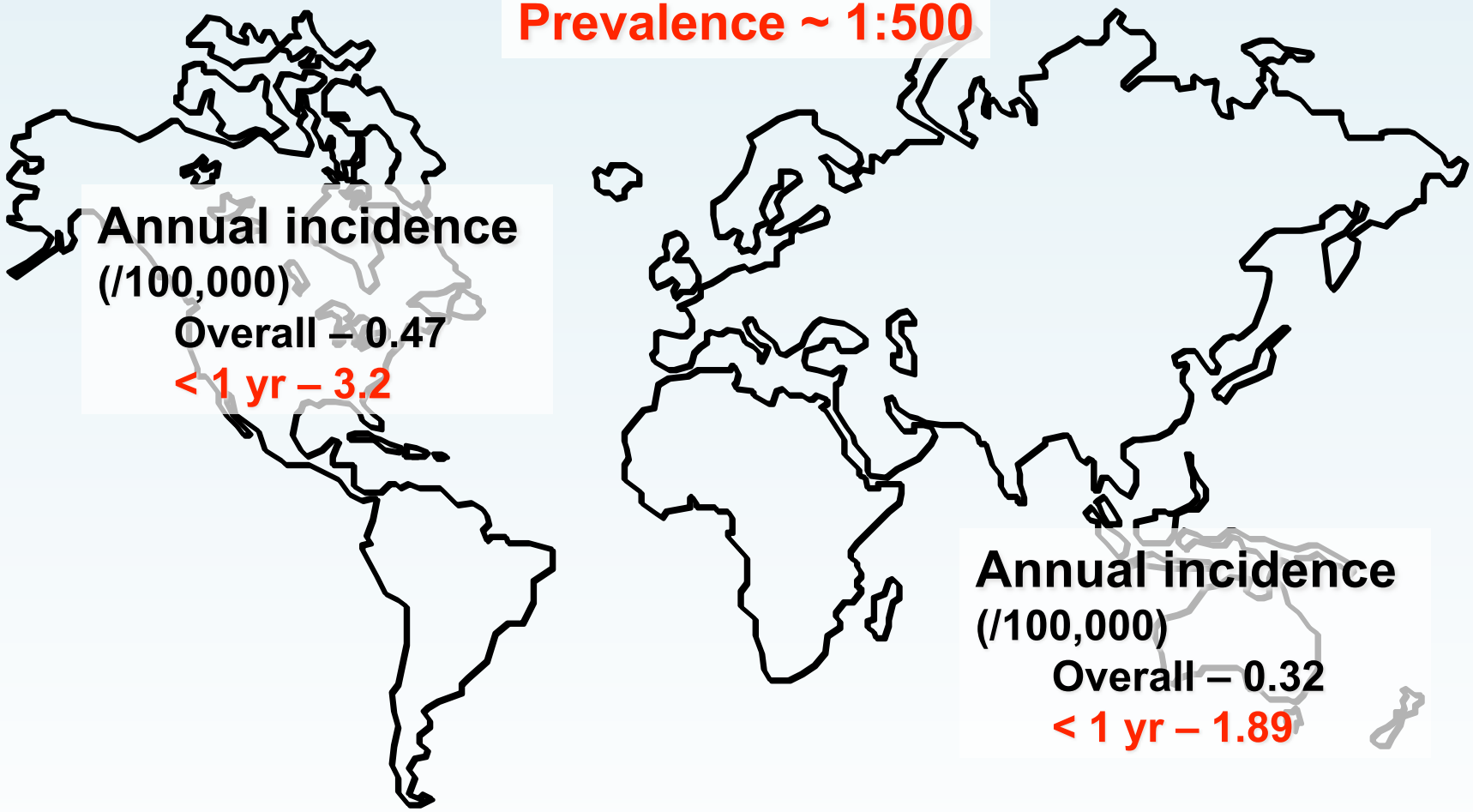
Definition

- **“Increased ventricular wall thickness or mass in the absence of loading conditions (hypertension, valve disease, congenital heart disease) sufficient to cause the observed degree of hypertrophy”**

ESC Classification of Cardiomyopathies, 2008

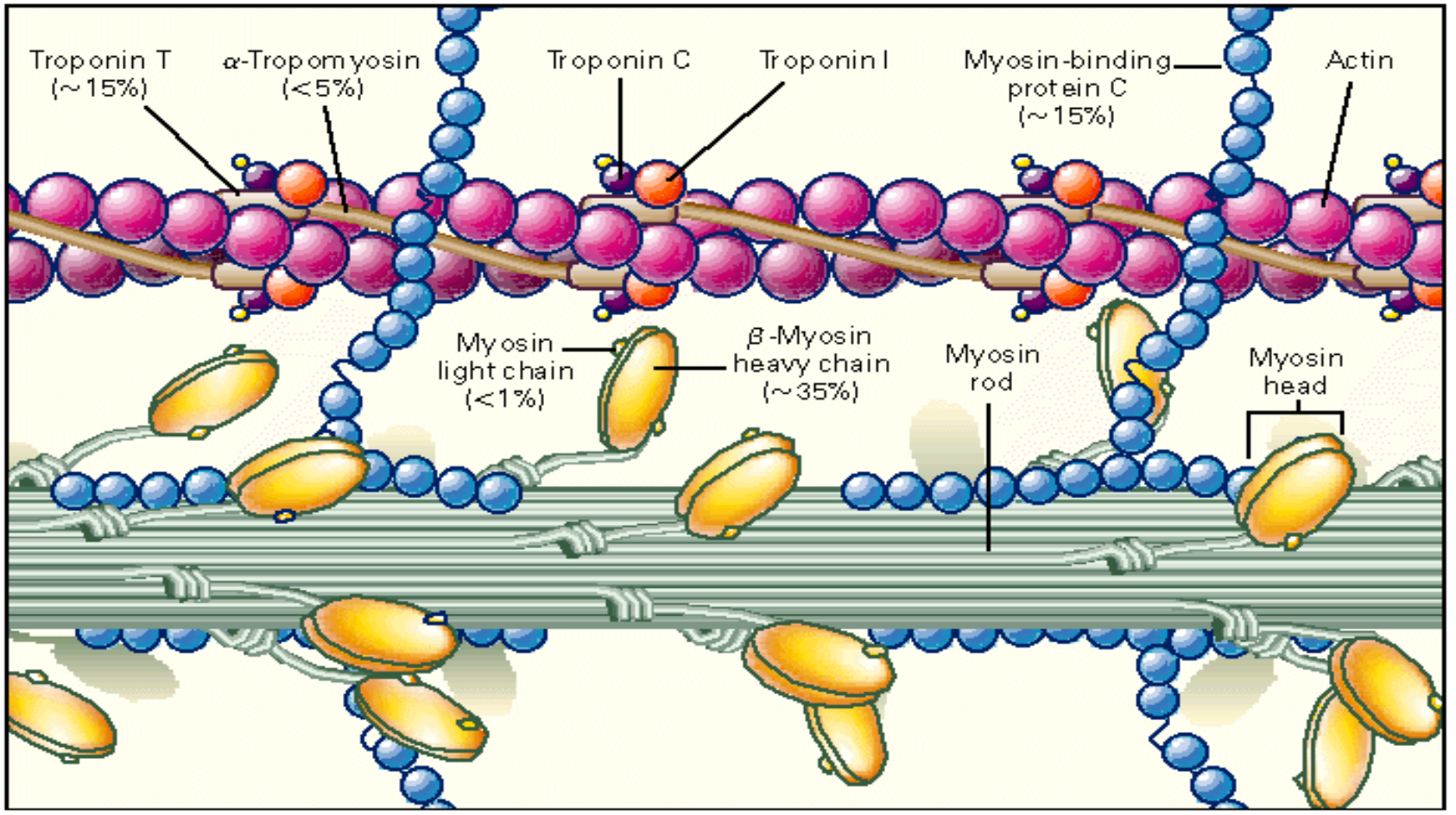
  - **Maximal LV wall thickness >2 SD above mean corrected for BSA +/- age**

Epidemiology

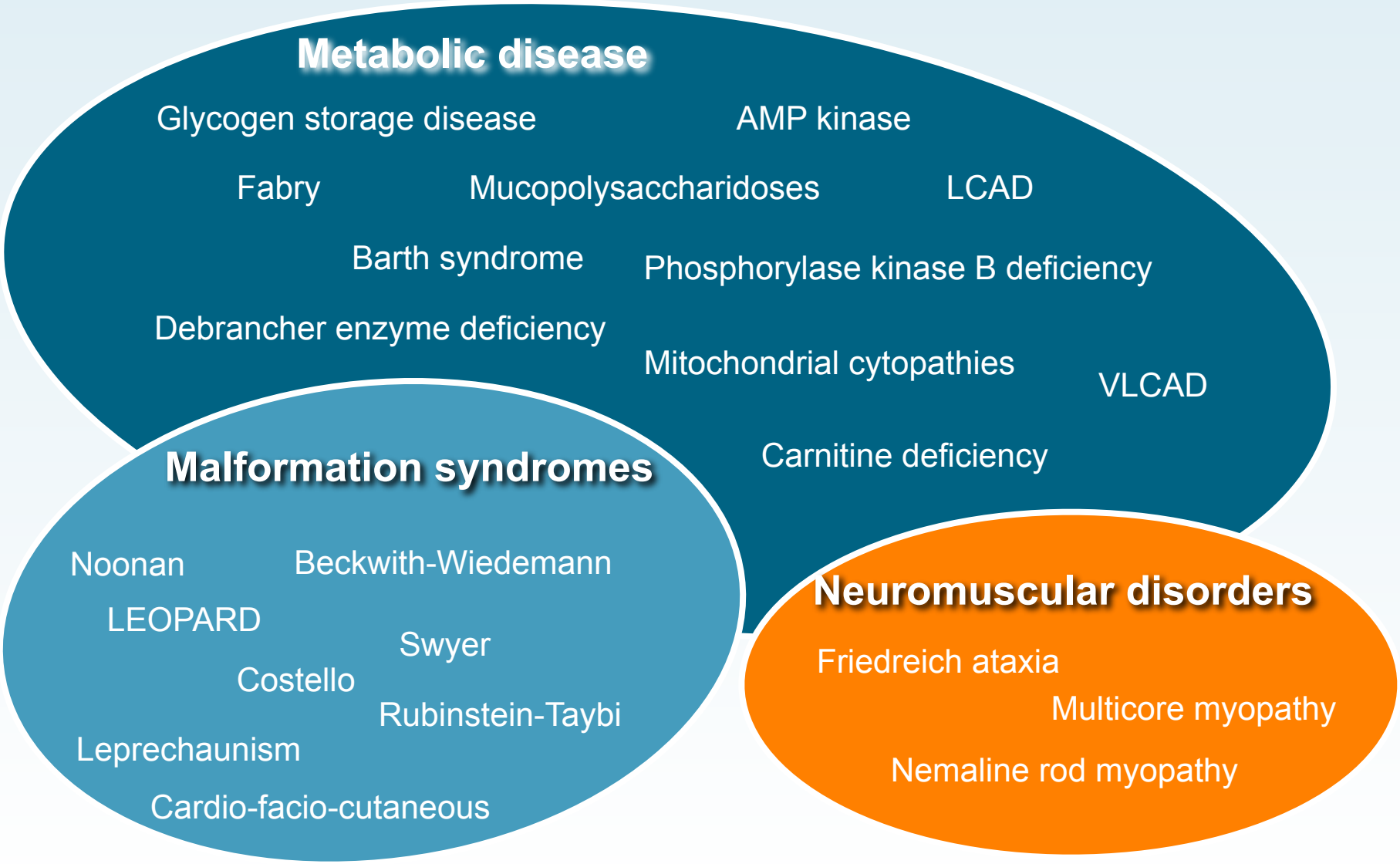


Familial HCM

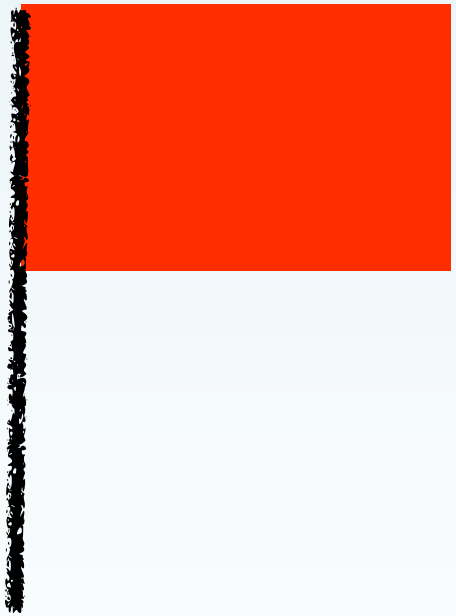
- >50% of HCM is familial



Causes of HCM in infants and children



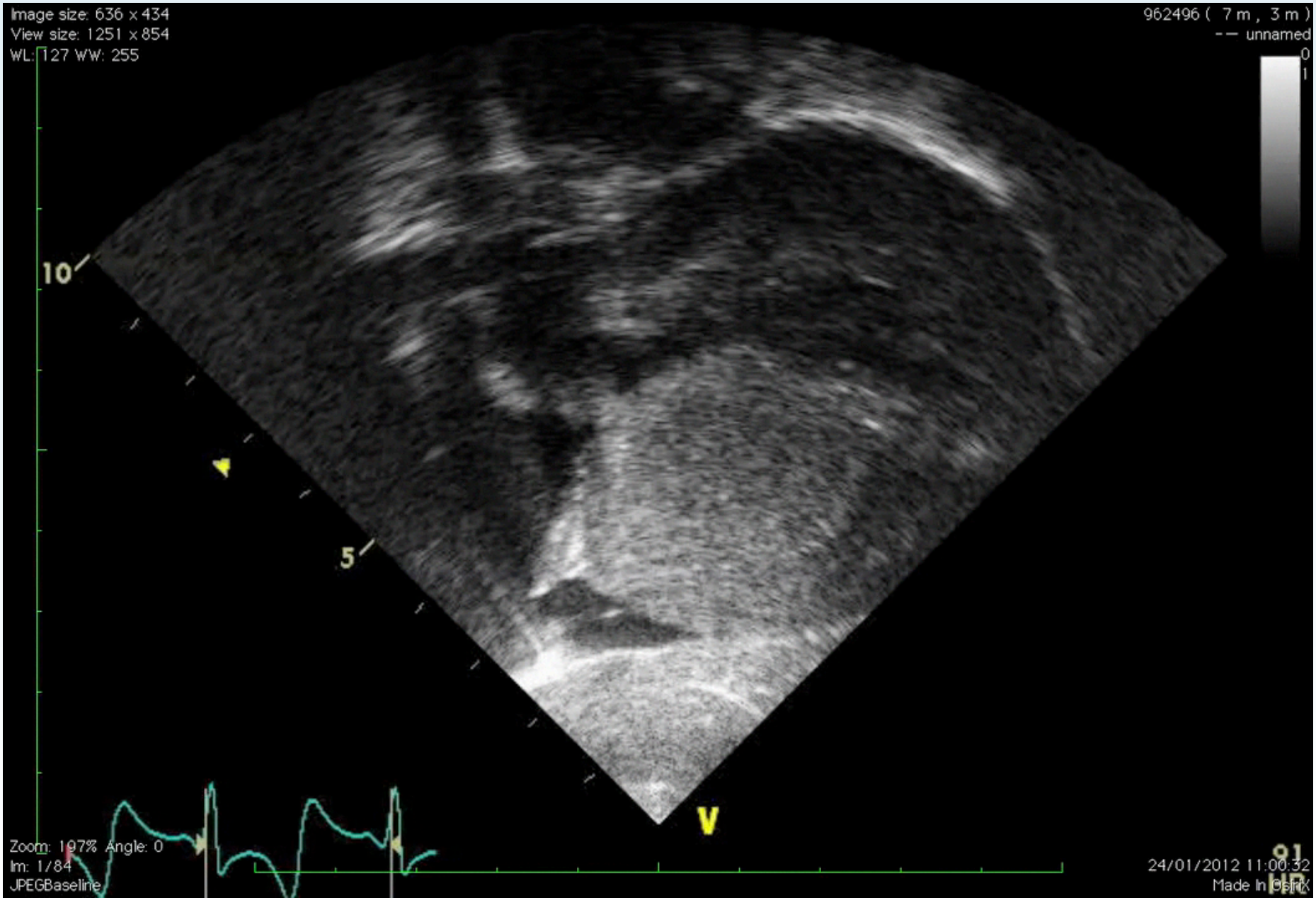
Echocardiographic “red flags”: Aetiology



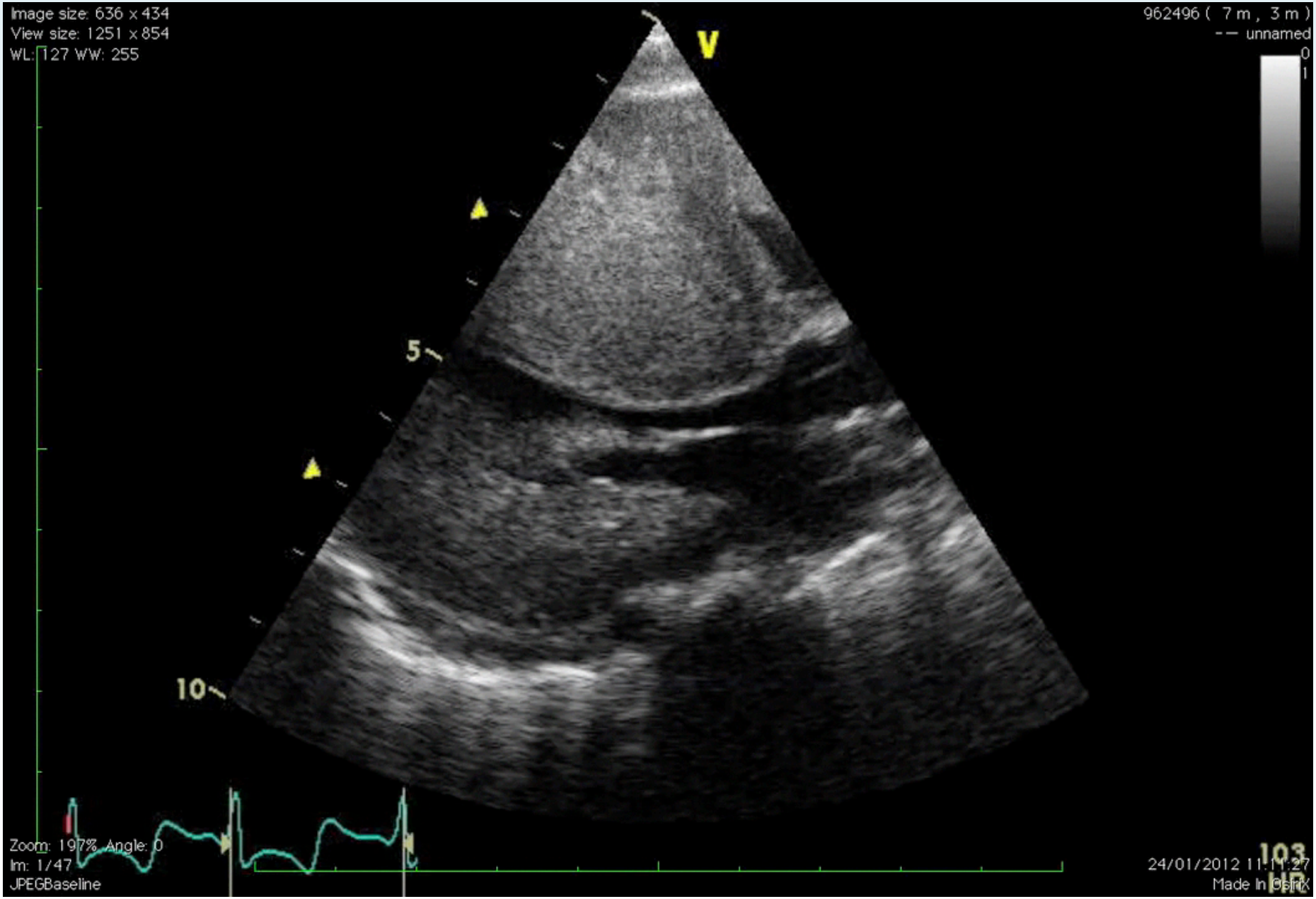
Echocardiographic clues to aetiology in paediatric HCM

	PRKAG2	Mitochondrial	Danon	GSD	Noonan	Friedreich ataxia
Concomitant RVH			✔	✔	✔	
Concentric LVH		✔		✔		✔
Extreme concentric LVH			✔	✔		
Global hypokinesia (with/without LV	✔	✔	✔	✔		(✔)

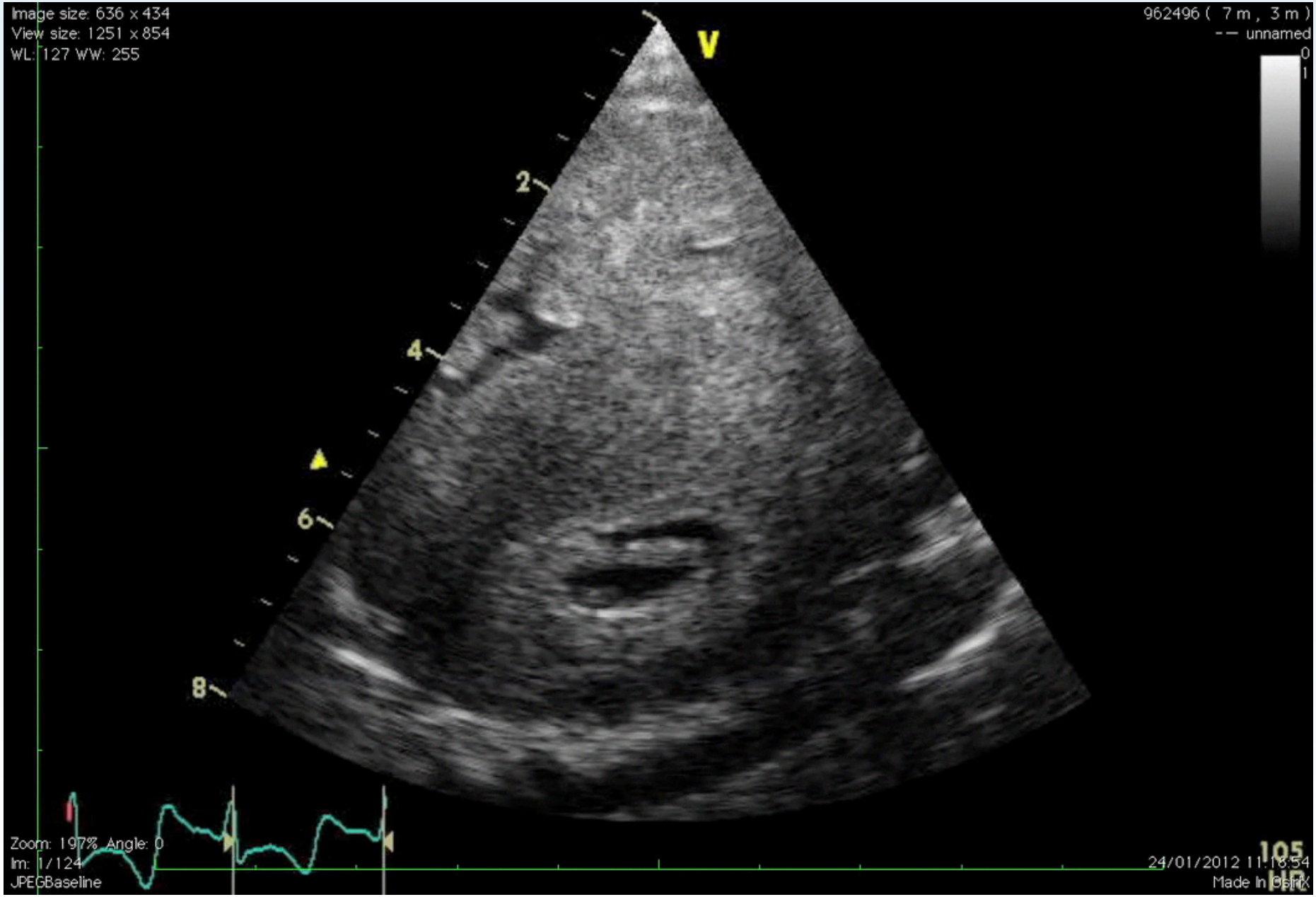
Pompes disease



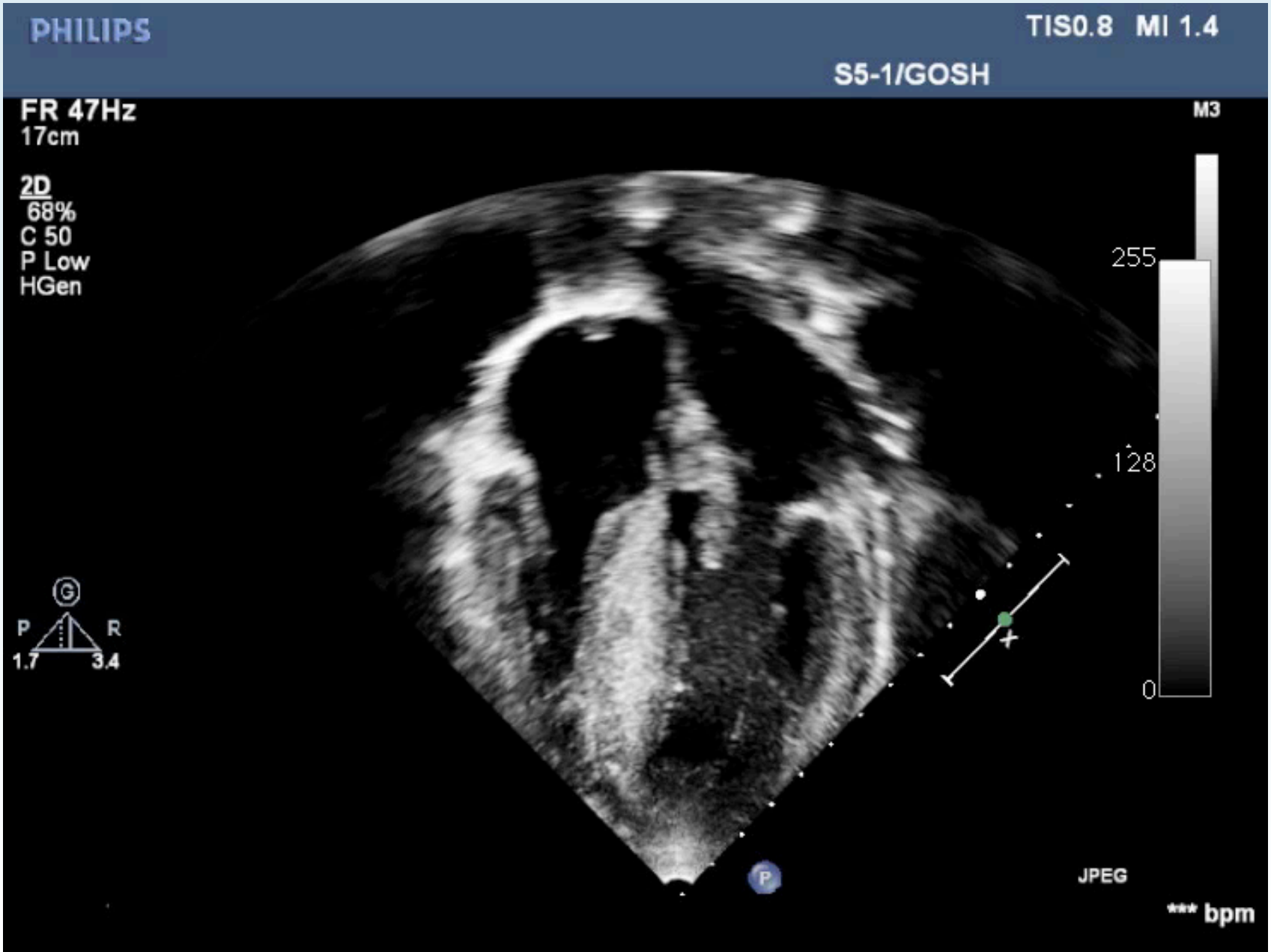
Pompes disease



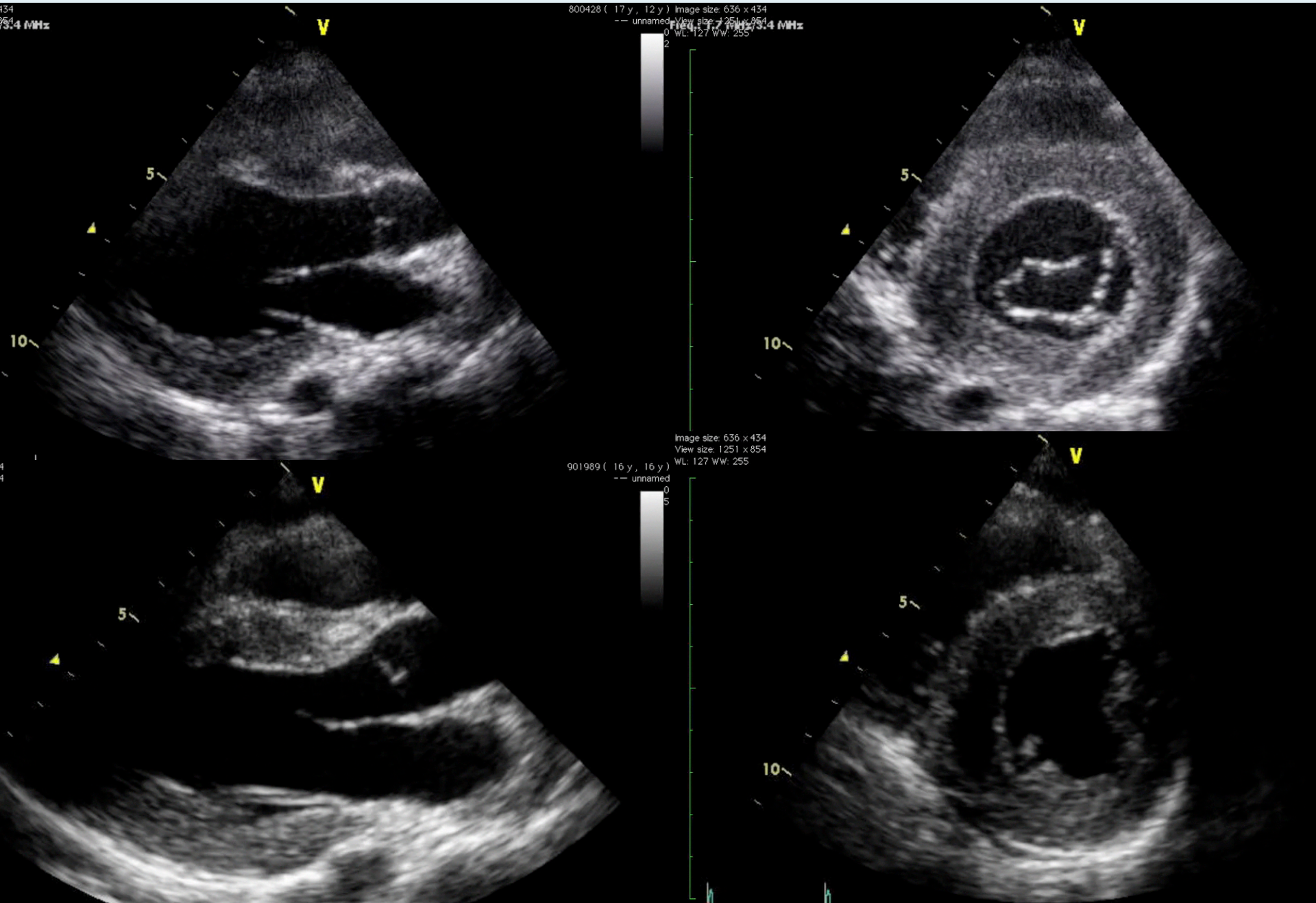
Pompes disease



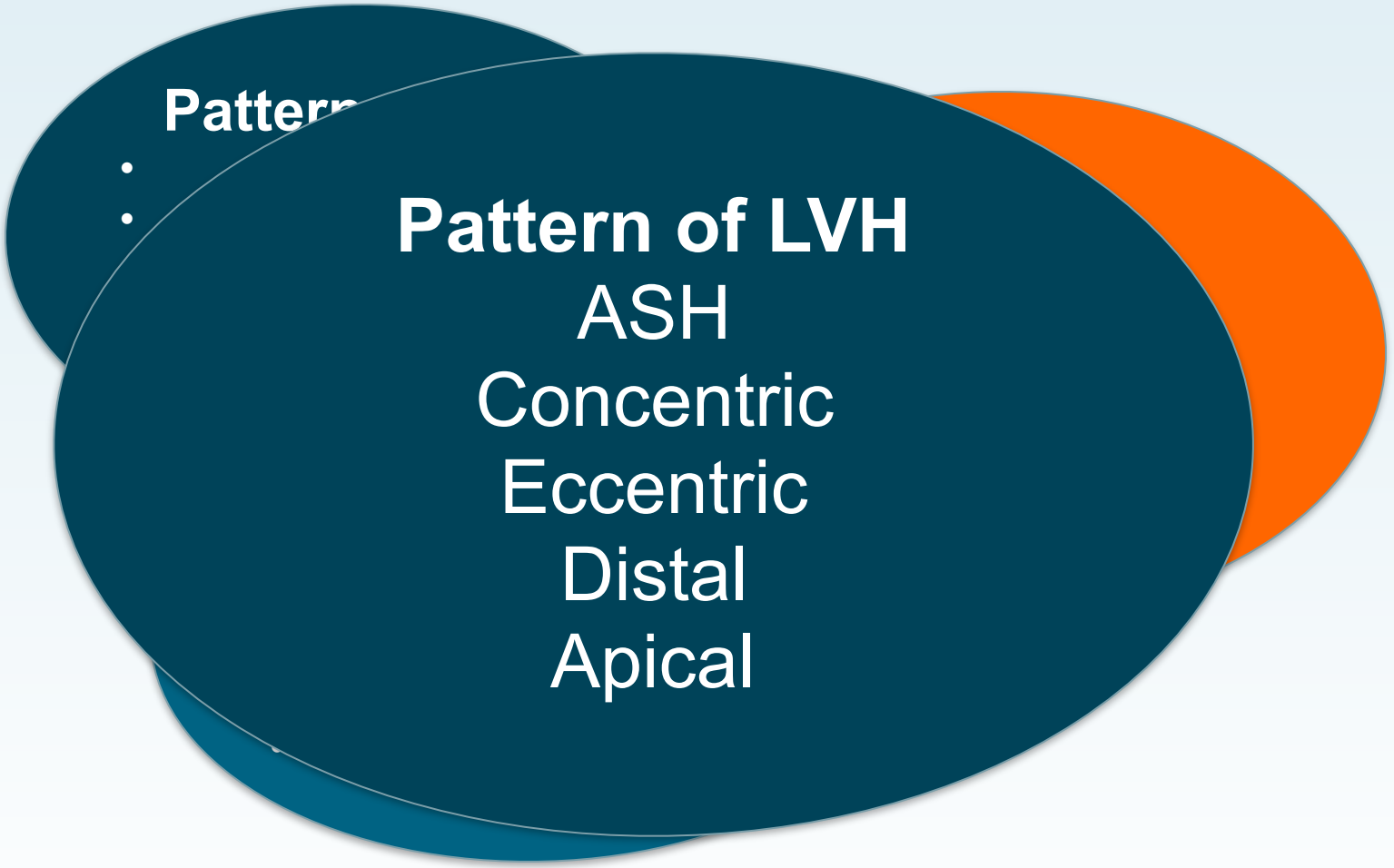
Biventricular hypertrophy



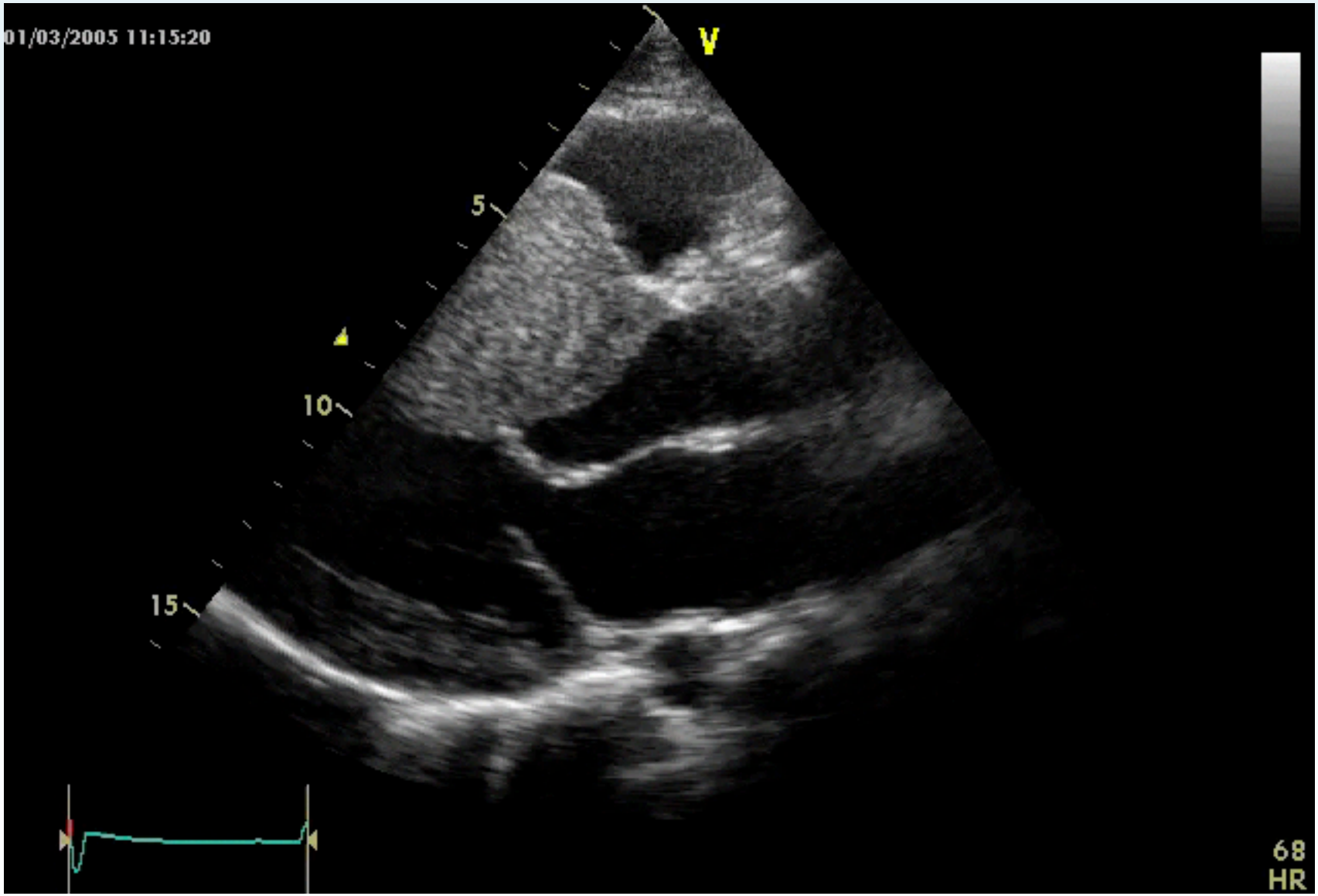
Concentric LVH



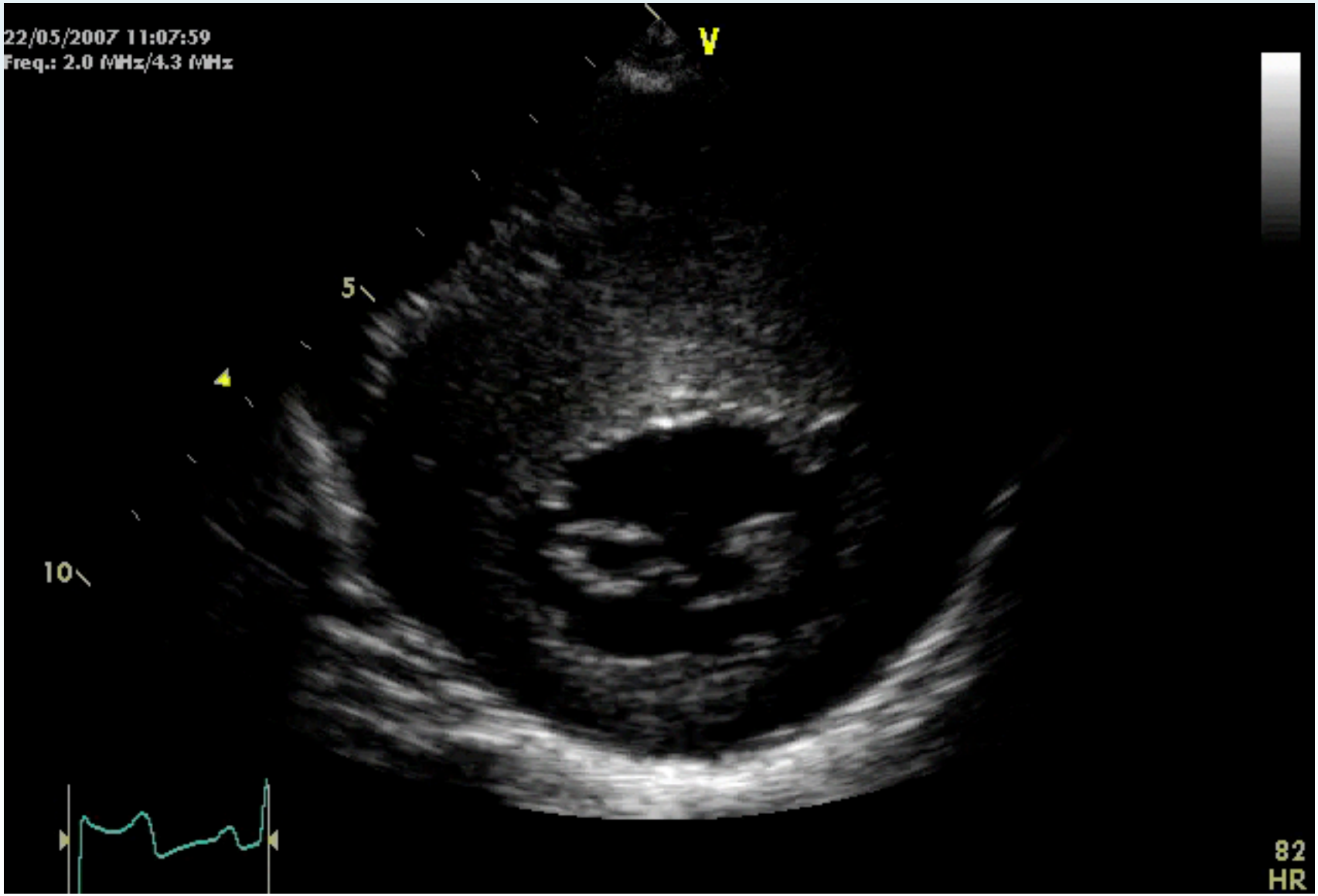
Echocardiography



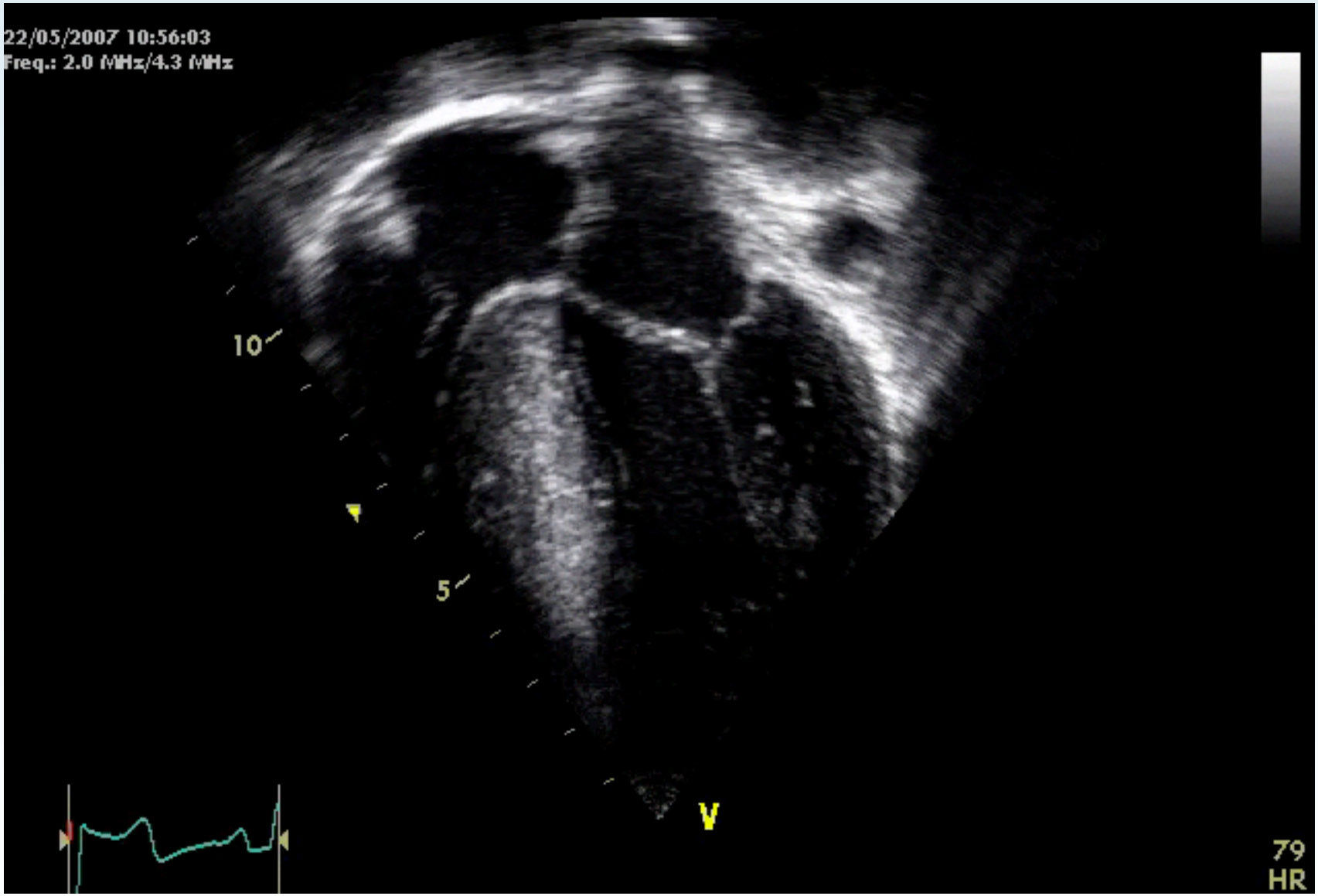
Asymmetric septal hypertrophy (ASH)



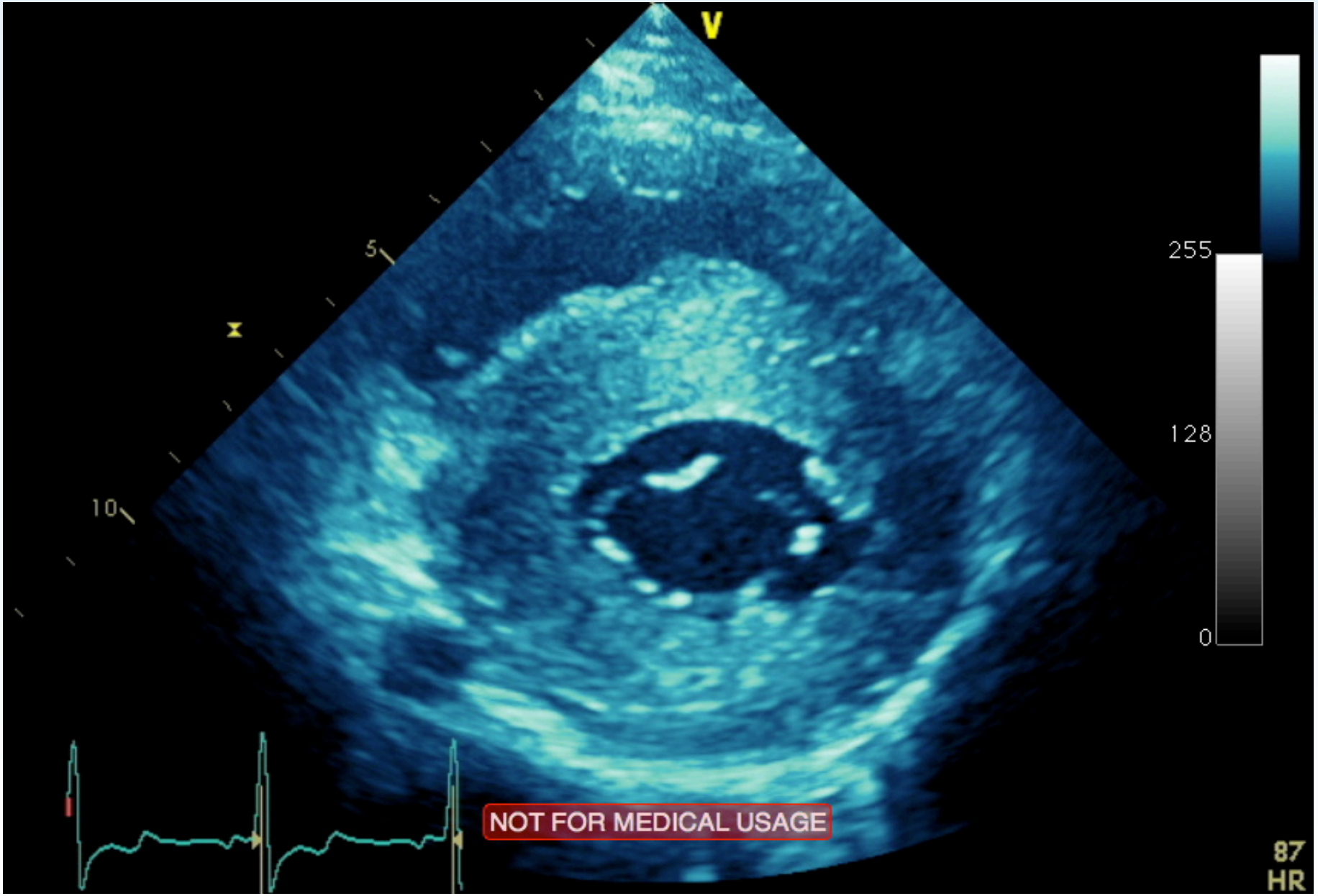
ASH

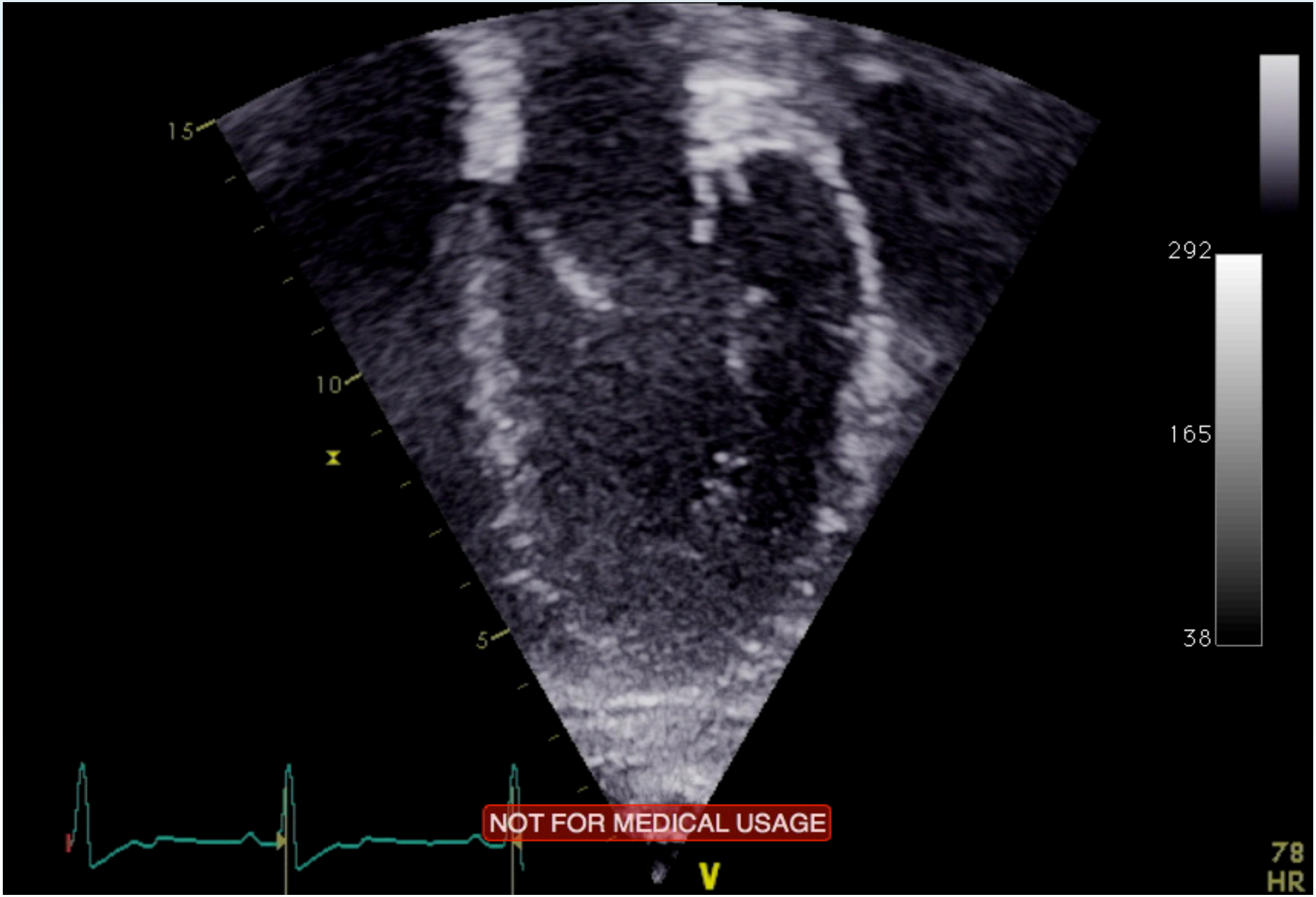


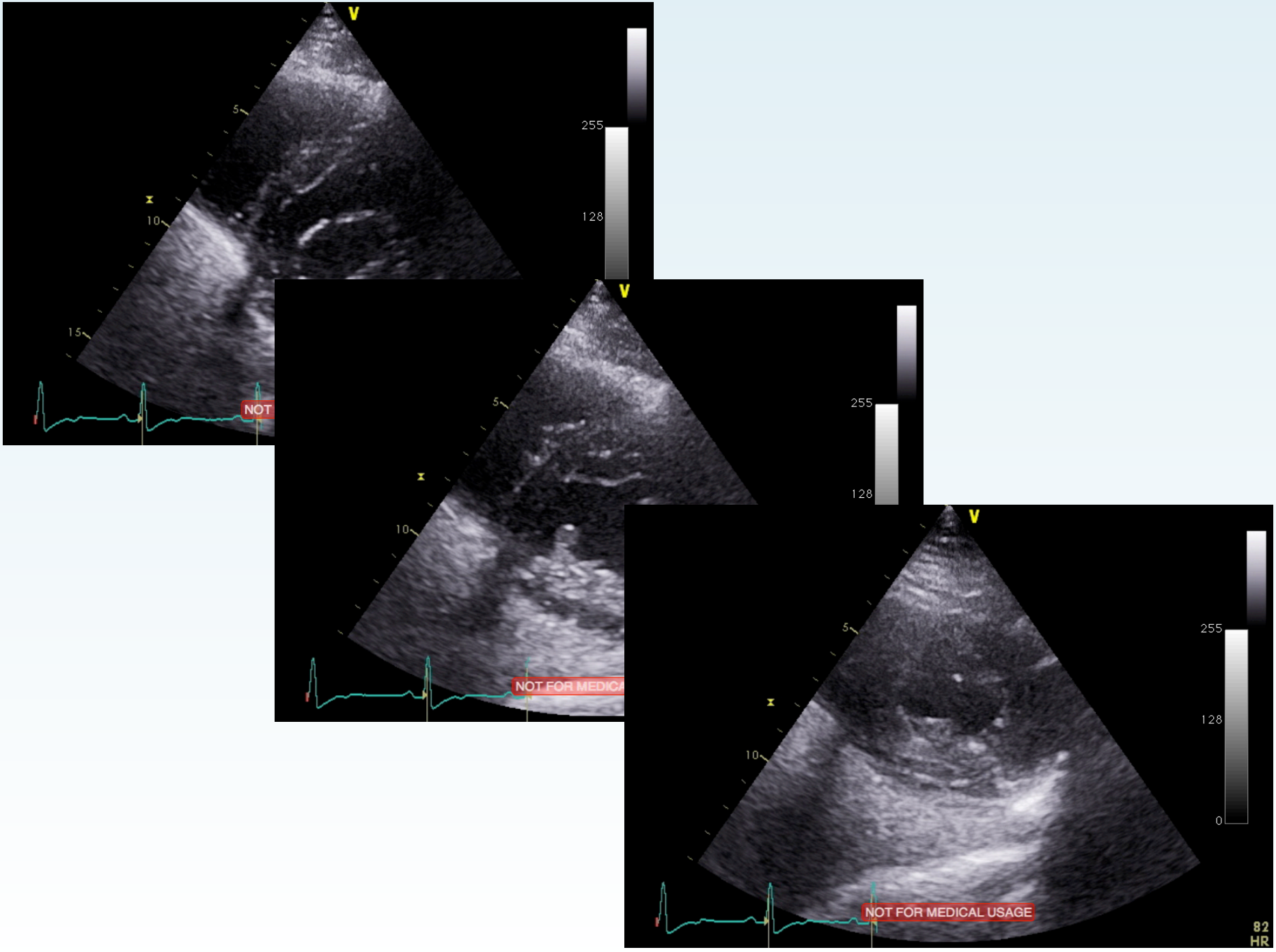
ASH



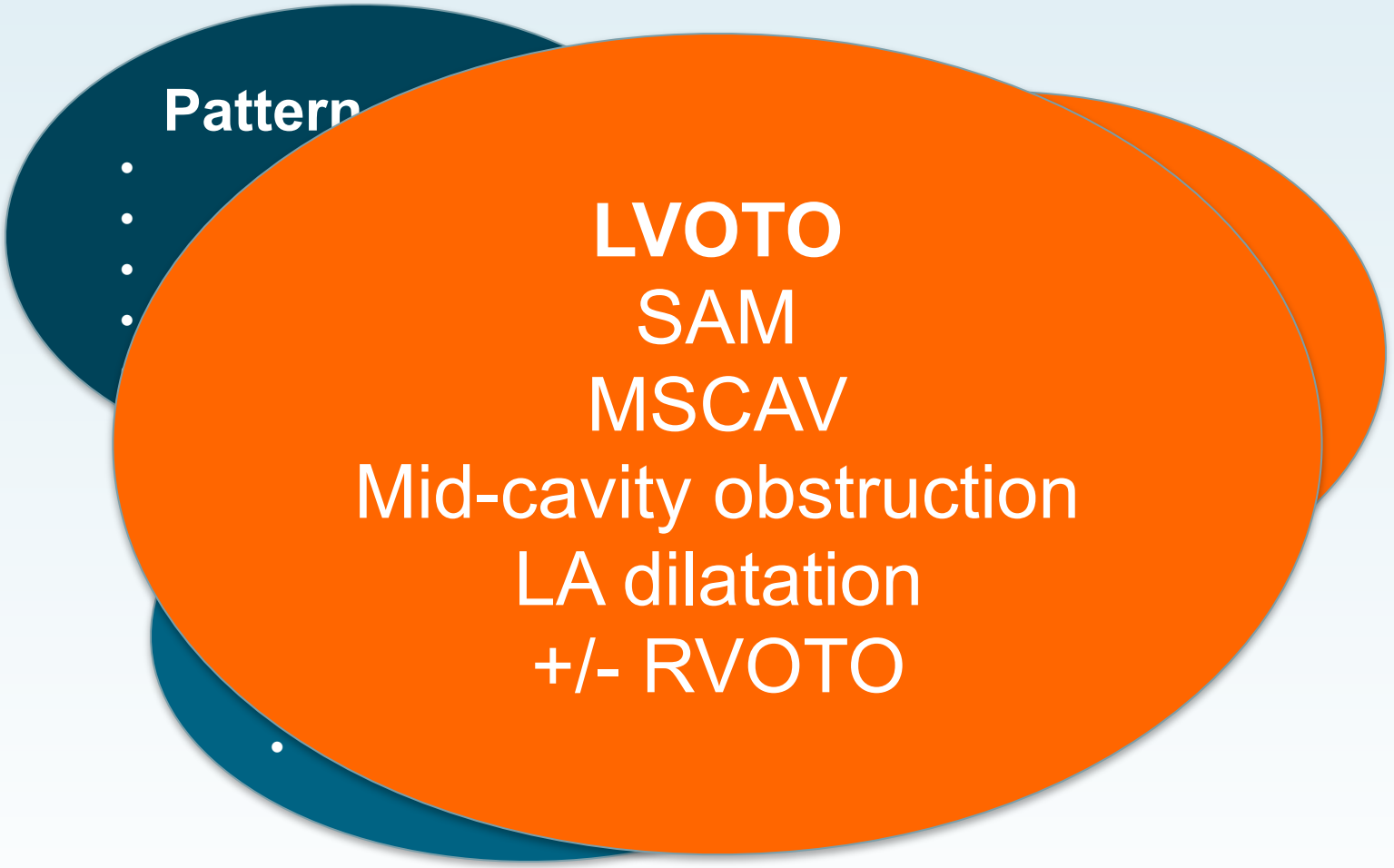
Concentric LVH

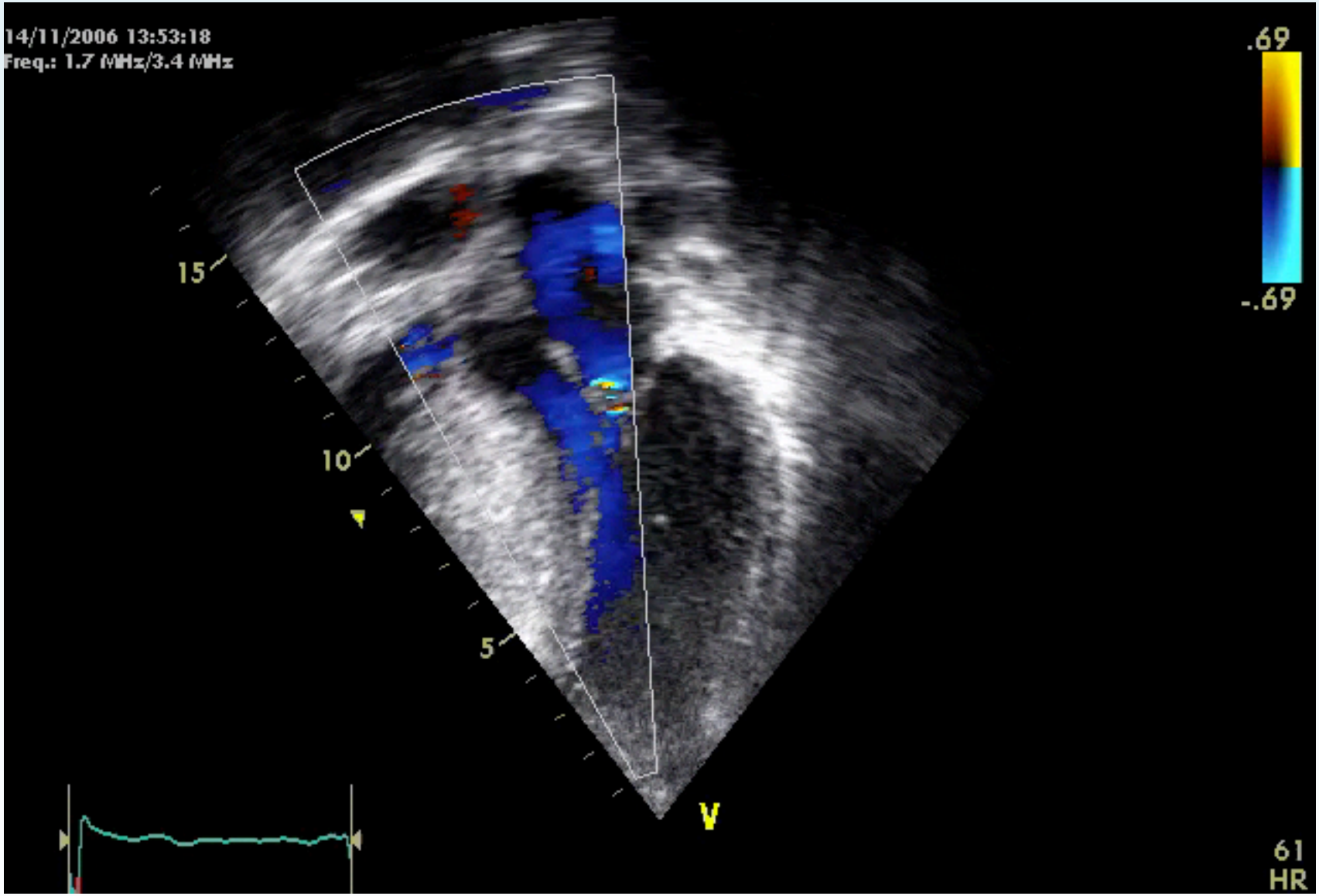


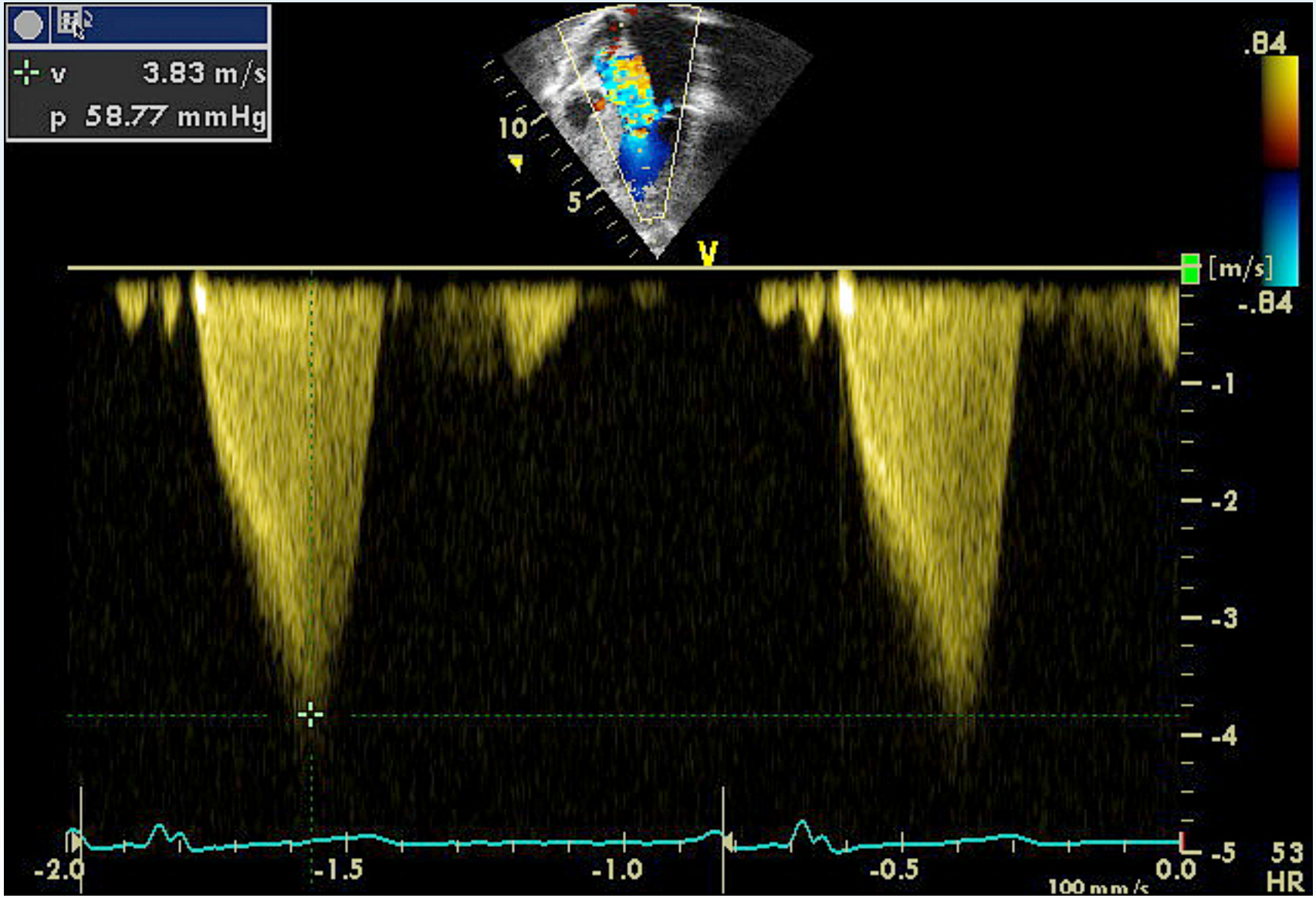

15
10
5
292
165
38
NOT FOR MEDICAL USAGE
78
HR

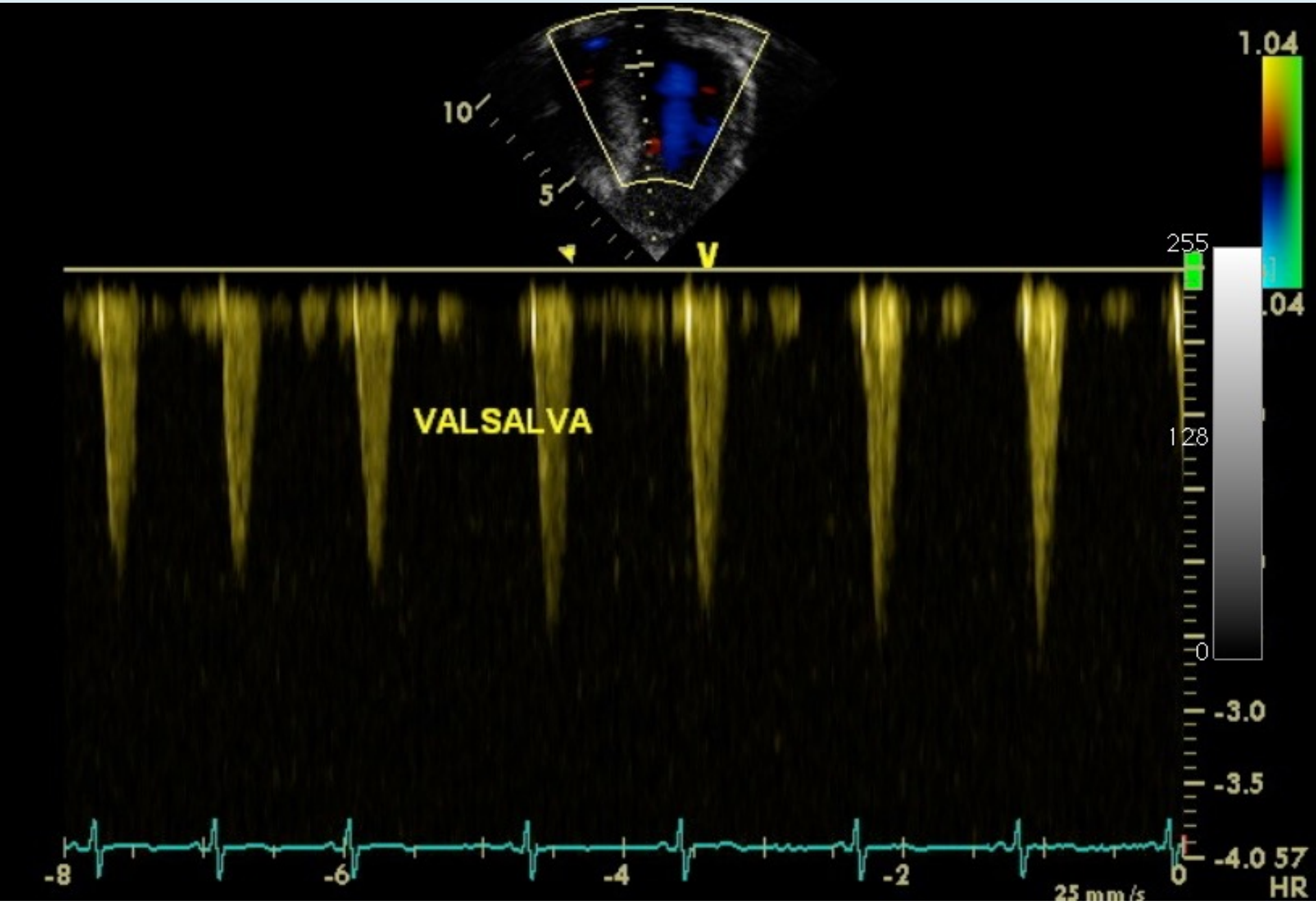

V
5
10
15
255
128
NOT
V
5
10
255
128
NOT FOR MEDICA
V
5
10
255
128
0
NOT FOR MEDICAL USAGE
82
HR

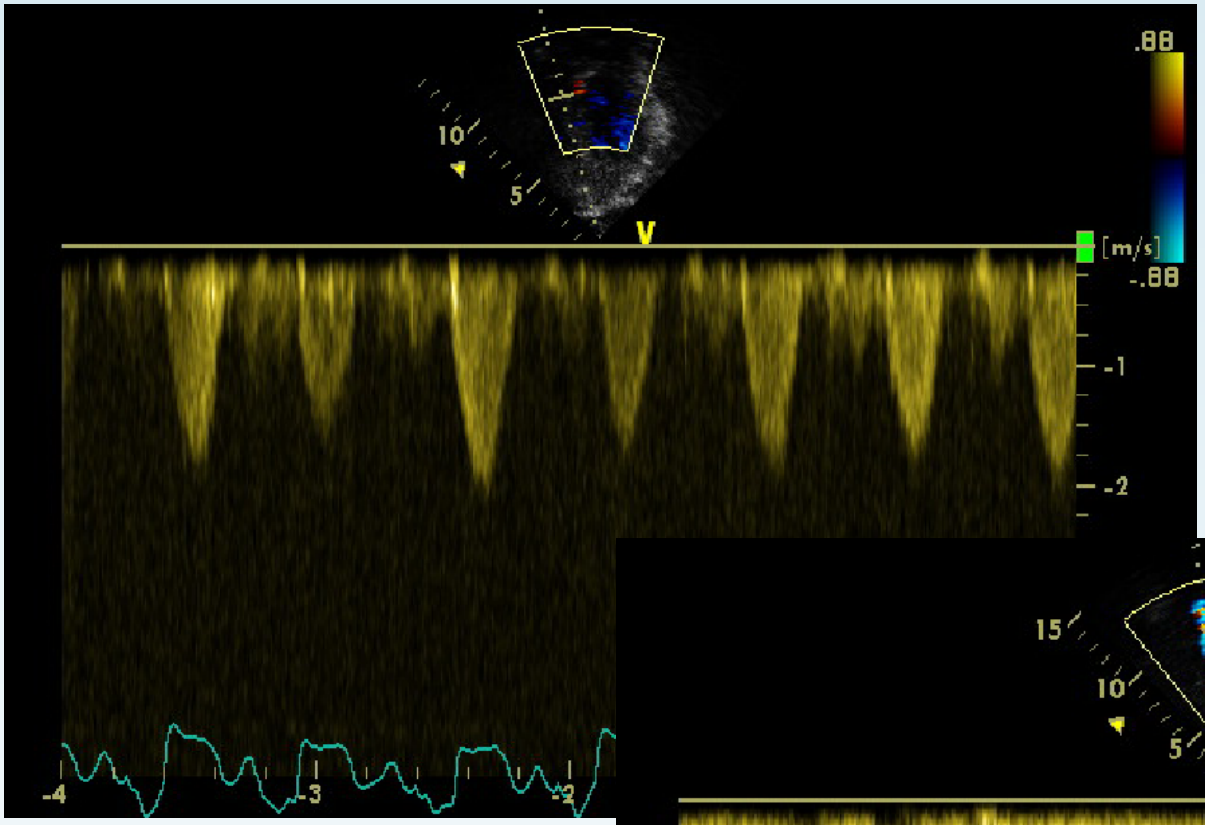
Echocardiography



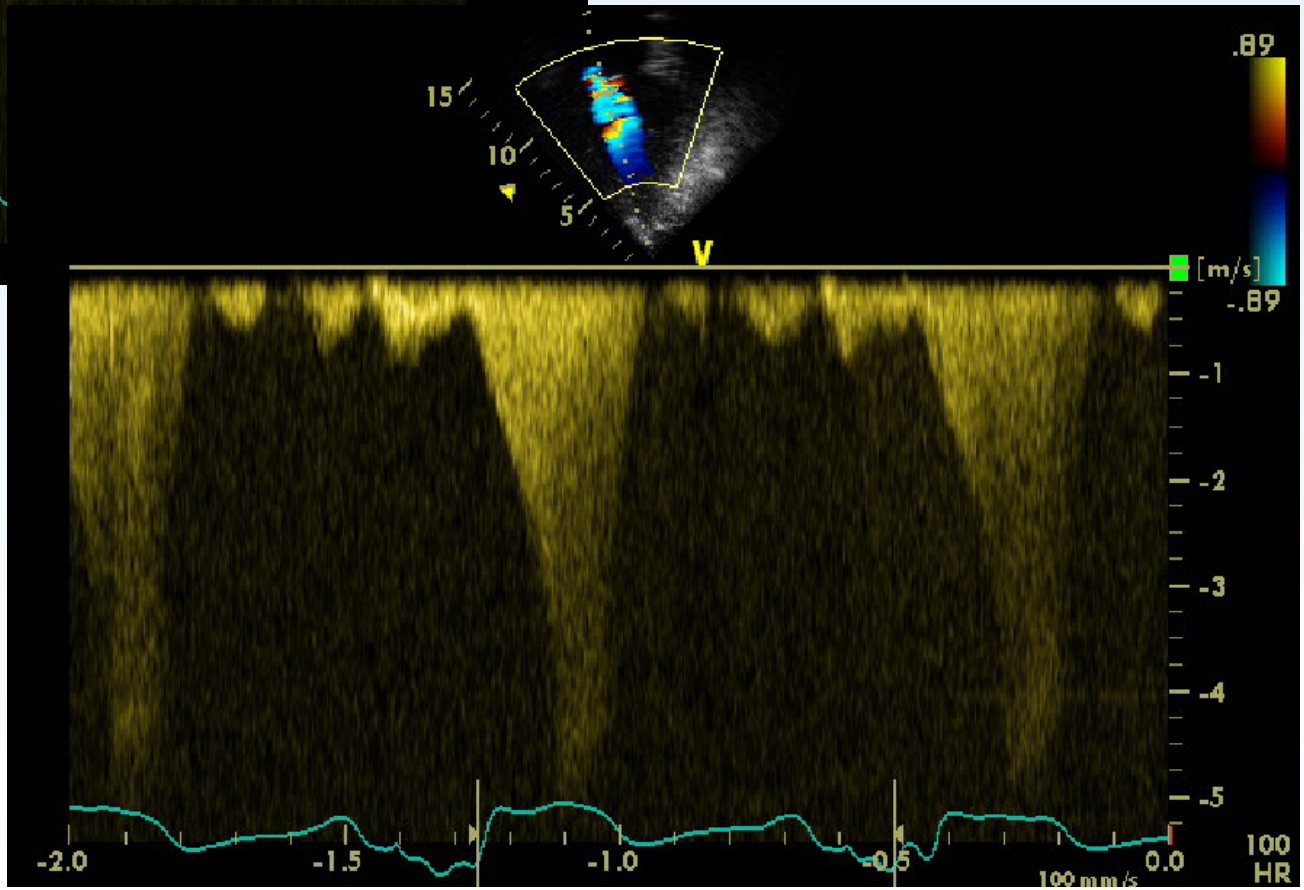

14/11/2006 13:53:18
Freq.: 1.7 MHz/3.4 MHz
.69
-.69
15
10
5
V
61
HR


v 3.83 m/s
p 58.77 mmHg
10
5
V
.84
[m/s]
-.84
-1
-2
-3
-4
-5
-2.0
-1.5
-1.0
-0.5
0.0
100 mm/s
53
HR

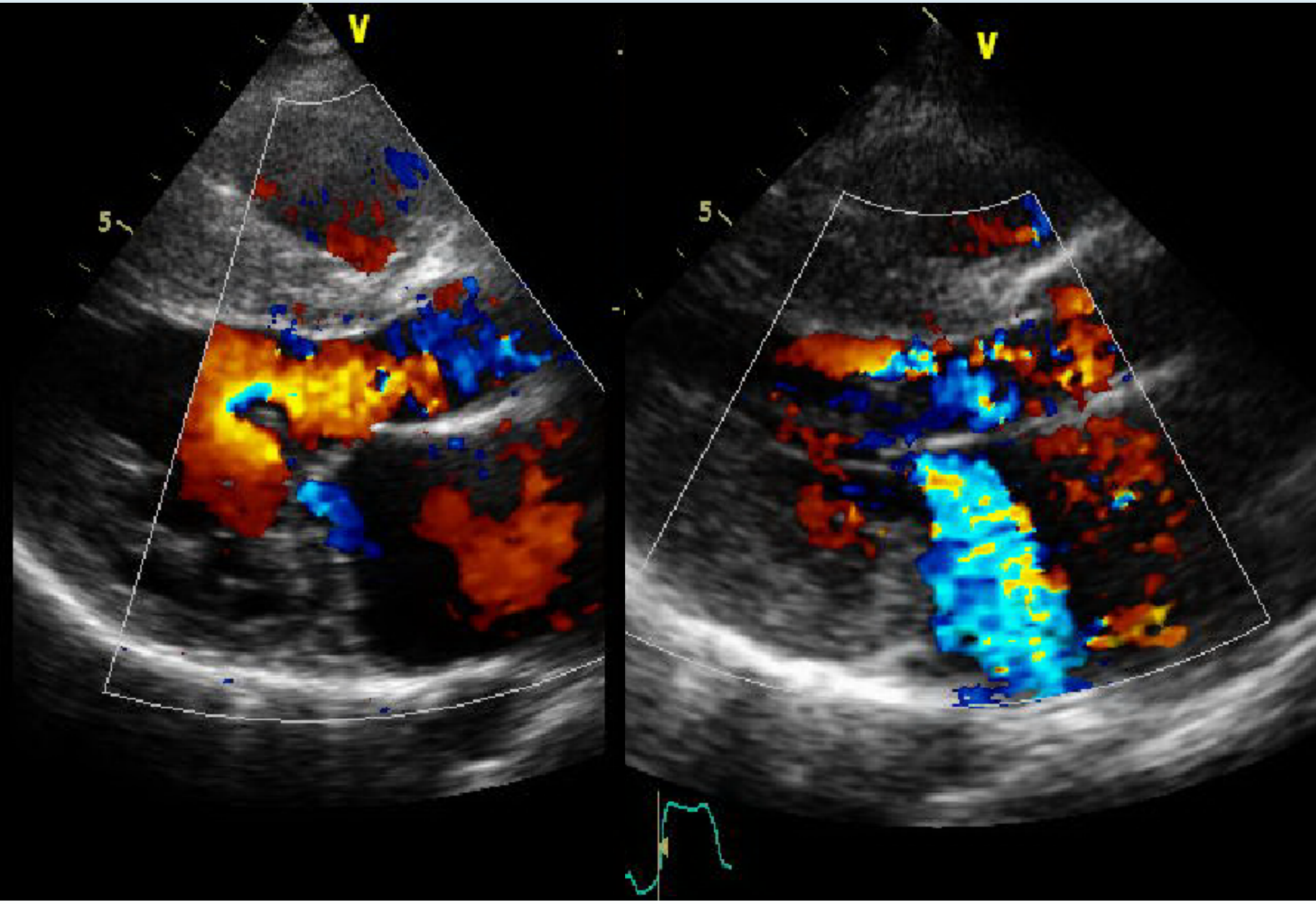

10
5
1.04
255
128
0
-3.0
-3.5
-4.0
57
HR
VALSALVA
-8
-6
-4
-2
0
25 mm/s

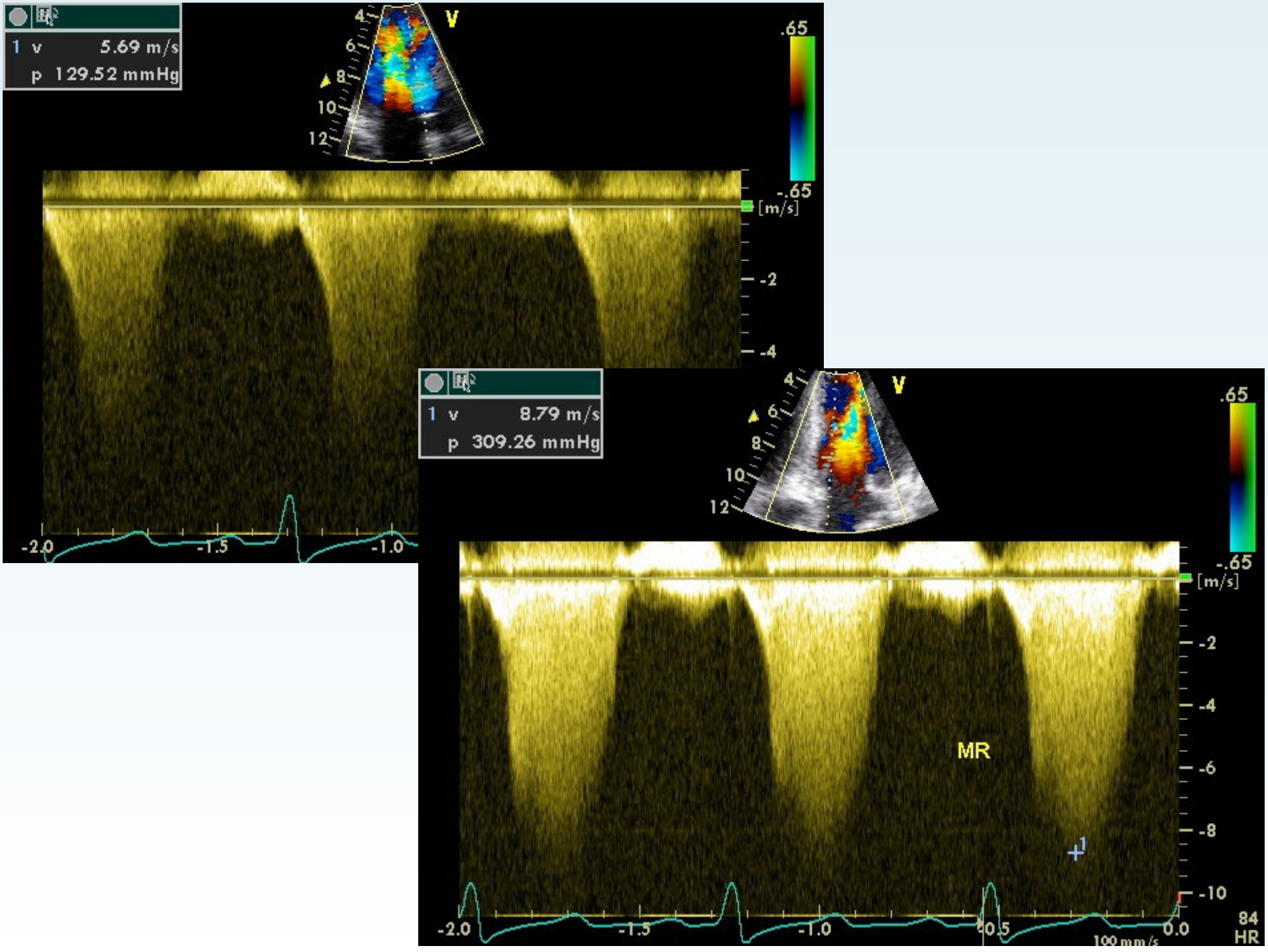


Rest

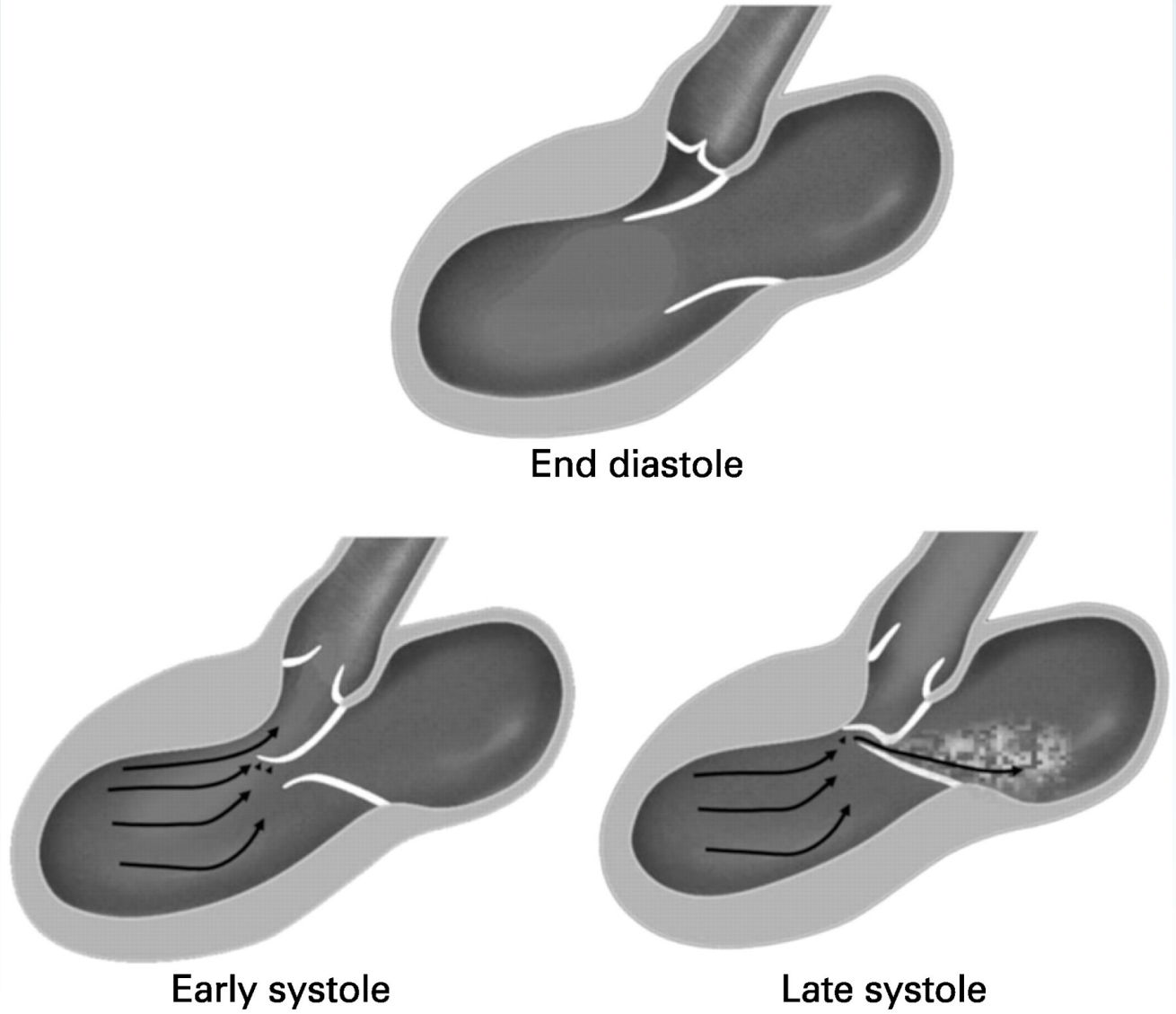


Exercise

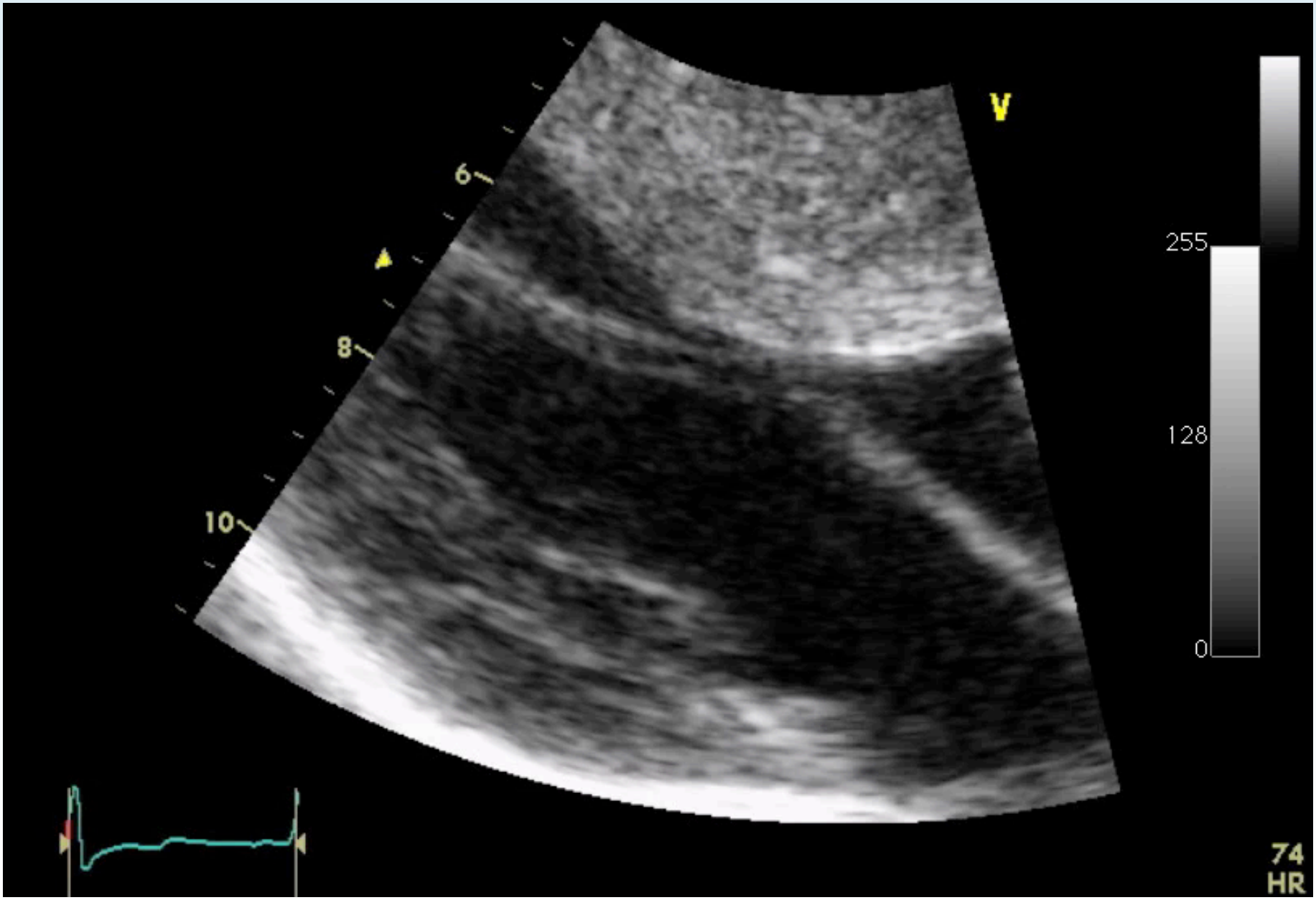

V
5
V
5

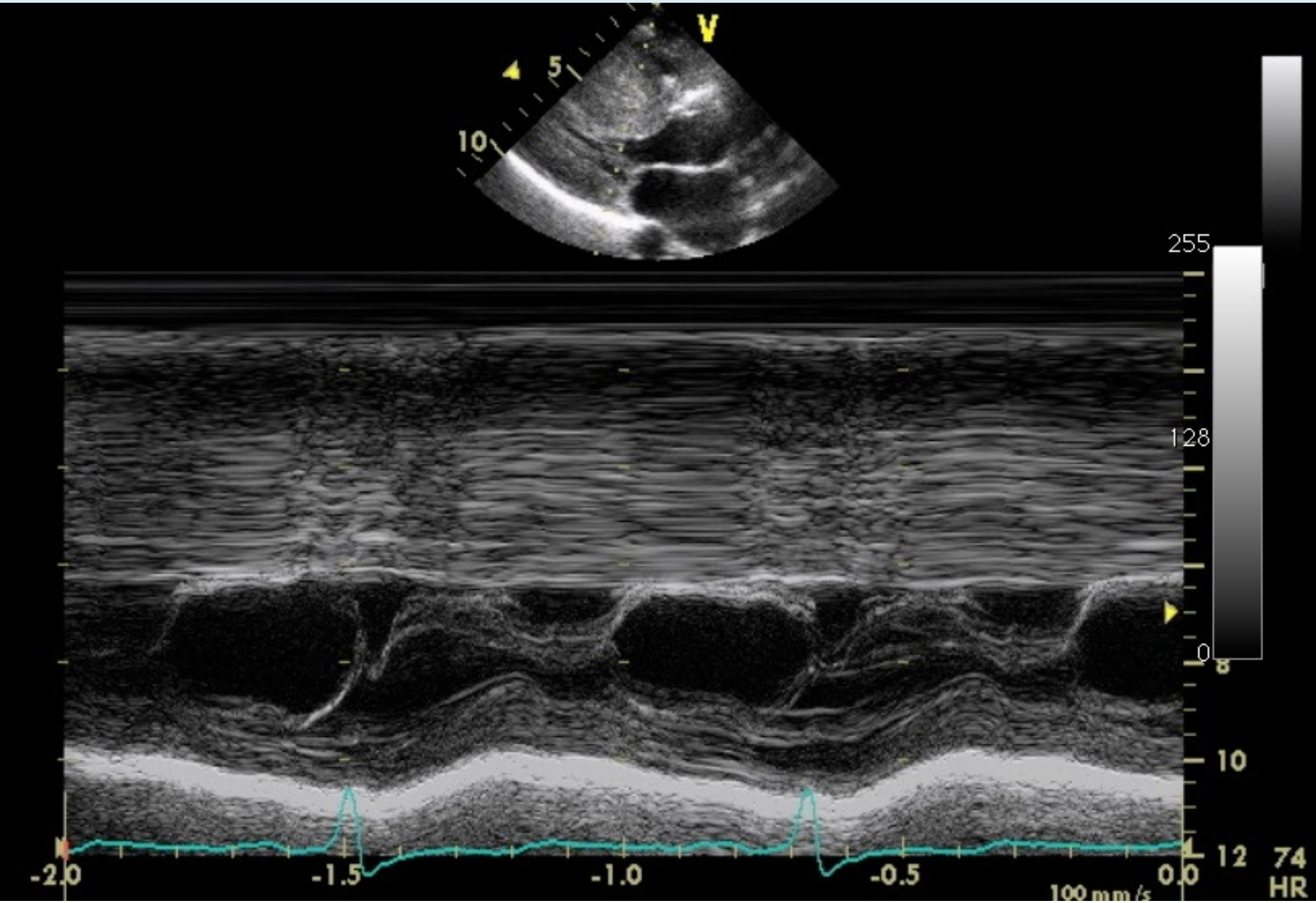

1 v 5.69 m/s
p 129.52 mmHg
V
.65
-.65
[m/s]
-2
-4
1 v 8.79 m/s
p 309.26 mmHg
V
.65
-.65
[m/s]
-2
-4
-6
-8
-10
MR
-2.0
-1.5
-1.0
-0.5
0.0
100 mm/s
84
HR

Mechanism of obstruction

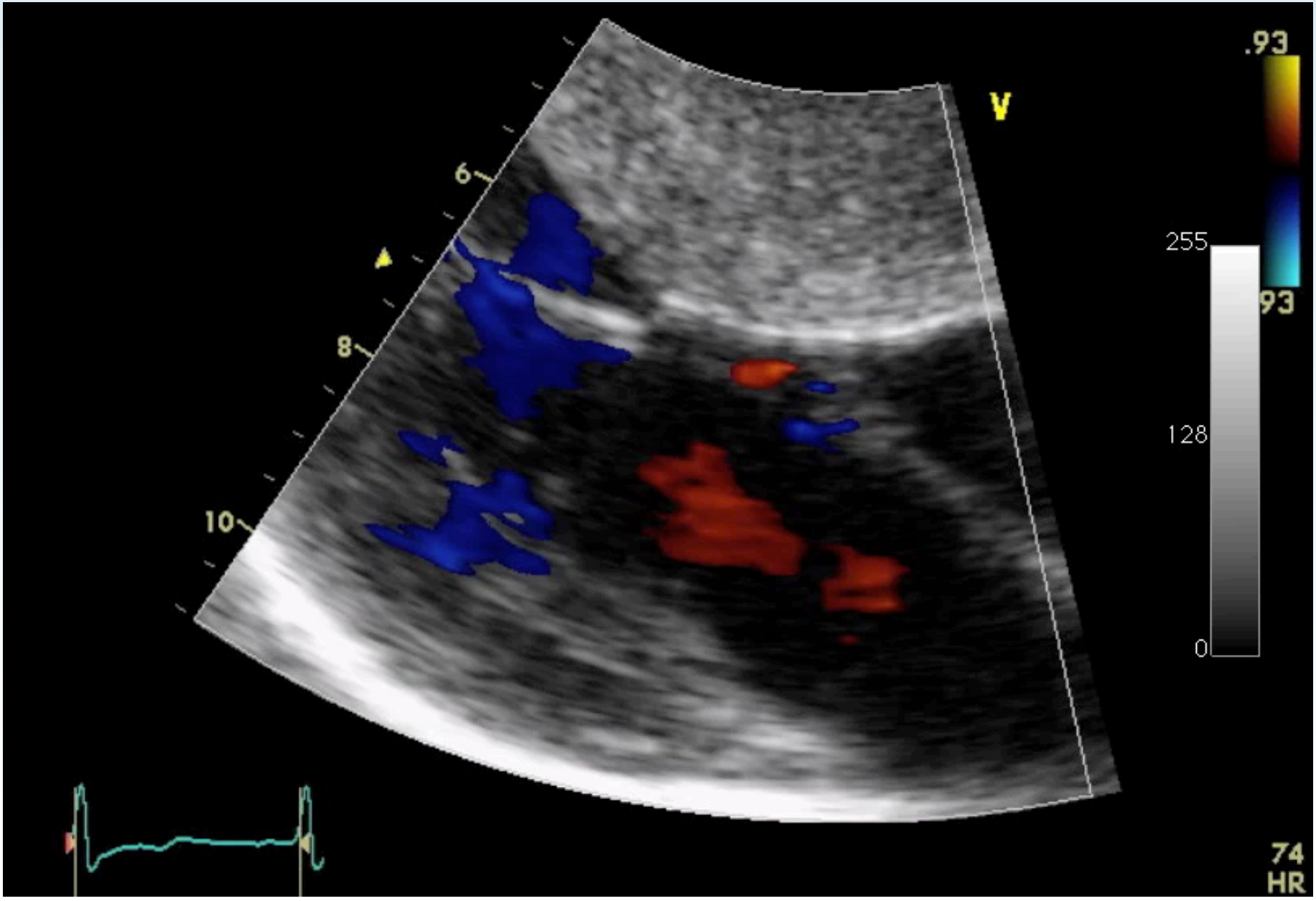


Systolic anterior motion (SAM)

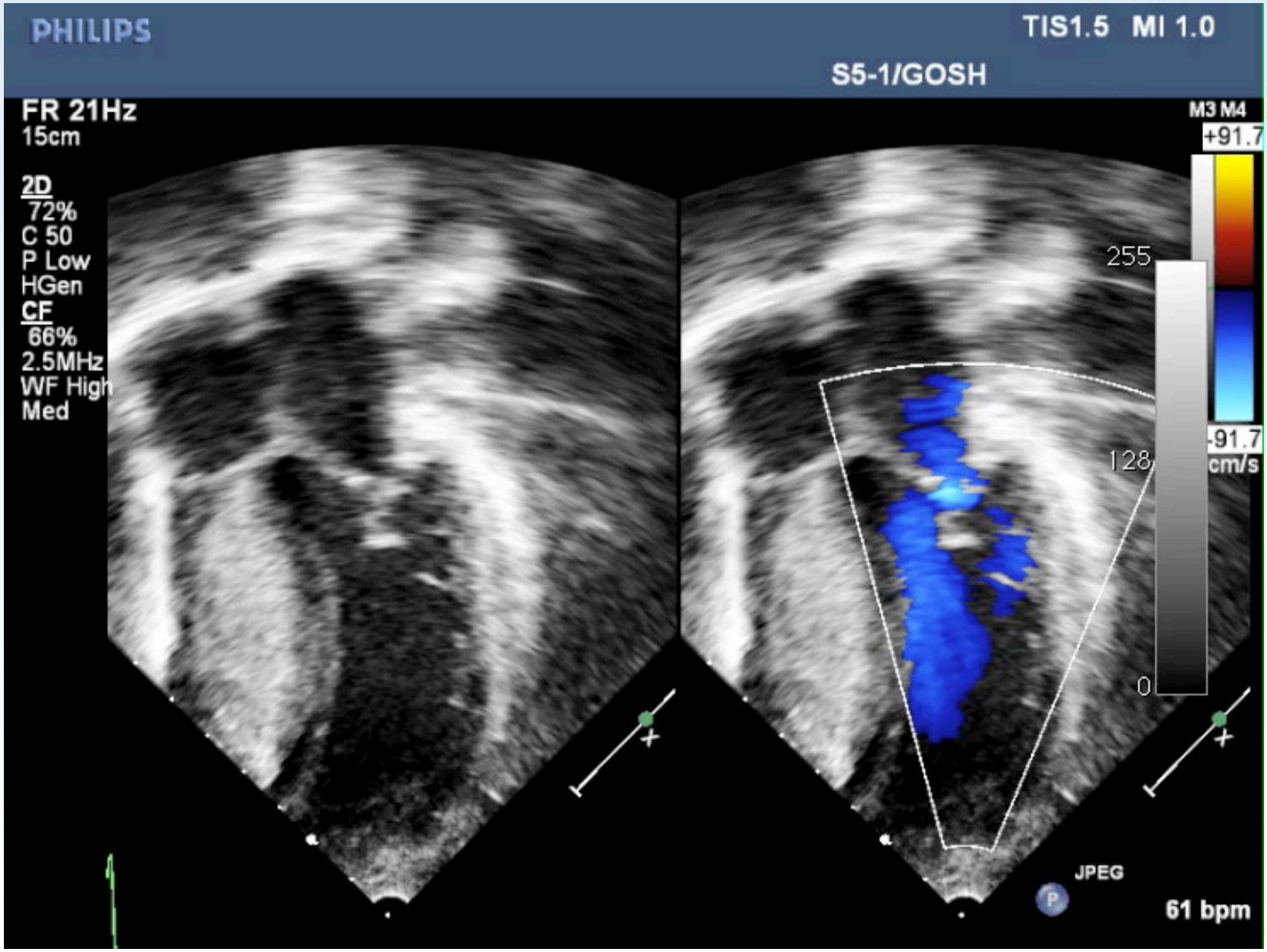



V
5
10
255
128
0
10
12
74
HR
-2.0
-1.5
-1.0
-0.5
0.0
100 mm/s

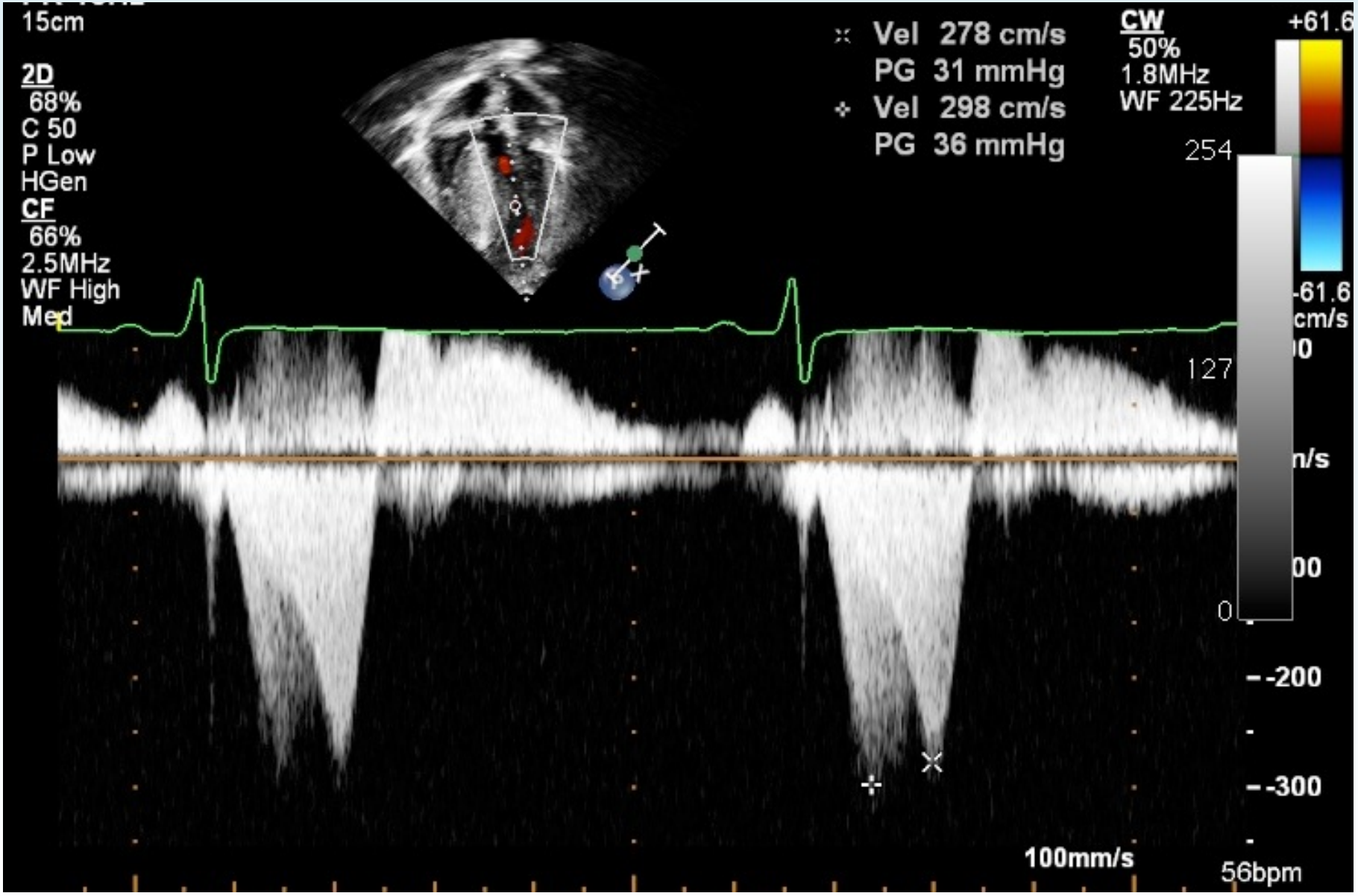
Posteriorly-directed MR

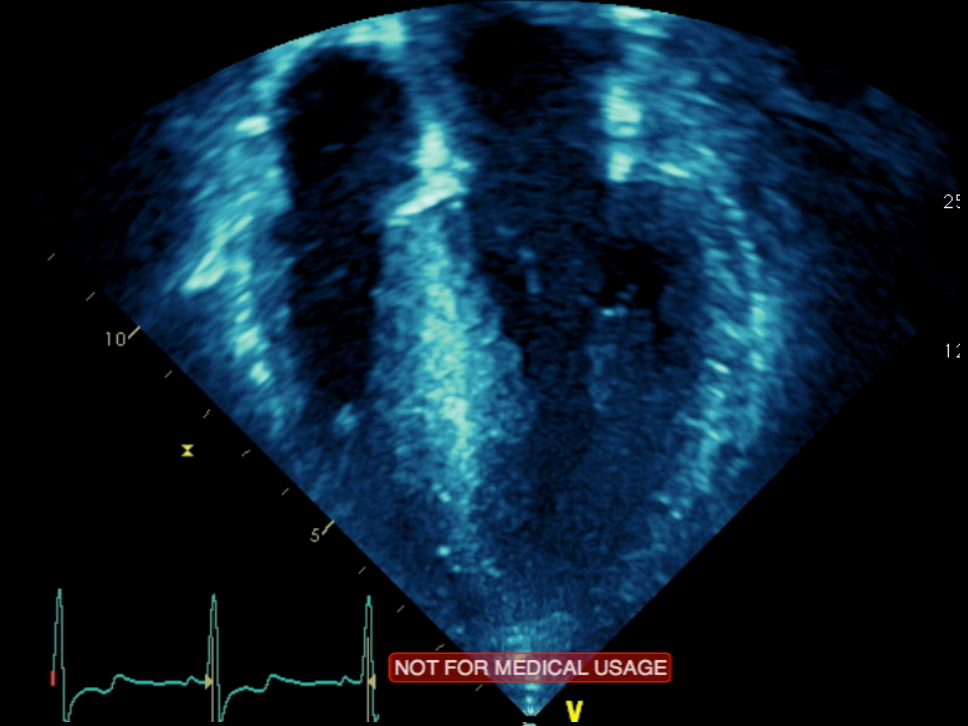


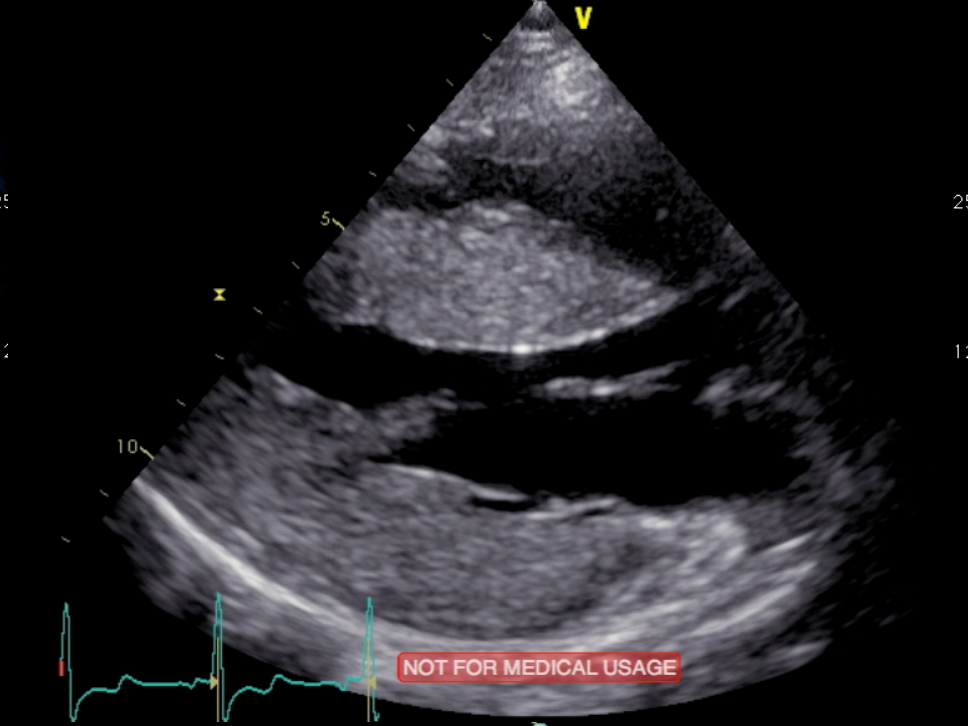
Mid-cavity obstruction

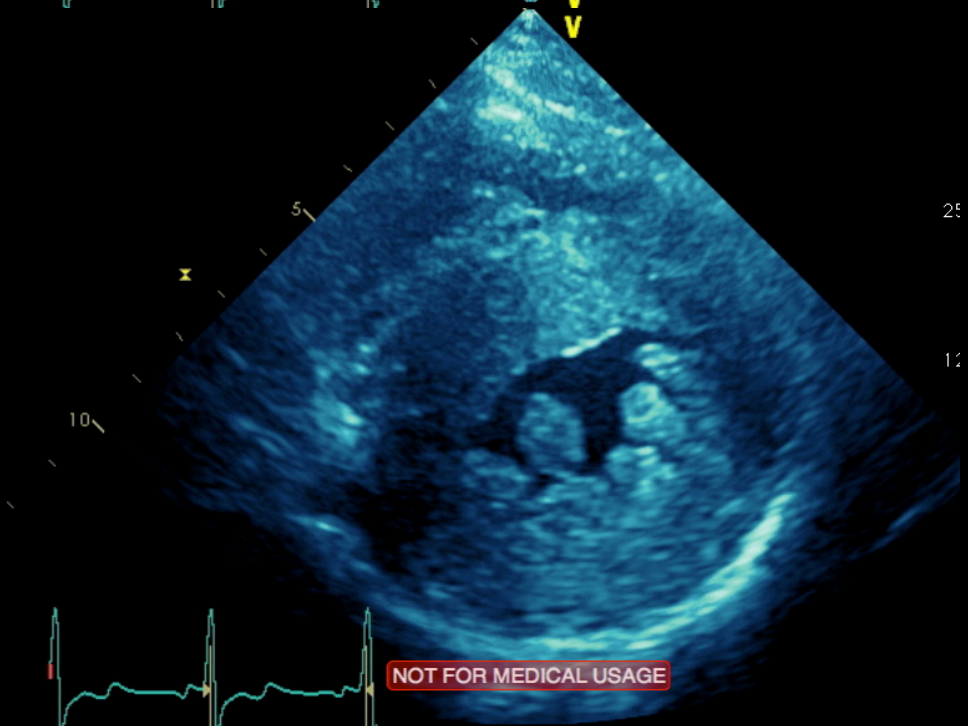


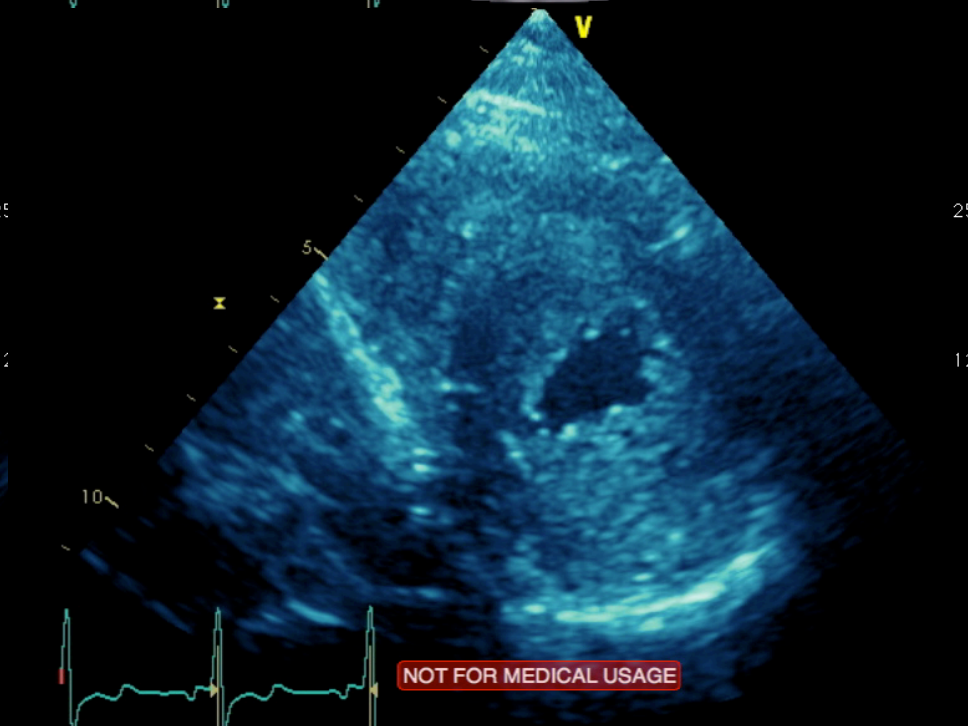
CW Doppler


15cm
2D
68%
C 50
P Low
HGen
CF
66%
2.5MHz
WF High
Med
Vel 278 cm/s
PG 31 mmHg
Vel 298 cm/s
PG 36 mmHg
CW
50%
1.8MHz
WF 225Hz
+61.6
254
127
0
-61.6
cm/s
-200
-300
100mm/s
56bpm

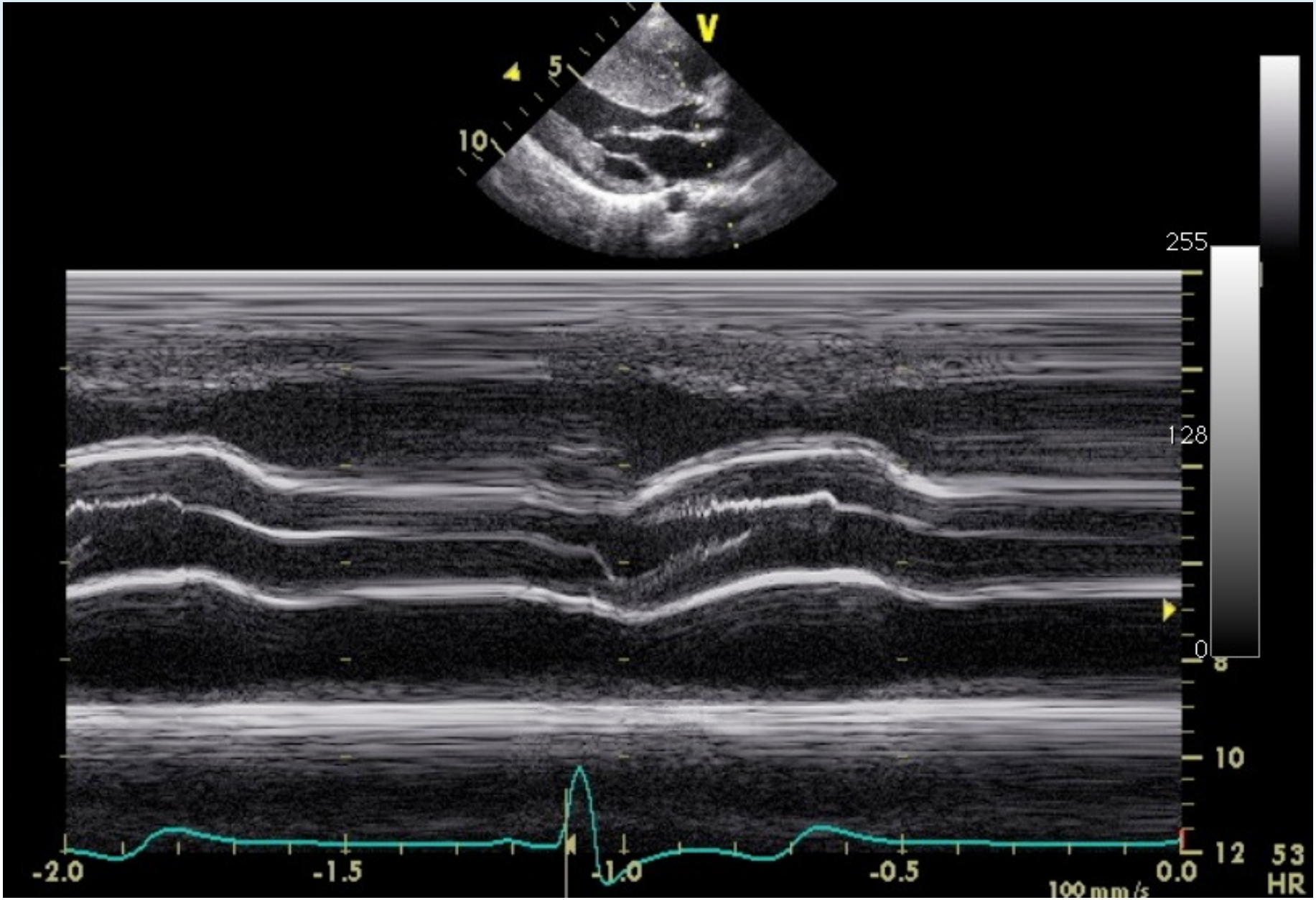

NOT FOR MEDICAL USAGE


NOT FOR MEDICAL USAGE

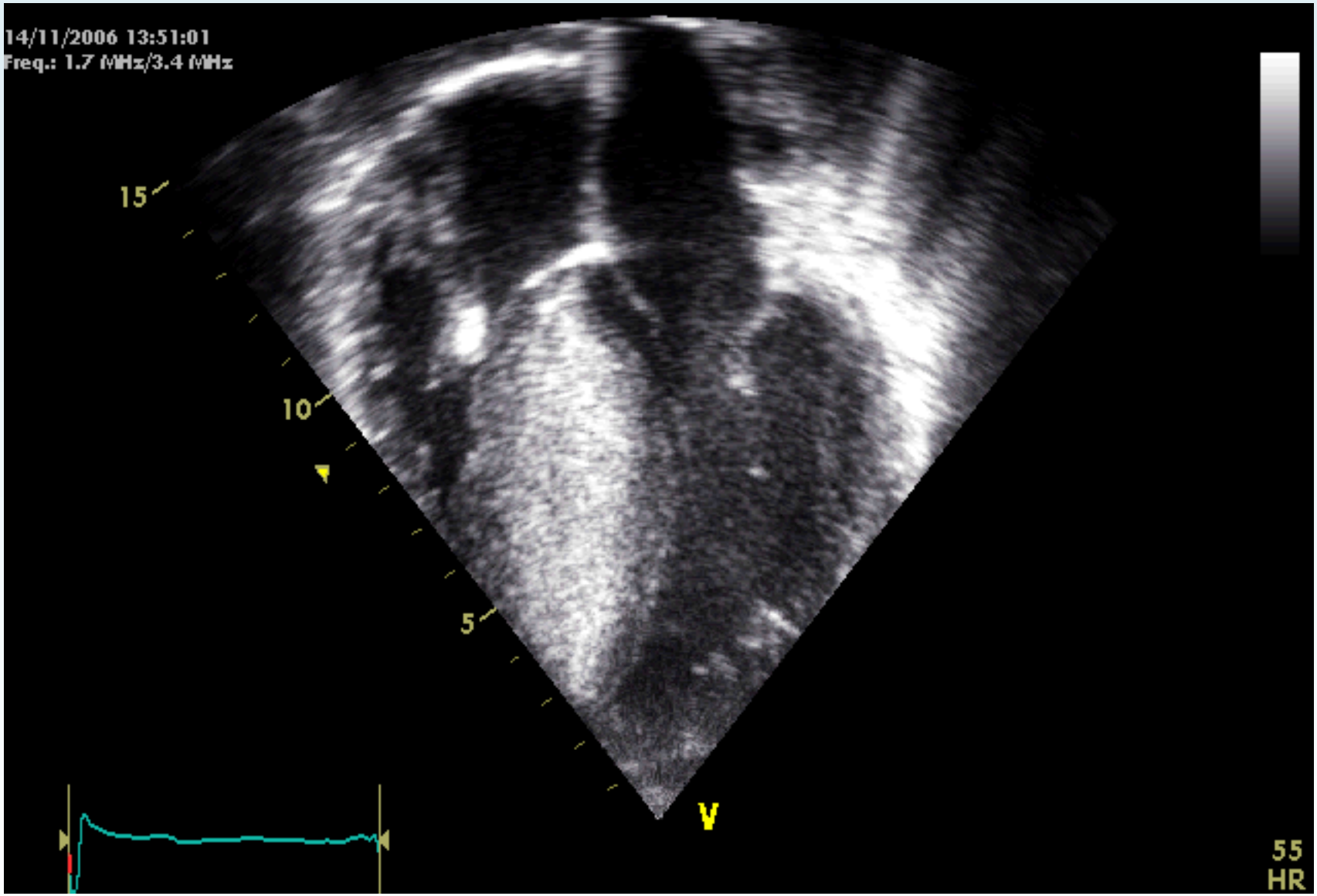

NOT FOR MEDICAL USAGE


NOT FOR MEDICAL USAGE

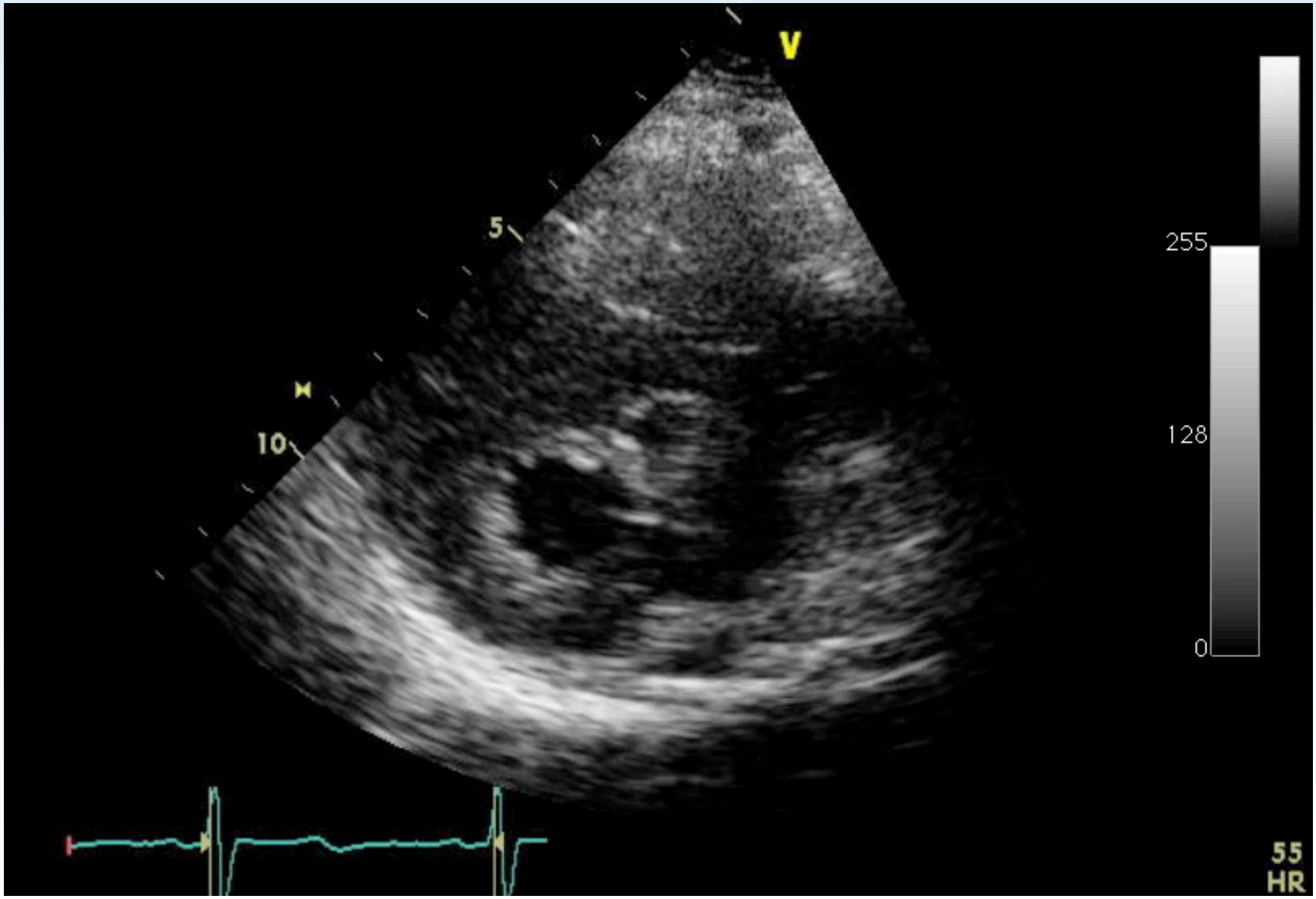
Mid-systolic closure of AoV



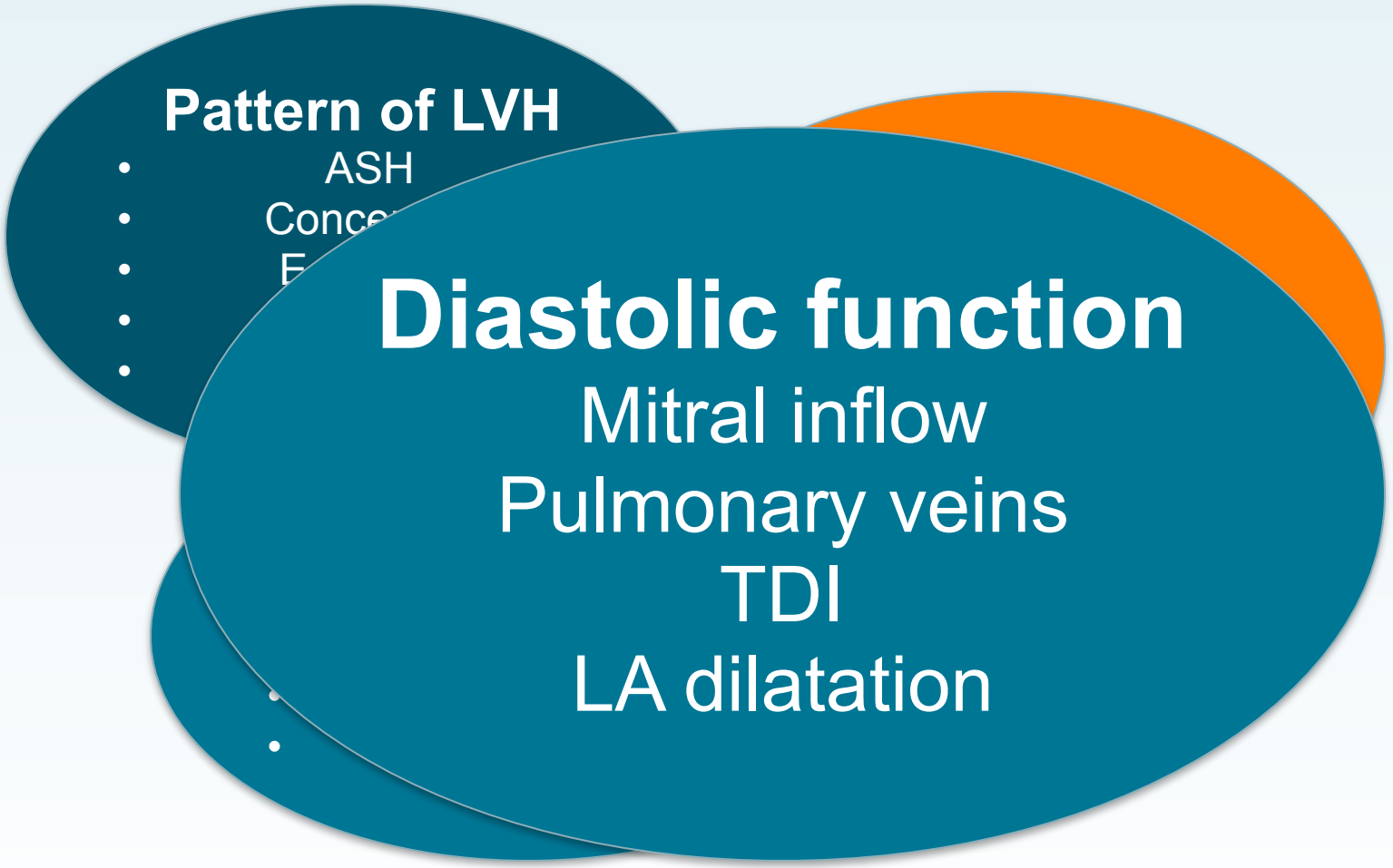
LA dilatation

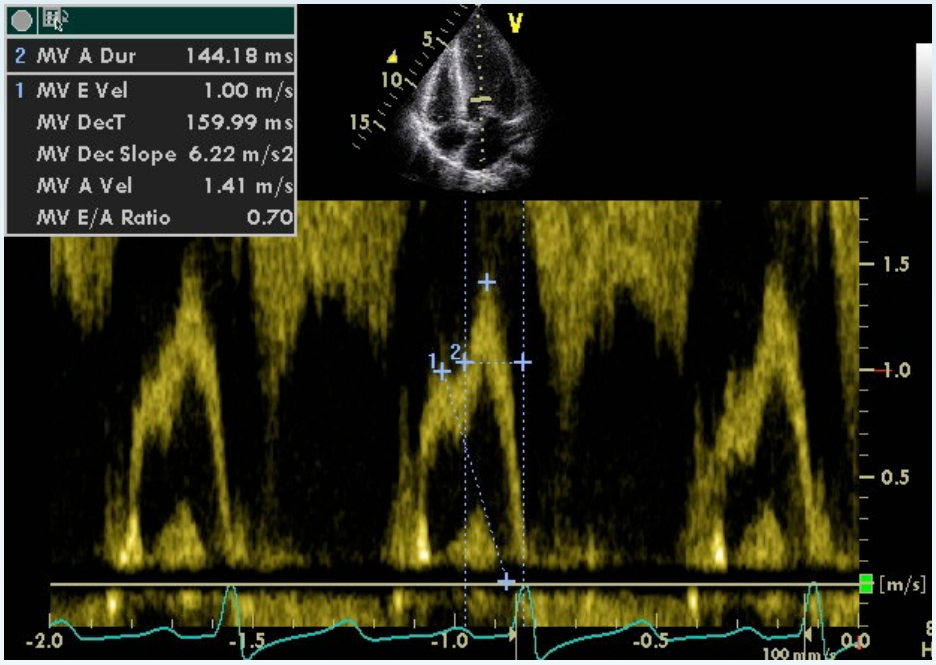


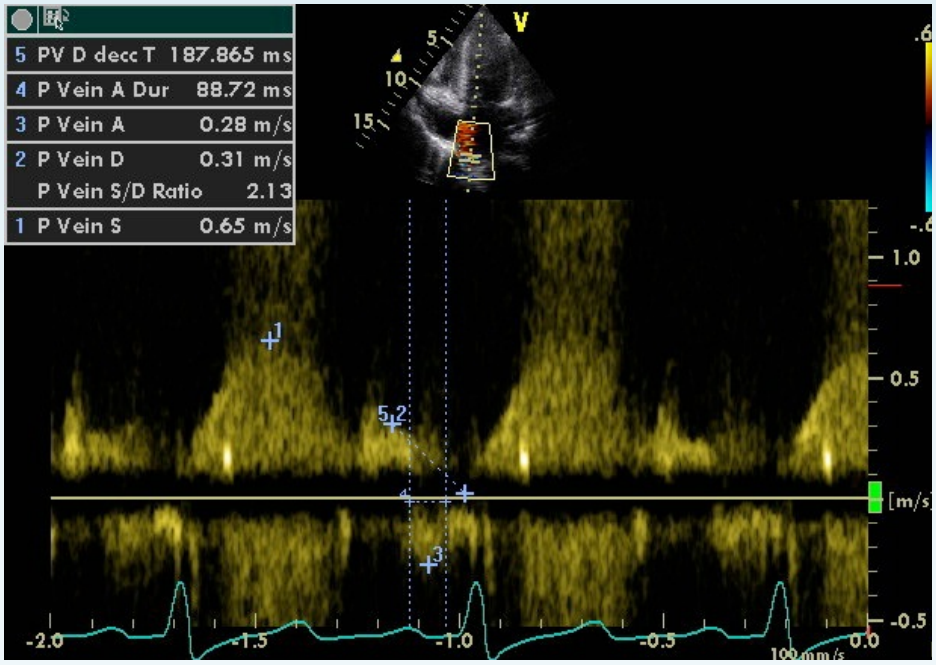
Post-myectomy

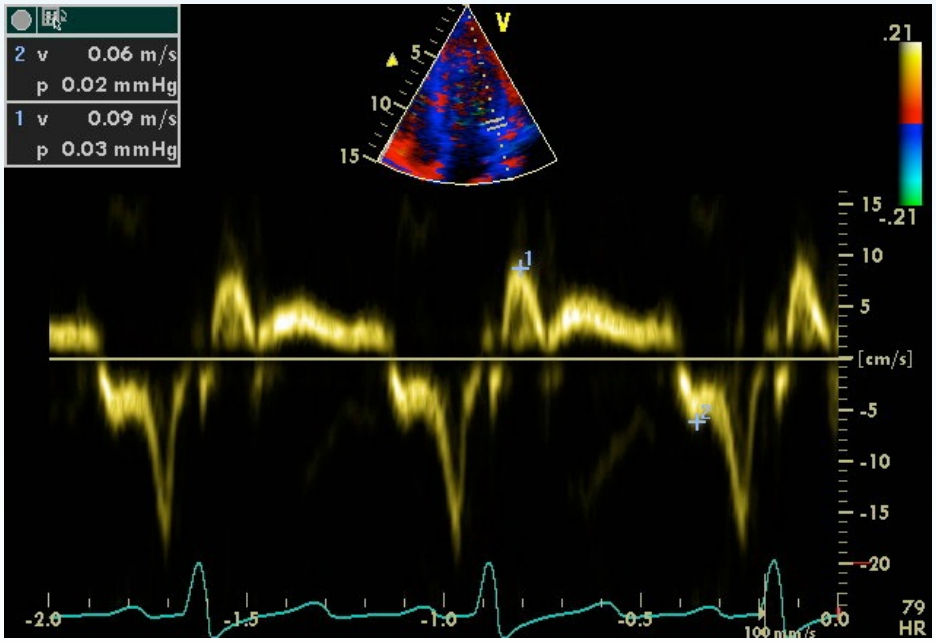


Echocardiography in HCM

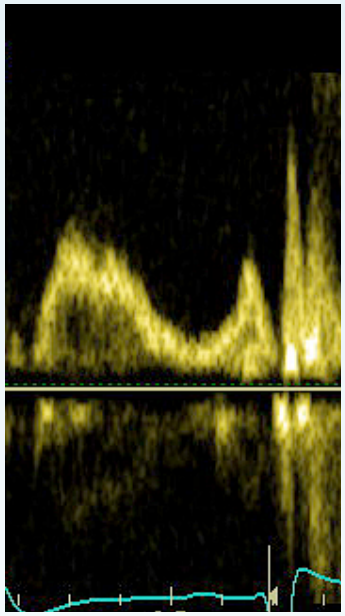


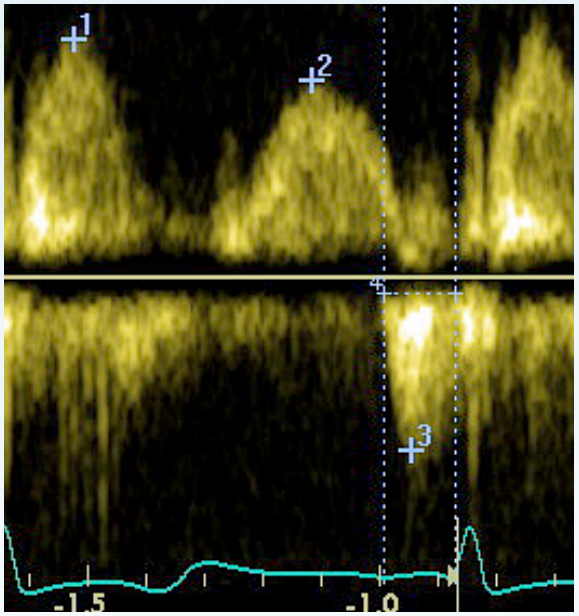


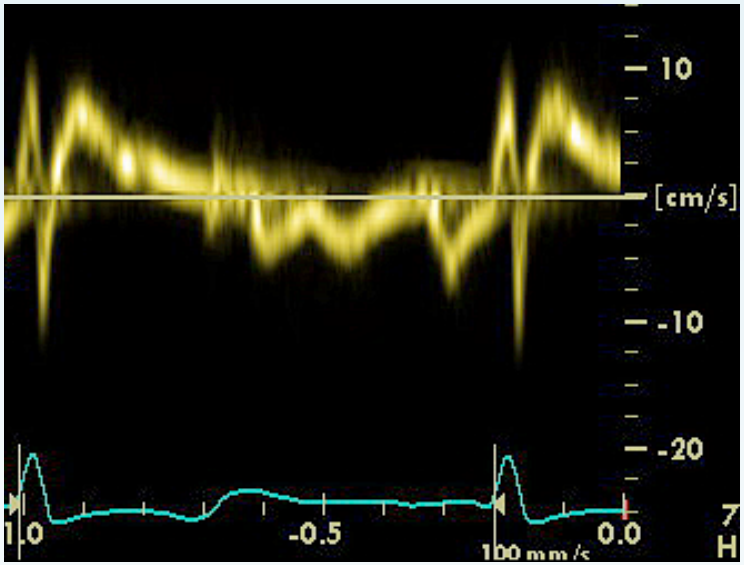




=> Impaired relaxation







“Pseudonormal” pattern

Dilated cardiomyopathy

Definition

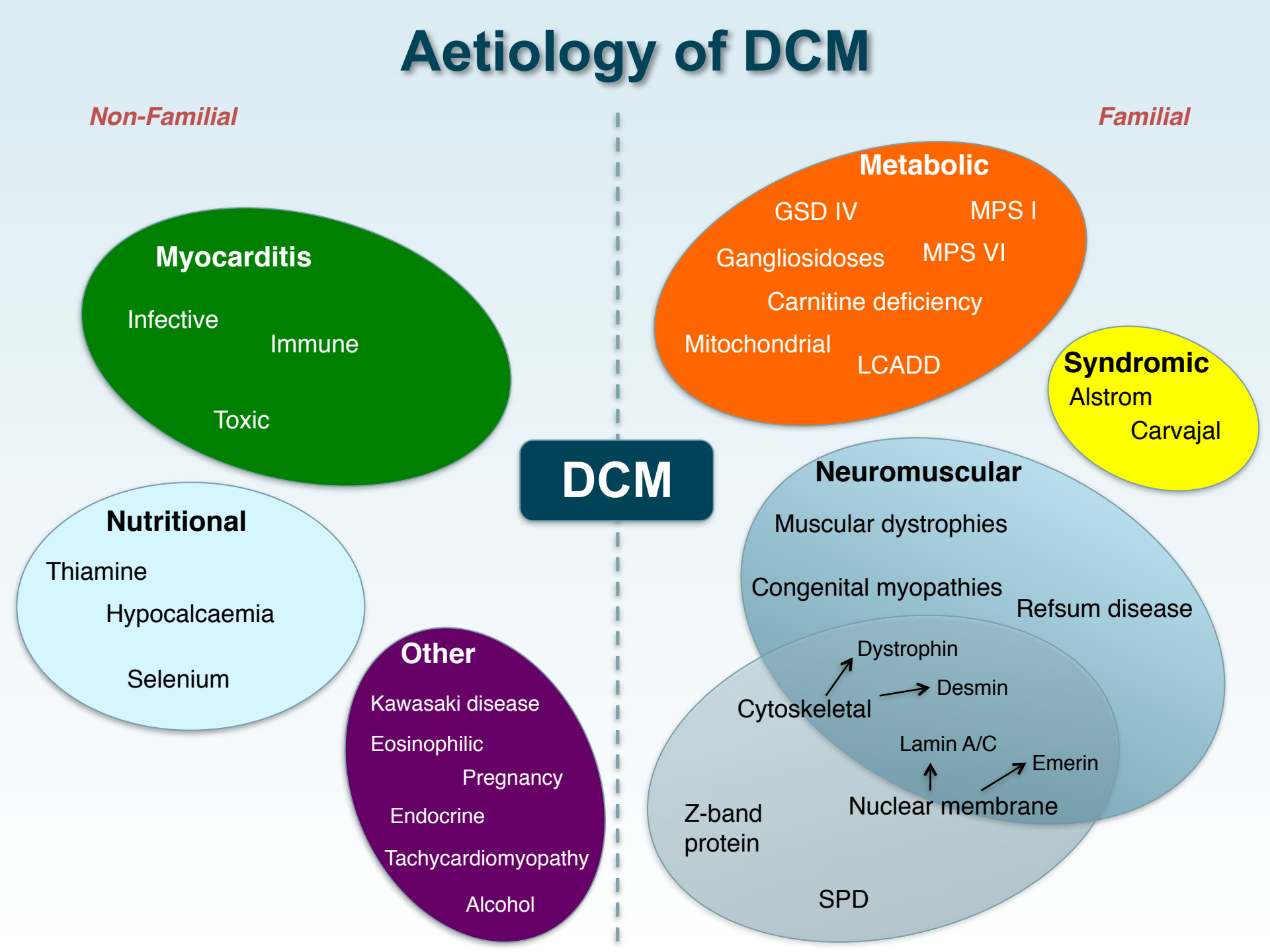
- **“Ventricular dilatation and left ventricular systolic dysfunction in the absence of abnormal loading conditions (hypertension, valve disease) or coronary artery disease sufficient to cause global systolic impairment.**
- **Right ventricular dilation and dysfunction may be present but are not necessary for the diagnosis. ”**

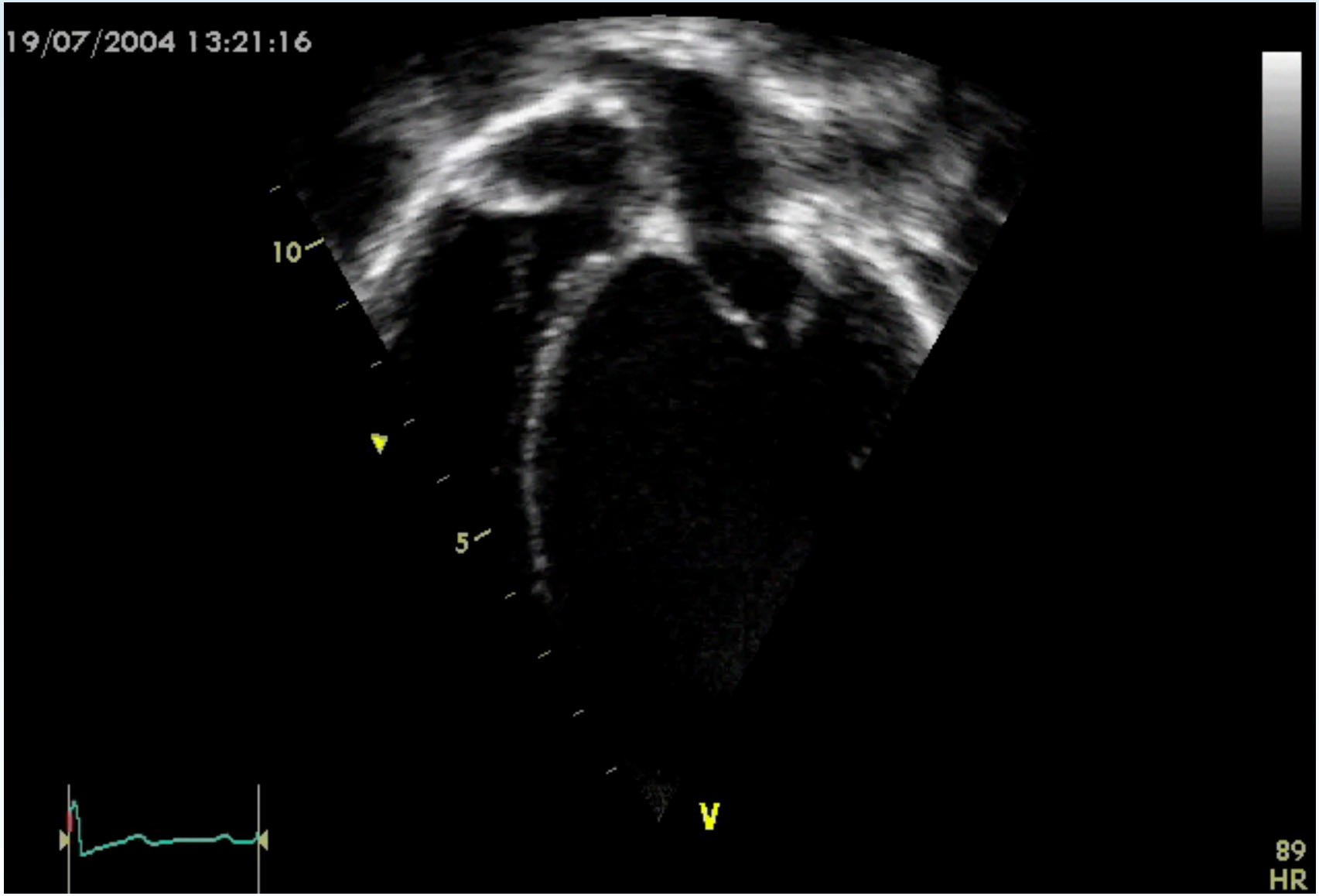
ESC Classification of Cardiomyopathies, 2008

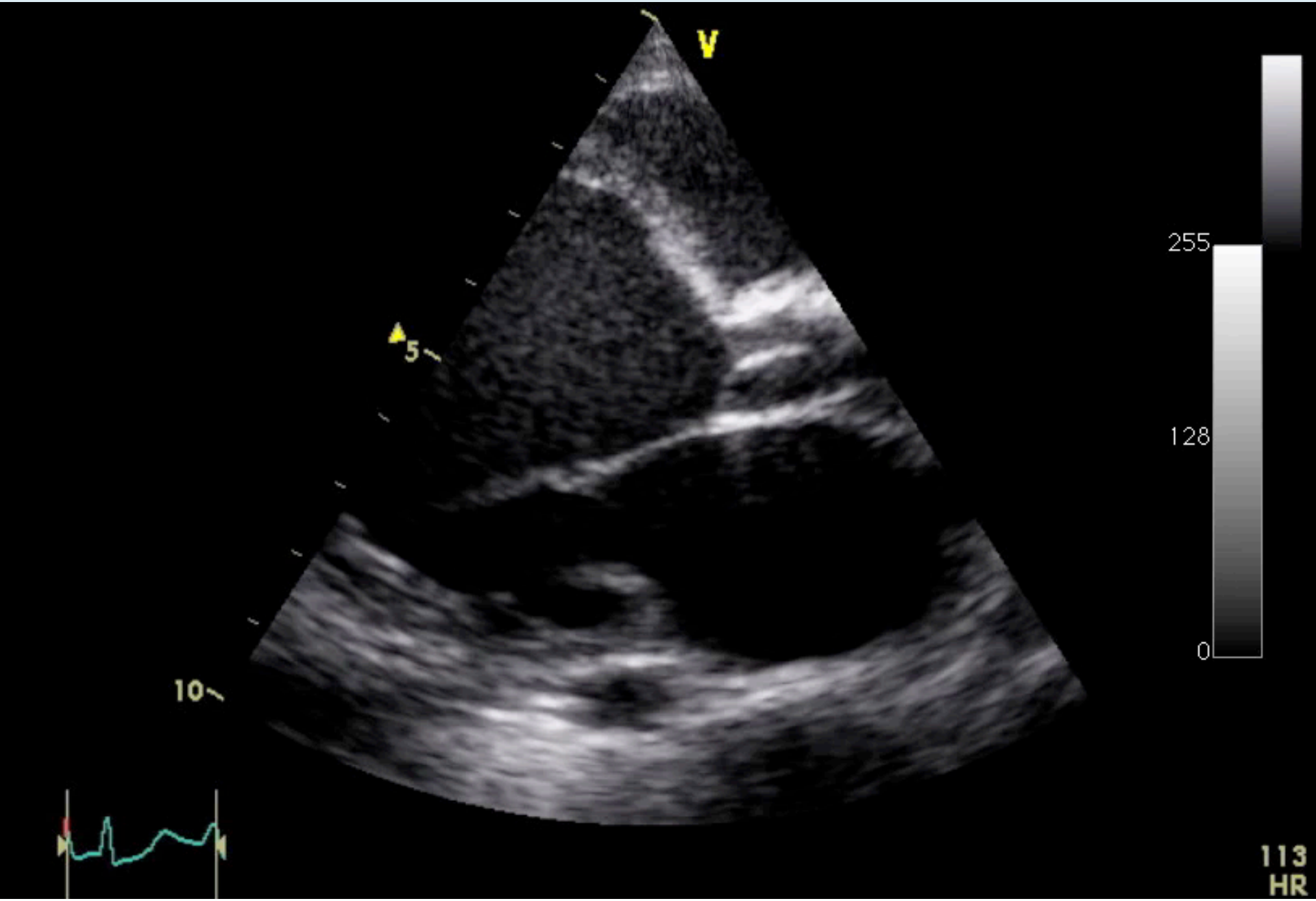
Epidemiology and aetiology

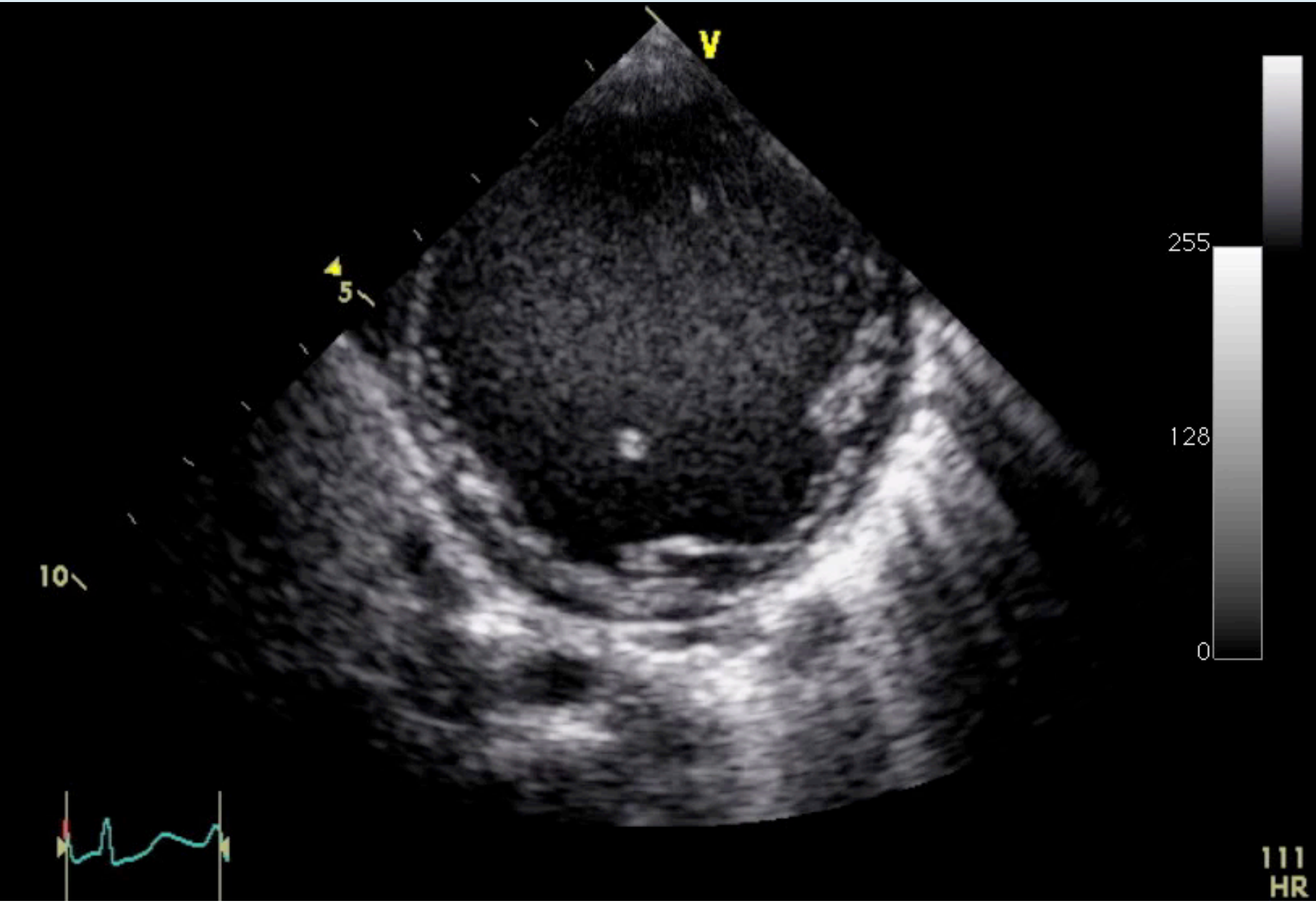
- Commonest cardiomyopathy in childhood
- Incidence 0.57/100,000
- Heterogeneous aetiology
  - Viral myocarditis
  - Metabolic
  - Neuromuscular
  - Syndromic
  - Acquired (e.g. nutritional deficiencies)
- Familial disease in approx. 25%
  - Autosomal dominant
  - X-linked (e.g. DMD)

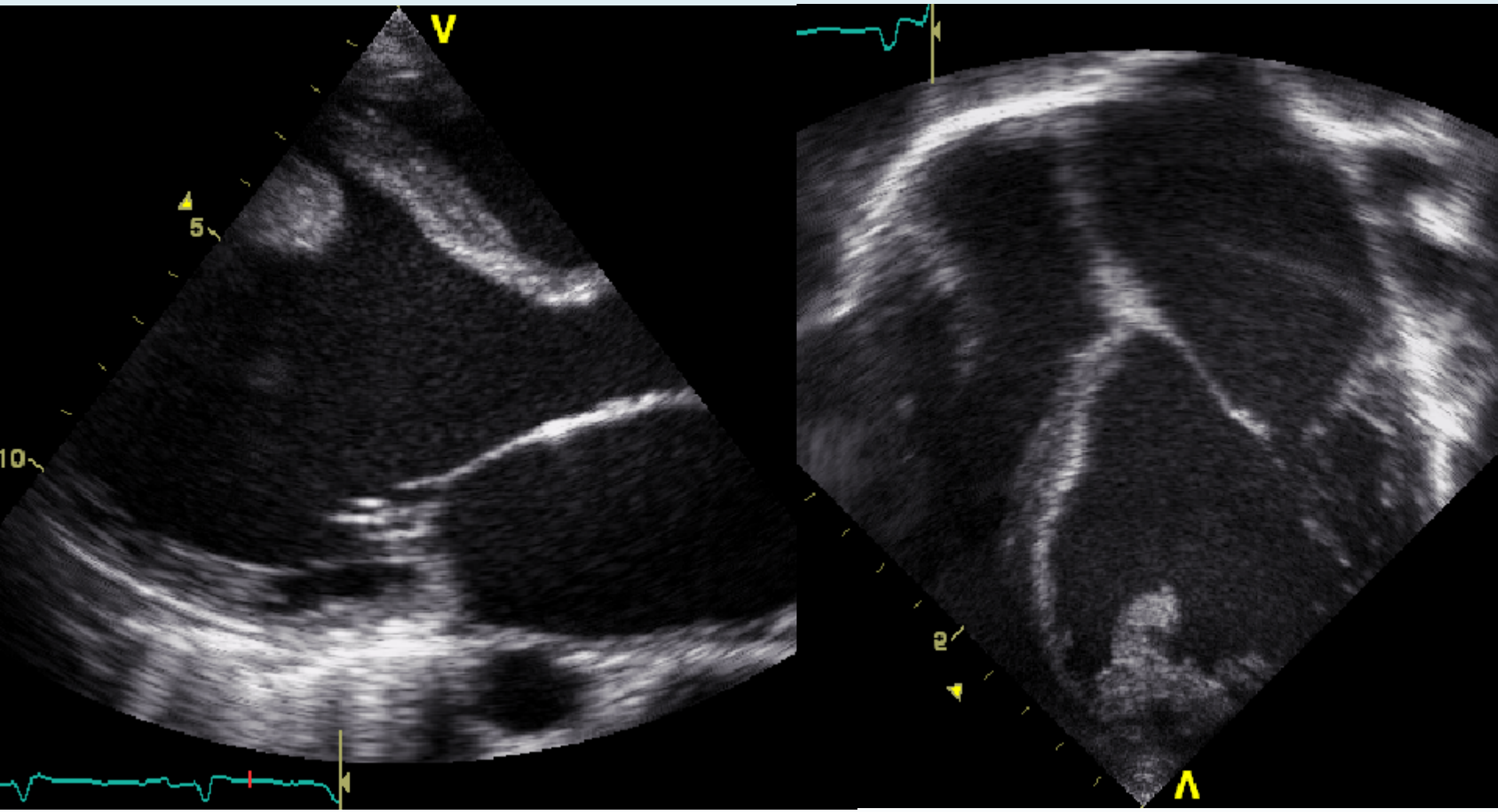
Aetiology of DCM

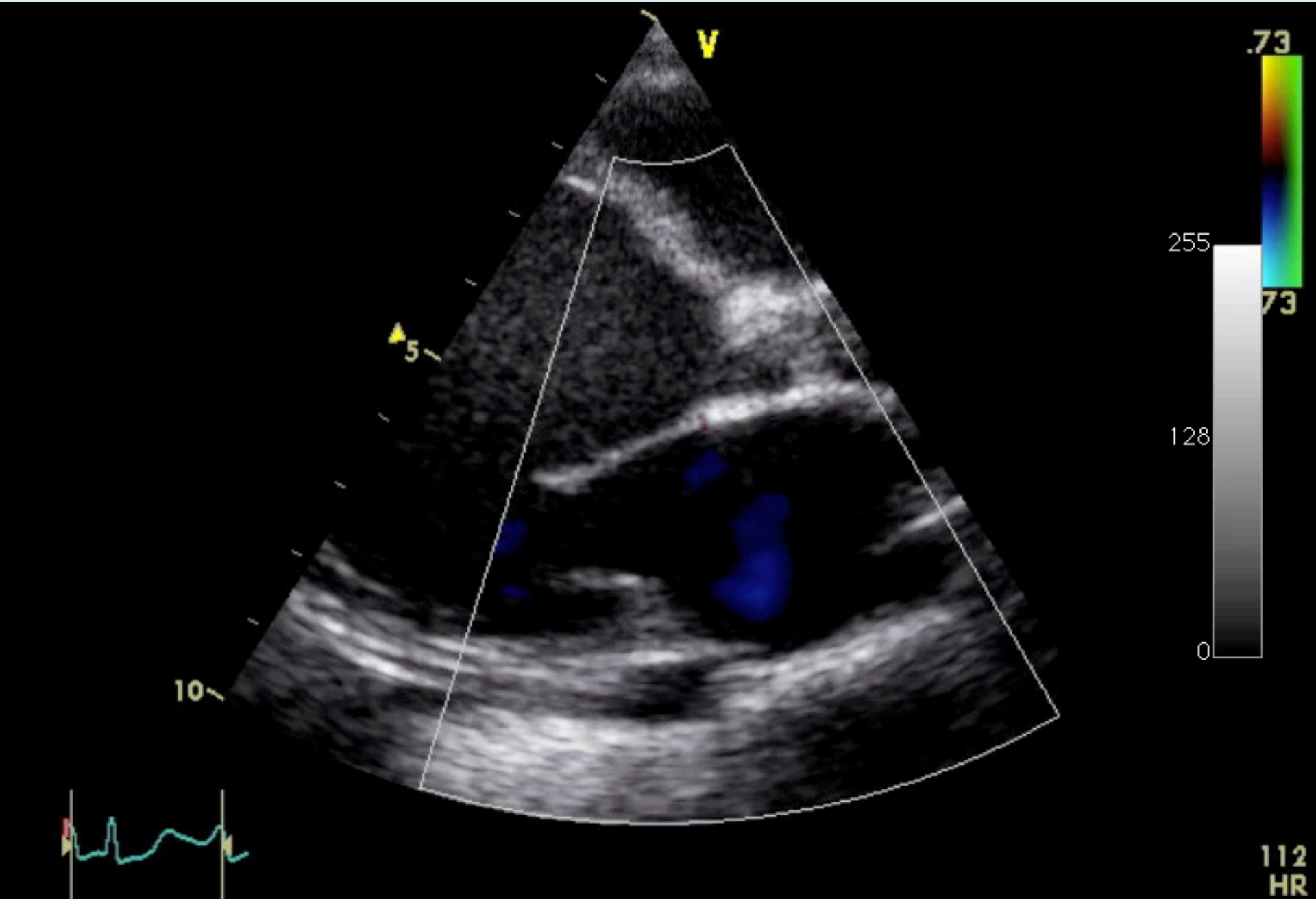


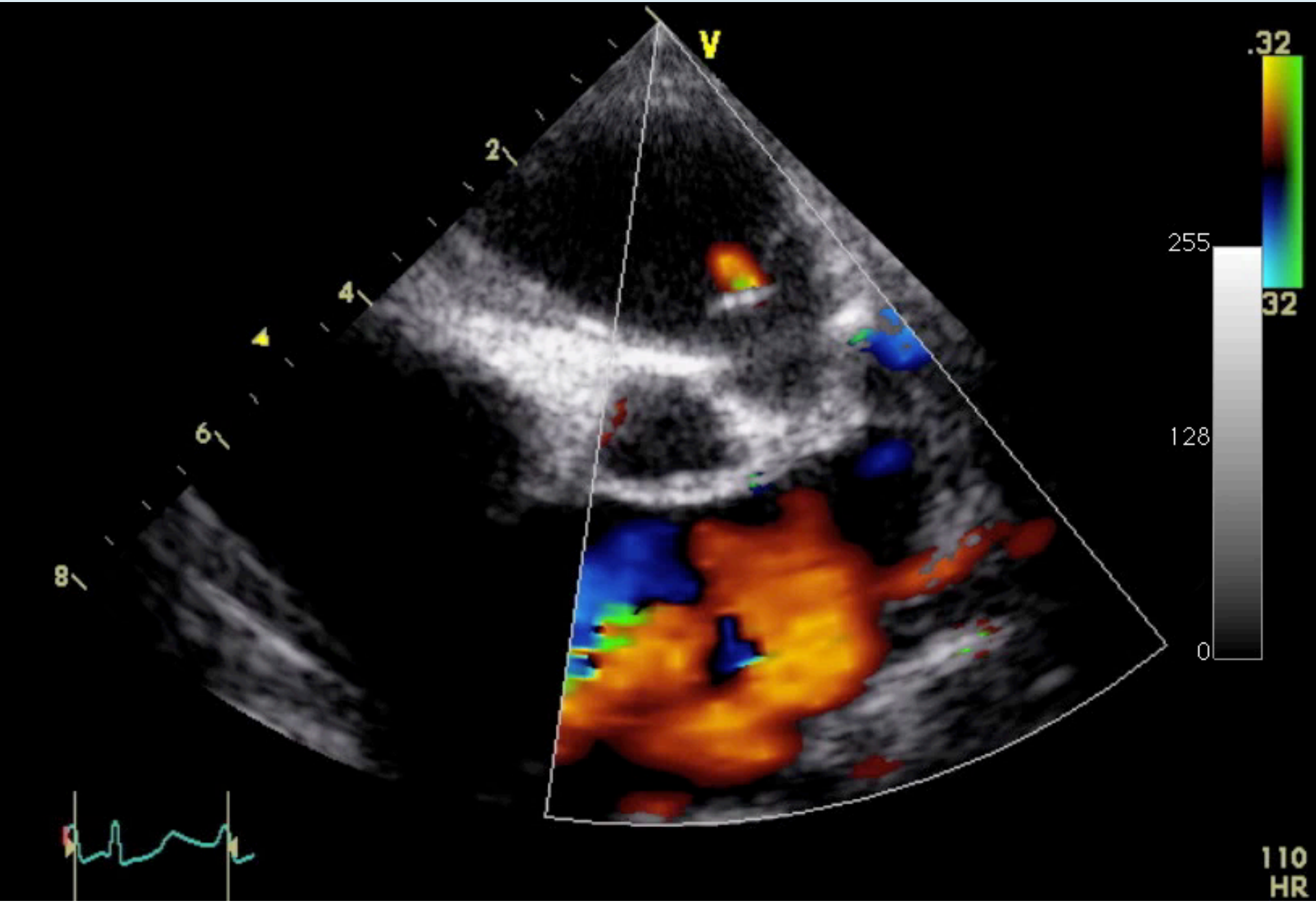

19/07/2004 13:21:16
10
5
89
HR


V
5
10
255
128
0
113
HR

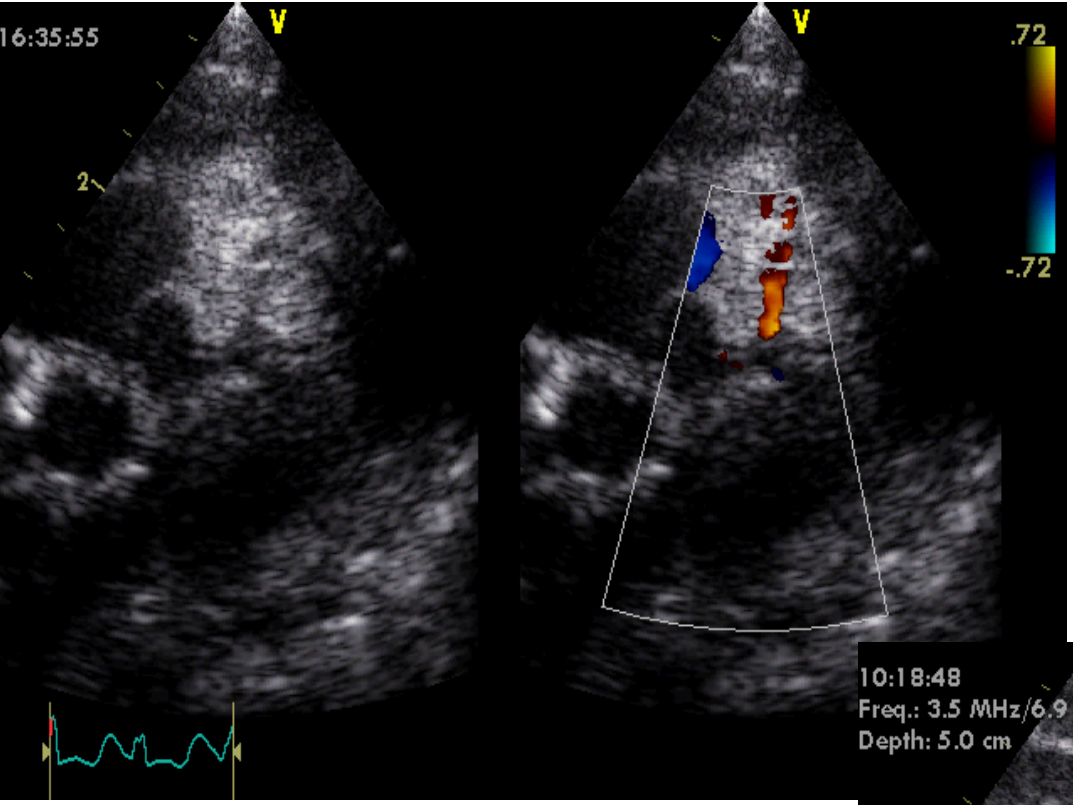

V
5
10
255
128
0
111
HR

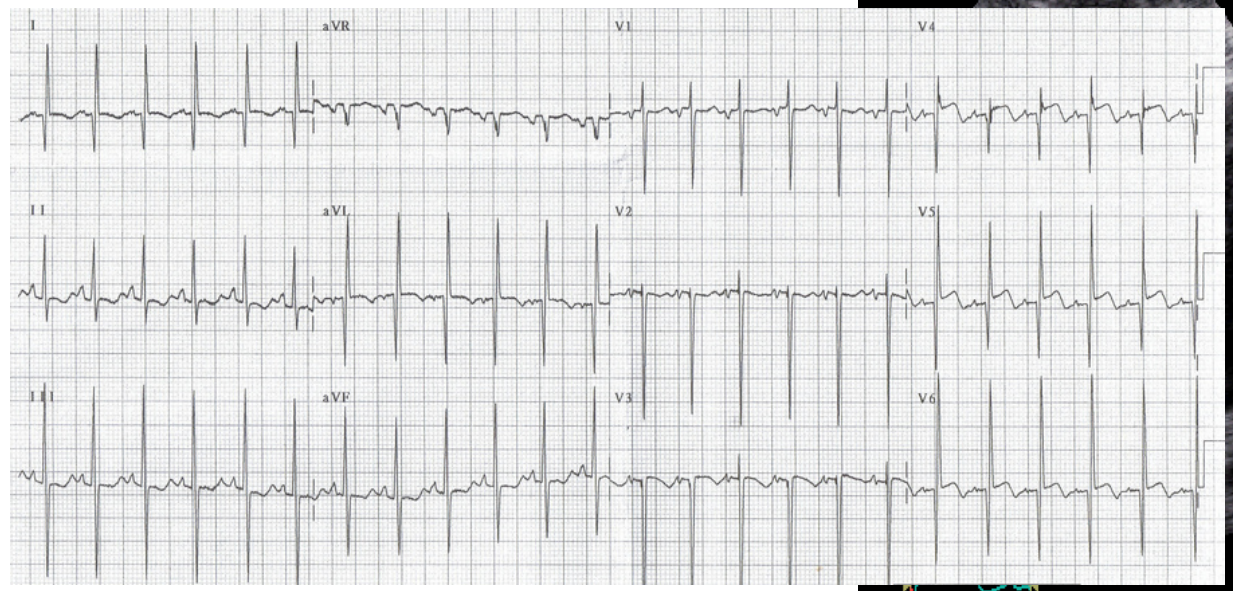

V
5
10
2
A

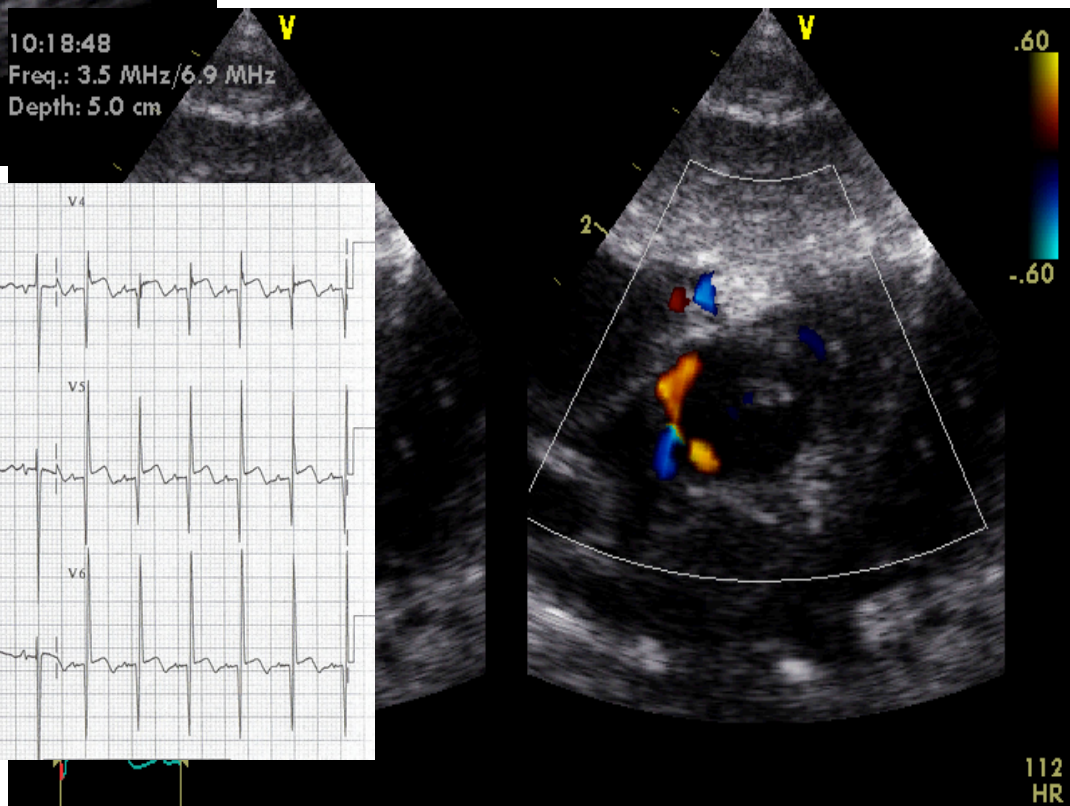

V
5
10
.73
255
128
0
73
112
HR


V
2
4
4
6
8
.32
255
32
128
0
110
HR

ALCAPA


16:35:55
.72
-.72


I
aVR
V1
V4
II
aVL
V2
V5
III
aVF
V3
V6


10:18:48
Freq.: 3.5 MHz/6.9 MHz
Depth: 5.0 cm
.60
-.60
112
HR

Restrictive Cardiomyopathy

Definition

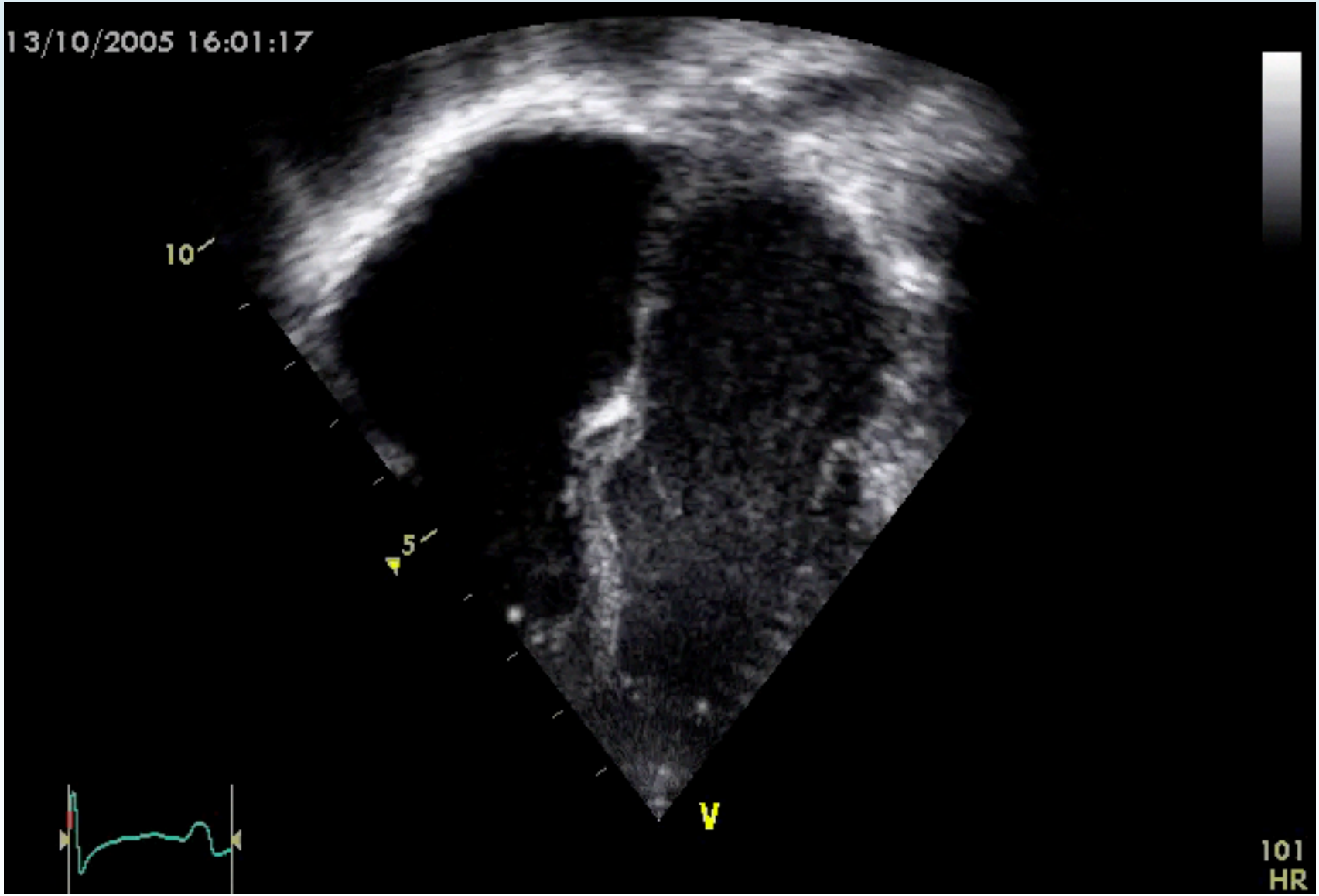
Restrictive ventricular physiology: ventricular filling in which increased myocardial stiffness causes ventricular pressure to rise precipitously with only small increases in volume

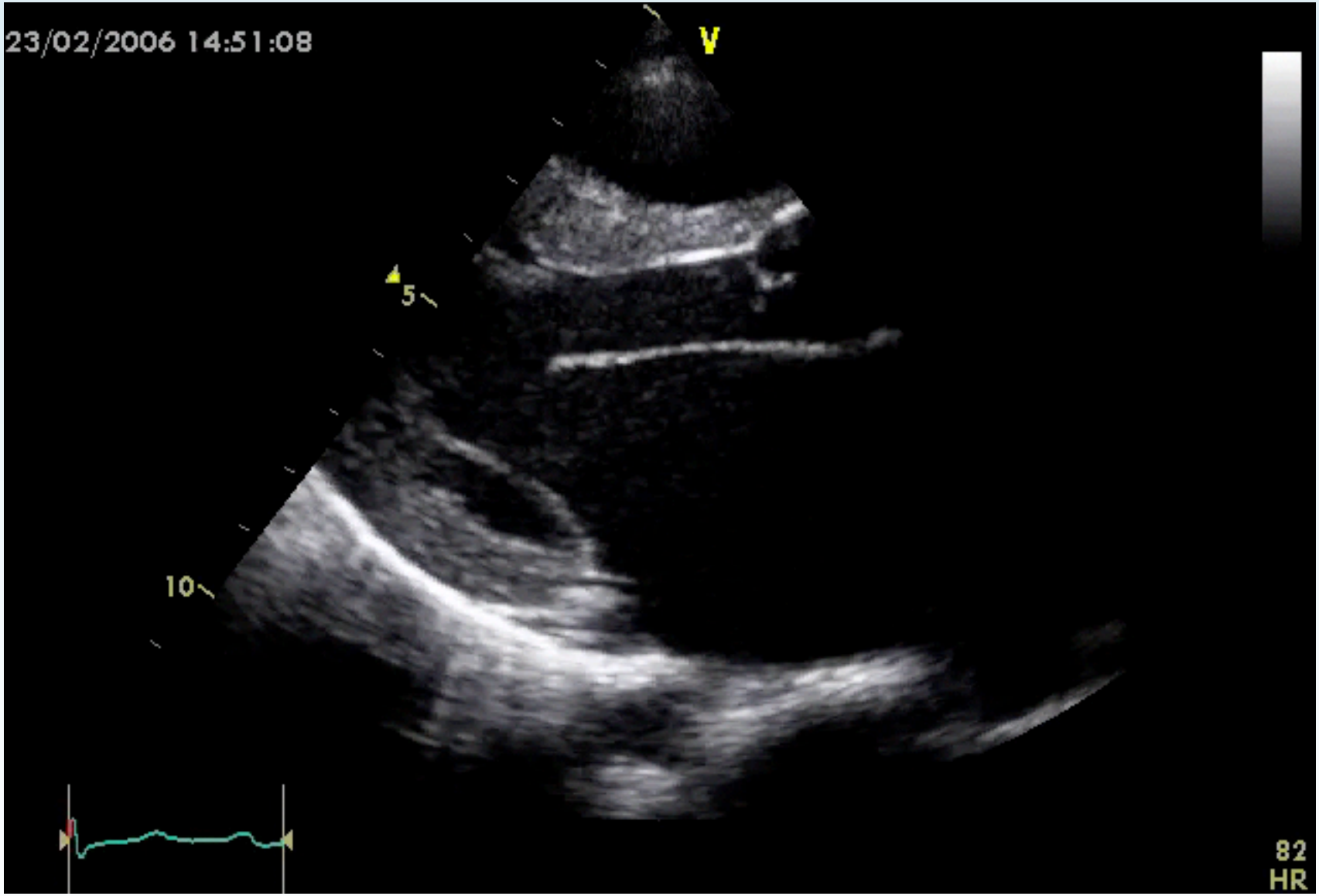
"Restrictive ventricular physiology in the presence of normal or reduced diastolic volumes (of one or both ventricles), normal or reduced systolic volumes, and normal ventricular wall thickness."

ESC classification 2008

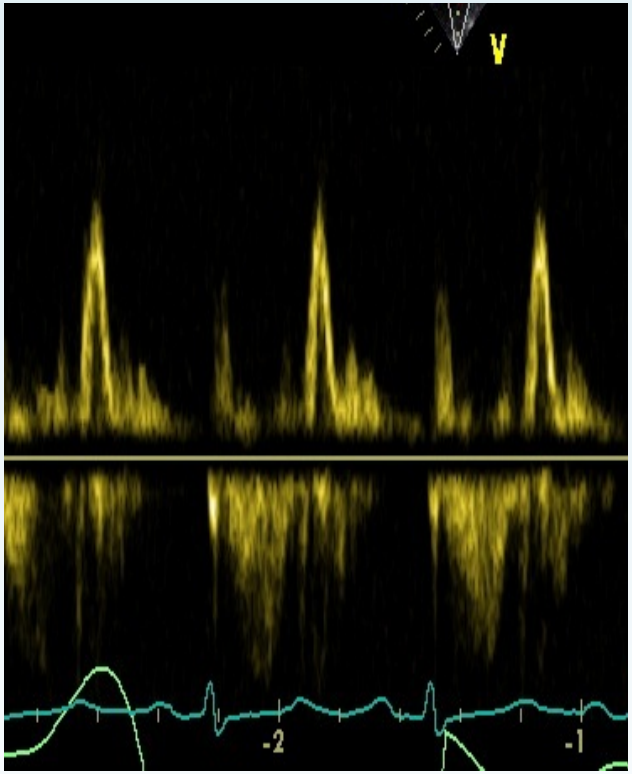
Prevalence

- Rarest of all cardiomyopathies
- <5% of all paediatric cardiomyopathies
- ~200 patients reported in paediatric literature


13/10/2005 16:01:17
10
5
V
101
HR


23/02/2006 14:51:08
V
5
10
82
HR

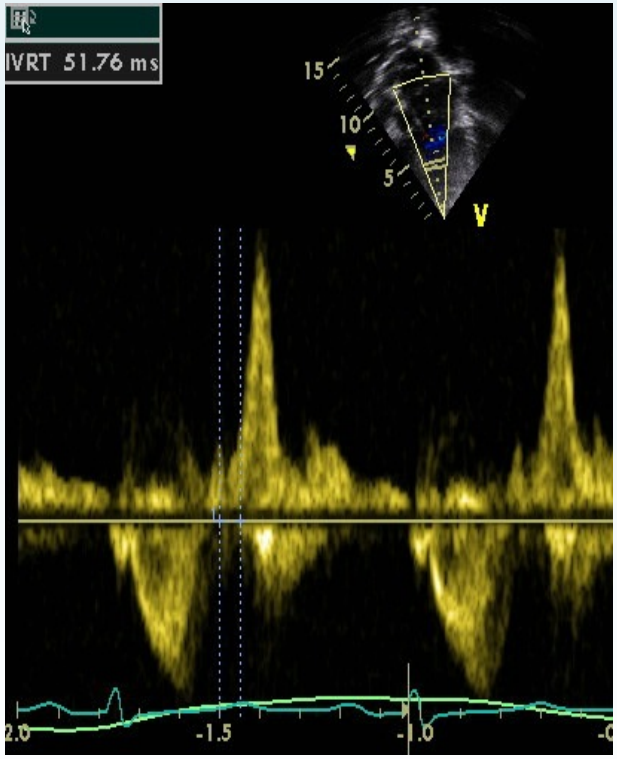
Echo – mitral inflow



E/A ≥ 2

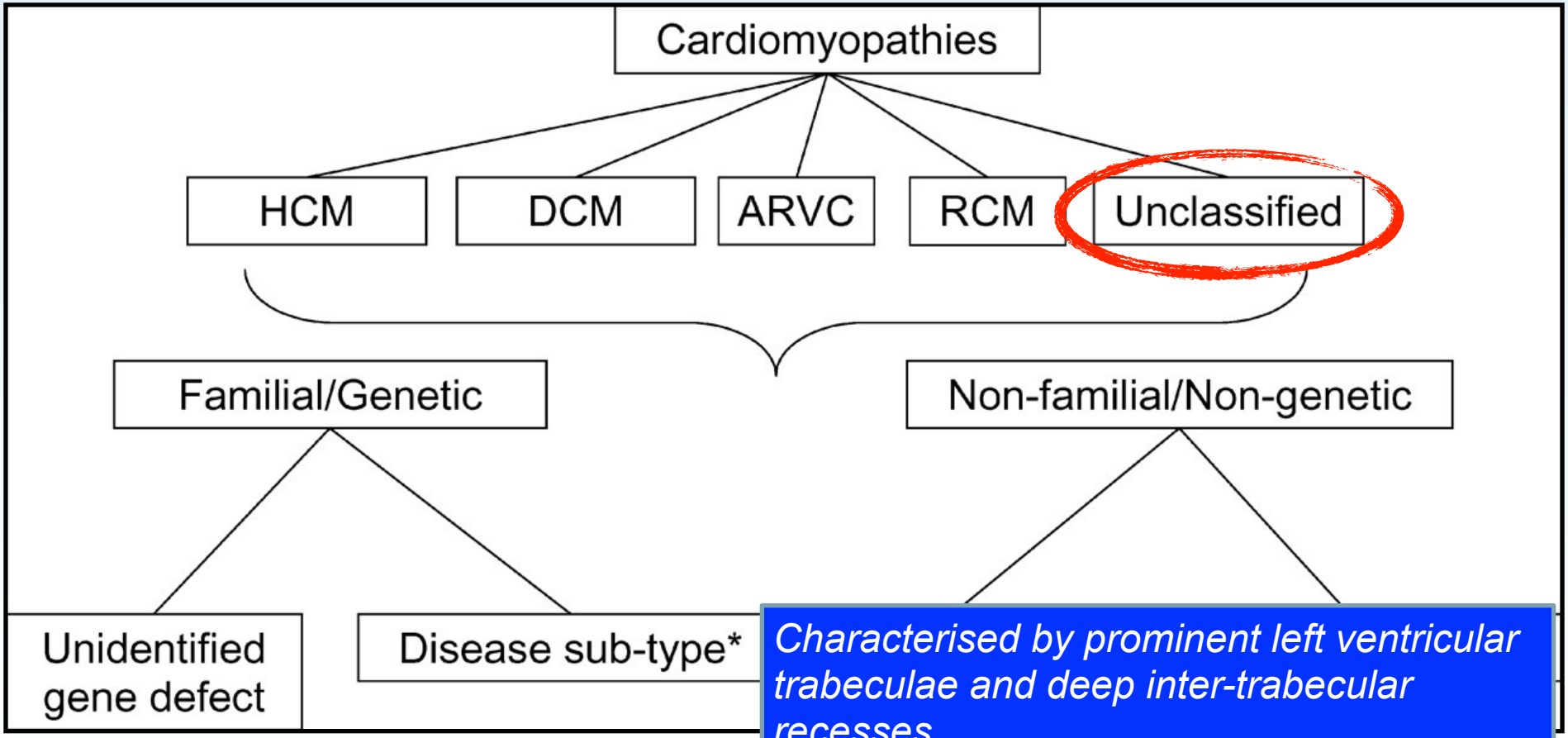
DT ≤ 150ms

IVRT ≤ 70ms

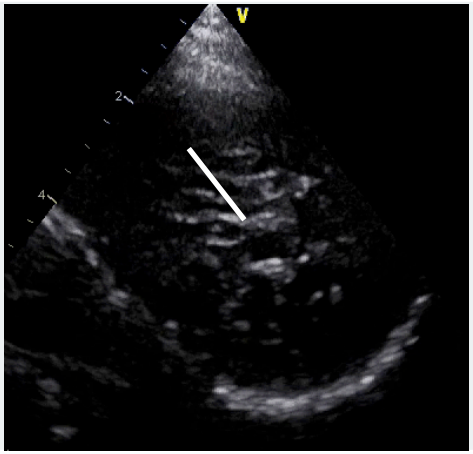
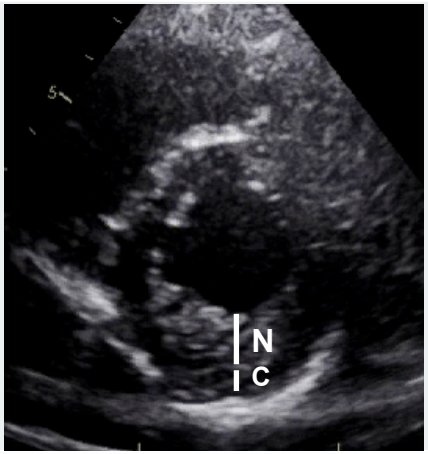
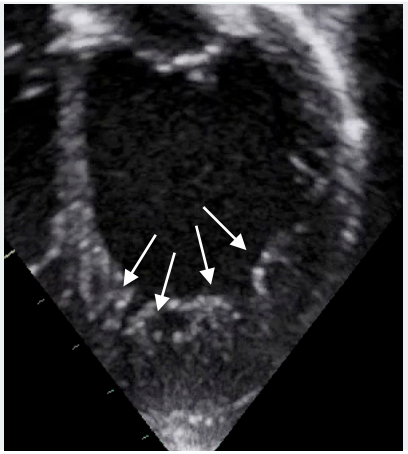


LEFT VENTRICULAR NON-COMPACTION

Classification - ESC



Echocardiographic criteria

Chin *et al.* (n=8)	Jenni *et al.* (n=34)	Stollberger *et al.* (n=62)
PSAx - end-diastole	PSAx - end-systole	A4Ch - end-diastole
Epicardial compacted layer (X); endocardial non-compacted layer (Y)	(i)Thick non-compacted layer (N); thin compacted layer (C) (ii)No coexisting cardiac abnormalities (iii)Numerous prominent trabeculations and deep recesses supplied by intraventricular blood	>3 trabeculations protruding from LV wall distal to papillary muscles in single imaging plane
X/Y <= 0.5	N/C >=2	N/C >=2
		

NB: LV function not included in any diagnostic criteria

Clinical classification

1) **Isolated** LVNC

2) LVNC associated with LV dilatation and dysfunction **at onset**
- Barth syndrome
- Tafazzinopathies

3) LVNC in hearts fulfilling diagnostic criteria for other **cardiomyopathies**
- HCM
- DCM
- RCM
- ARVC

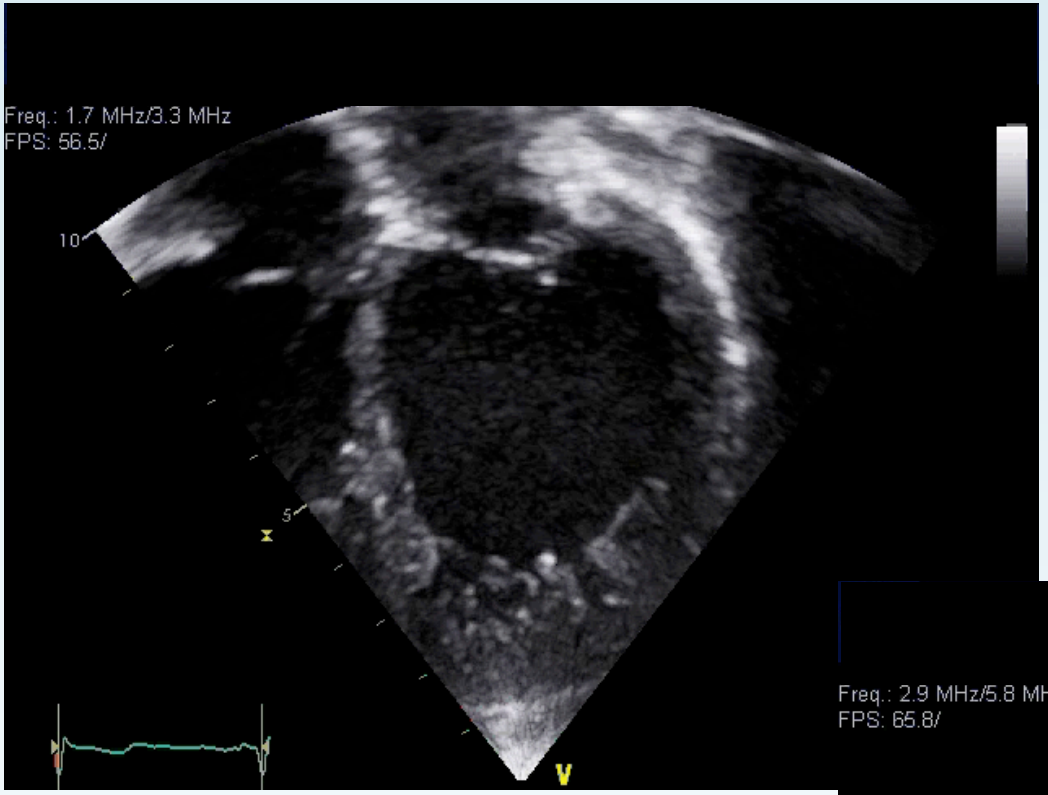
4) LVNC associated with **congenital heart disease**

5) **Syndromes** with LVNC
- Monogenic (e.g. Danon; Fabry)
- Complex (e.g. chromosomal abnormalities)

6) **Acquired** (+/- reversible) LVNC
- Athletes
- Pregnancy
- Sickle cell anaemia
- Chronic renal failure
- Myopathies

7) **Right ventricular**/biventricular non-compaction

Adapted from Arbustini *et al.* JACC 2016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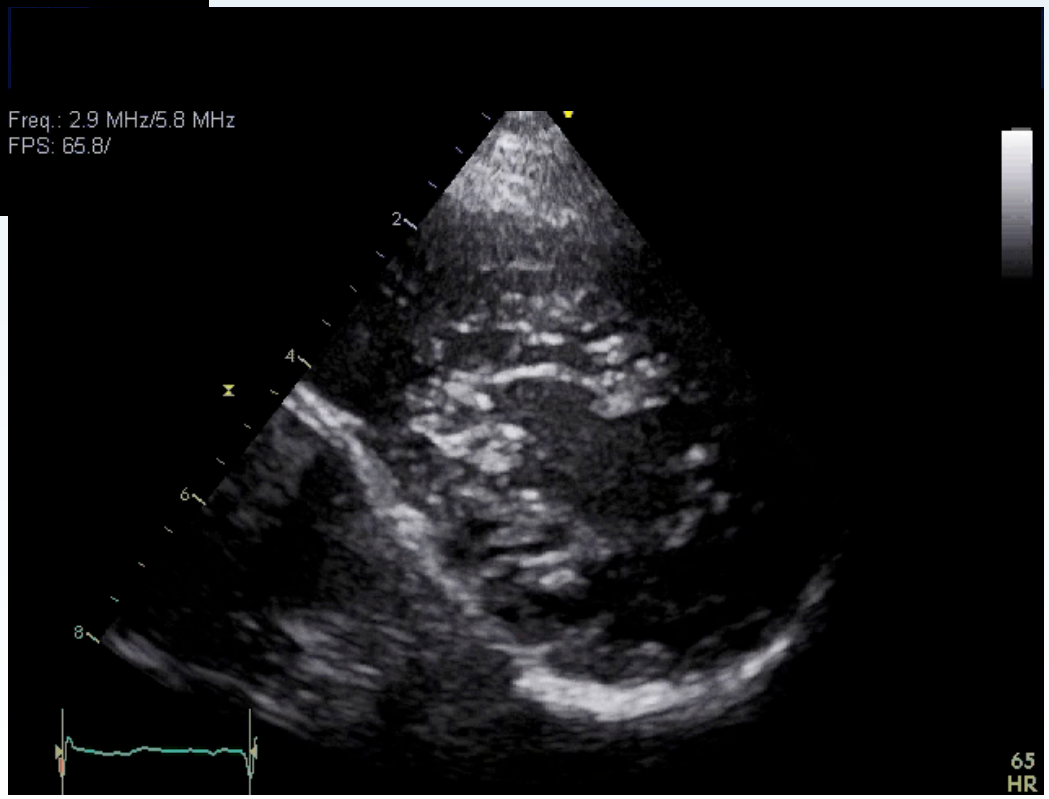
FPS: 56.5/
10
5


Freq.: 2.9 MHz/5.8 MHz
FPS: 65.8/
2
4
6
8
65
HR