Myotonic Dystrophy and The Heart

HOWARD M. ROSENFELD, MD FACC

Chief Division of Pediatric Cardiology

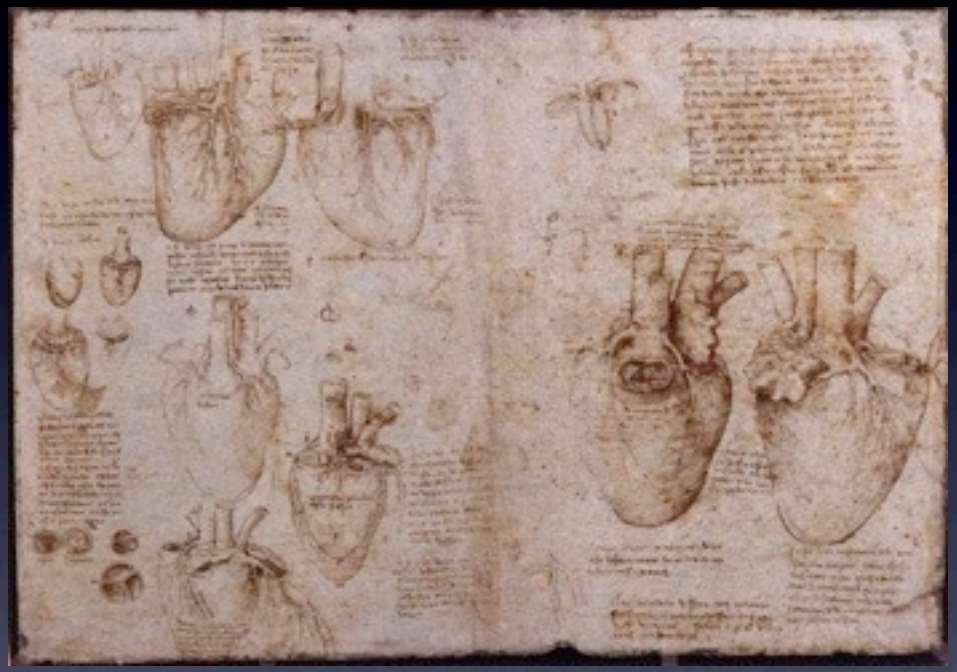
Children's Hospital and Research Center Oakland

Pediatric Cardiology Medical Group-East Bay, Inc

Overview

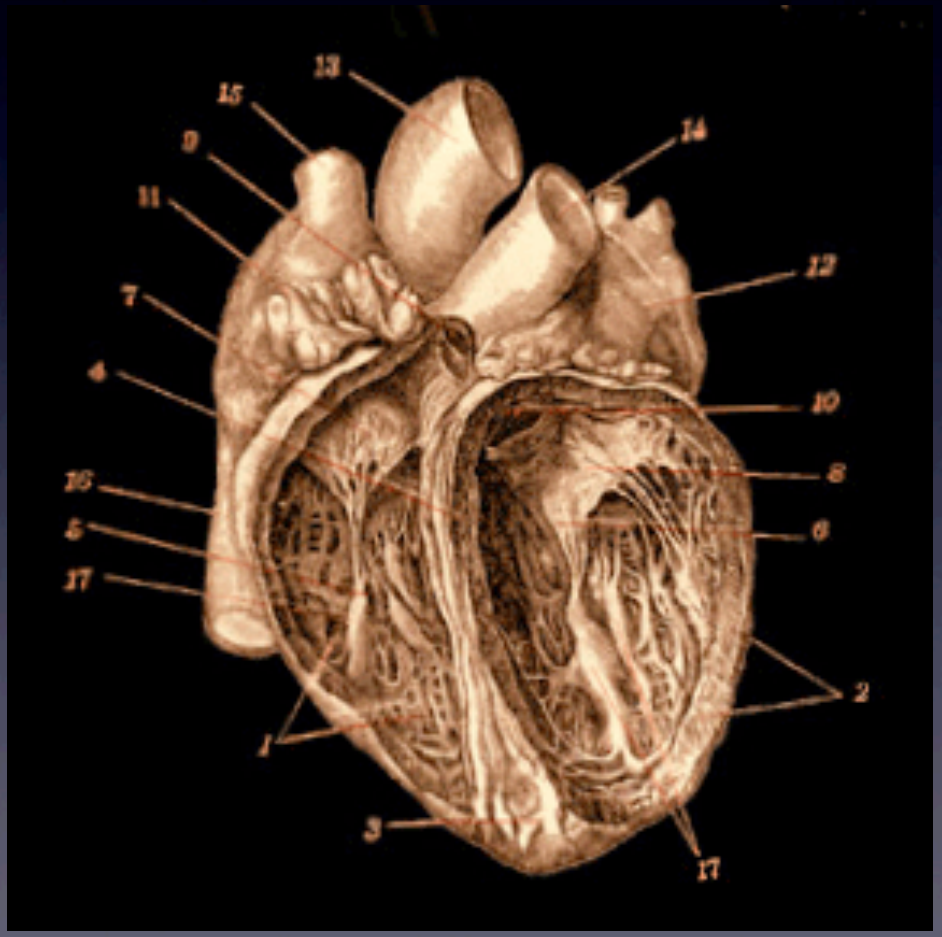
- Cardiac Anatomy/ Function
- Cardiac testing
- Cardiac Manifestations/ Symptoms
- Rhythm Disturbances
- & Management
- “level playing field”
- Myotonic Dystrophy I and II

Egyptian god of the dead, Anubis weighing a heart/soul

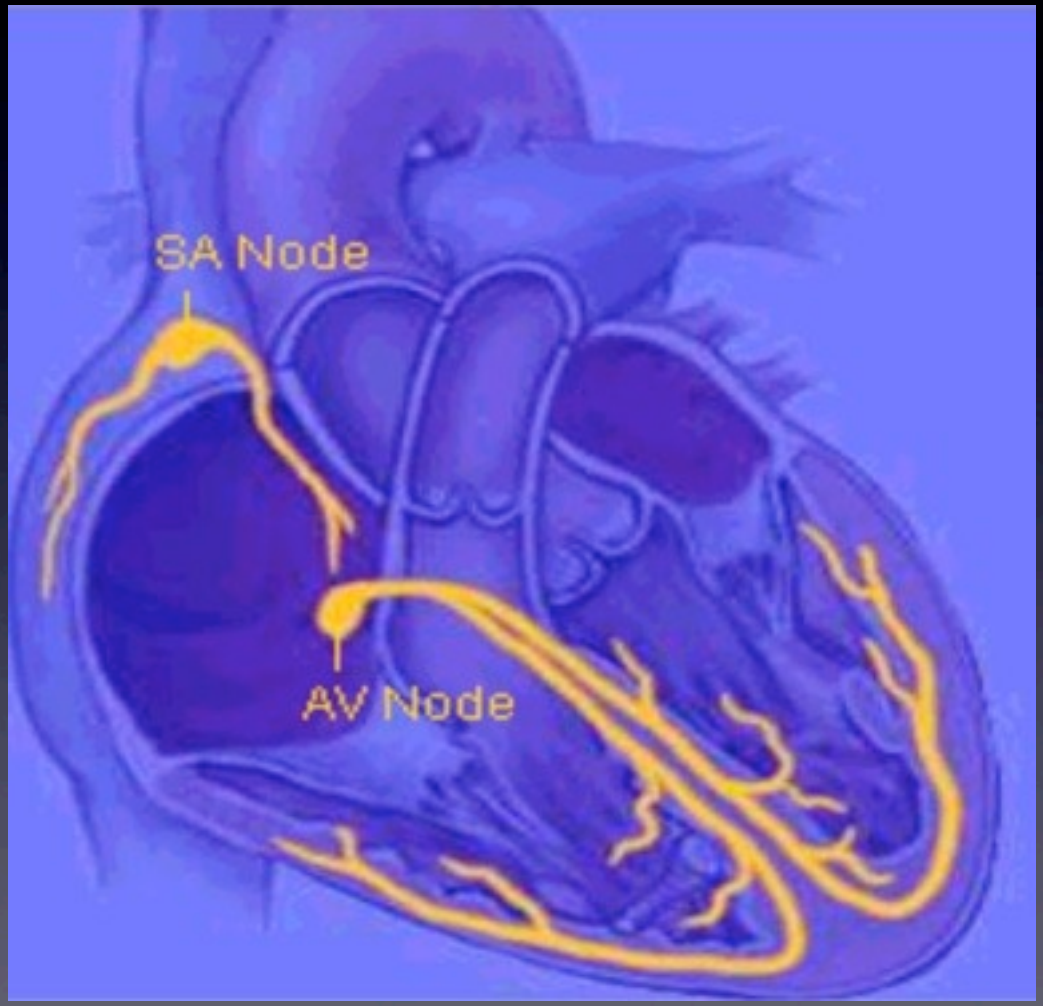


Leonardo da vinci
1510

Heart as a pump



Cardiac Electrical System

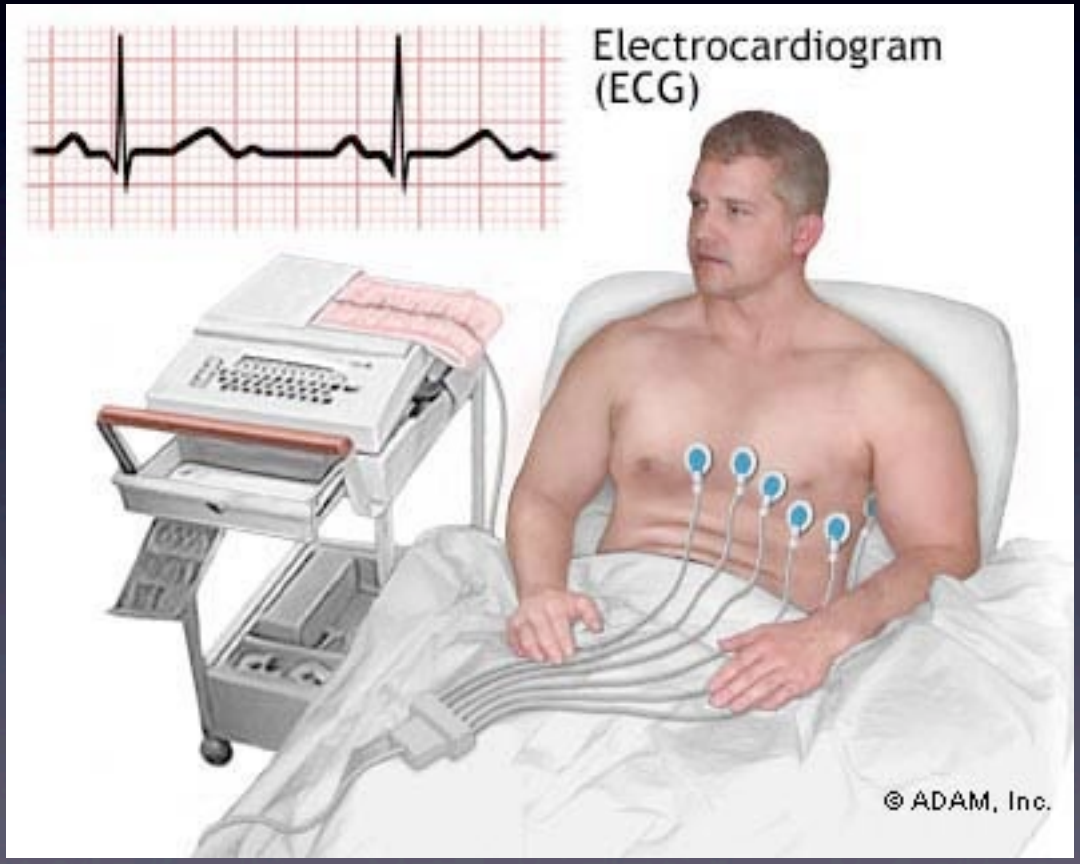


Cardiac Electrical System

Iodine absorbtion micro CT scanner

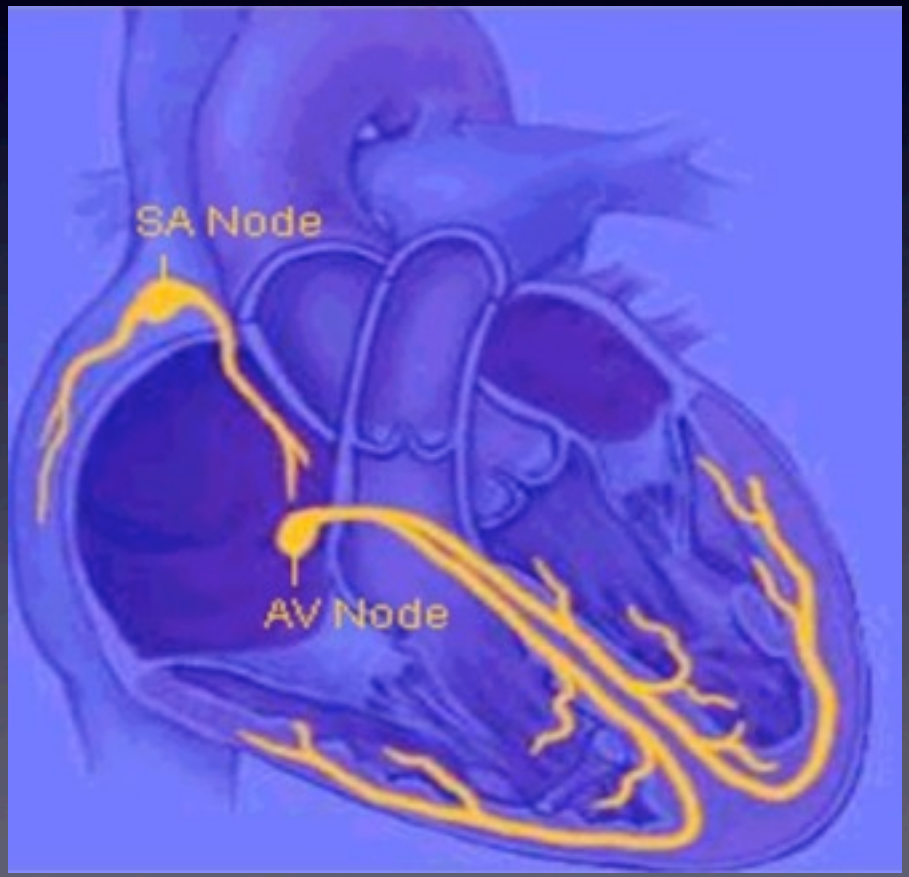
Department of Musculoskeletal Biology, Institute of Ageing & Chronic Disease, University of Liverpool

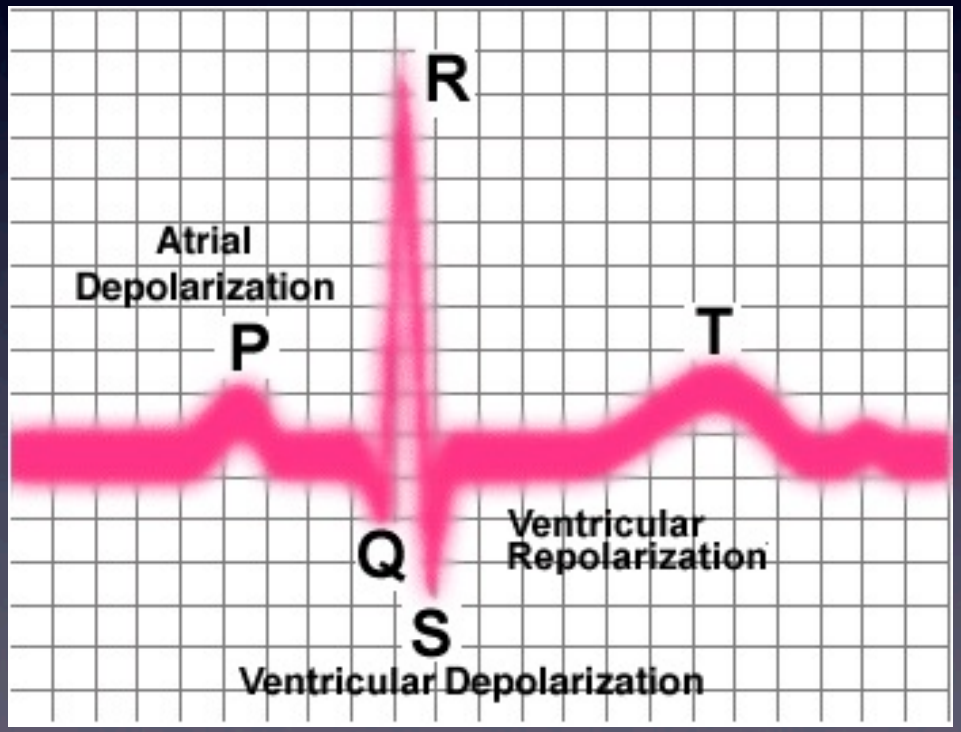
ECG/EKG



EKG Elements

"Sinus beat"

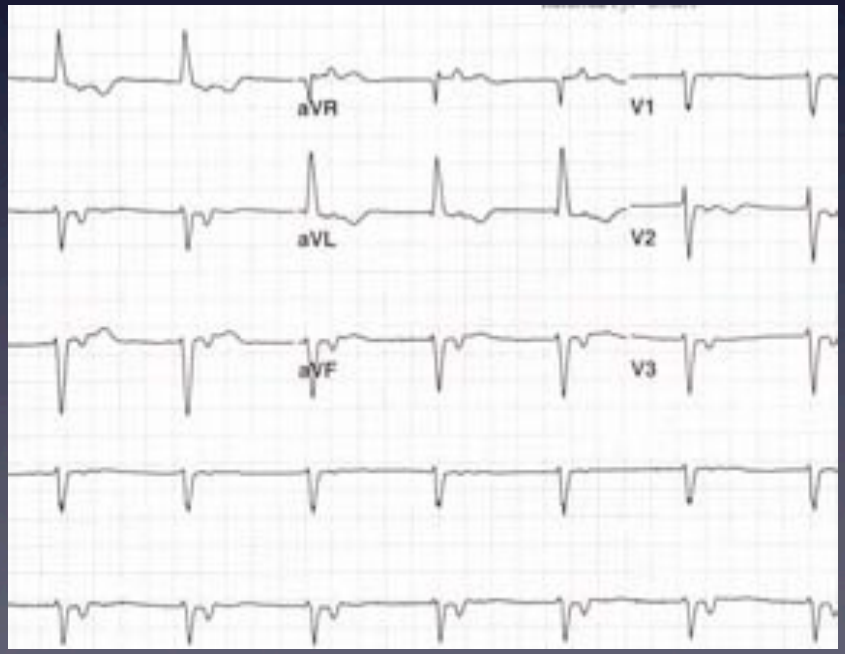




Rhythm

- Sinus rhythm requires

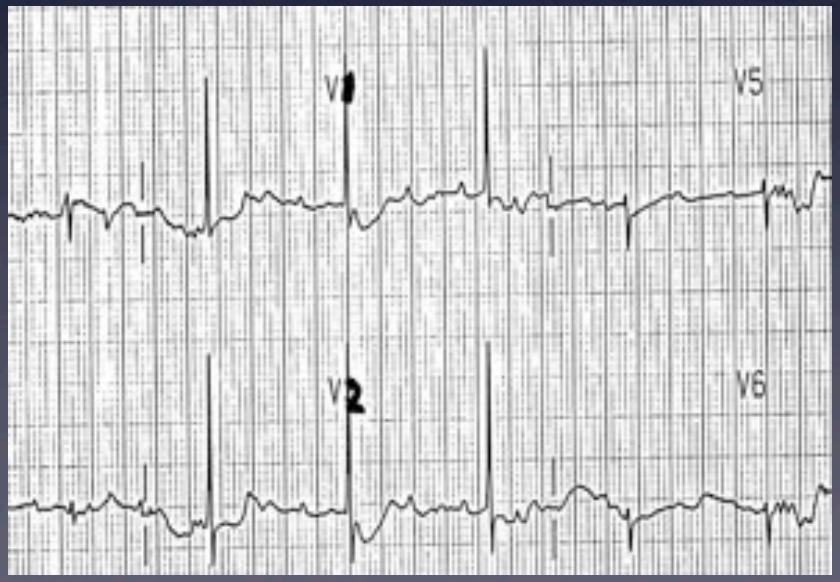
  1) P wave preceding every QRS



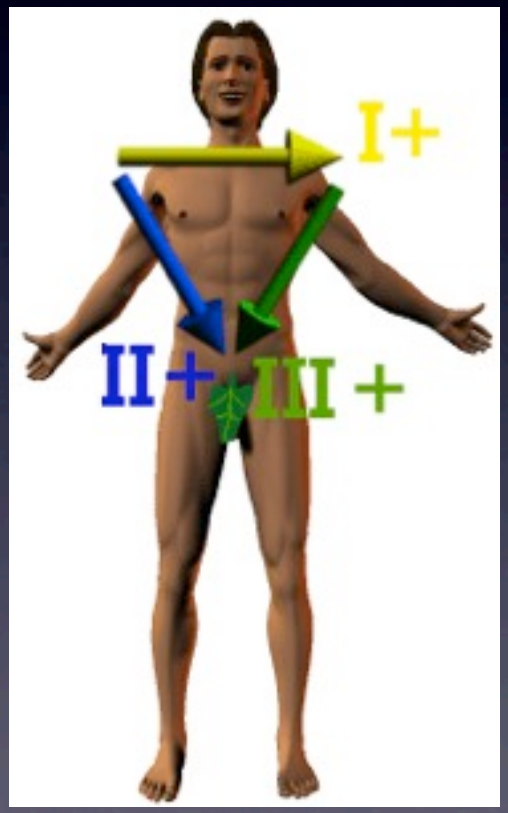
Rhythm

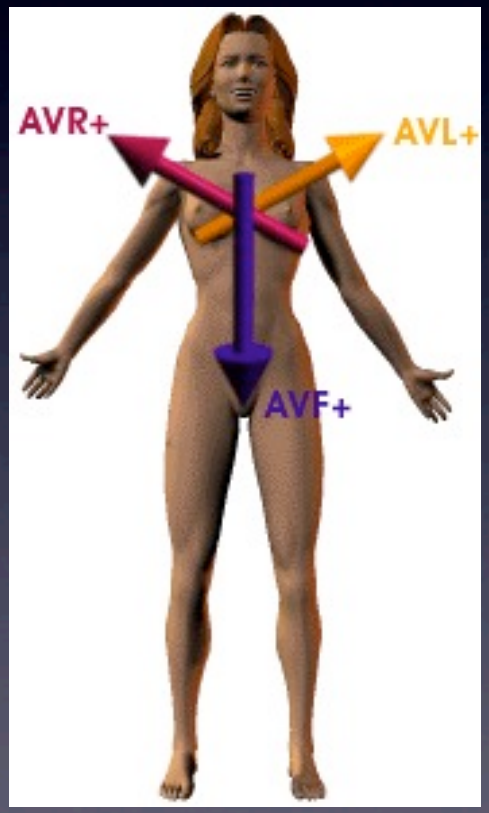
- Sinus rhythm requires

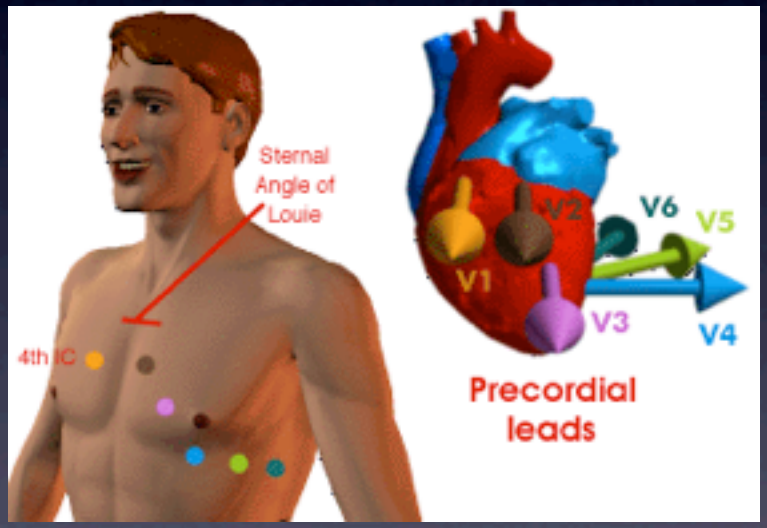
2) QRS following each P wave



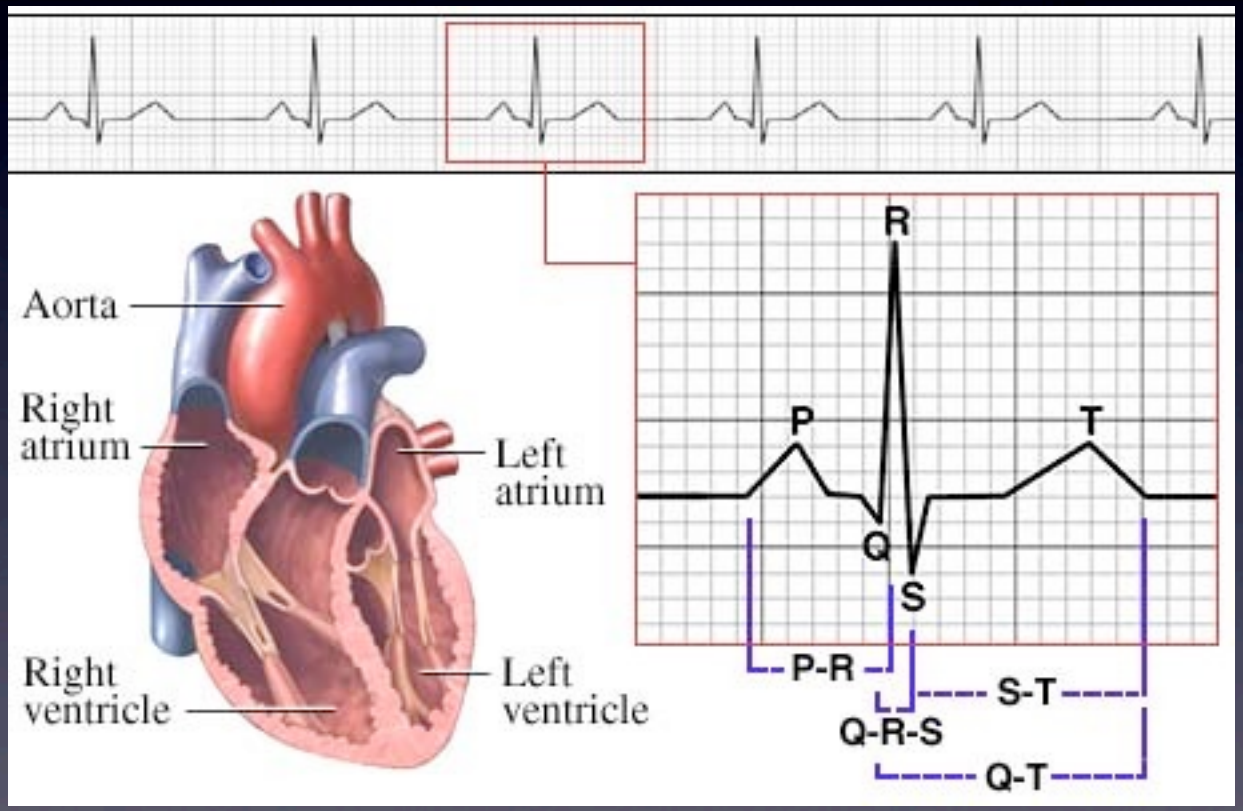
Axes


I+
II+
III +


AVR+
AVL+
AVF+


Sternal
Angle of
Louie
4th IC
V1
V2
V3
V4
V5
V6
Precordial
leads

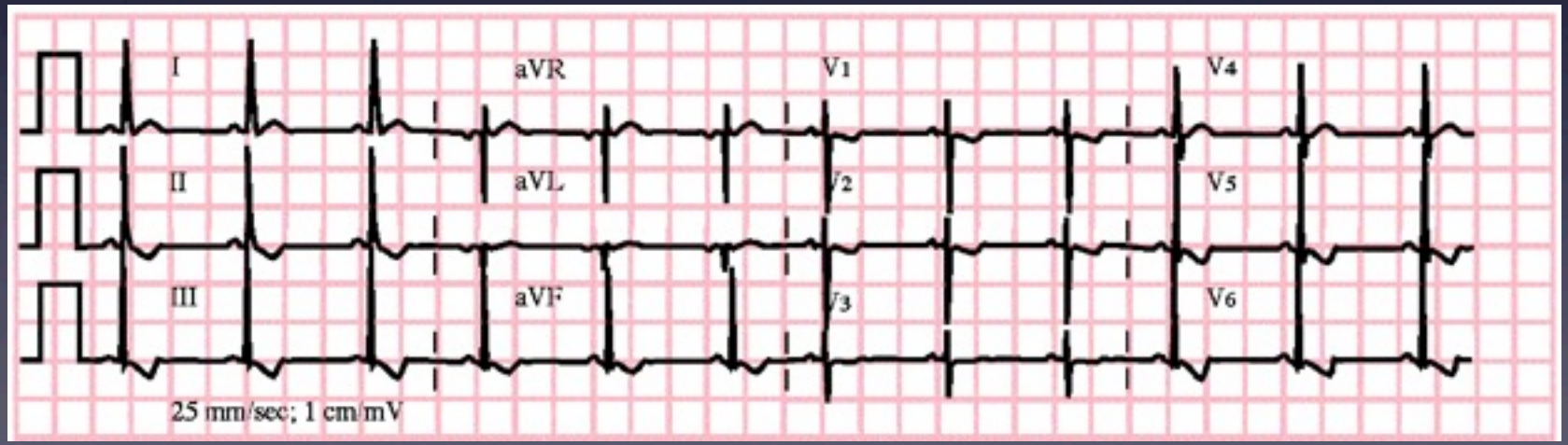
Intervals


Aorta
Right atrium
Left atrium
Right ventricle
Left ventricle
R
P
T
Q
S
P-R
S-T
Q-R-S
Q-T

FORCES

left sided

- **Ventricular hypertrophy**

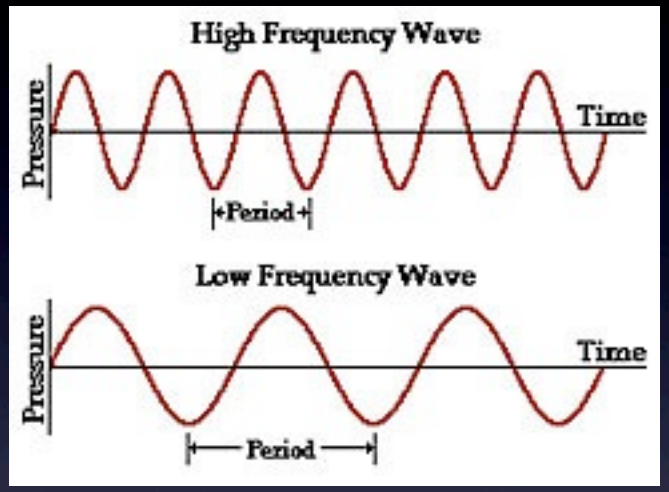


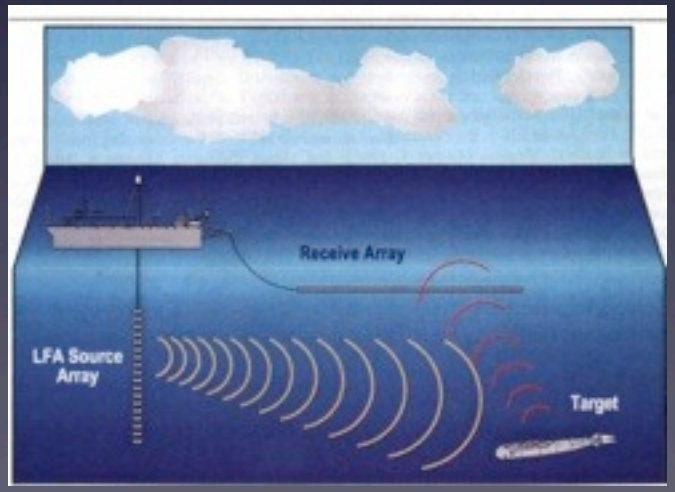
Holter monitoring

Event monitoring

Echocardiogram

Echocardiography: Physics



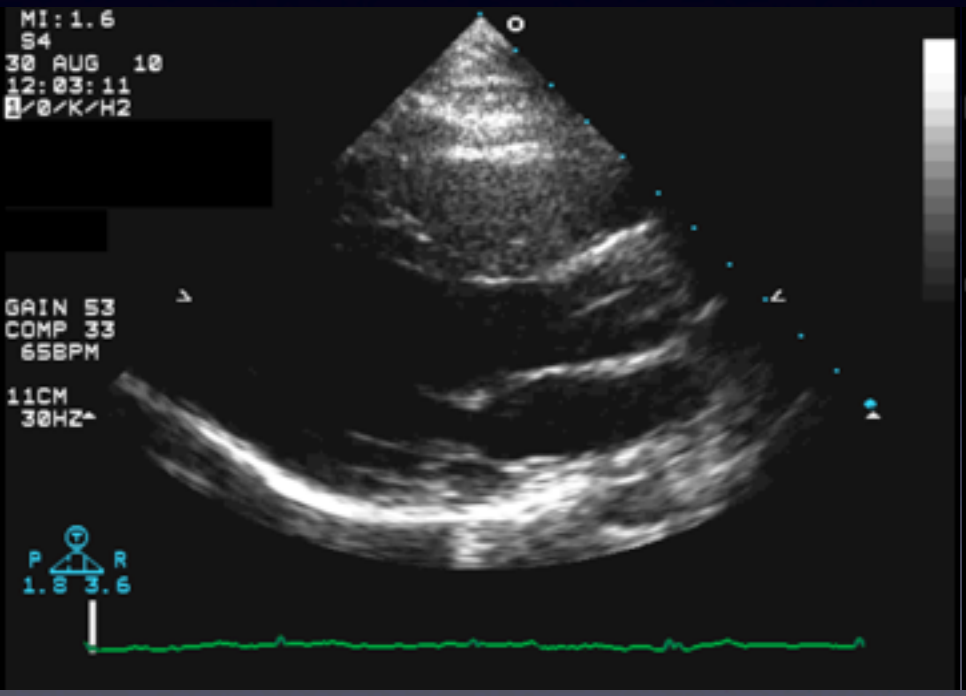


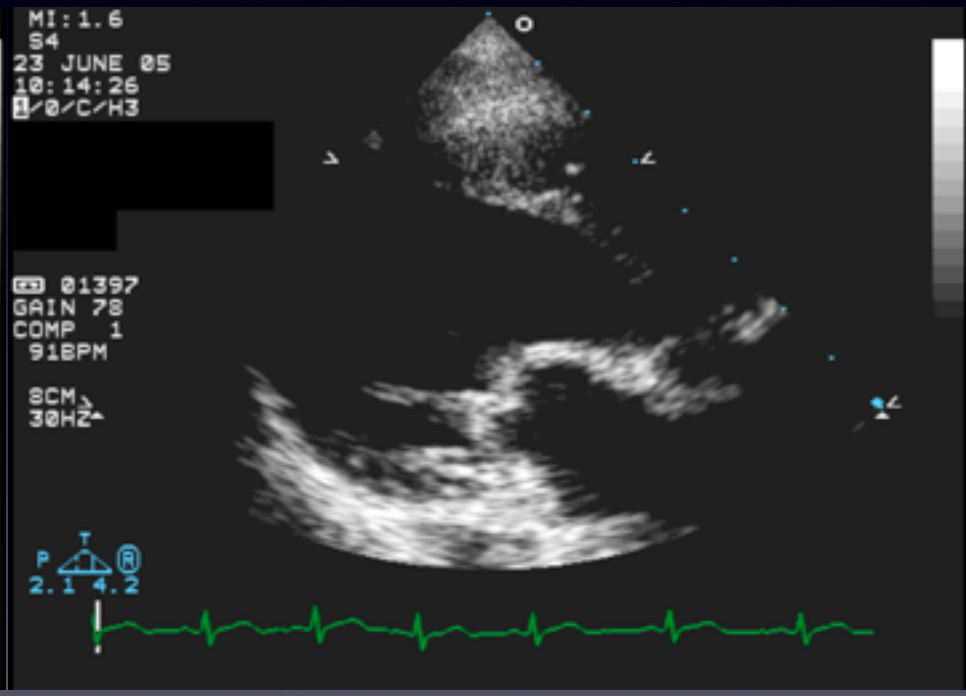
- Sound travels in waves of compression and decompression through a transmitting medium (water, tissue)
- These high frequency sound waves travel in straight lines and are either reflected or transmitted at changes in medium

Echo “Planes”

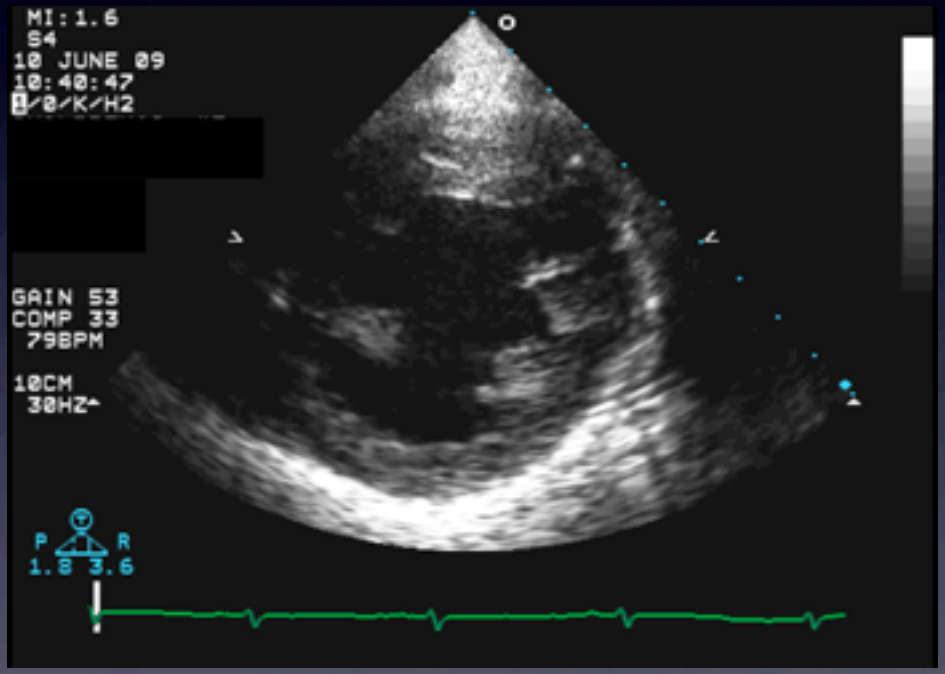
- **The plane of the transducer determines the “cut” of heart imaged**

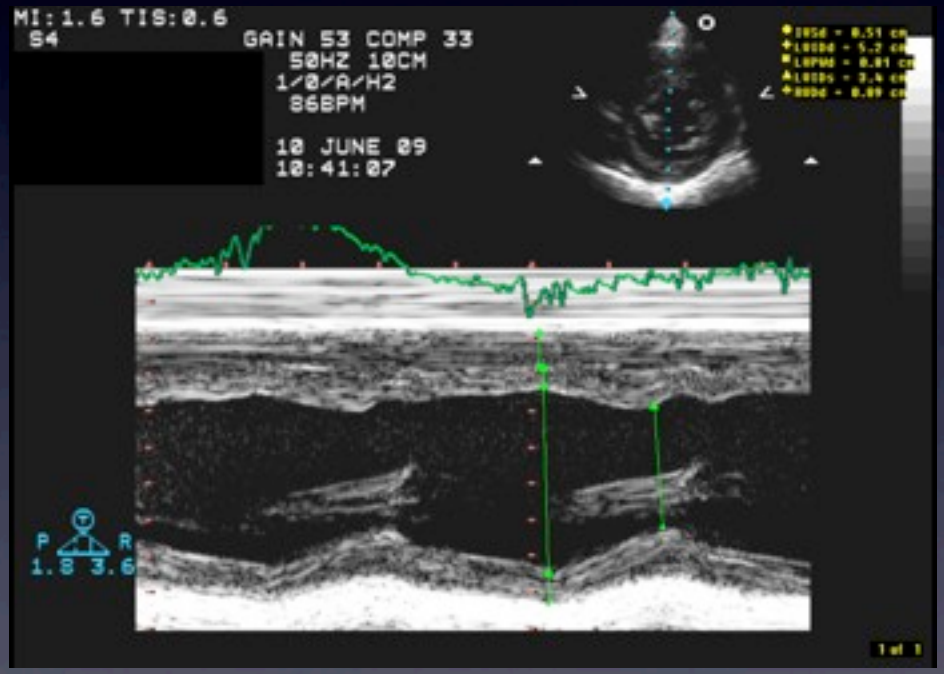
Valve Pathology



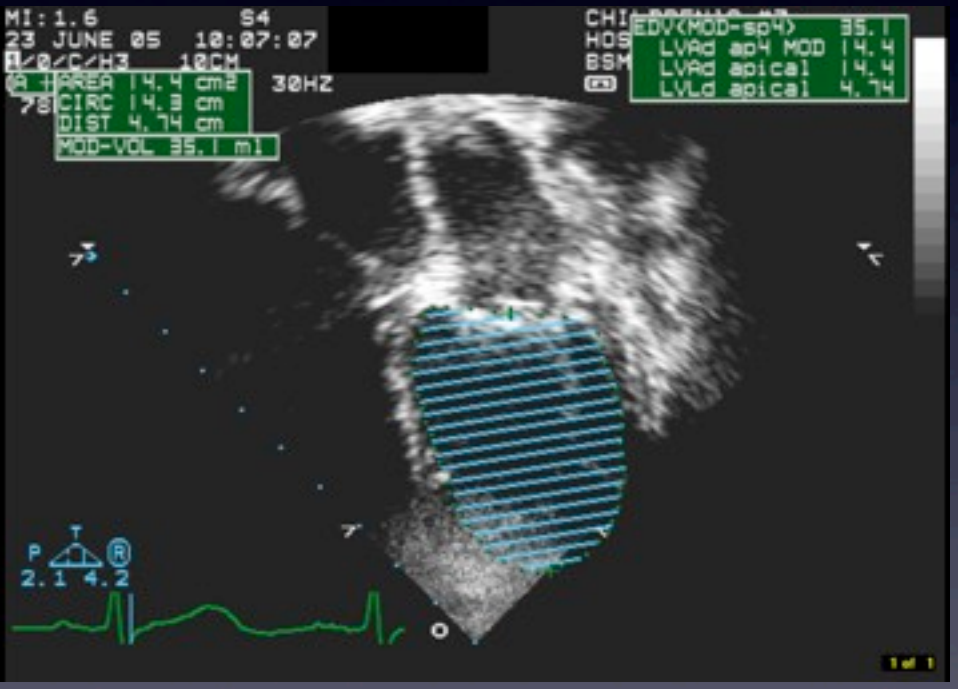


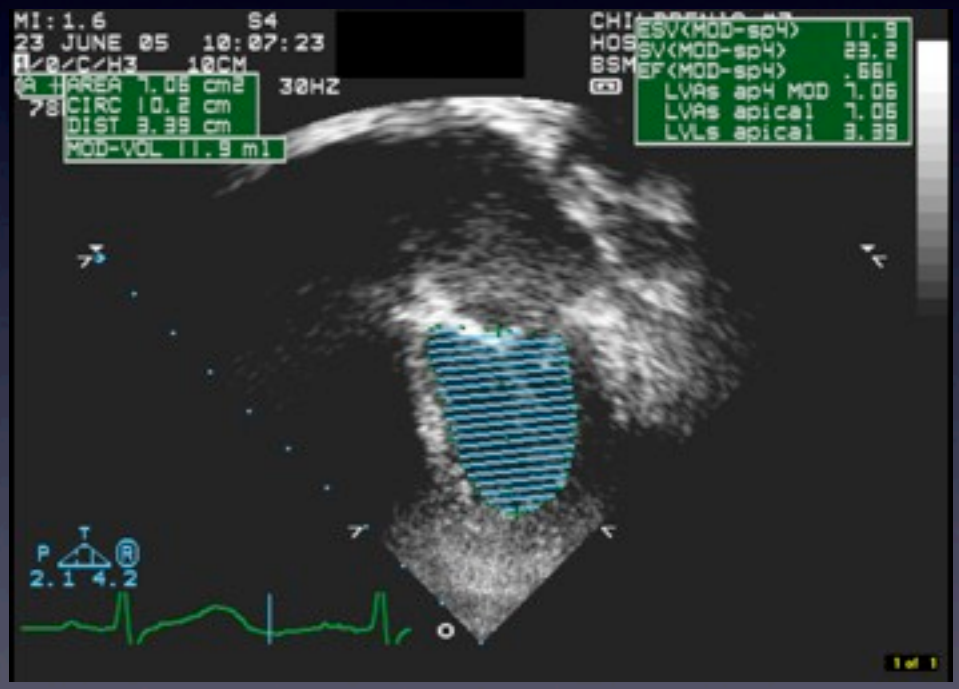
Echocardiography funciton



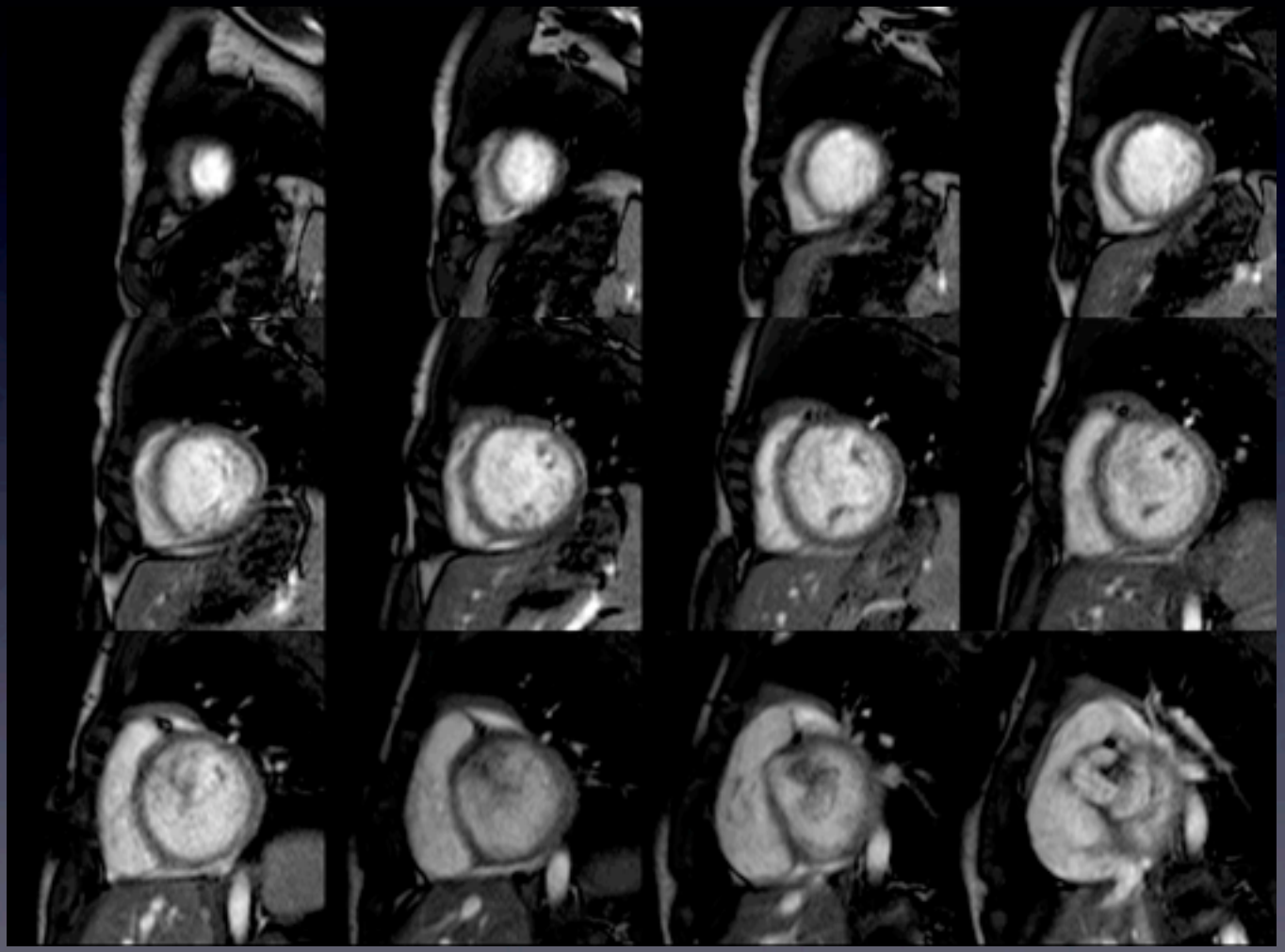


Echocardiography funciton





MRI for cardiac funciton/ morphology



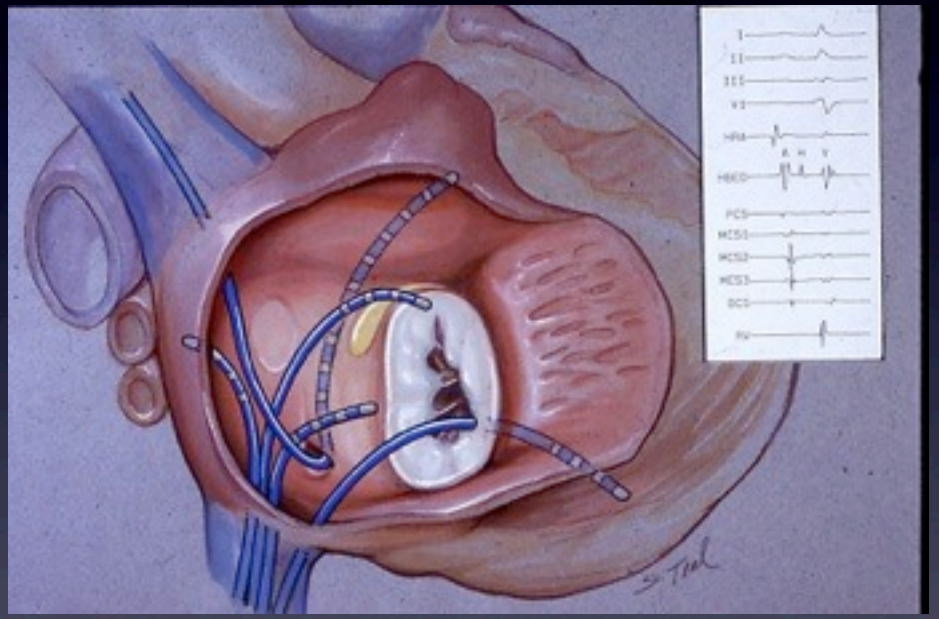
Free breathing cine imaging LV short axis stack

Catheterization

Werner Forssman 1929

Catheterization

Electrophysiology testing



MD: Cardiac Manifestations

- Pump related: Dilated Cardiomyophathy- progressive muscle weakness leading to cardiac enlargement and congestive heart failure (pump failure) or arrhythmia
- Electrical related: Conduction systemic disease- atrial arrhythmias, complete conduction block or ventricular fibrillation/ tachycardia leading to sudden cardiac death

Symptoms of **arrhythmia**

- Palpitations
- shortness of breath/exercise intolerance
- dizziness/lightheadedness
- presyncope/ syncope

Symptoms of **arrhythmia**

- Dysautonomia or Autonomic dysfunction: Included in the differential with hypotension, postural orthostatic tachycadia (POTS), or vasovagal dizziness/syncope

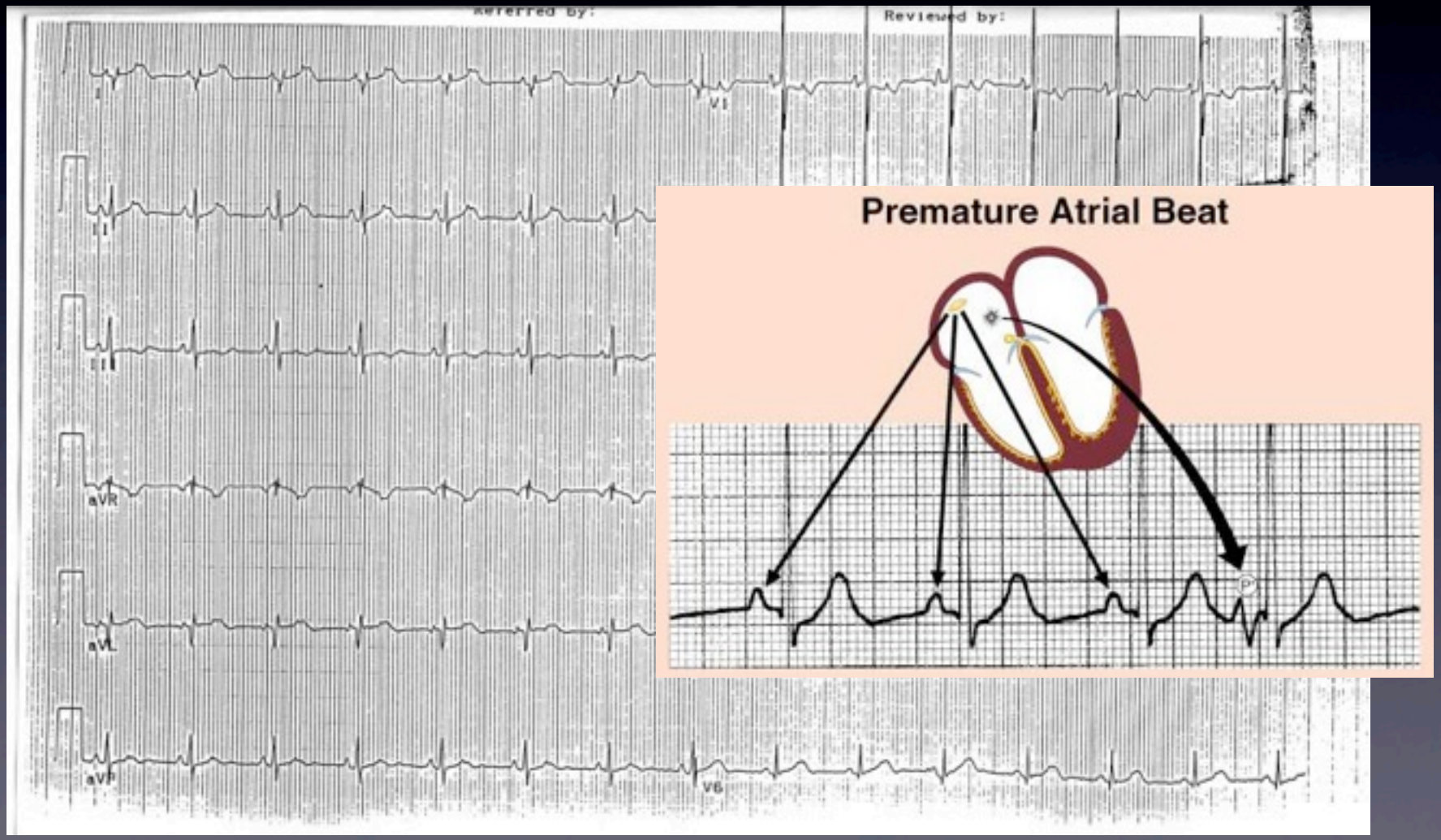
Single Extra-beats

PAC-premature atrial complex: conducted on non conducted

PVC-premature ventricular complex

Slow Cardiac rhythms

- **Premature atrial beats with block**



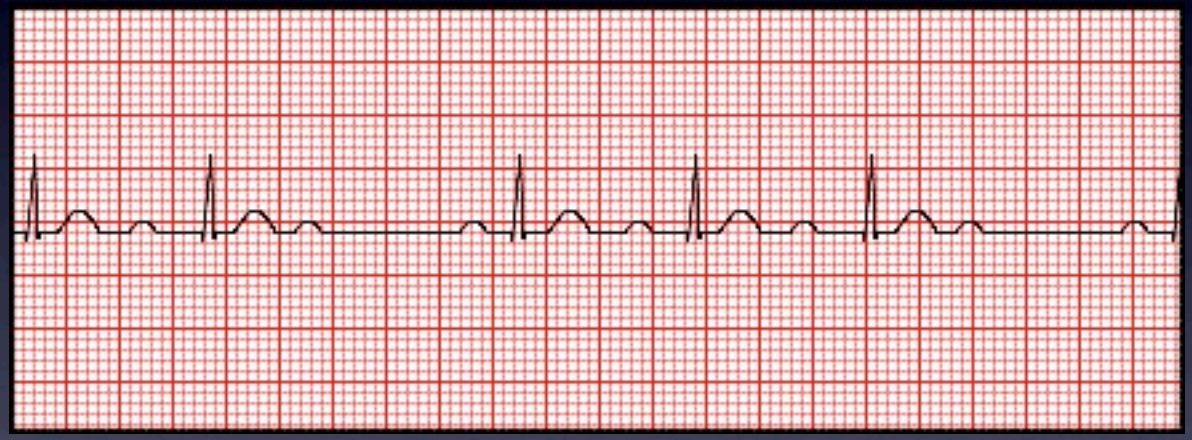
Atrioventricular block- types:

- first degree AV block- PR prolongation

Atrioventricular block- types:

- second degree AVB- dropped beats

Mobitz type 1
Wenckebach



Mobitz type 2

Atrioventricular block-types:

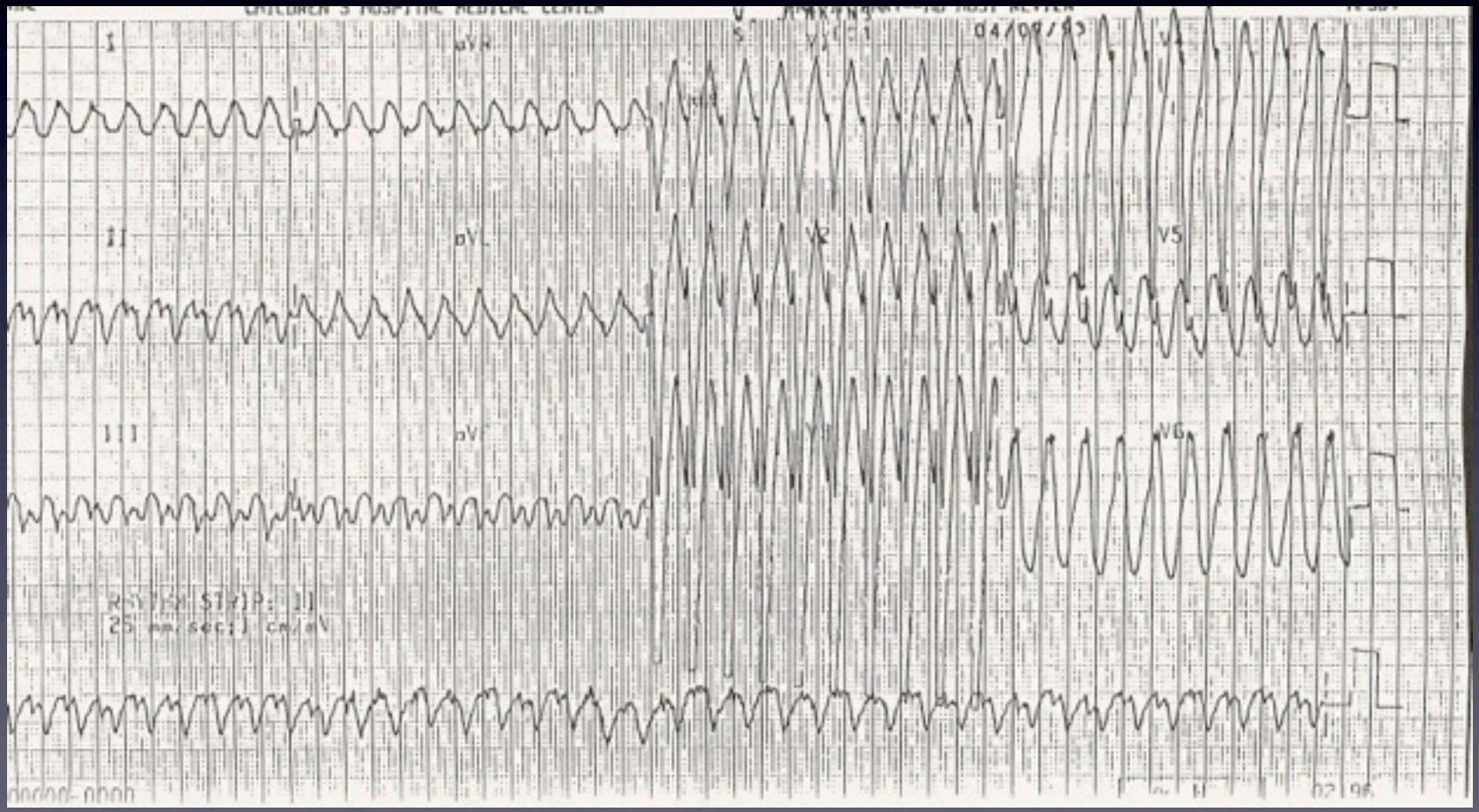
- Third degree AV block-complete heart block

Fast Cardiac Rhythms

- **Atrial flutter**

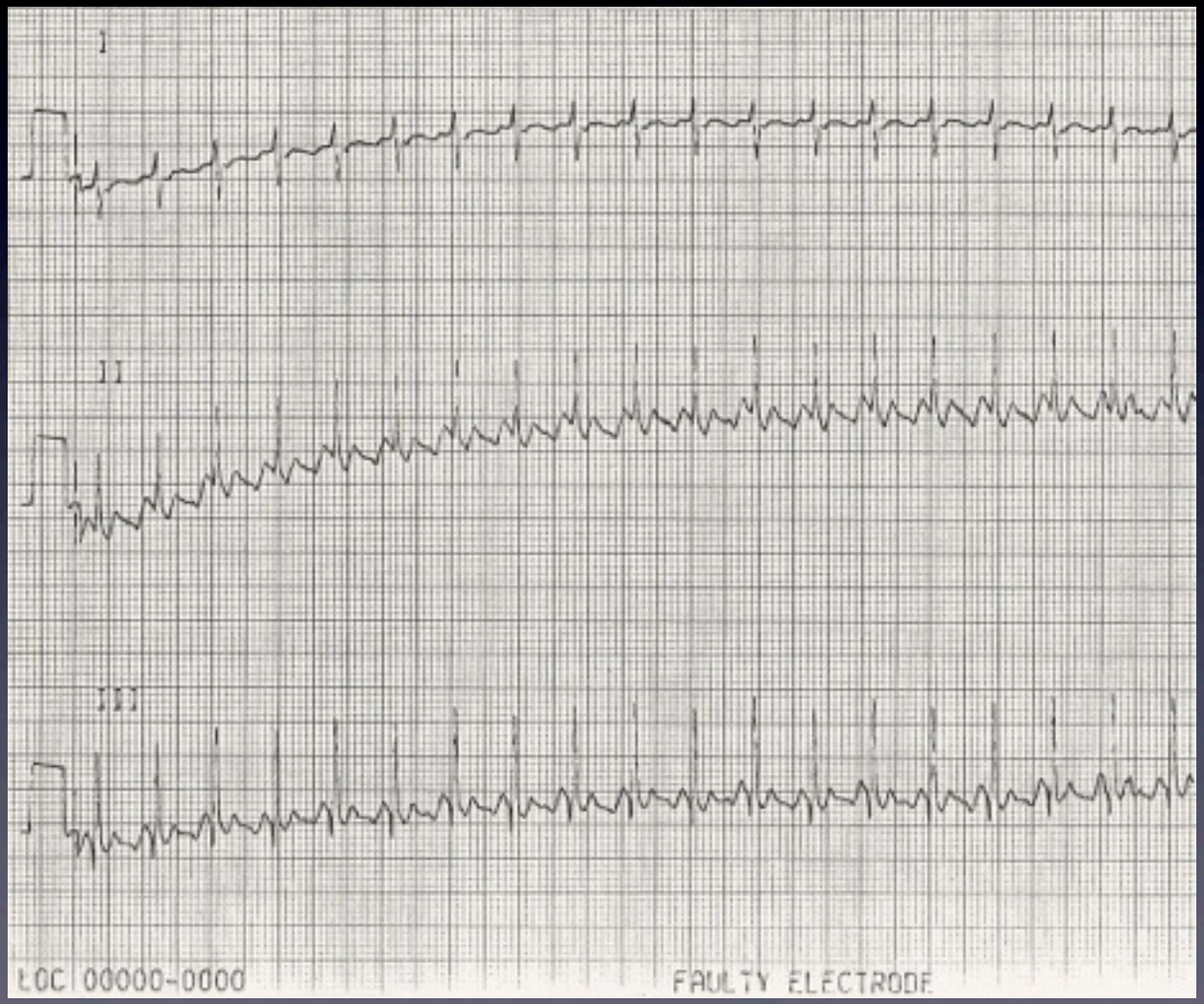
Fast Cardiac Rhythms

- **Atrial flutter with 1:1 conduction and aberrancy**



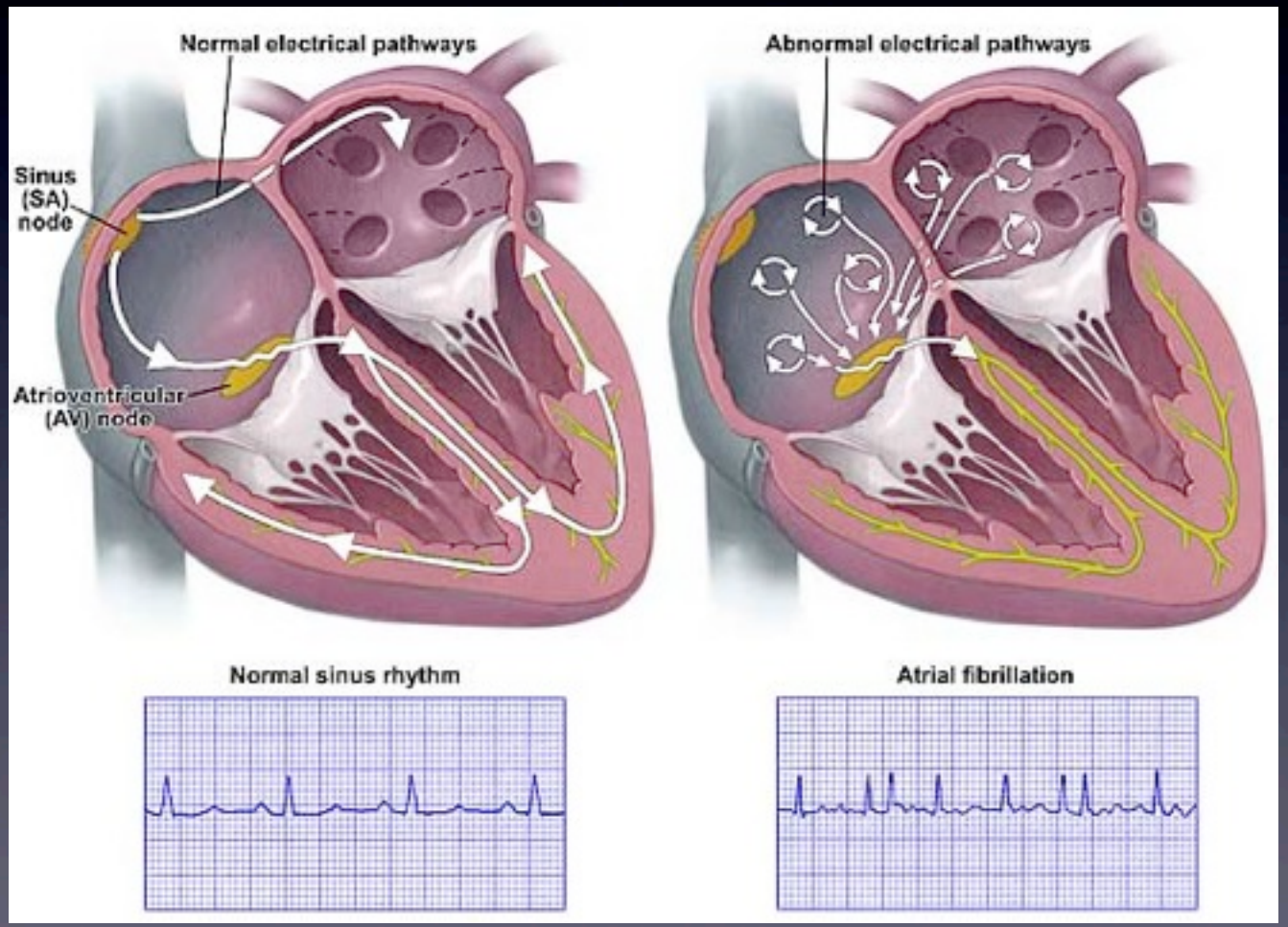
Fast Cardiac Rhythms

- **Atrial flutter- 2:1 block**



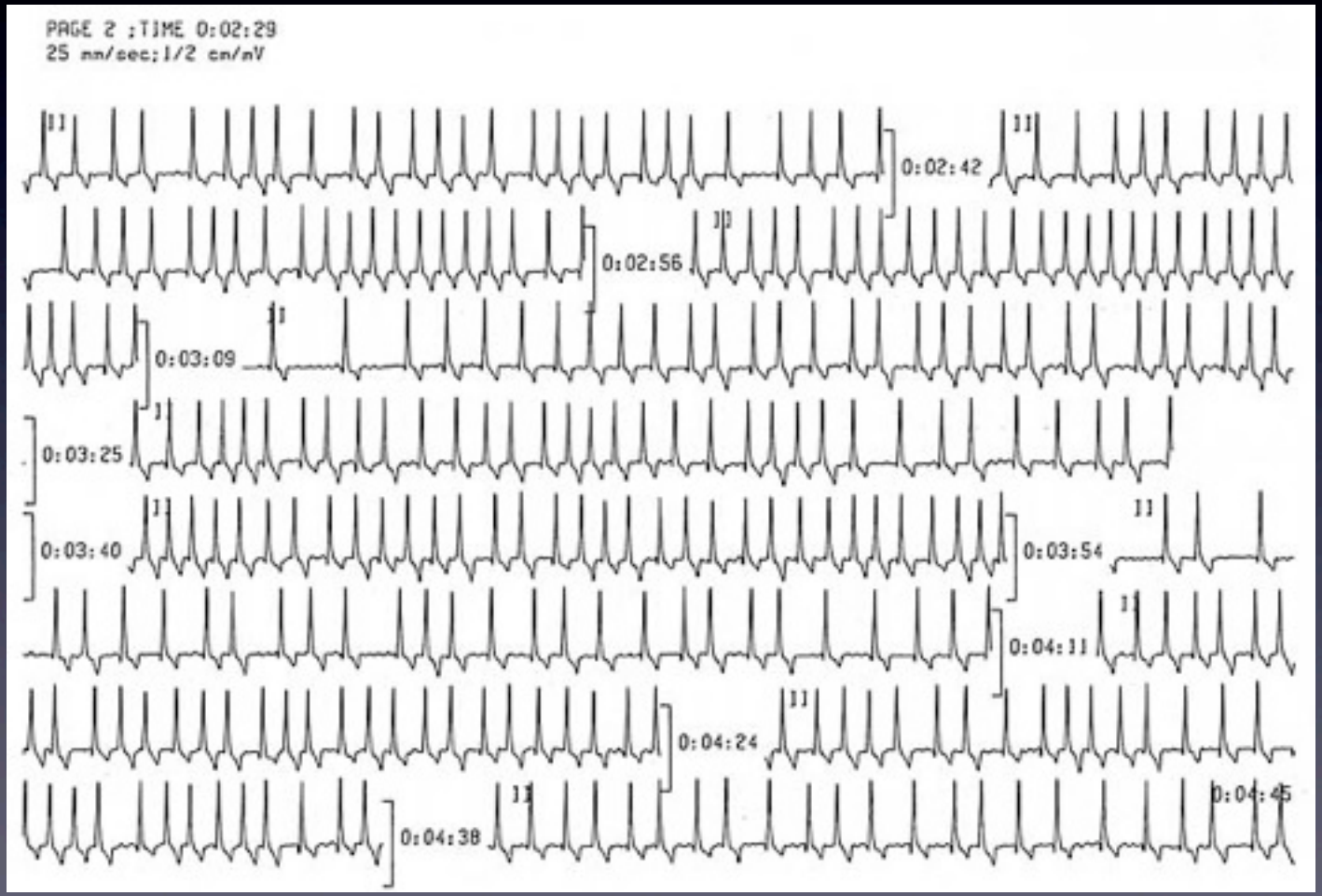
Fast Cardiac Rhythms

- **Atrial fibrillation**



Fast Cardiac Rhythms

- **Atrial fibrillation**

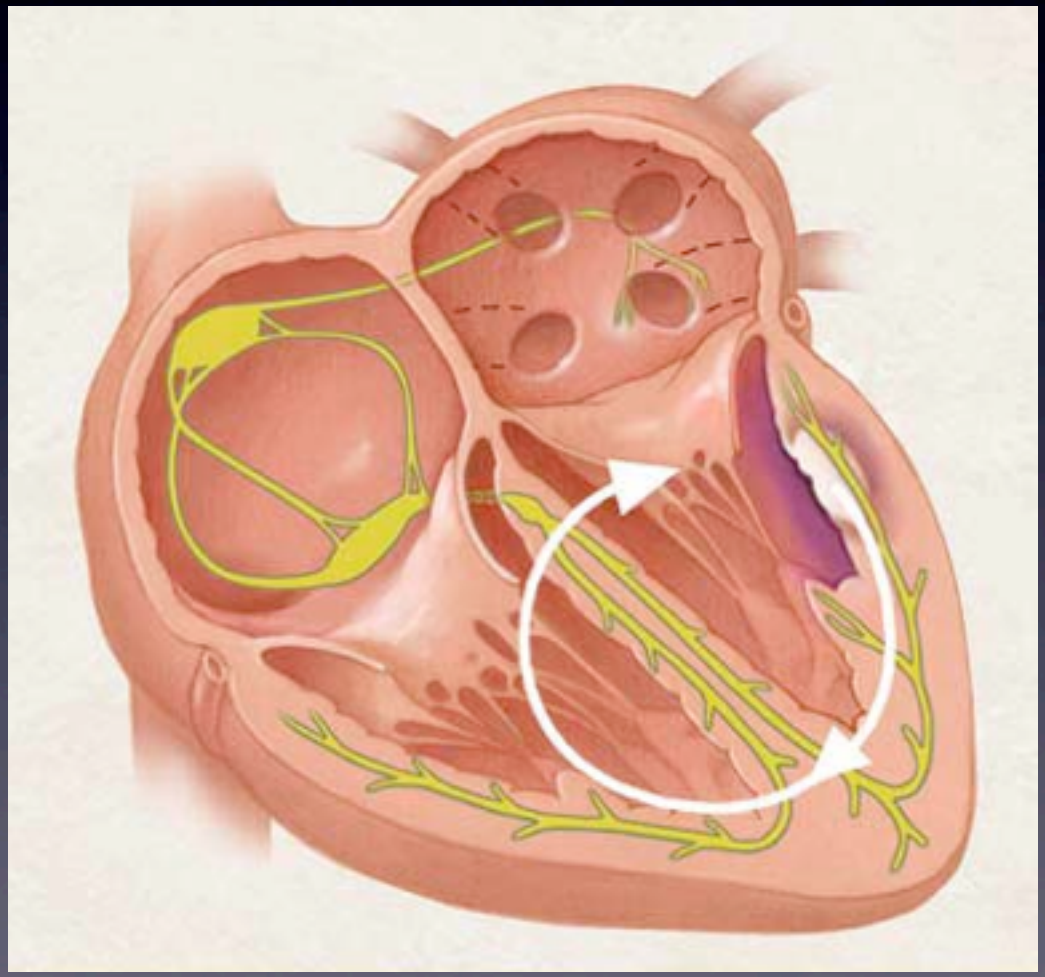


Fast Cardiac Rhythms

- **atrial flutter: cardioversion**

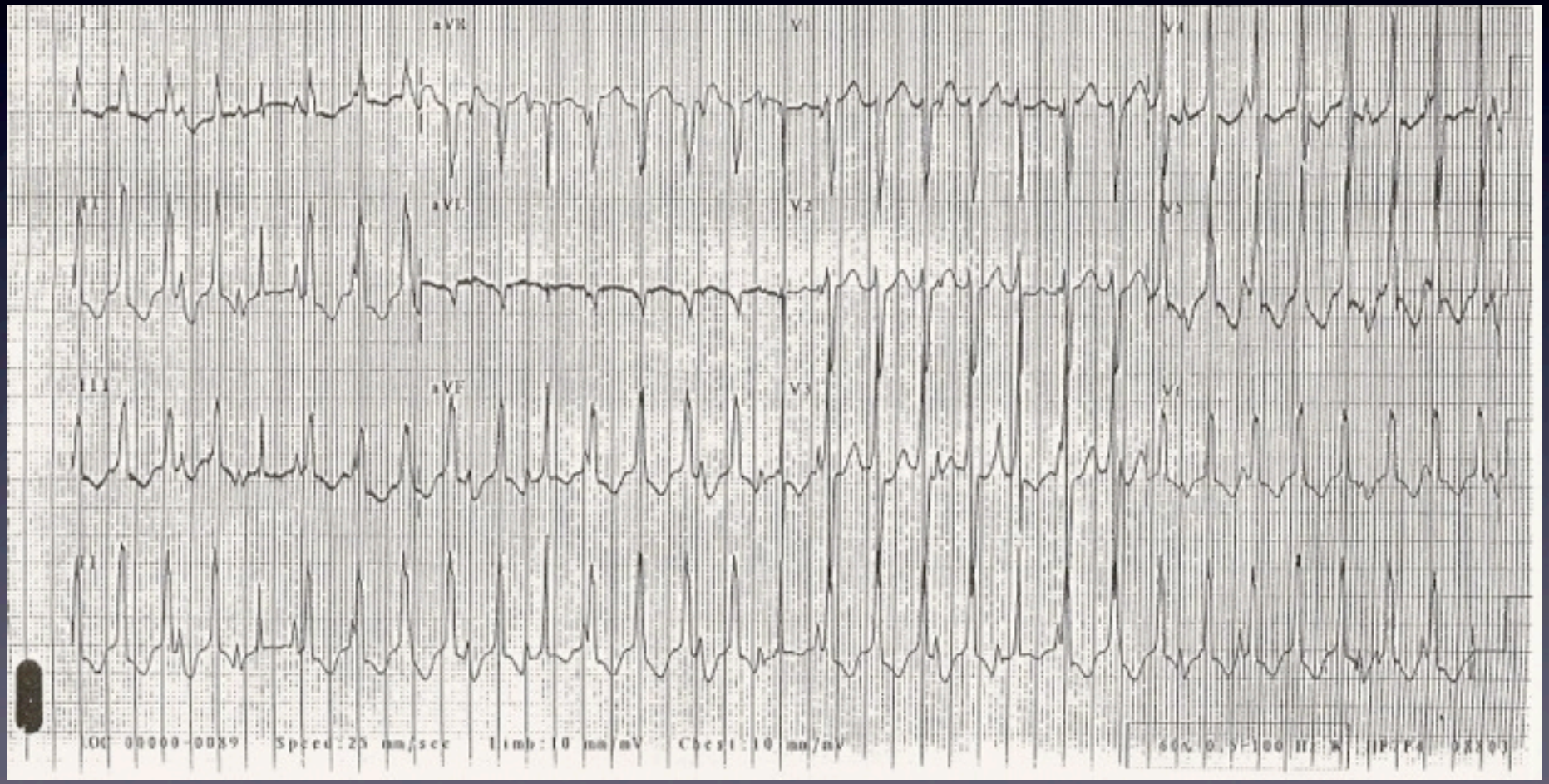
Fast Cardiac Rhythms

- ventricular tachycardia



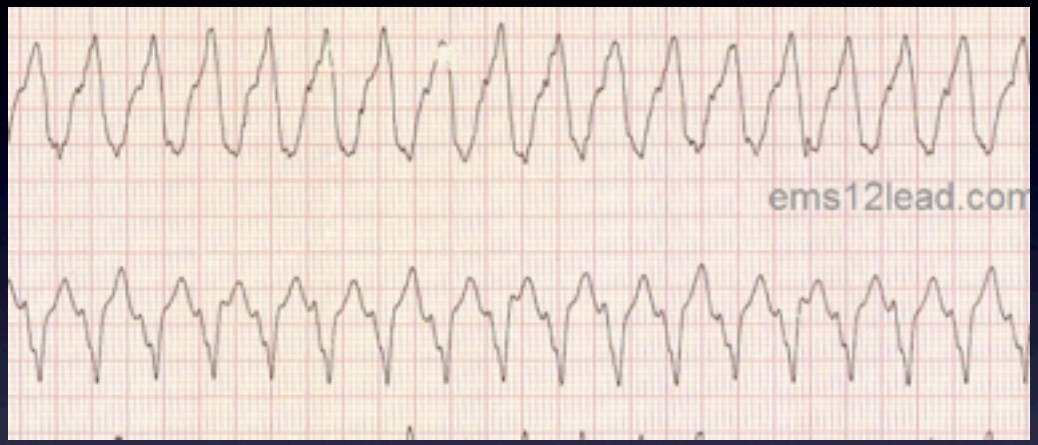
Fast Cardiac Rhythms

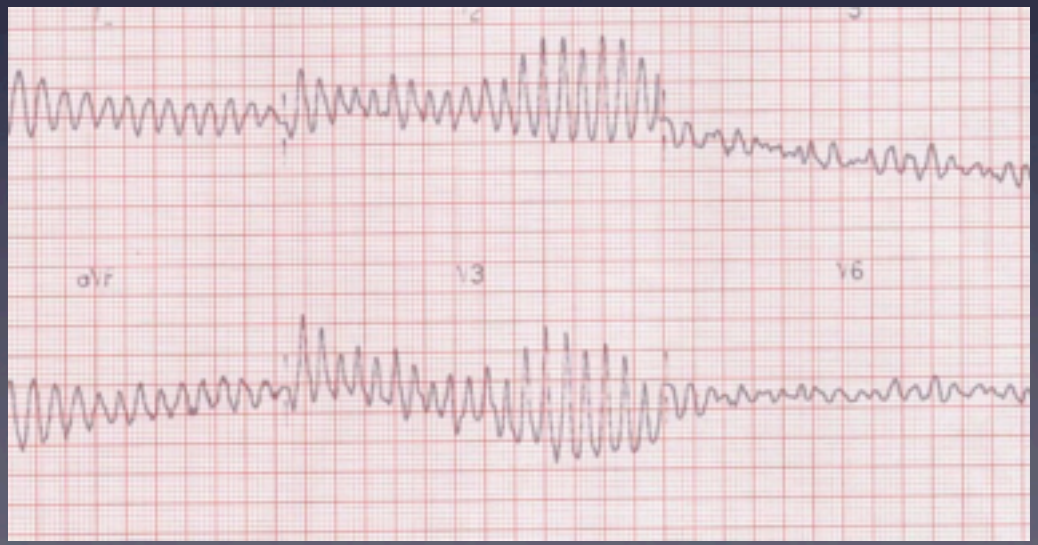
- perfusing ventricular rythm



Fast Cardiac Rhythms

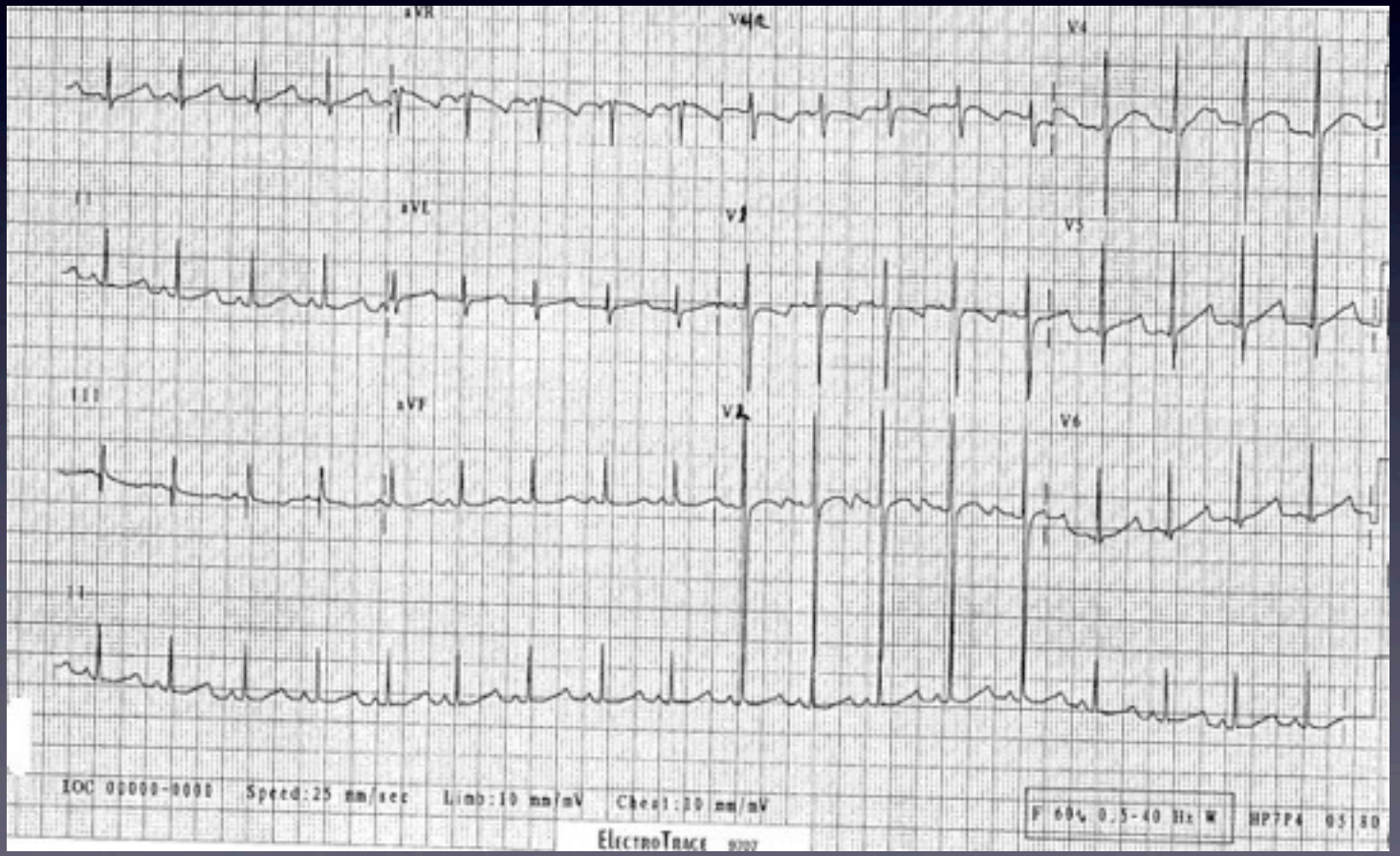
Nonperfusiong ventricurlar ryhthm





Rhythm Disturbances

- **Prolongation of the corrected QT interval**



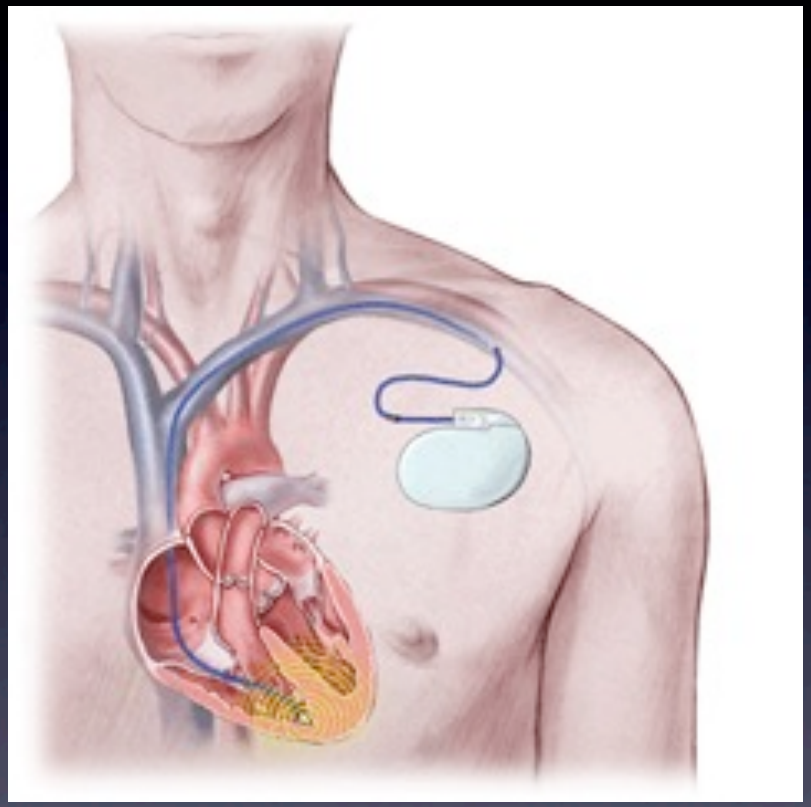
Cardiac Evaluation

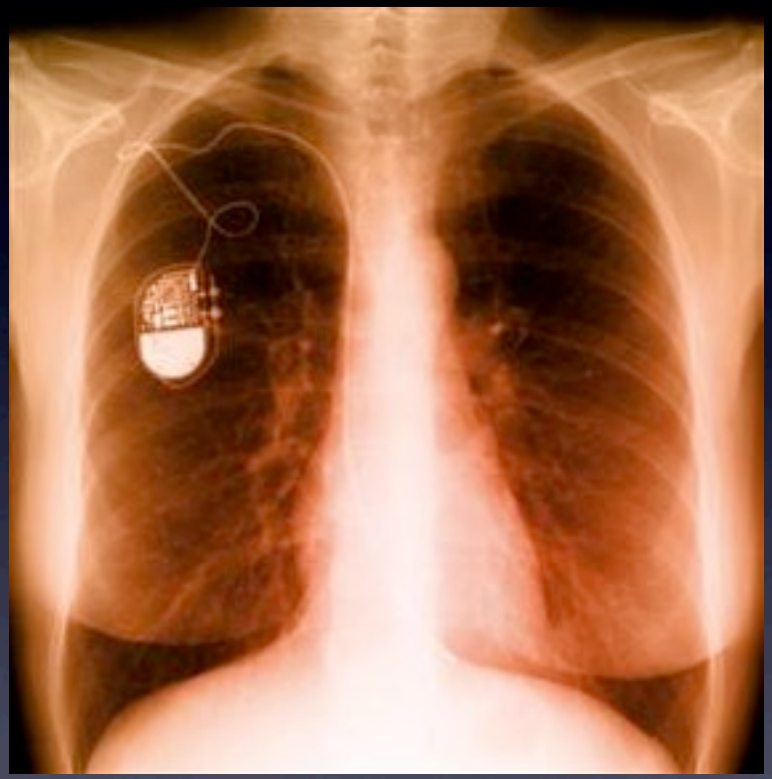
- Annual Cardiology visit
- Annual EKG
- Holter monitoring as needed
- Echocardiography (every 2-5 years or as needed based on functional concerns)
- Catheterization
- Electrophysiology Testing

Arrhythmia and Sudden death in MD

- Normal EKG = low risk for sudden cardiac death over a 5 year period
- Arrhythmias in younger patients more frequently tachyarrhythmias than conduction block
- Endpoints of Sudden cardiac death or pacemaker implantation:
  - associated with prolonged baseline PR interval & QTc
  - Looser association- advanced age/muscle imapirment.
  - No association with number of CTG repeats

Pacemaker & AICD





Pacemaker-slow rhythms and block

Defibrillator- slow rhythms/block & fast rhythms

N Engl J Med. 2008 Jun 19;358(25):2688-97.

Electrocardiographic abnormalities and sudden death in myotonic dystrophy type 1.

Groh WJ, Groh MR, Saha C, Kincaid JC, Simmons Z, Ciafaloni E, Pourmand R, Otten RF, Bhakta D, Nair GV, Marashdeh MM, Zipes DP, Pascuzzi RM.

Department of Medicine, Krannert Institute of Cardiology, Indiana University, Indianapolis 46202, USA. wgroh@iupui.edu

- 406 adult MD1 patients
- 5.7 year follow-up 27 sudden deaths
- SD Associated with
  - Severe EKG abnormality: non-sinus rhythm, PR greater than 240msec, QRS interval greater than 120msec, 2nd or 3rd degree AVB
  - Atrial tachyarrhythmias

Indication for pacemaker therapy

- 2nd and 3rd degree AV block
  Class I indication: Condition in which permanent pacing is definitely beneficial, useful, and effective. Implantation of a cardiac pacemaker is acceptable and necessary.
- 1st degree AV block
  Class IIB indication: Condition in which the usefulness, efficacy of permanent pacing is less well established by evidence/opinion.

JAMA. 2012 Mar 28;307(12):1292-301.

Electrophysiological study with prophylactic pacing and survival in adults with myotonic dystrophy and conduction system disease.

Wahbi K, Meune C, Porcher R, Bécane HM, Lazarus A, Laforêt P, Stojkovic T, Béhin A, Radvanyi-Hoffmann H, Eymard B, Duboc D.

Pierre et Marie Curie-Paris 6 University, Myology Institute, Pitié-Salpêtrière Hospital, 75013 Paris, France. karim.wahbi@cch.aphp.fr

- Is an invasive strategy with electrophysiolgic studies and prophylactic permanent pacing in MD1 patients with infranodal conduction delays superior to a noninvisive stragety?
- Conclusion: Among patients with MD1, an invasive strategy was associated with a higher rate of 9 year survival when compared with a noninvasive strategy.

Defibrillator Therapy

- Secondary prevention: preventing sudden cardiac death following the survival of an initial event
- Primary prevention: Preventing sudden cardiac death before the occurance of an initial event

J Cardiovasc Electrophysiol. 2011 Dec;22(12):1369-75. doi: 10.1111/j.1540-8167.2011.02200.x. Epub 2011 Oct 28.

Pacemaker and implantable cardioverter-defibrillator use in a US myotonic dystrophy type 1 population.

Bhakta D, Shen C, Kron J, Epstein AE, Pascuzzi RM, Groh WJ.

Division of Cardiology, Department of Medicine, Krannert Institute of Cardiology Department of Biostatistics, Indiana University, Indianapolis, Indiana, USA.

- Study: Assessment in MD1 of implant rates, indications, and outcomes for patients receiving pacemakers or implantable cardioverter-defibrillators
- Conclusion: MD1 patients commonly receive antiarrhythmic devices. The risk of VT/VF and sudden death suggests that AICDs rather than pacemakers should be considered for these patients.

What can you do??

- Be aware of symptoms of heart disease: fatigue, SOB, CP, palpitations, dizziness and syncope
- Regular EKG and cardiologist involvement
- Be knowledgable and a good self advocate regarding cardiac disease
- Research therapeutic options carefully

Flecainide in MD??

J Clin Neuromuscul Dis. 2005 Sep;7(1):25-8.

A cautionary tale: the risks of flecainide treatment for myotonic dystrophy.

Gorog DA, Russell G, Casian A, Peters NS.

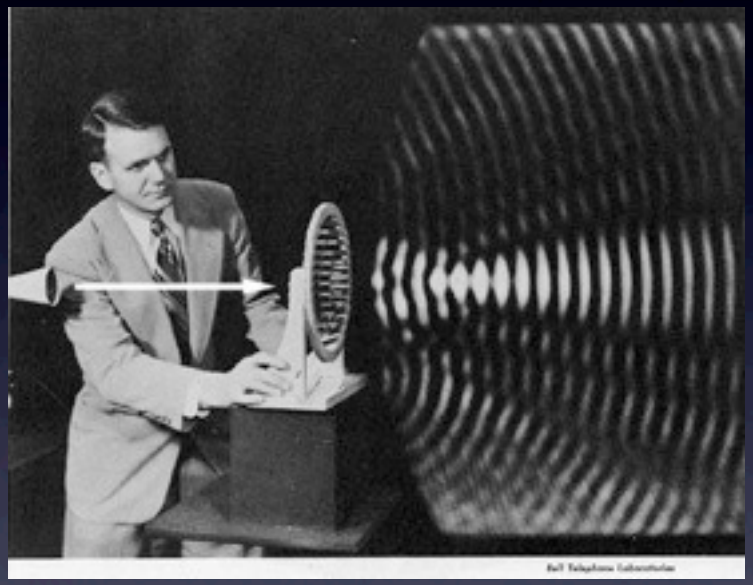
From the Waller Cardiac Department, St. Mary's Hospital, Imperial College, London, UK.

Abstract

Myotonic dystrophy (MD) is associated with important cardiac abnormalities, and 30% of deaths are attributable to cardiac causes, predominantly arrhythmias. Sodium channel blockers have been used to improve muscle strength and relaxation in MD. Flecainide is a potent selective blocker of the mutant sodium channel in myotonia and inhibits the abnormal noninactivating sodium current in both painful myotonia congenita and painless MD with a resultant improvement in muscle relaxation. We describe the case of a 41-year-old woman with MD who developed ventricular tachycardia (VT) while taking flecainide to improve her muscle strength. Flecainide was discontinued and VT could not subsequently be induced. Although flecainide is an effective antiarrhythmic agent, it may also be proarrhythmic, particularly in patients at risk for VT. We recommend careful cardiac assessment, risk stratification, and consideration of high-risk patients for early screening electrophysiological studies, especially if considering use of a class 1 antiarrhythmic agent.

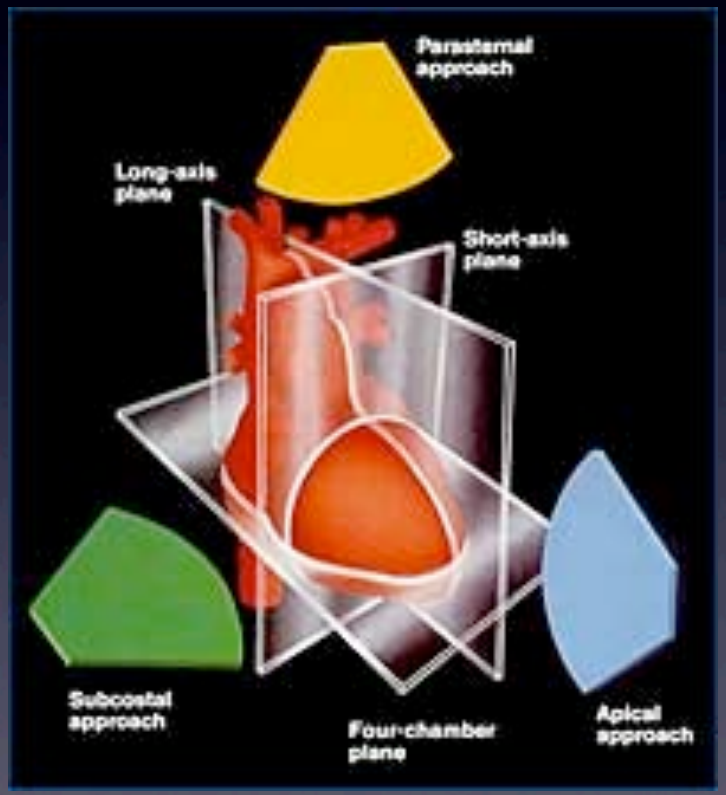
Type 1C - sodium channel blocker

Physics of echocardiography



- Ultrasound = sound waves like audible sound
- Audible sound 15-20 kilohertz (15-20,000 cycles/second)
- Medical ultrasound 1-12 megahertz (1-12,000,000 cycles/second)

Echo Imaging



- **“Planes” of sound cut through the heart to provide slices of anatomy**
- **Wavelengths, less than a millimeter, are capable of resolving fine anatomic structures**

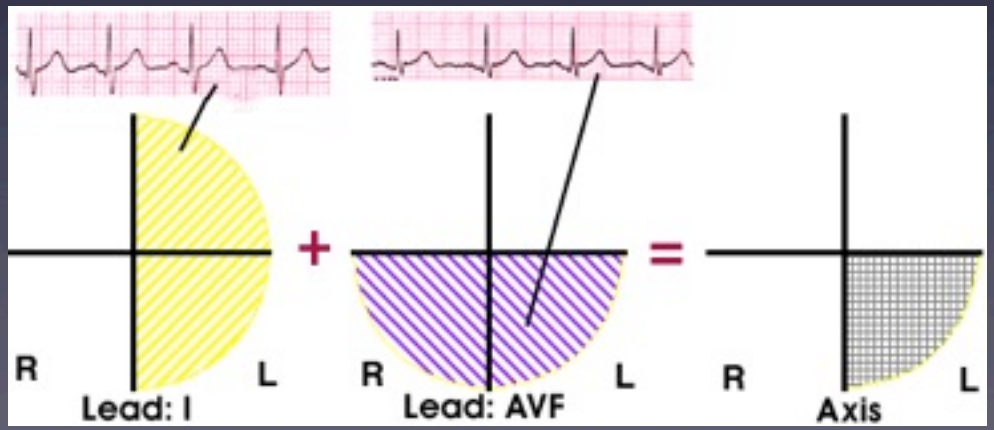
Basic Principles:EKG

- EKG elements
- Rate
- Axes
- Rhythm
- Intervals
- forces

Rhythm

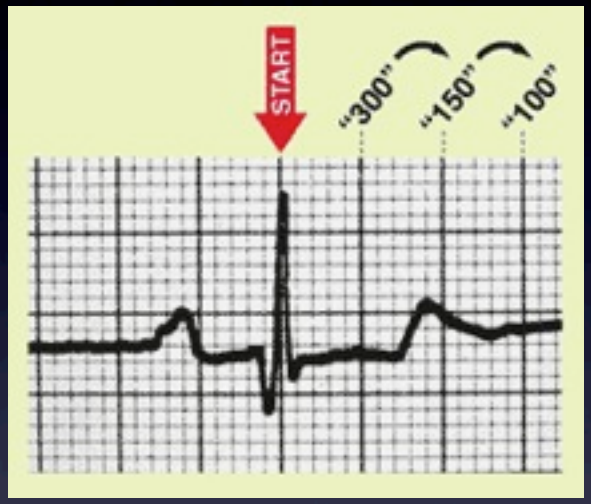
- Sinus rhythm requires

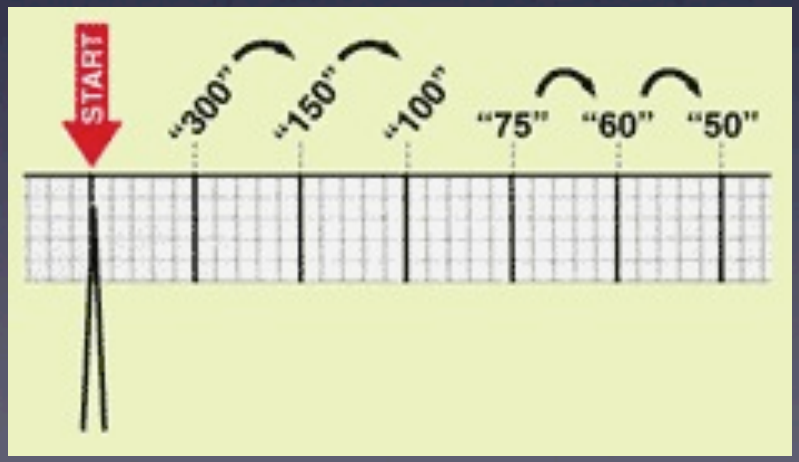
3) Appropriate P wave axis

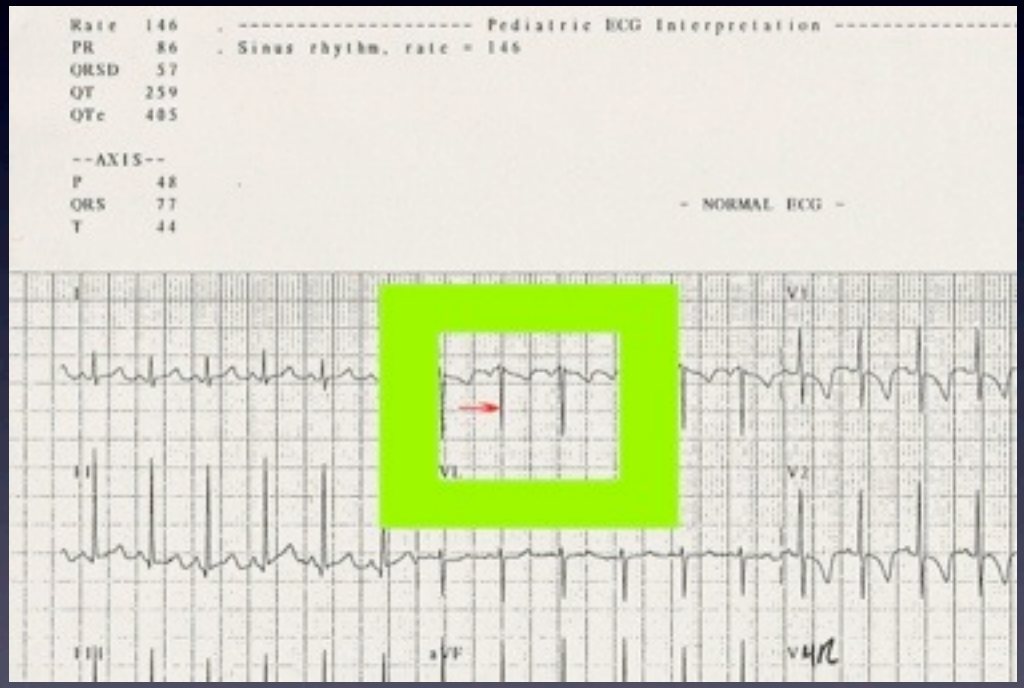


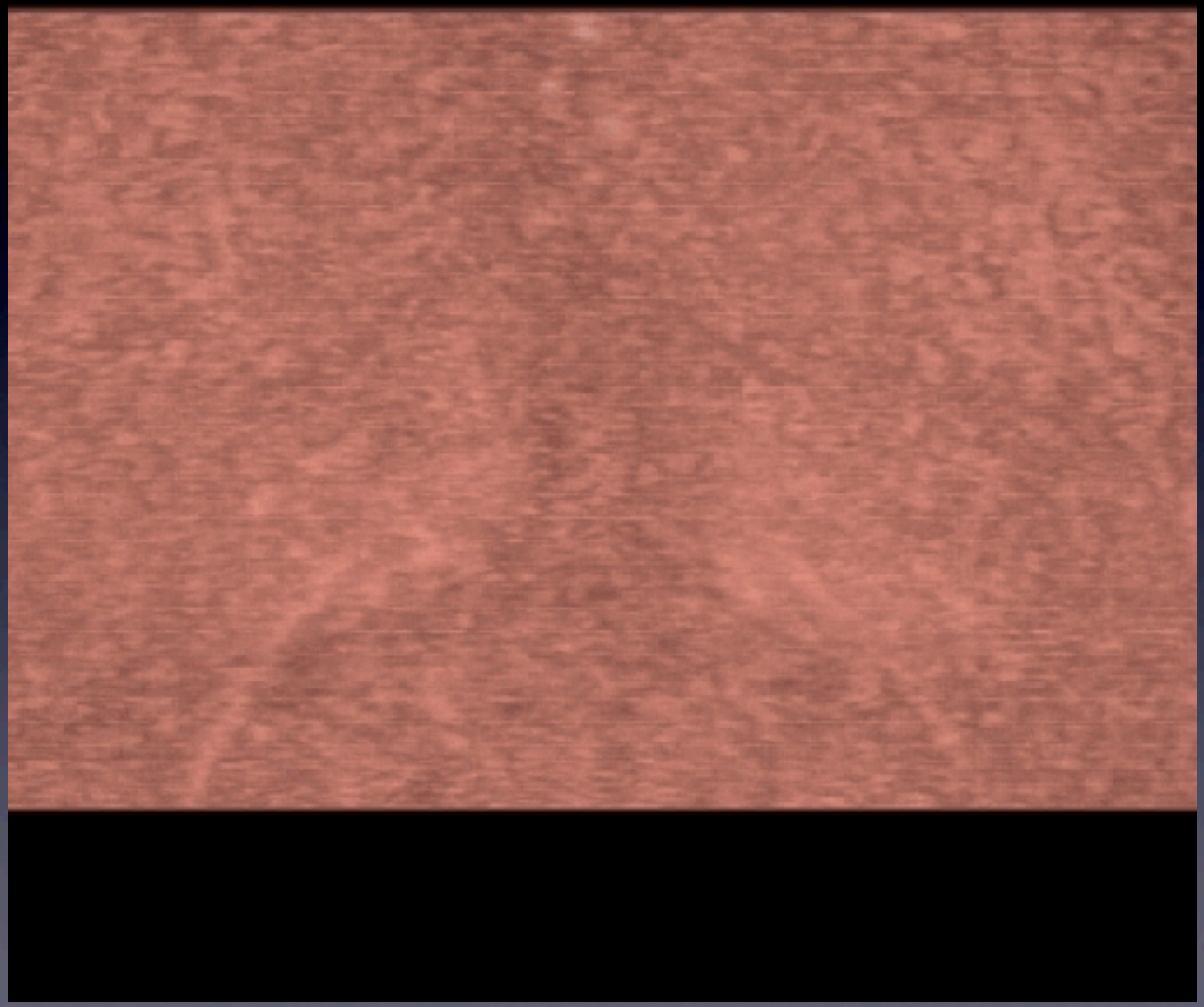
Sinus axis

Rate


START
"300"
"150"
"100"

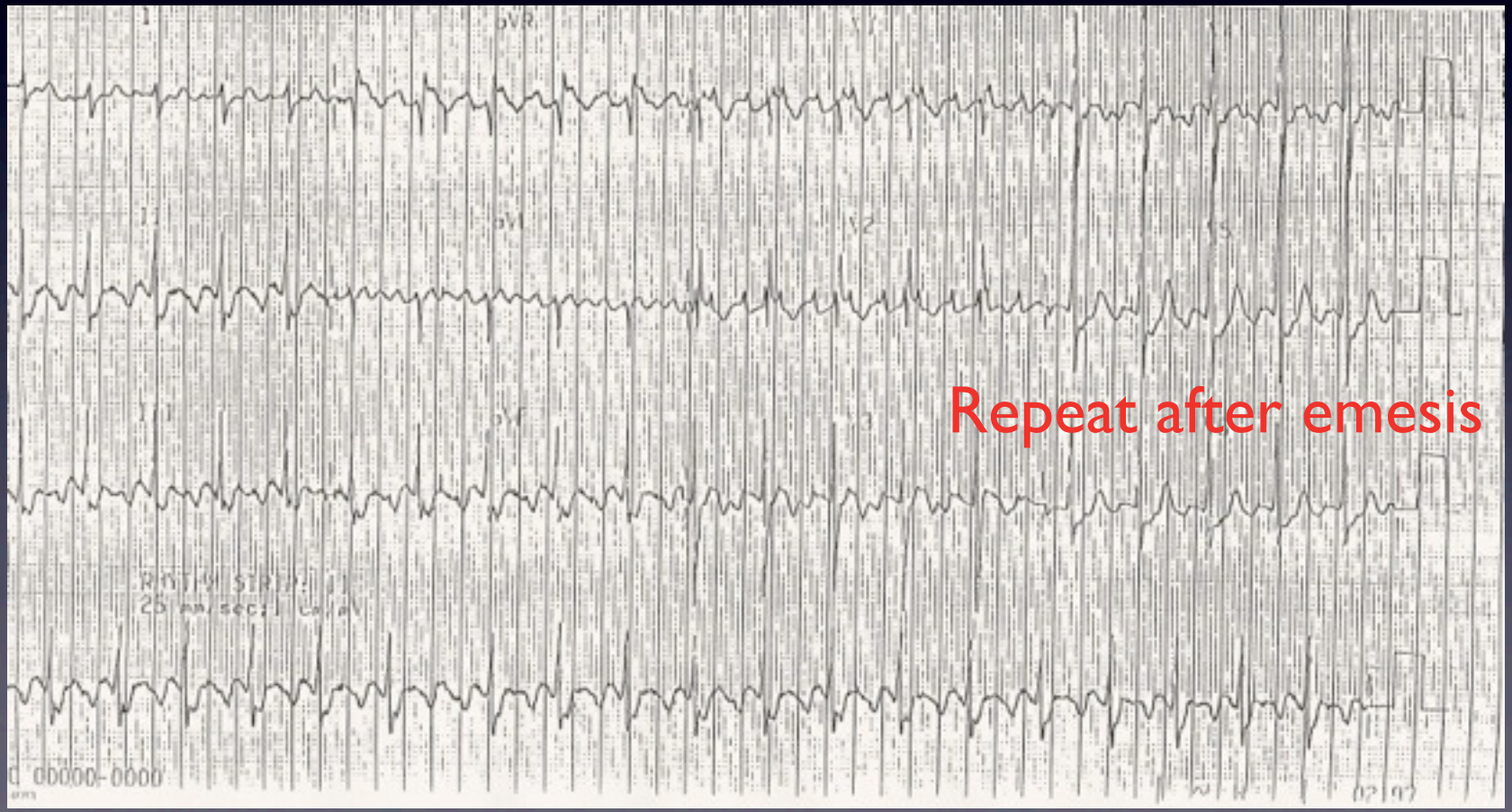

START
"300"
"150"
"100"
"75"
"60"
"50"


Rate 146
PR 86
QRSD 57
QT 259
QTc 405
--AXIS--
P 48
QRS 77
T 44
Pediatric ECG Interpretation
Sinus rhythm, rate = 146
- NORMAL ECG -
I
II
III
aVF
V1
V2
V4R



Fast Cardiac Rhythms

- **atrial flutter with 2:1 block**



Slow Cardiac rhythms

- **Complete heart block**