BSHG Brussels 2025 Cardiology concepts for geneticists

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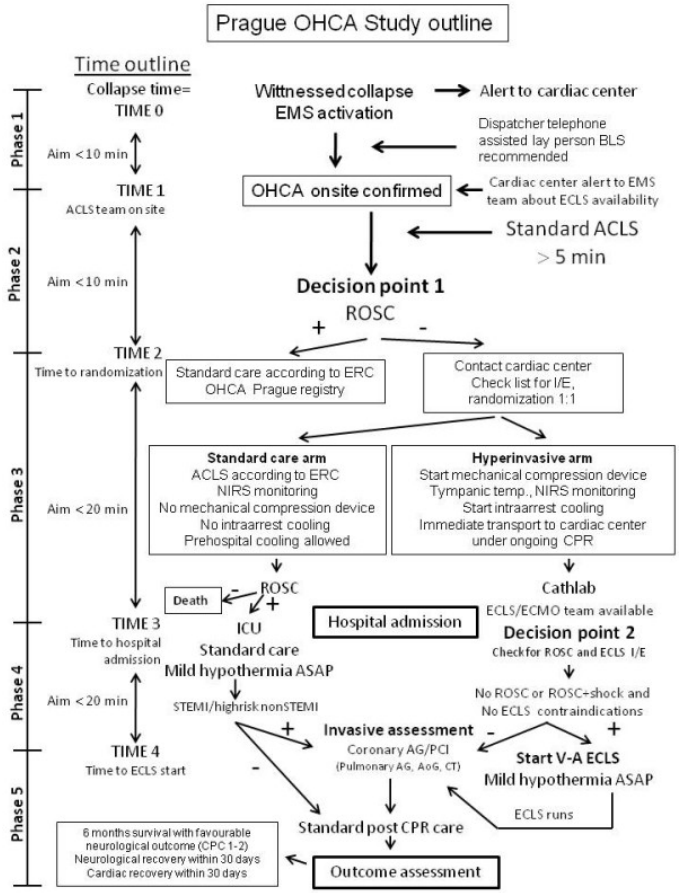
Barriers to overcome when working together with a cardiologist

Problem 1

Doctors (and cardiologists in particular) LOVE abbreviations

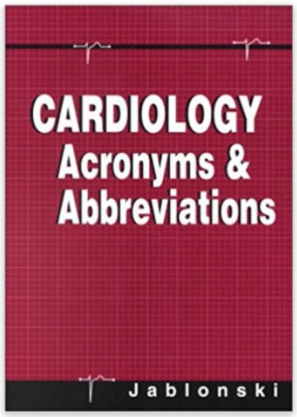


Only 17 Abbreviations: ACLS: advanced cardiac life support; AG: angiography; ASAP: as soon as possible; BLS: basic life support; CPC: cerebral performance category; CPR: cardiopulmonary resuscitation; CT: computed tomography; ECLS: extracorporeal life support; EMS: emergency medical service; ERC: European Resuscitation Council; ICU: intensive care unit; I/E: inclusion/exclusion; NIRS: near infrared spectroscopy; OHCA: out of hospital cardiac arrest; ROSC: return of spontaneous circulation; STEMI: ST elevation acute myocardial infarction; TTE: transthoracic echocardiography

Problem 1

Doctors (and cardiologists in particular) LOVE abbreviations

Filling BOOKS with abbreviations



Cardiology Acronyms & Abbreviations Paperback – 28 Sep 2001

by Stanley Jablonski (Author)

1 review from the U.S.

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This spinoff from the Dictionary of Medical Acronyms & Abbreviations, 4th ed. focuses entirely on cardiology terminology. It consists entirely of terms, tests, and clinical trials related to cardiovascular medicine. Indispensable reference for all health care professionals, and other users of medical information related to cardiology.

Problem 2

SCD is the most important abbreviation in life & in cardiology

Problem 2

SCD is the most important abbreviation in life & in cardiology

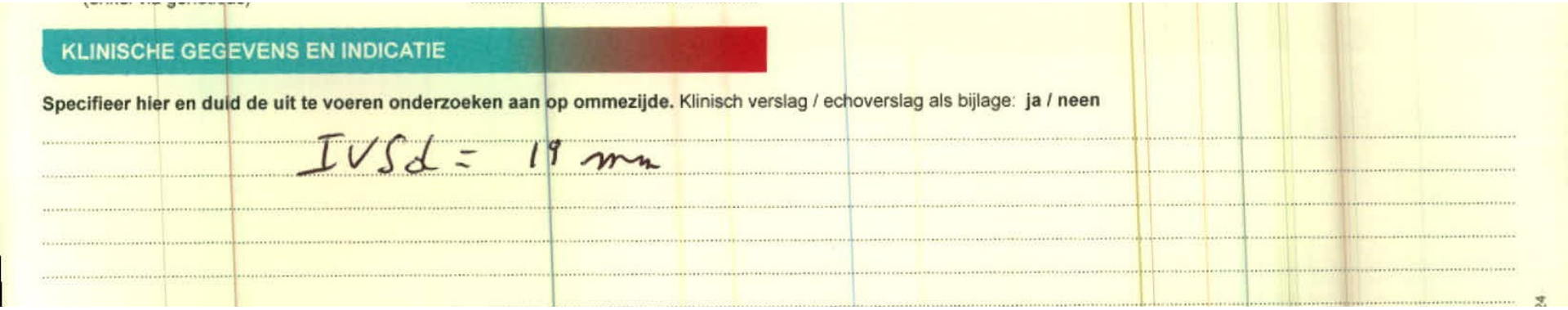
…and it is hereditary in up to 80% of the young making it your problem too !

Problem 3

Doctors (and cardiologists in particular) WRITE as little as possible

Clinical data are missing, phenotype insufficiently described

Complicates the interpretation of genetic variants

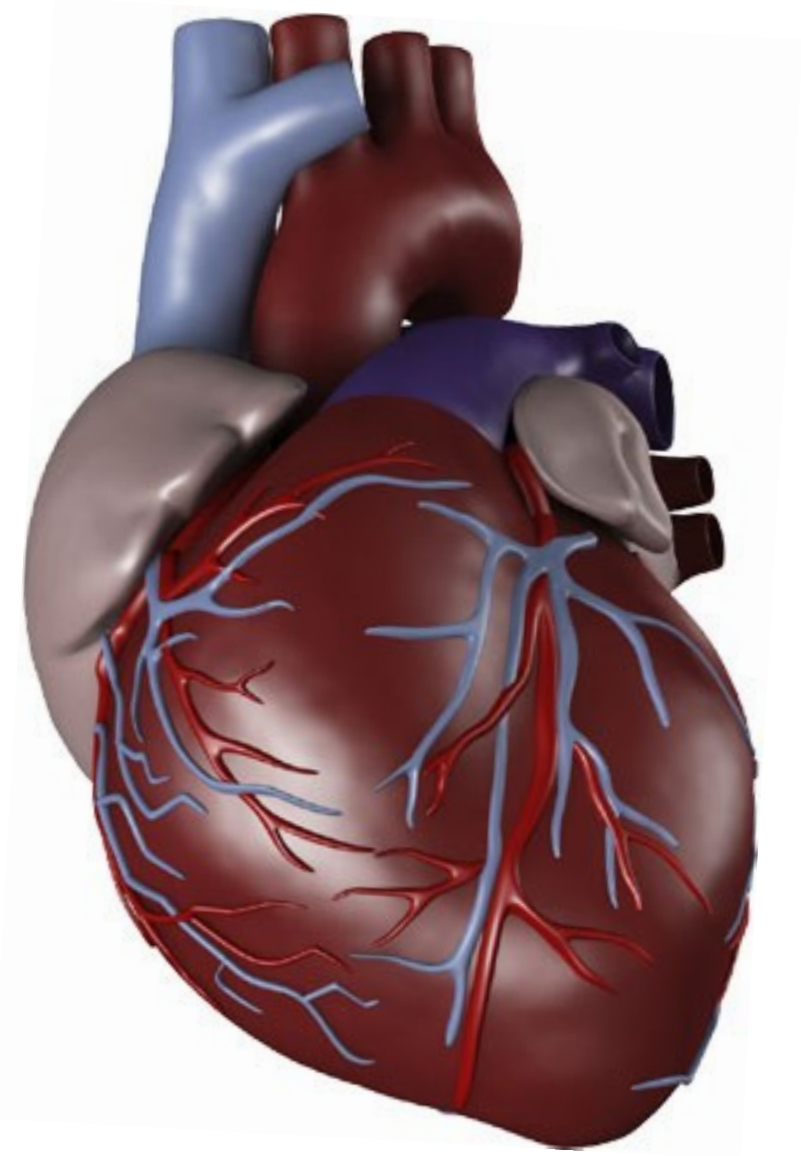


Bringing both worlds together

1. Anatomy of the heart
2. Normal function of the heart
3. Introduction to cardiac diseases
4. Strutural heart disease
5. Electrica heart disease

1.Anatomy of the heart

Anatomy of the heart



The Heart

Motor

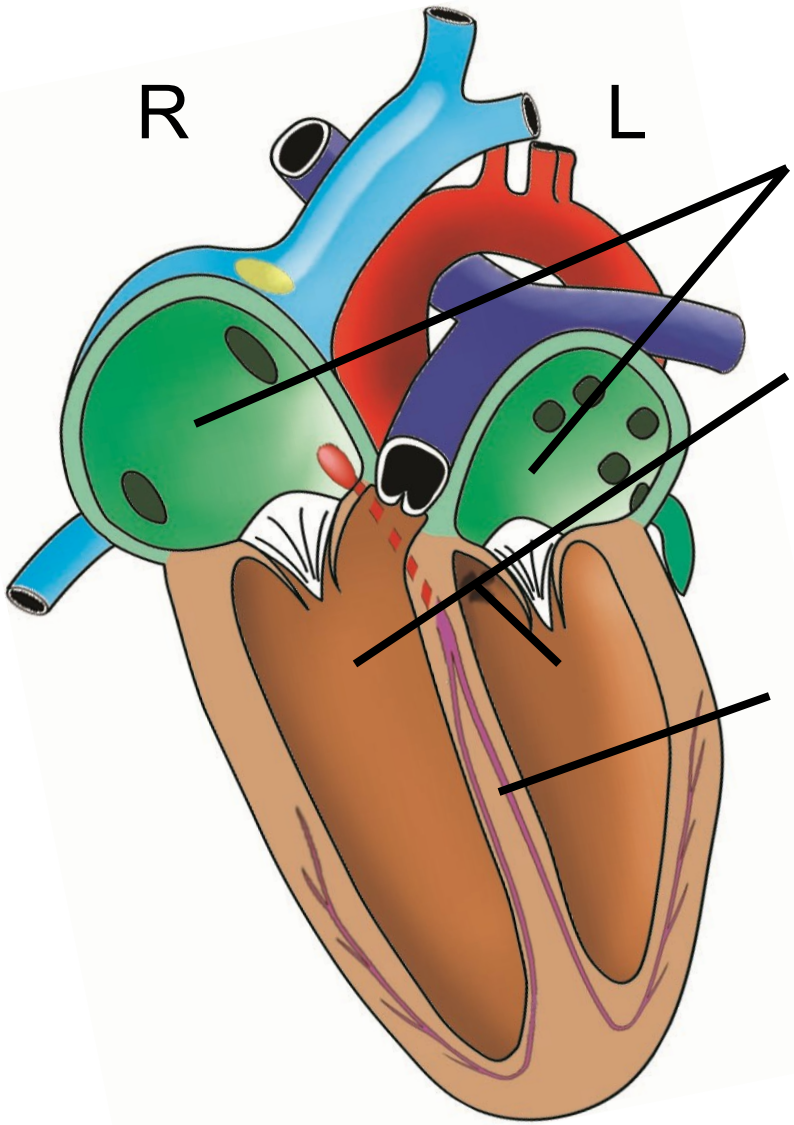
Muscular pump

100.000 beats/day

Organ perfusion

- O_2 to organs
- CO_2 to lungs

Anatomy of the heart



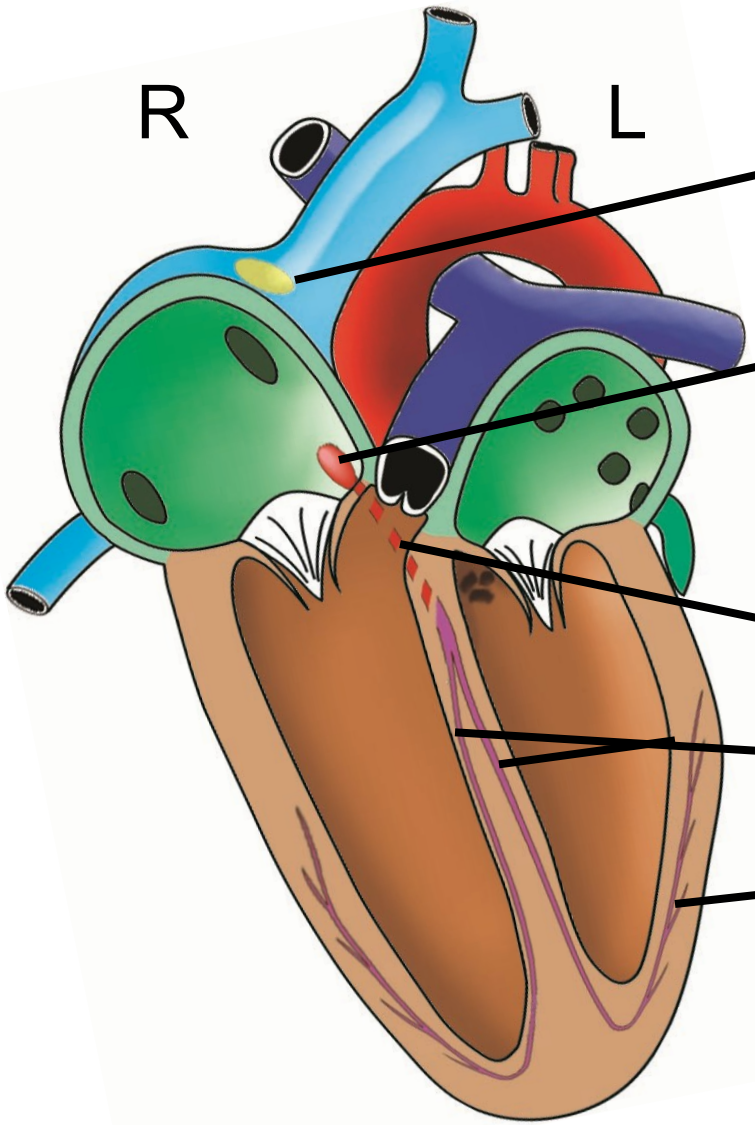
4 Chambers

Atria (RA & LA)

Ventricles (RV & LV)

Interventricular septum (IVS)

Anatomy of the heart



Conduction system

Sinus node (SN)

AV node (AVN)

His Purkinje system:

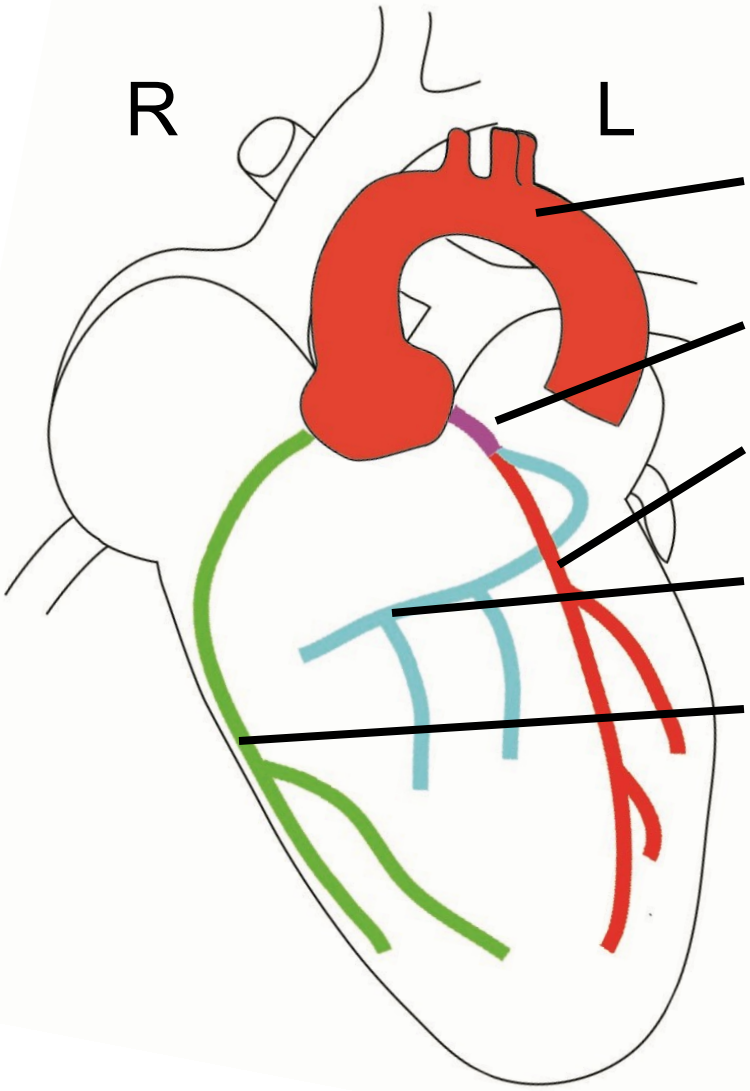
His Bundle

Bundle branches

Purkinje network

Anatomy of the heart

Coronary arteries



Aorta

Left Main Coronary Artery (LMCA)

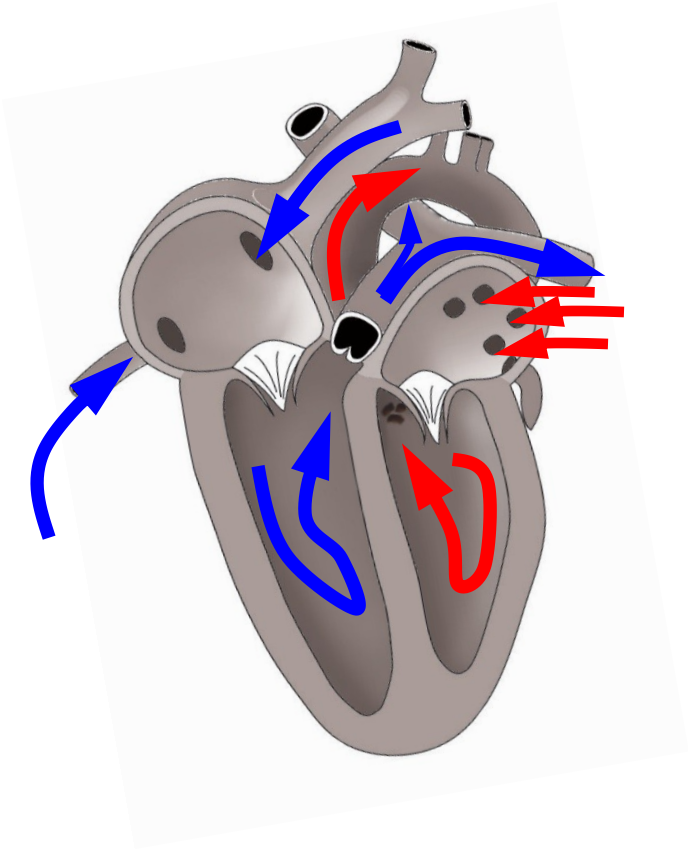
Left Anterior Descendent (LAD)

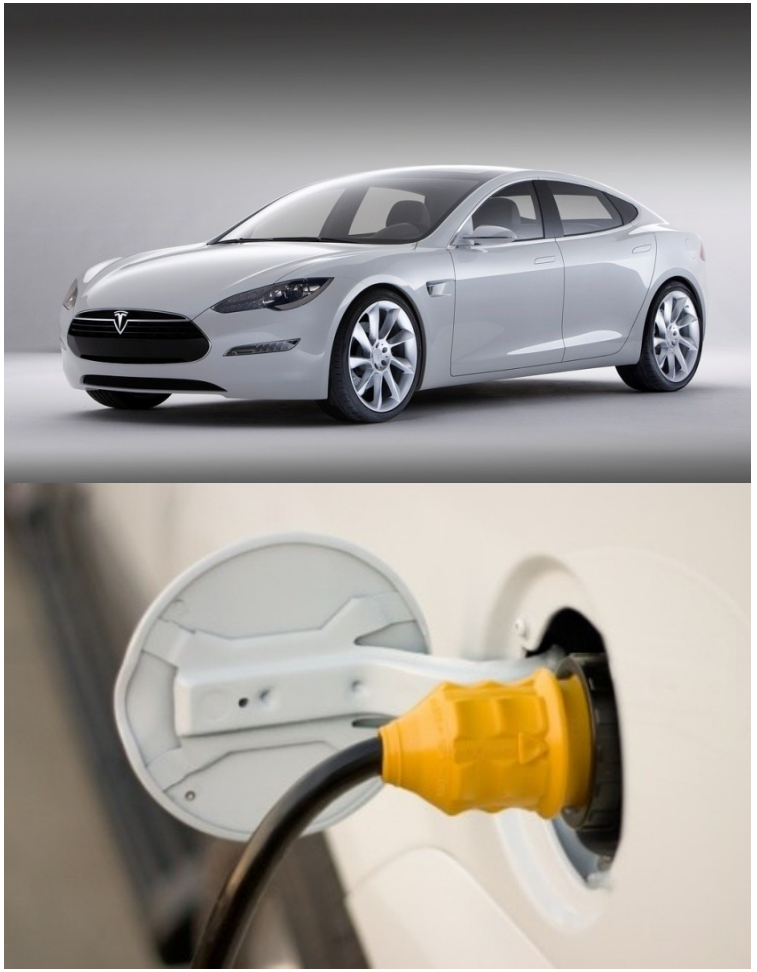
Ramus CircumfleXus (RCX)

Right Coronary Artery (RCA)

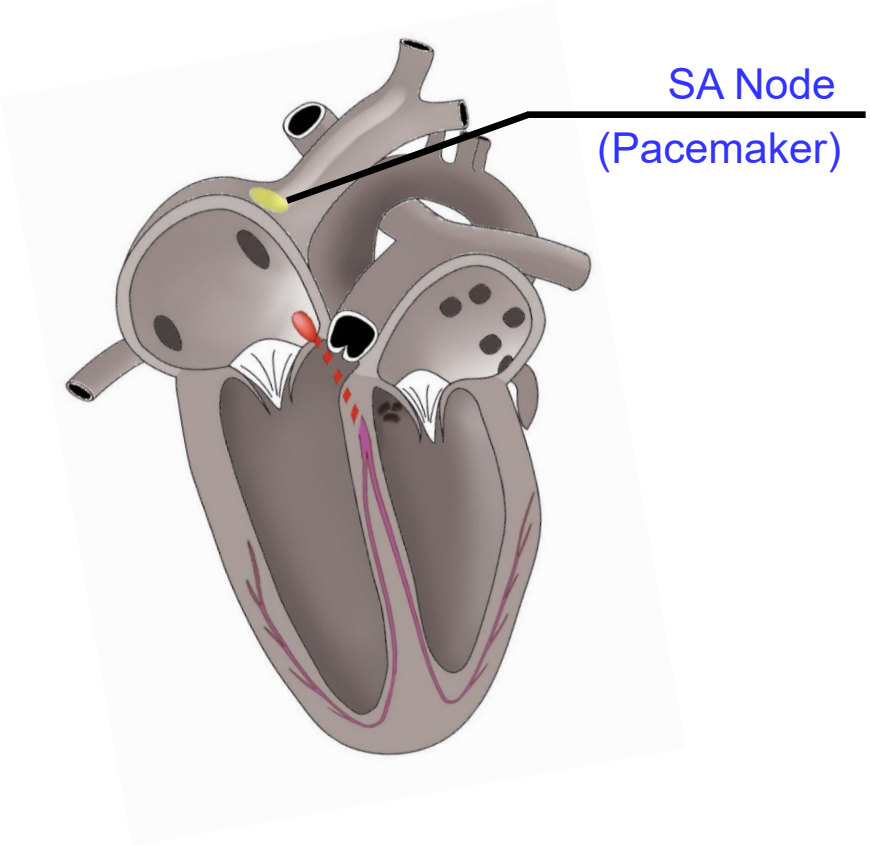
2. Normal function of the heart

Normal function of the heart





Normal function of the heart



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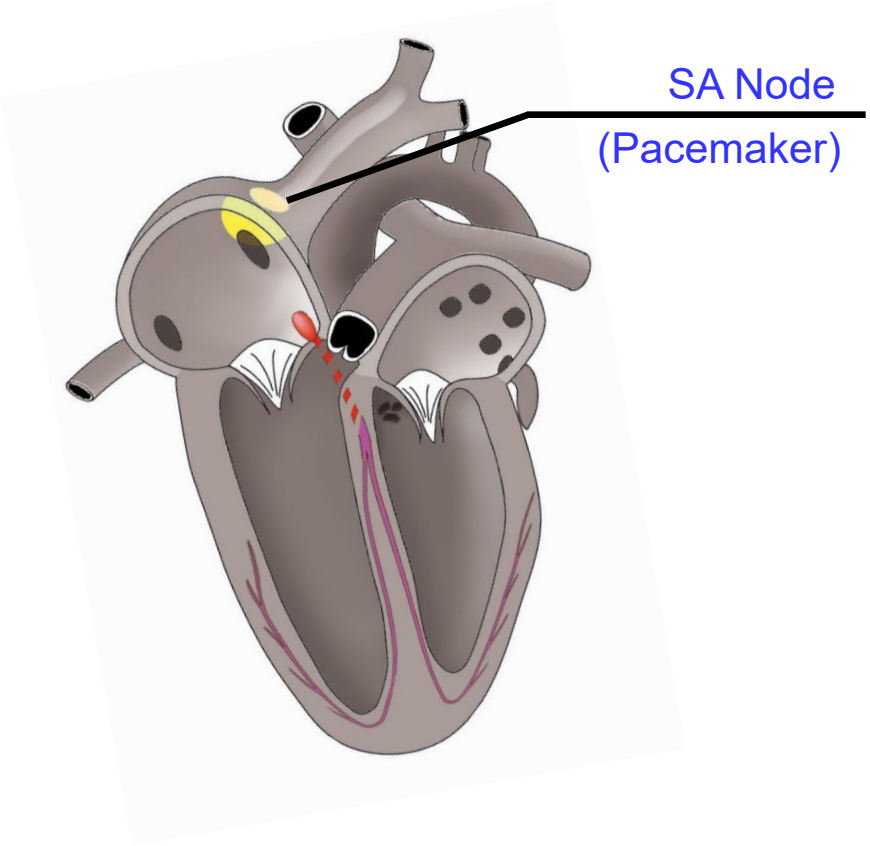

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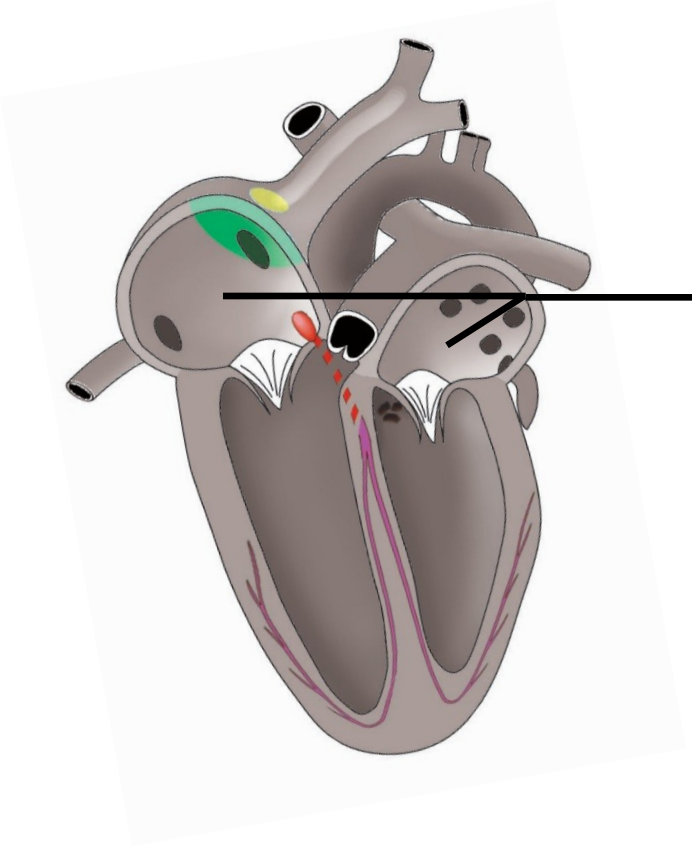
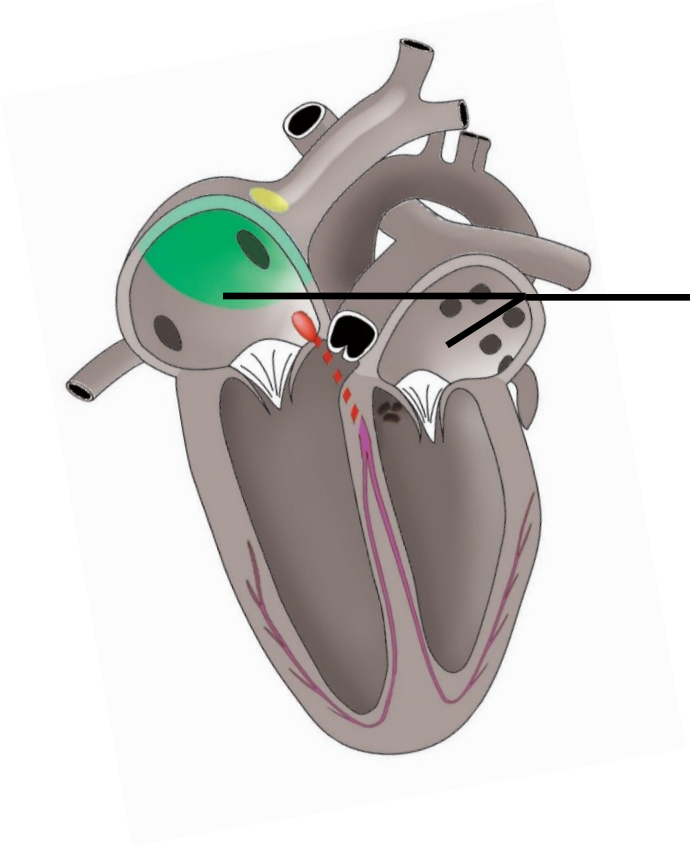


Muscle conduction velocity ~0.5m/s

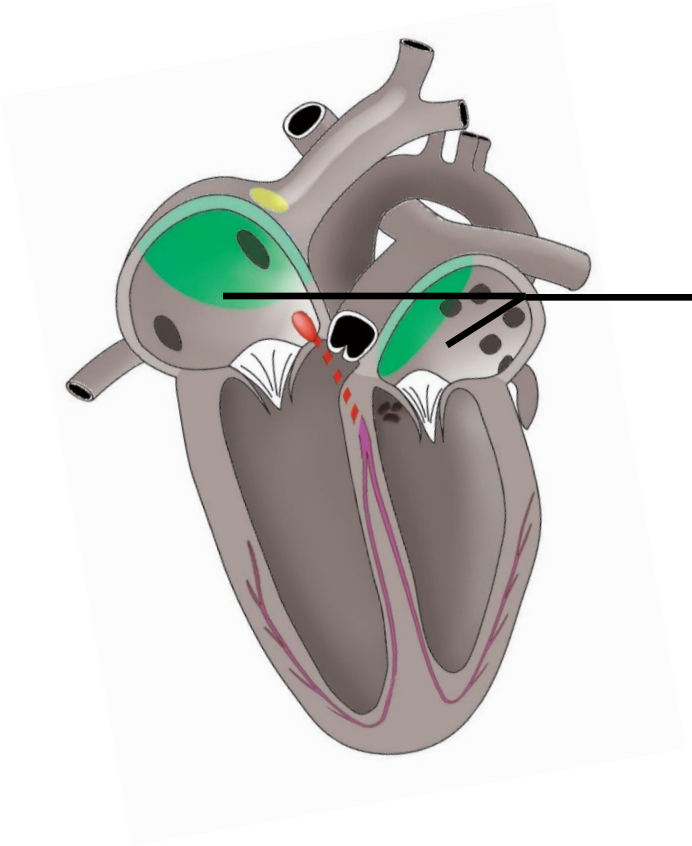
Normal function of the heart



Atrium

Muscle conduction velocity ~0.5m/s

Normal function of the heart



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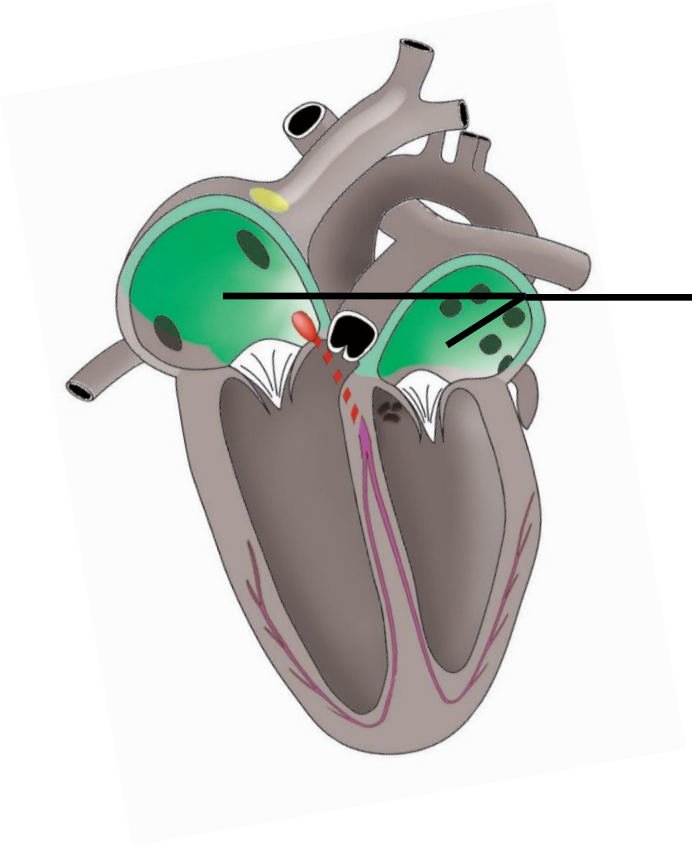

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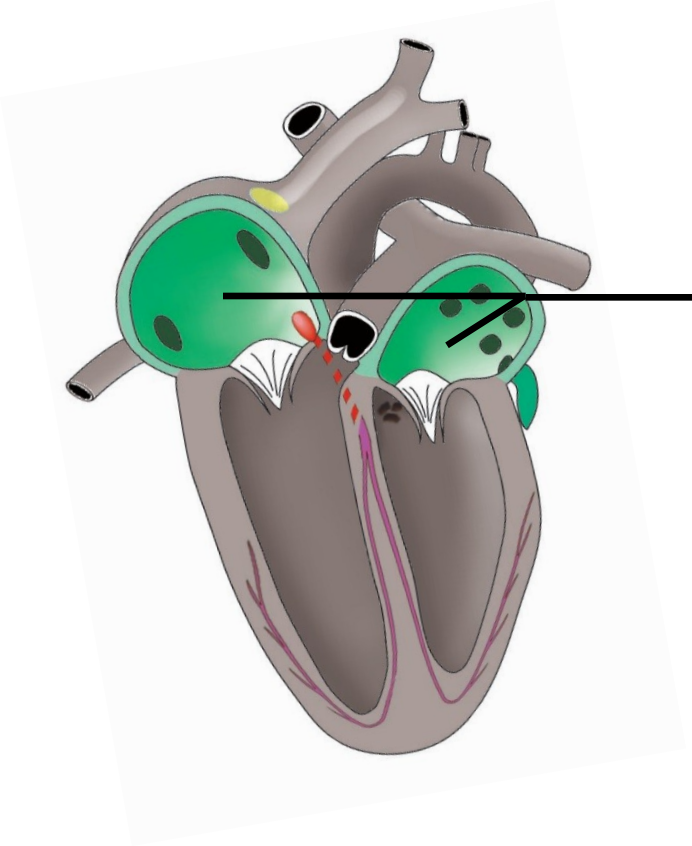

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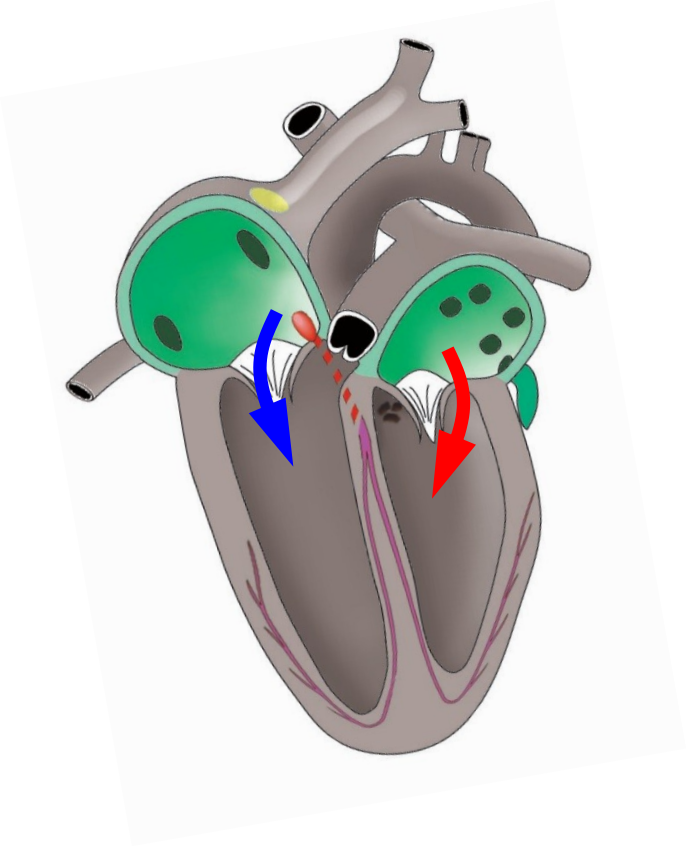

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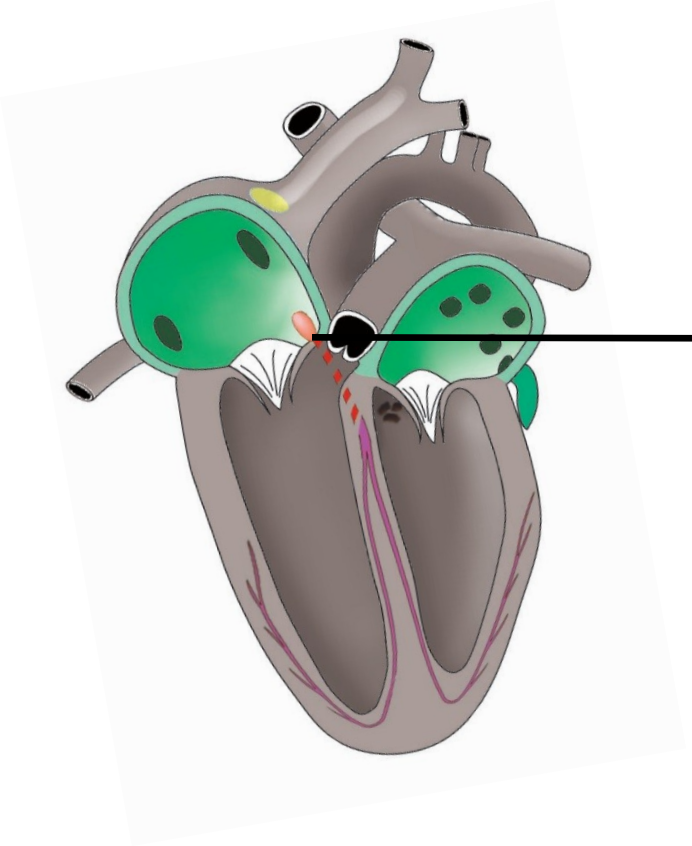
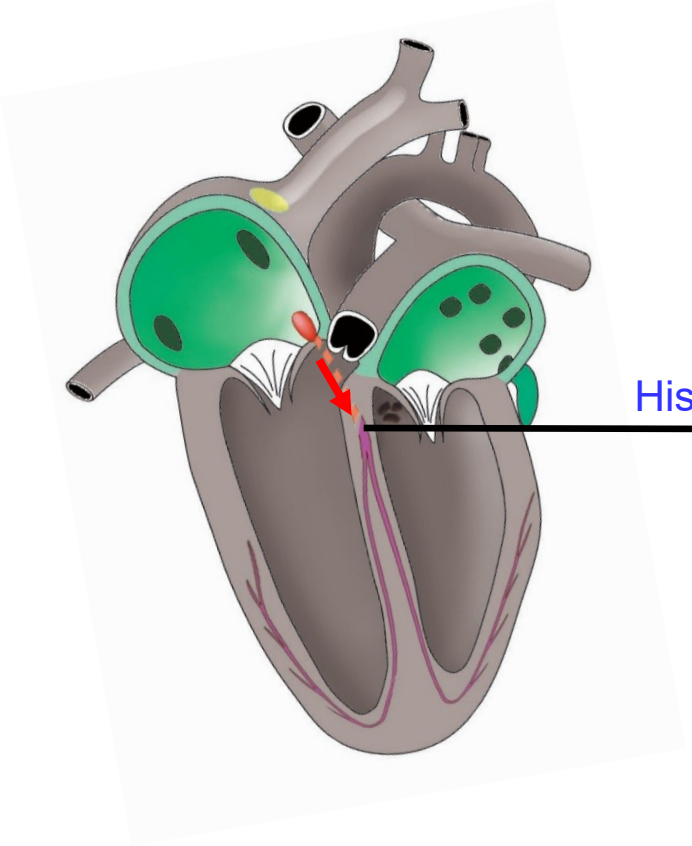


AV Node

Conduction slowing
~0.05m/s
= filter for excessive atrial activity

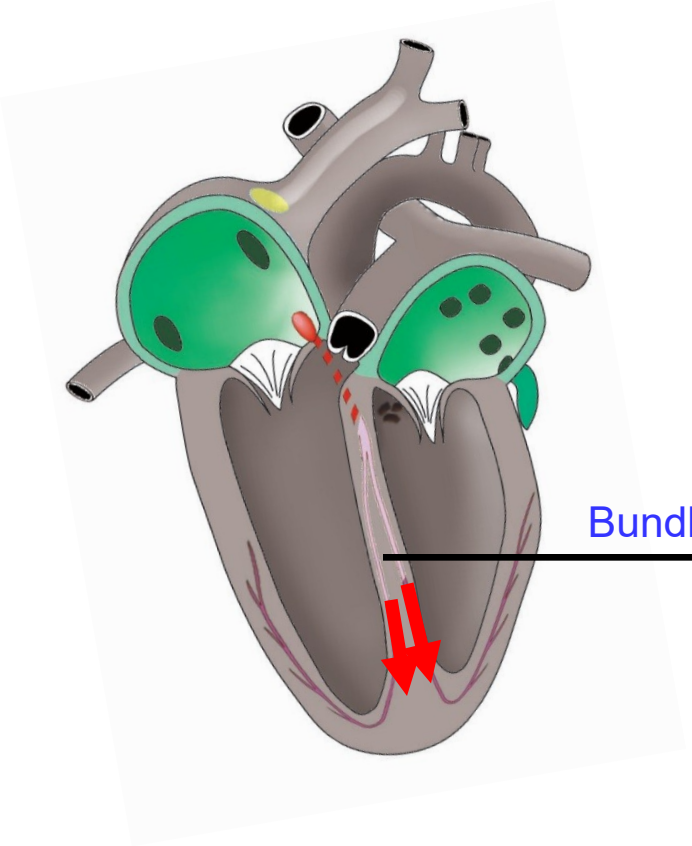
Normal function of the heart



His bundle

Fast conduction
~2m/s

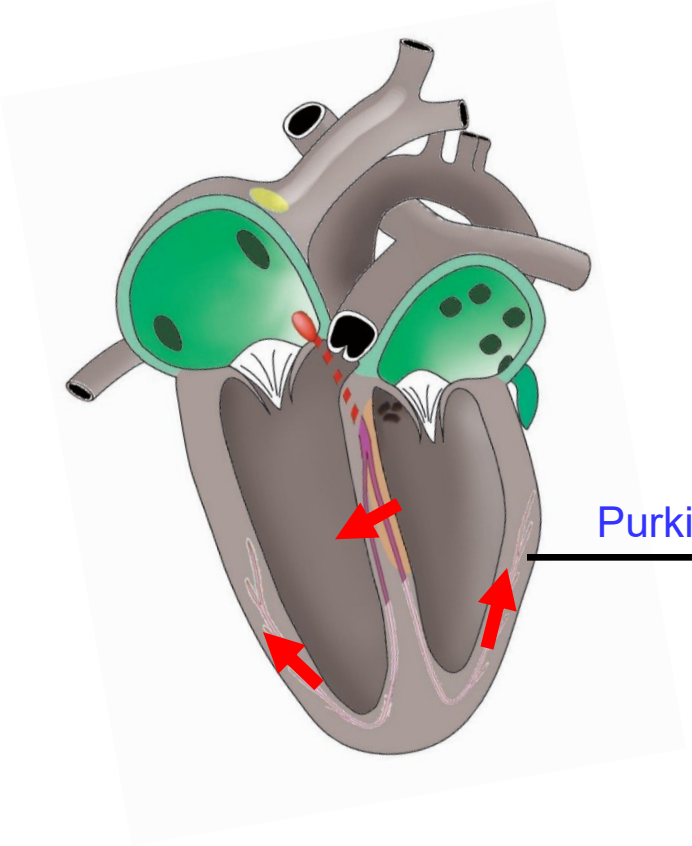
Normal function of the heart



Bundle branches

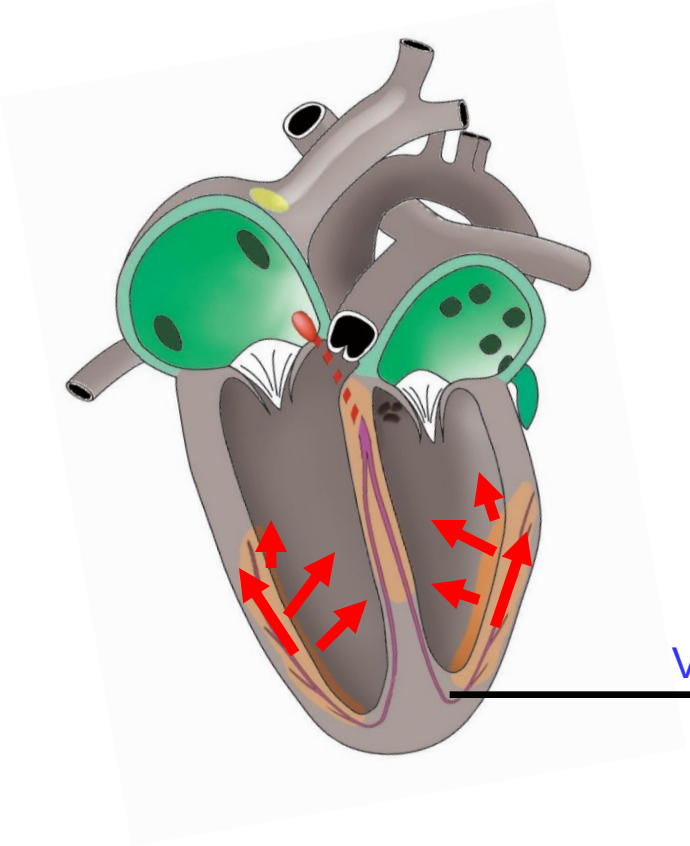
Fast conduction
~2m/s

Normal function of the heart



Purkinje Fibre — Fast conduction ~4m/s

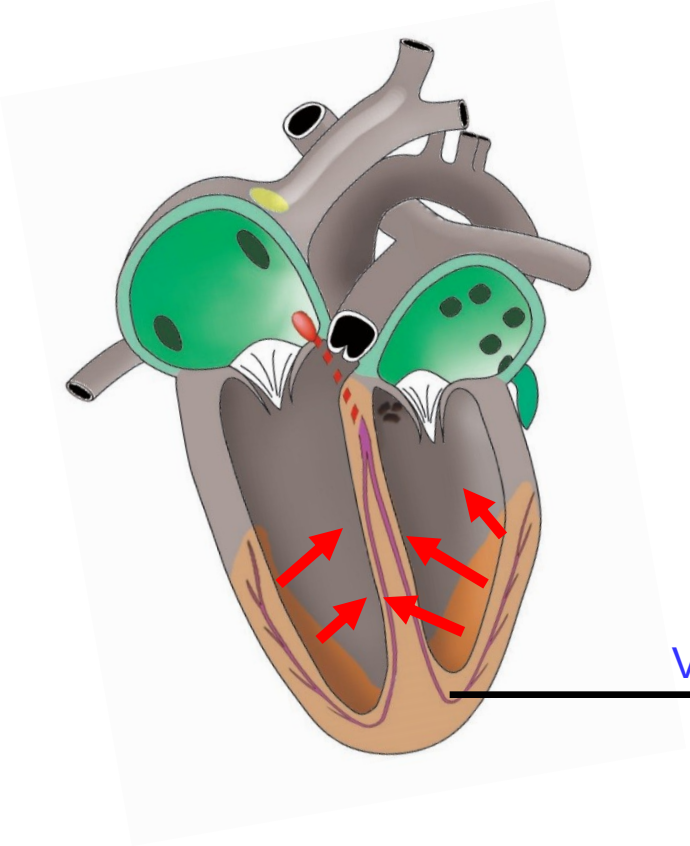
Normal function of the heart



Ventricle

Muscle conduction velocity ~0.5m/s

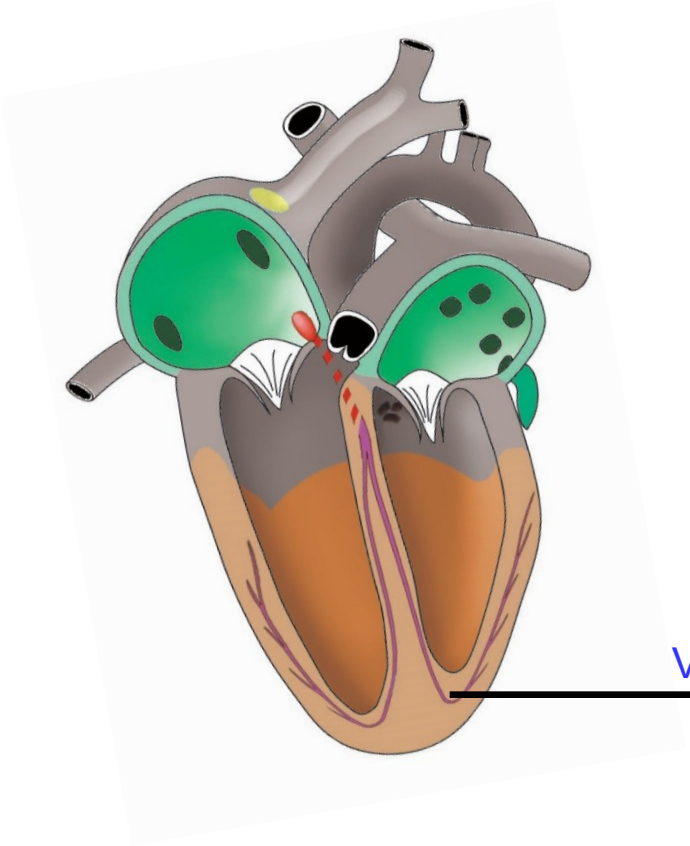
Normal function of the heart



Ventricle

Muscle conduction velocity ~0.5m/s

Normal function of the heart



Ventricle

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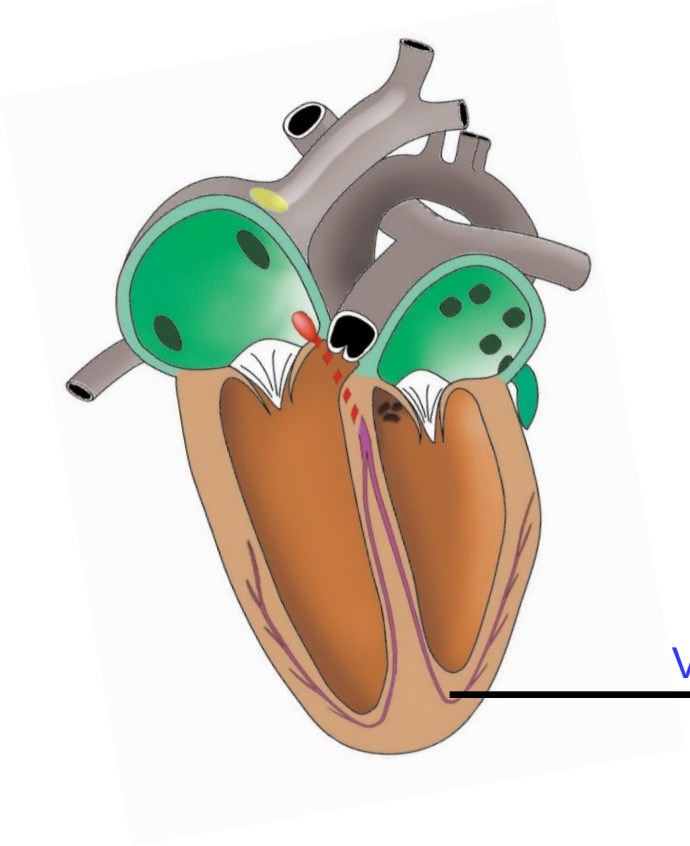
Normal function of the heart



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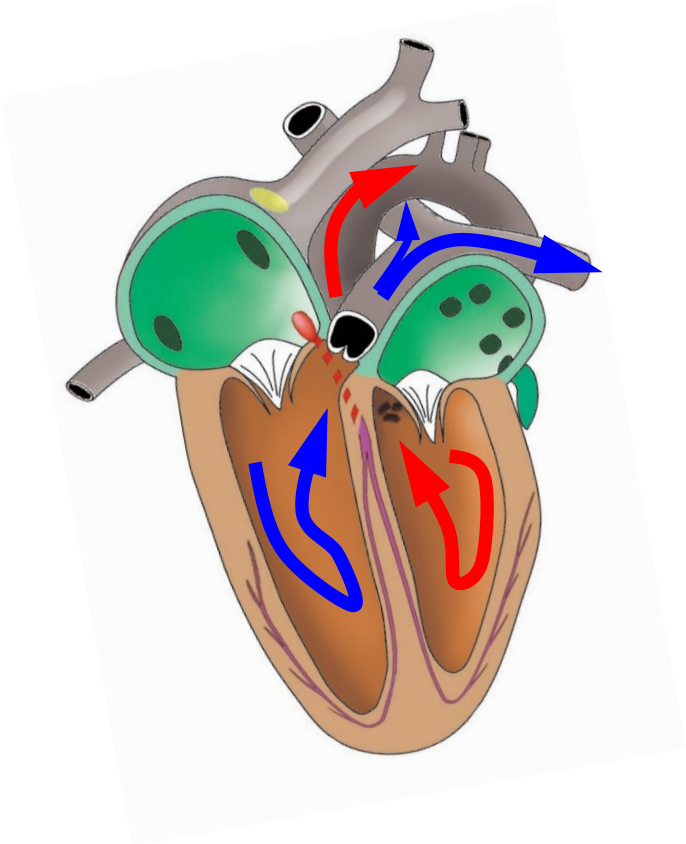
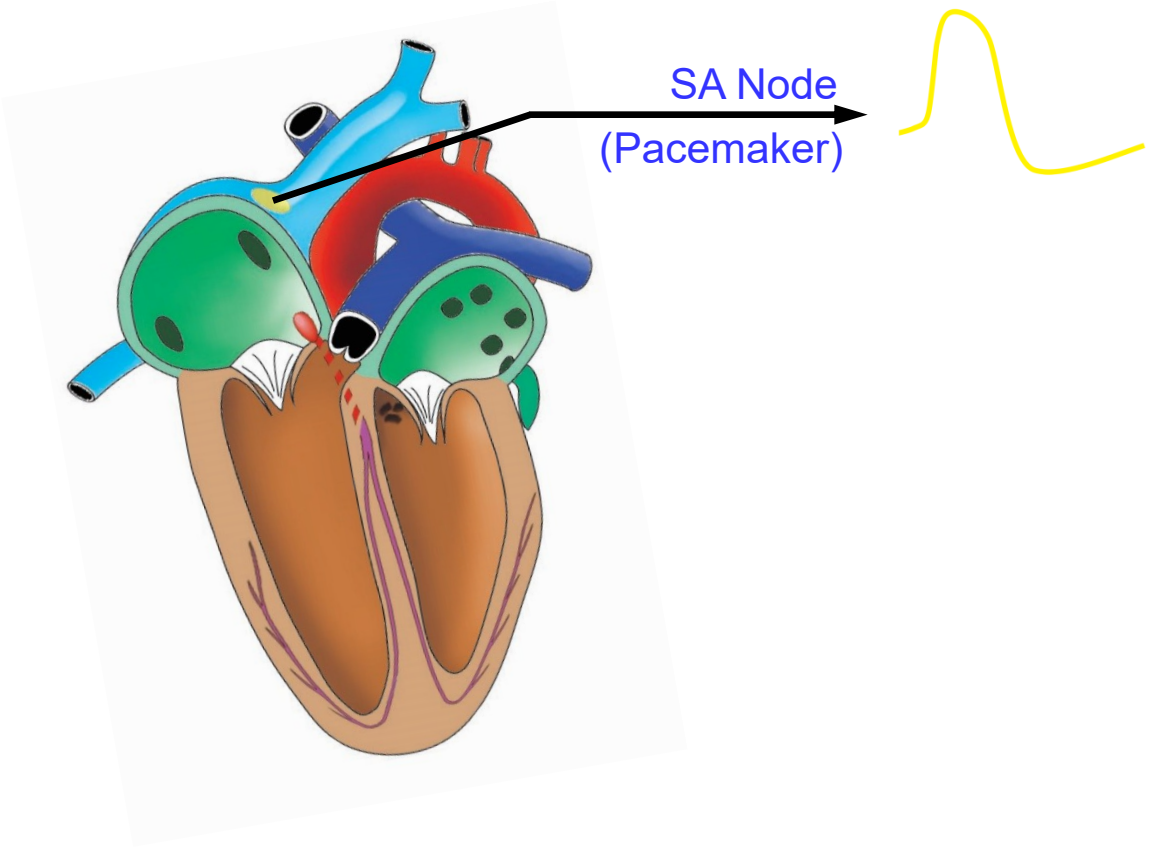


Electrical functioning of the heart

Action potential (AP)



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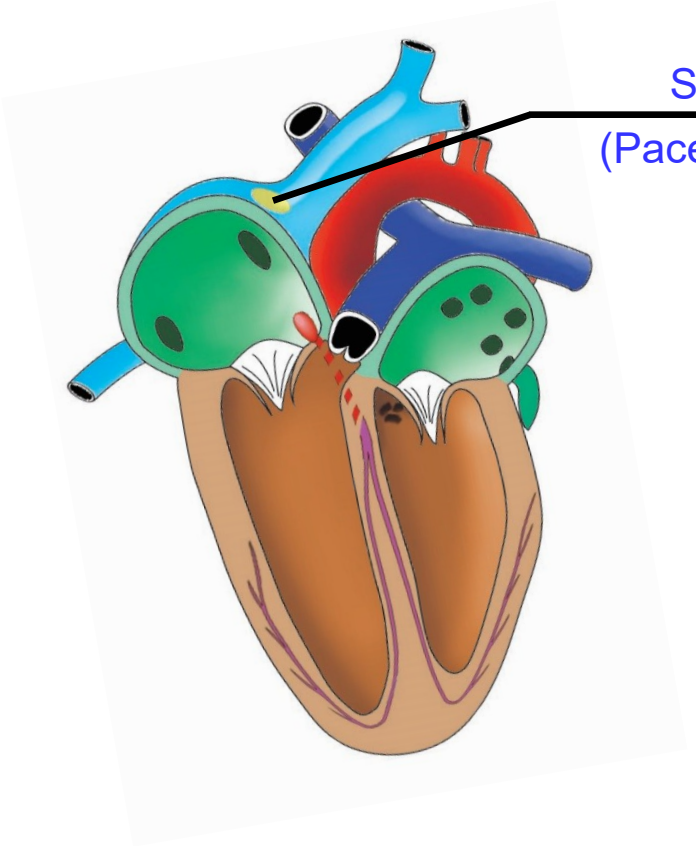
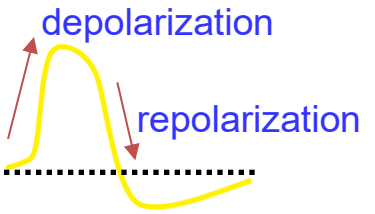


SA Node
(Pacemaker)

Action potential (AP)



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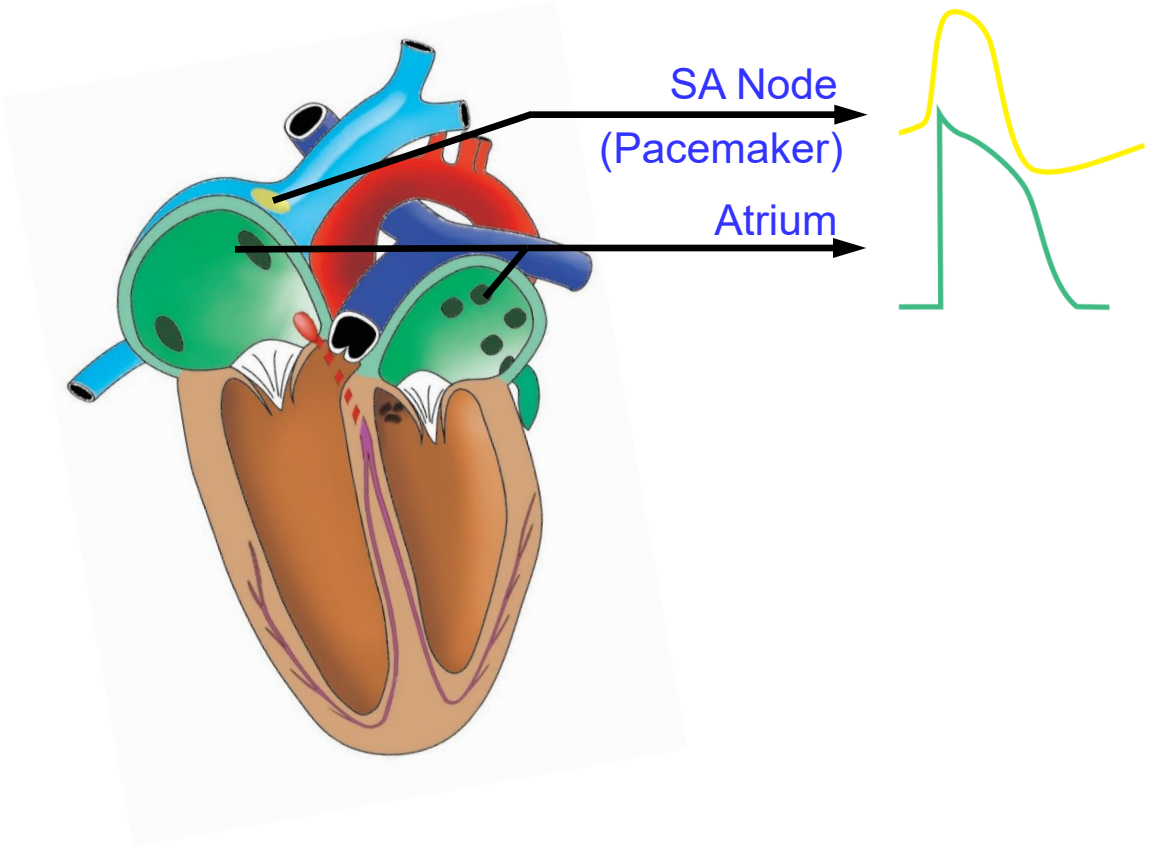

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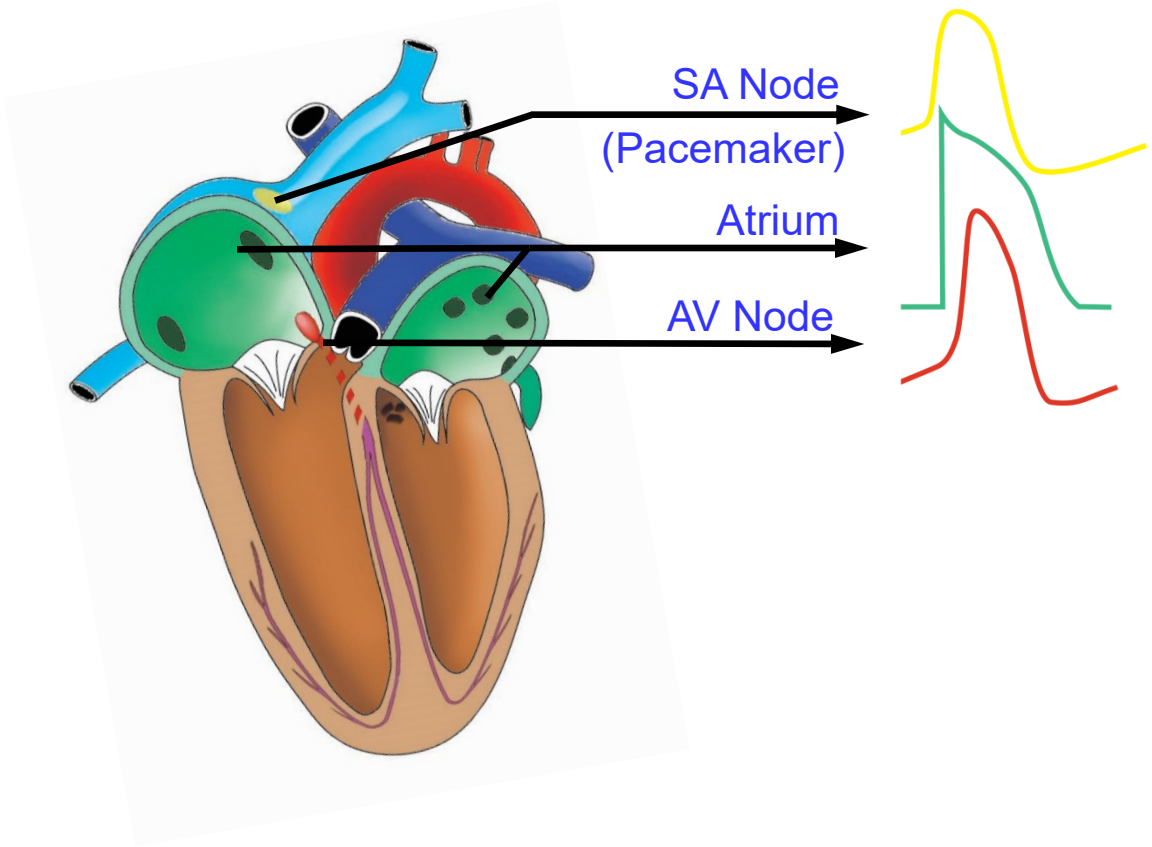

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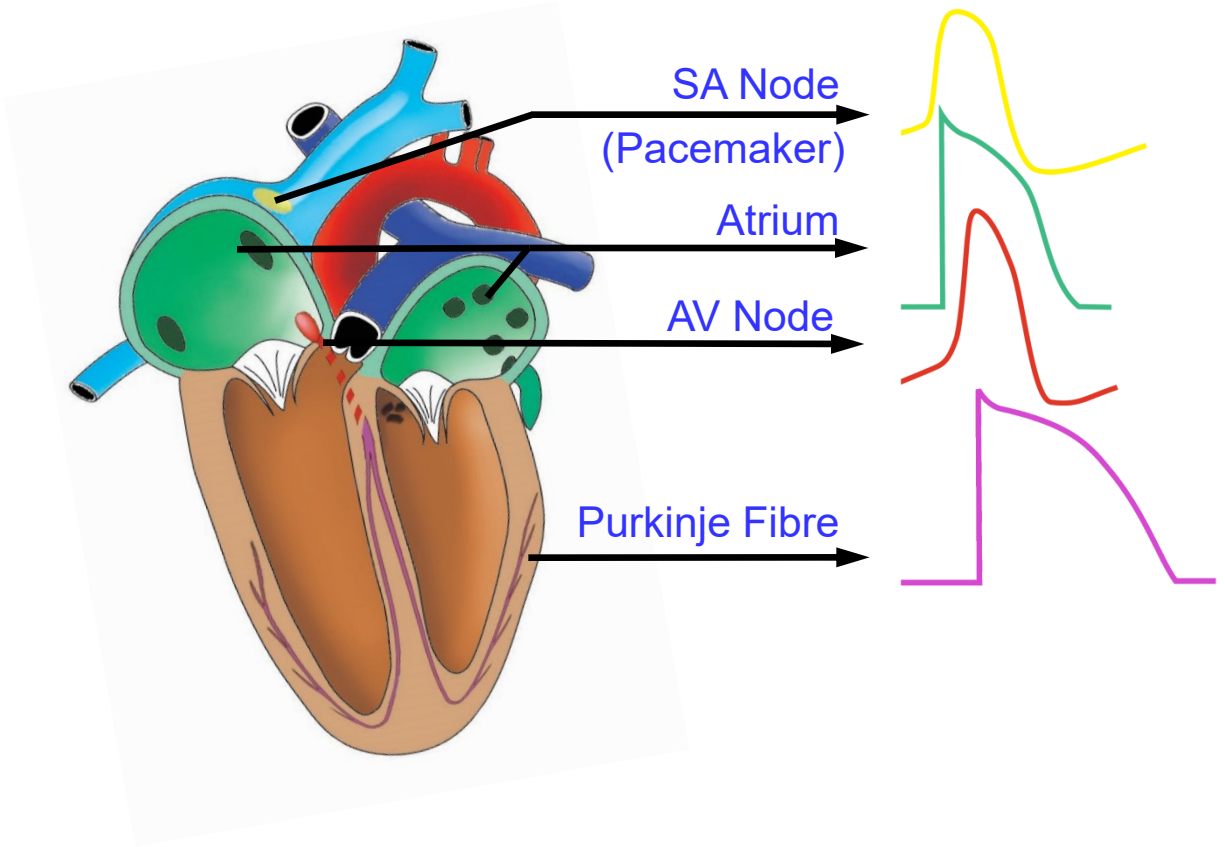

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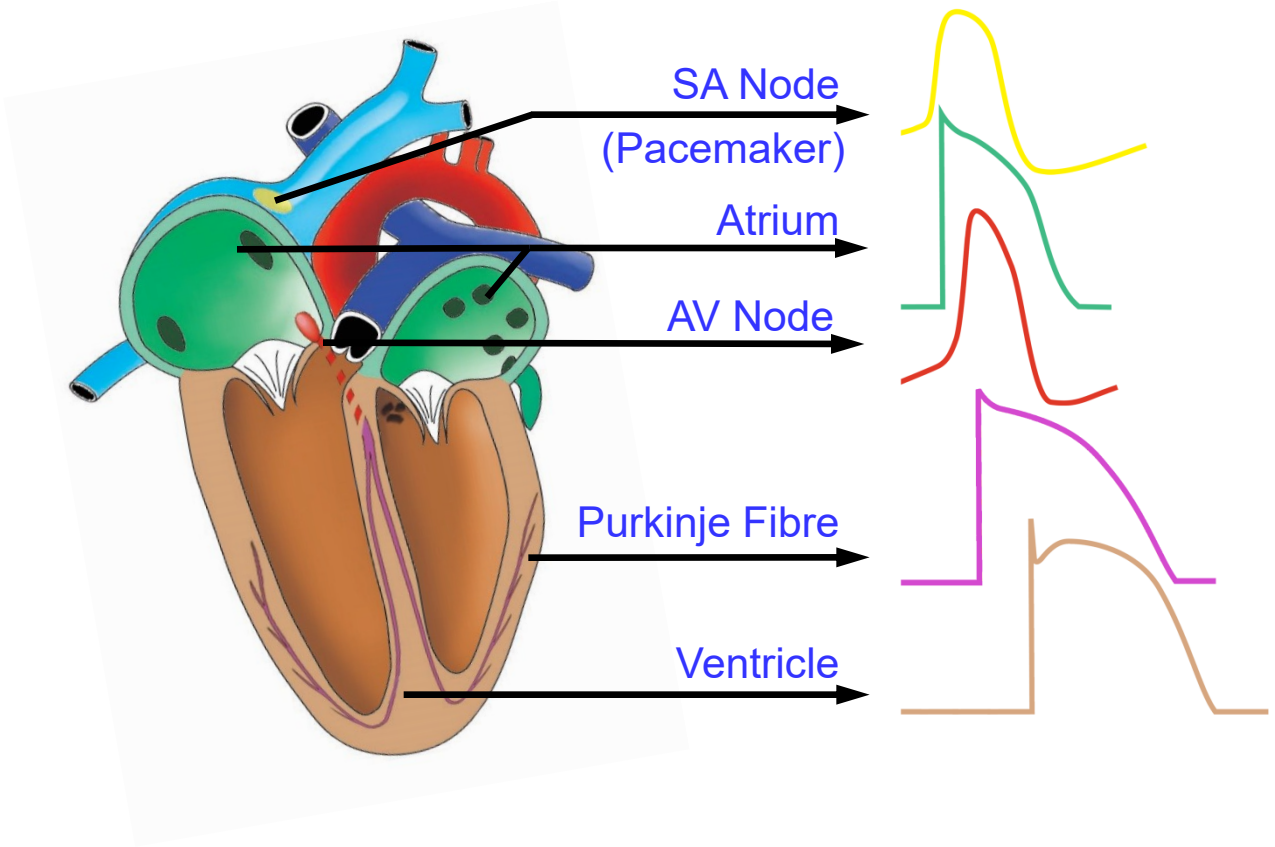

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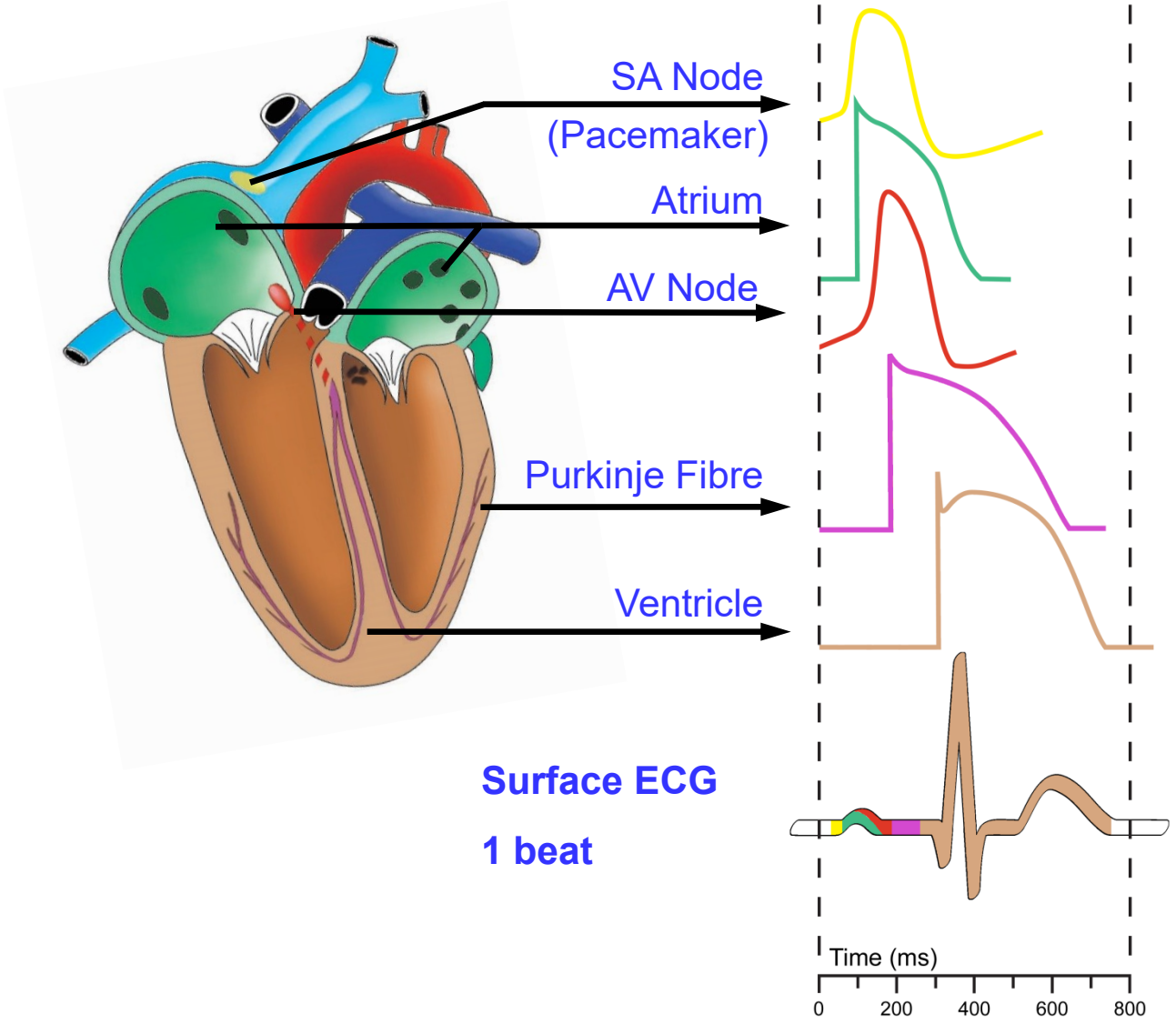

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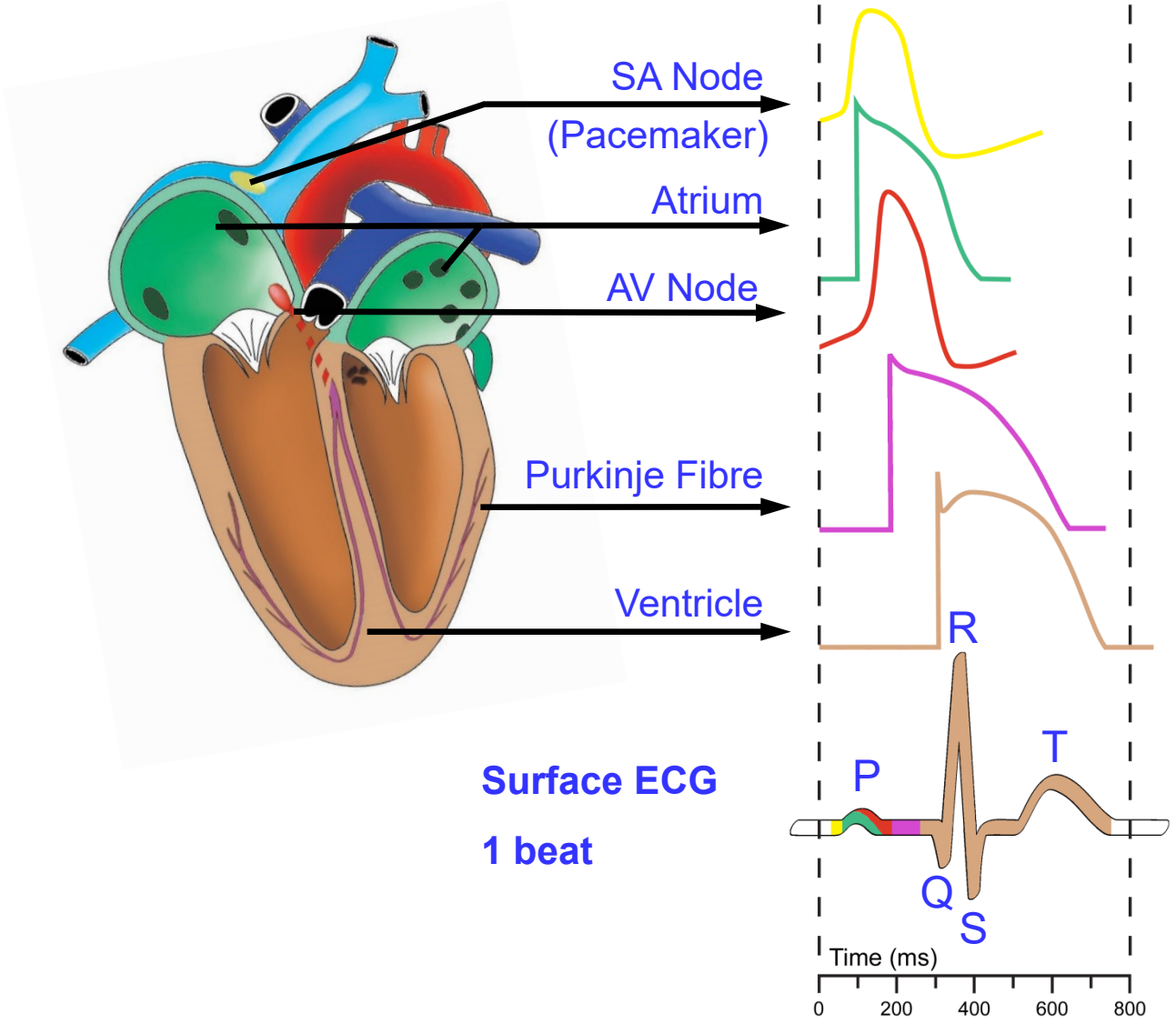

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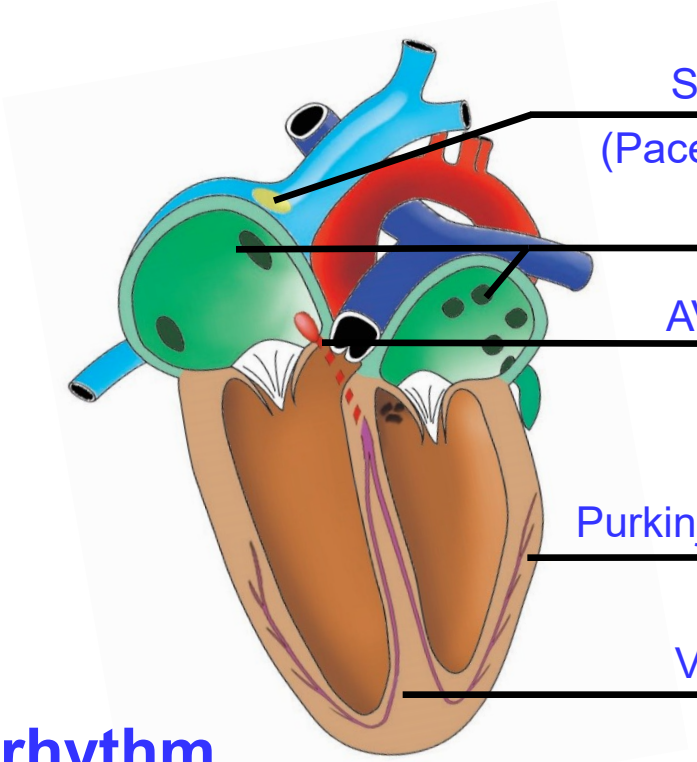
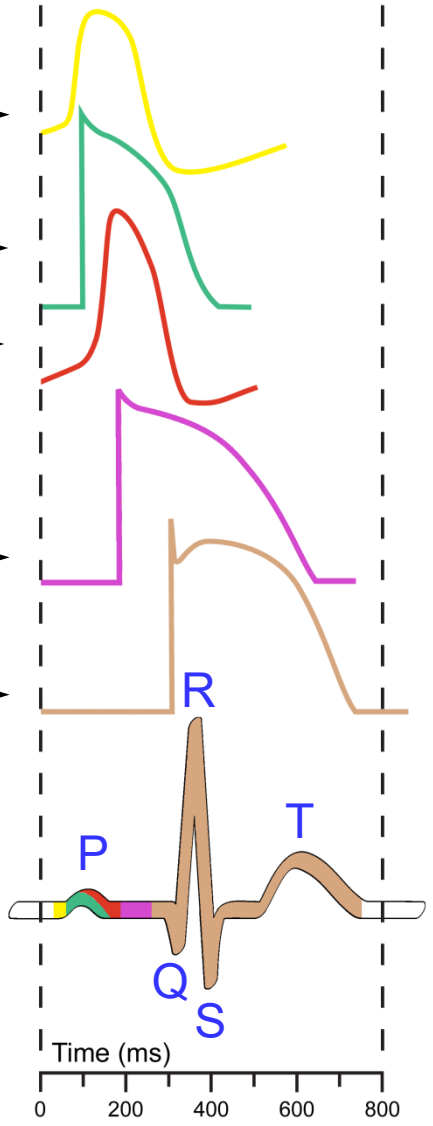
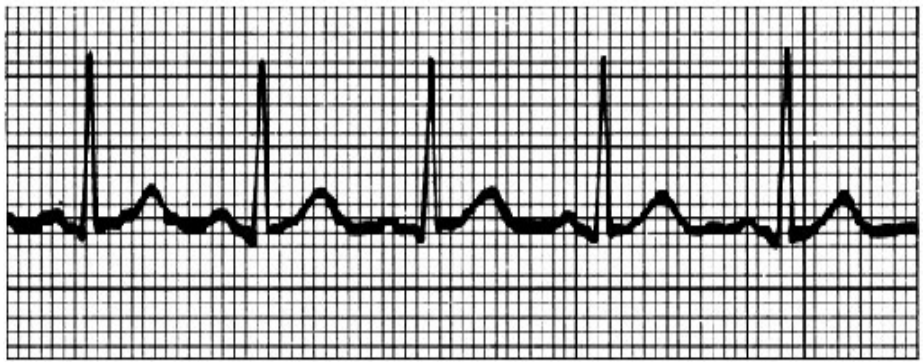


Action potential (AP)



Sinus rhythm

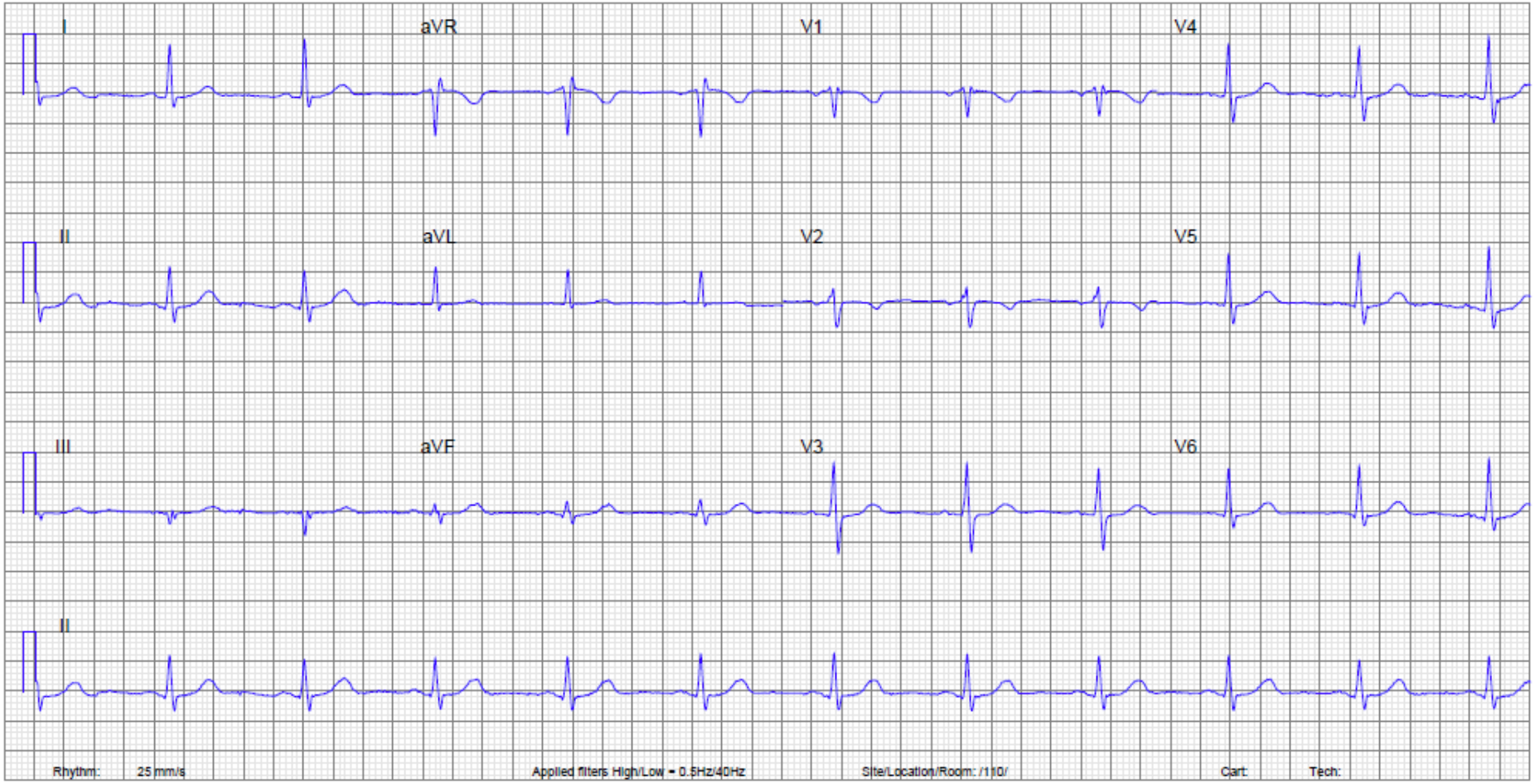


Surface ECG

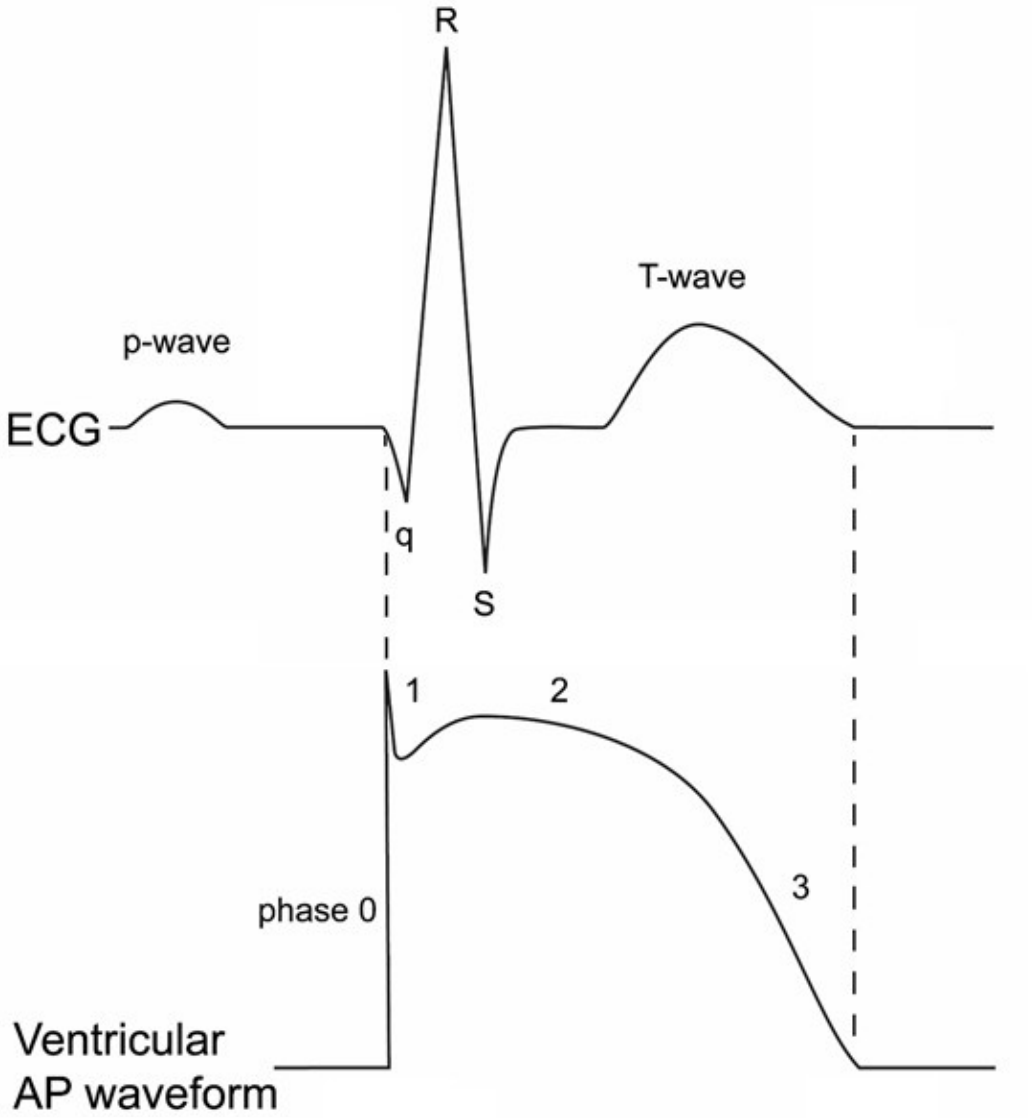
1 beat

Electrical functioning of the heart

ECG = 12-lead recording of the sum of all vectors
depolarization and repolarization of the entire heart

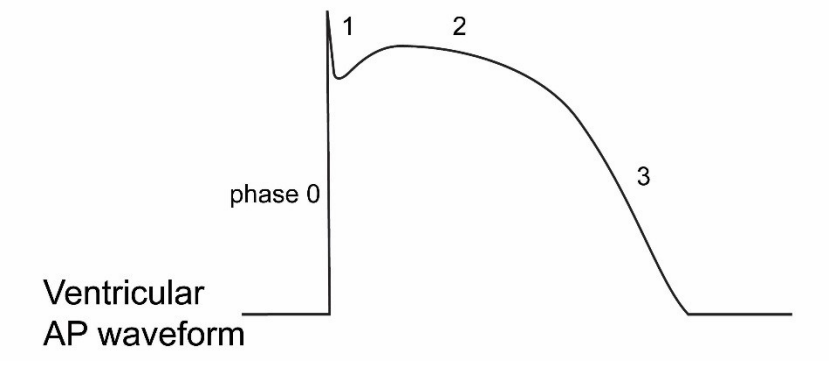


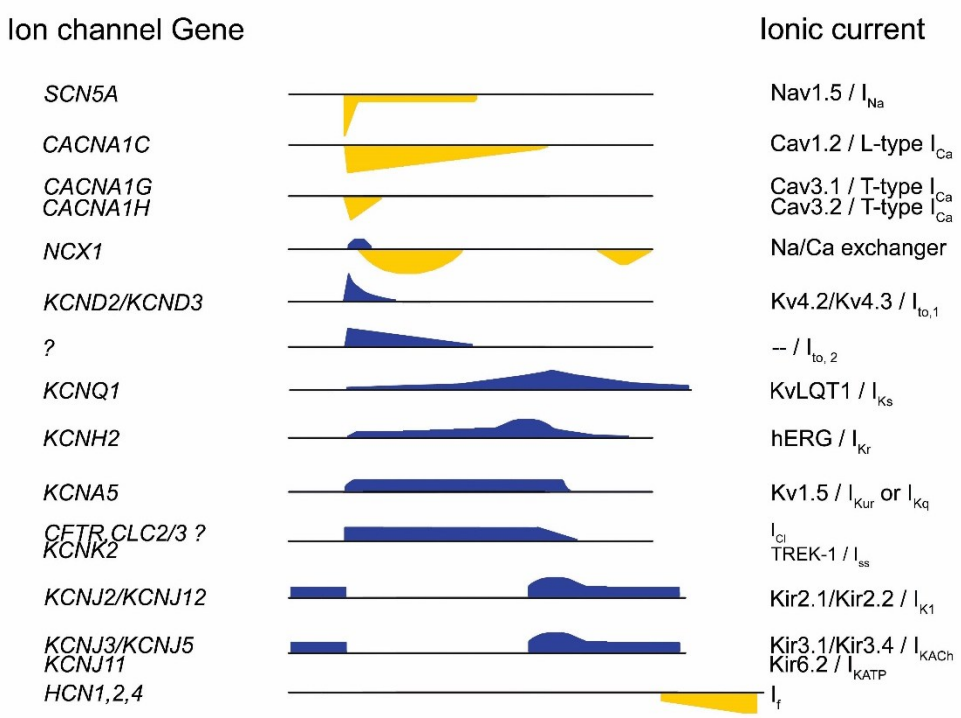
Electrical functioning of the heart



Electrical functioning of the heart

The cardiac action potential (AP)





Electrical functioning of the heart

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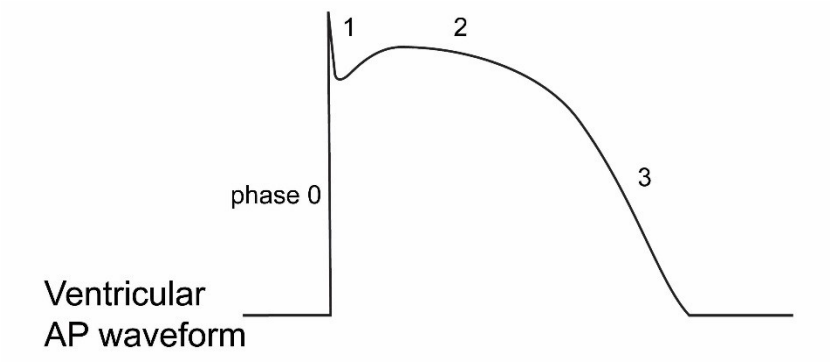
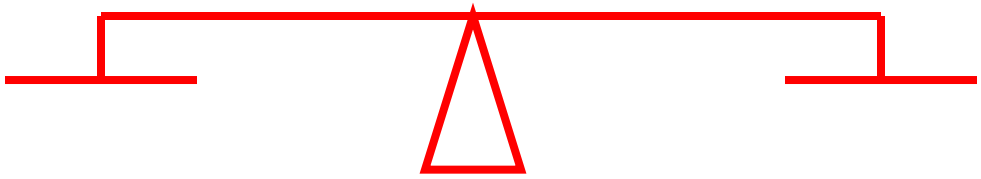




Depolarization

I_{Na+} (SCN5A)

I_{Ca2+} (α_{1c}, α_{1H}, Exch.)

Repolarization

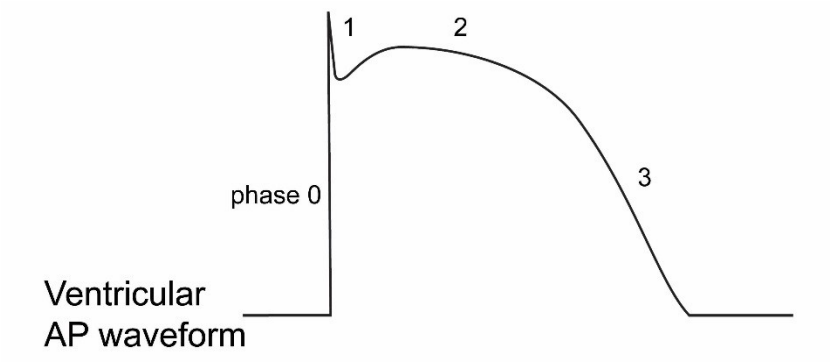
I_{to} (Kv4.2/4.3)

I_{Kr}, I_{Ks} (KvLQT1,hERG)

I_{K1} (Kir 2.1)

Electrical functioning of the heart

The cardiac action potential (AP)



Depolarization

Na^{+}/Ca^{2+}

Repolarization

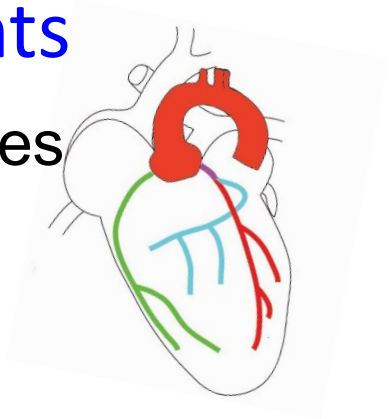
K^{+}

3. Introduction to cardiac disease

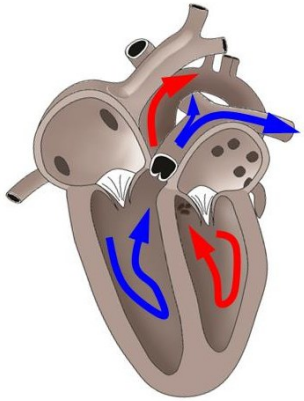
Introduction to cardiac disease

3 Main components

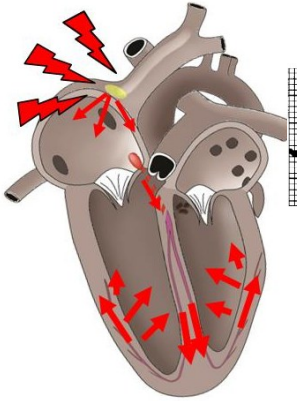
1. Coronary arteries

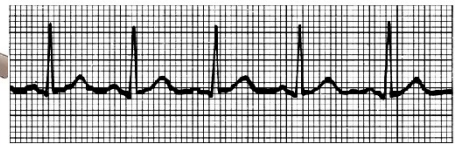


2. Pump function



3. Electrical function

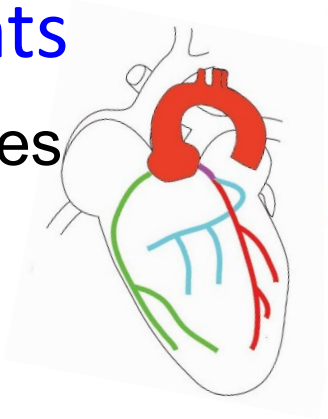


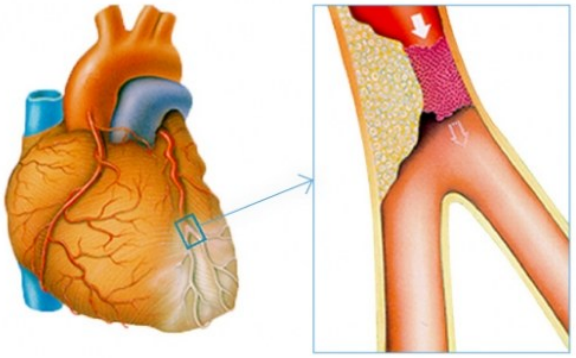


Introduction to cardiac disease

3 Main components

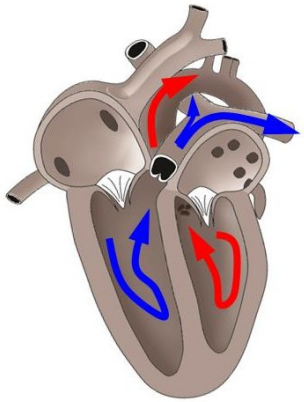
1. Coronary arteries



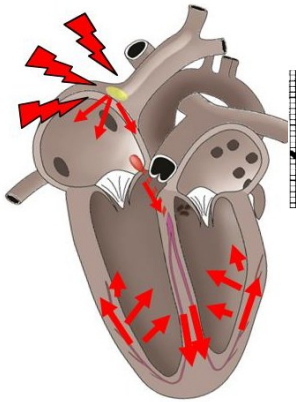


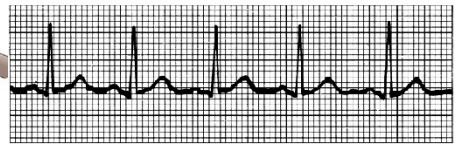
Coronary artery disease (CAD)

2. Pump function



3. Electrical function

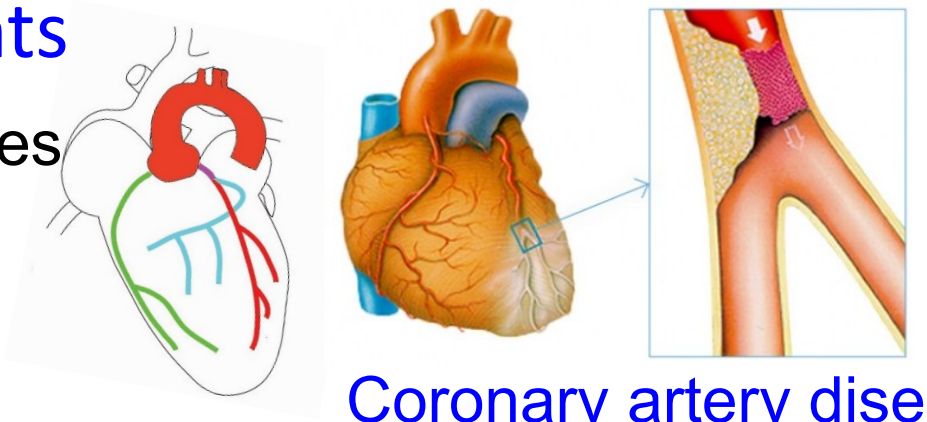




Introduction to cardiac disease

3 Main components

1. Coronary arteries



Coronary artery disease (CAD)

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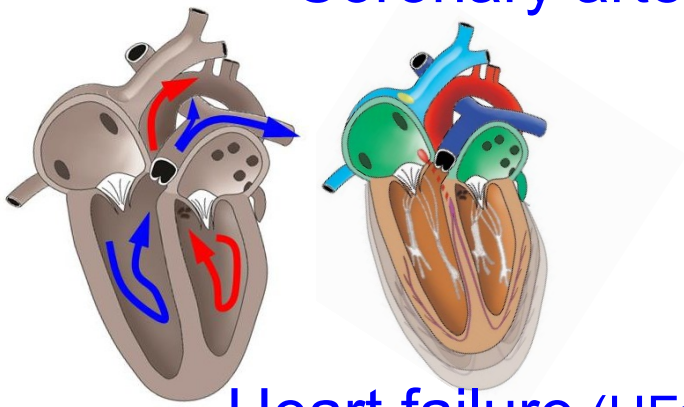
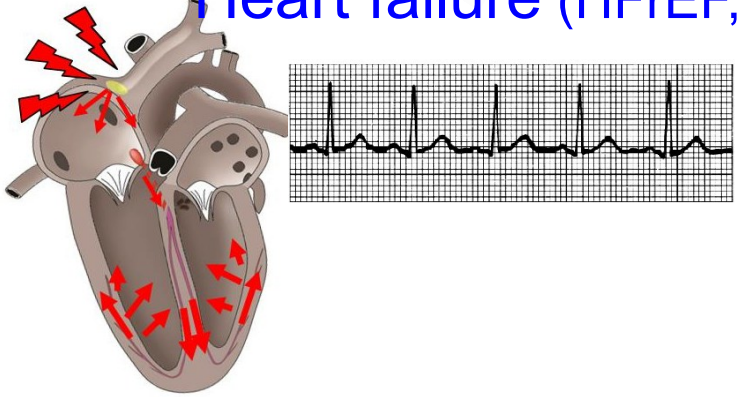


Heart failure (HFrEF, HFmrEF, HFpEF)

3. Electrical function



Introduction to cardiac disease

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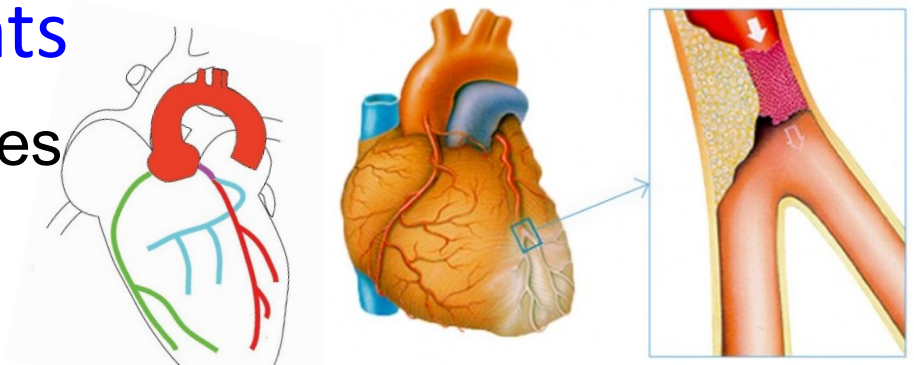

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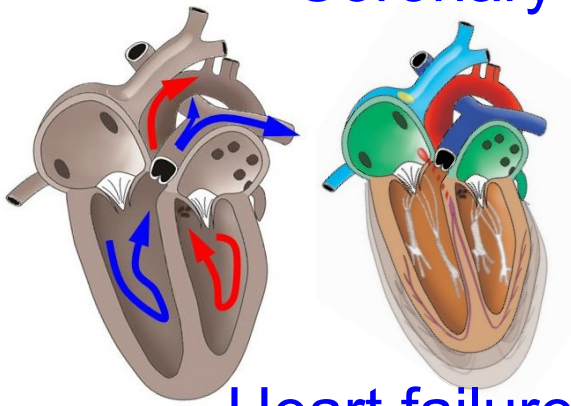

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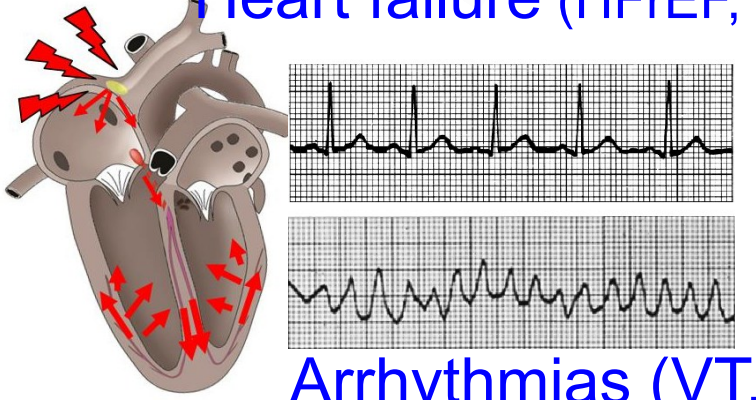

Arrhythmias (VT, TdP, VF,…)

Introduction to cardiac disease

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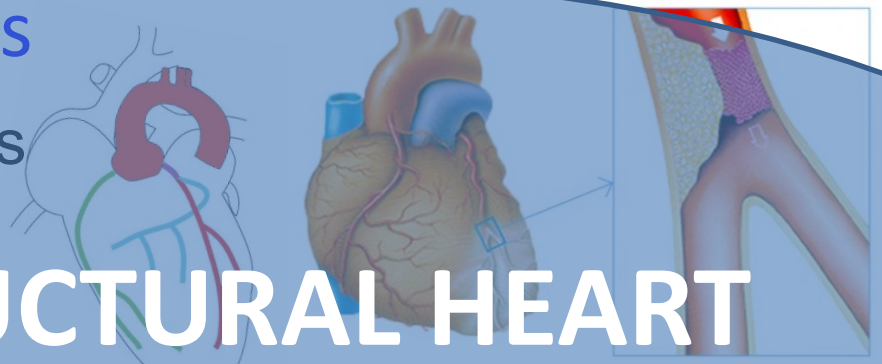
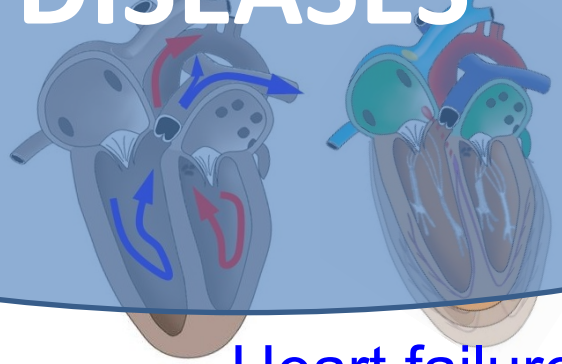


STRUCTURAL HEART DISEASES

Coronary artery disease (CAD)

2. Pump function



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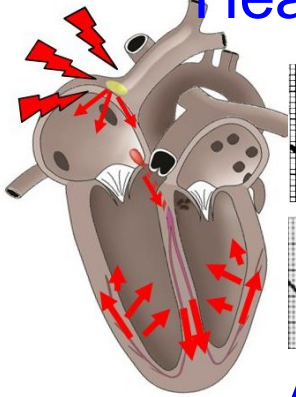

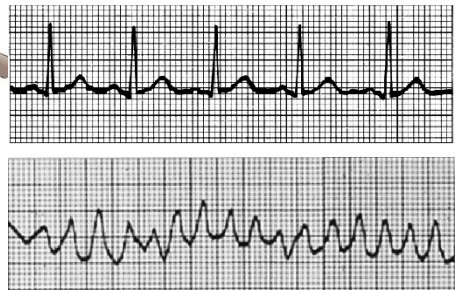

Arrhythmias (VT, TdP, VF,…)

Introduction to cardiac disease

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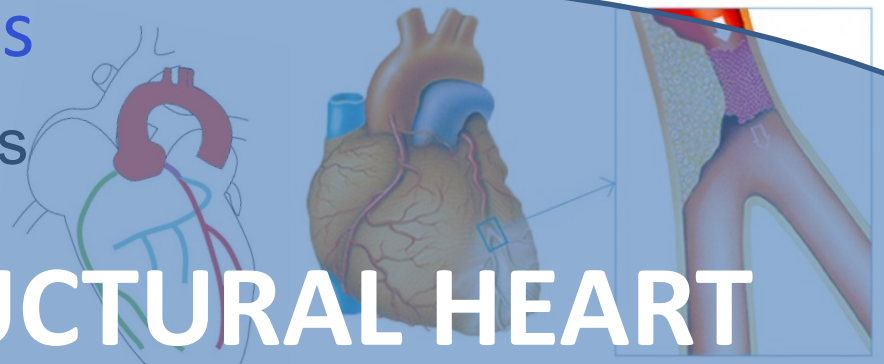
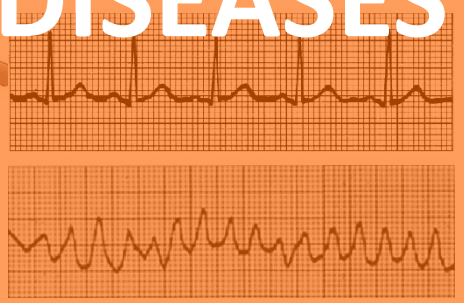


STRUCTURAL HEART DISEASES

Coronary artery disease (CAD)

Heart failure (HFrEF, HFmrEF, HFpEF)

ELECTRICAL DISEASES



Arrhythmias (VT, TdP, VF,…)

Introduction to cardiac disease

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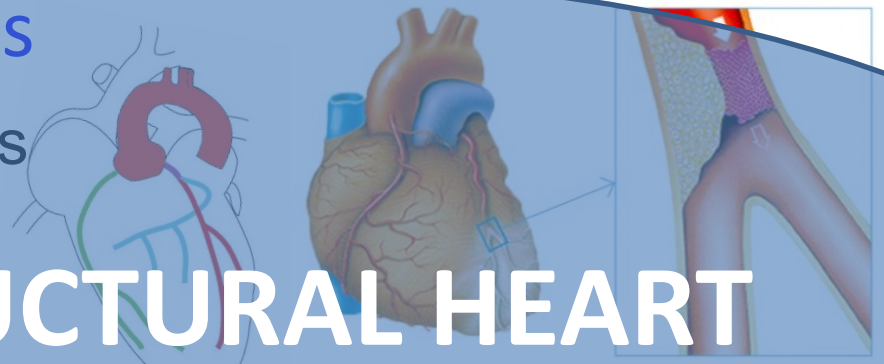
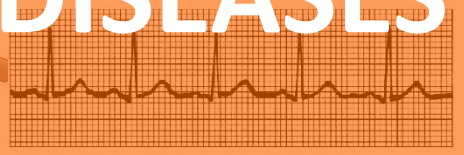


STRUCTURAL HEART DISEASES

Coronary artery disease (CAD)

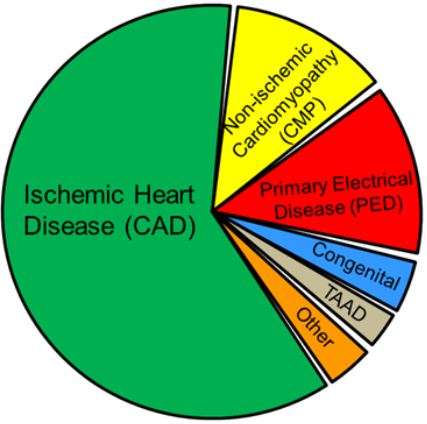
Heart failure (HFrEF, HFmrEF, HFpEF)

ELECTRICAL DISEASES



Arrhythmias (VT, TdP, VF,…)

Introduction to cardiac disease



Sudden Cardiac Death

- Structural Heart Disease (CMP)
  - Ischemic Heart Disease (CAD)
  - Congenital Heart Disase
  - Non-ischemic cardiomyopathy
    - DCM / HCM ACM / RCM
  - Fam WPW
  - Sarcoidosis Myocarditis
- Thoracic Aortic Aneurysm & Dissection (TAAD)
- Primary Electrical Disease (PED)
  - Commotio cordis
  - LQTS, SQTS, CPVT, PCCD, MEPPC, IVF, J-wave syndromes: BS, ERS

Structural disease → **Non-structural disease**

4. Structural heart disease

Structural heart disease

Congenital Heart Disease (CHD)

Acquired
Rarely hereditary

Coronary artery disease (CAD)

Acquired
Gene predisposition

Cardiomyopathy (CMP)

Hereditary
Acquired

- Hypertrophic CMP (HCM)
- Dilated CMP (DCM)
- Restrictive CMP (RCM)
- Arrythmogenic CMP (ACM)
- Unclassified

Detectable changes in cardiac structure and/or function

Congenital Heart Disease

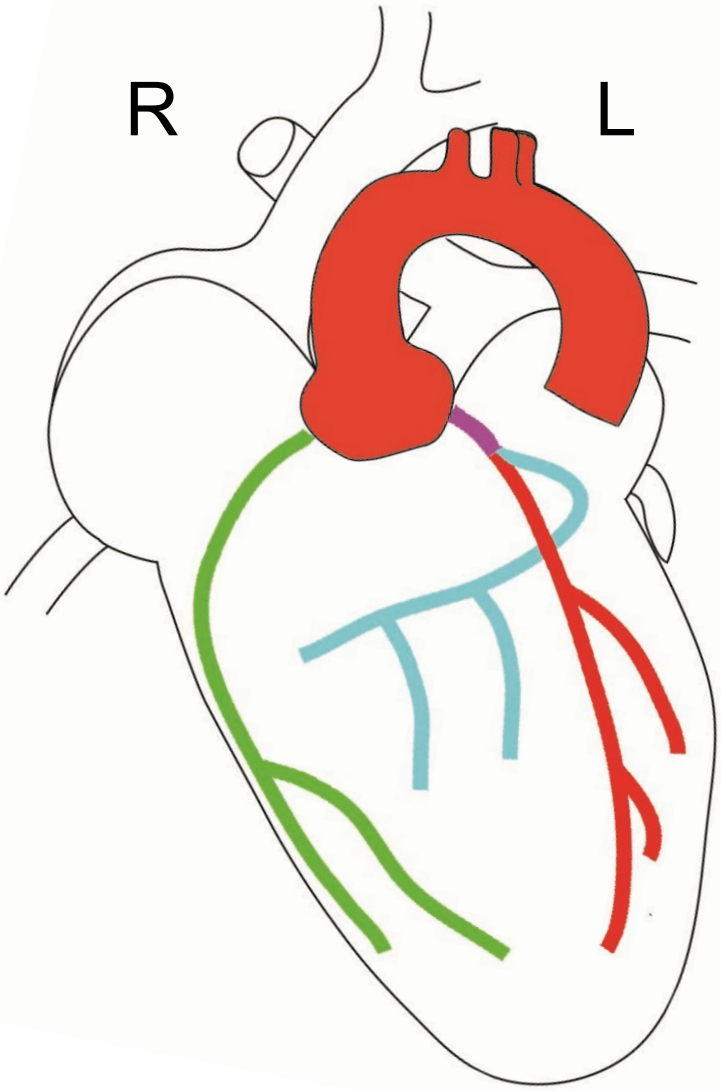
Congenital Heart Disease (CHD)

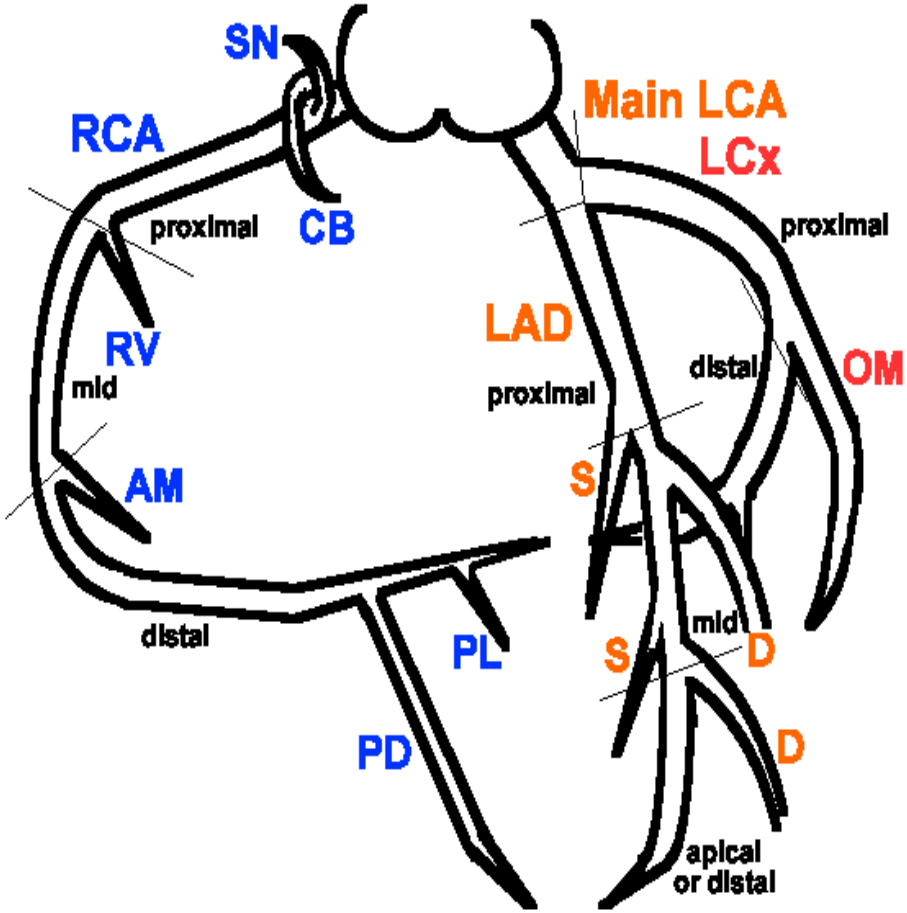
3% Mendelian single gene defect
5-15% Chromosomal defect

Atrial Septum Defect (ASD)
Ventricular Septal Defect (VSD)
Patent Ductus Arteriosus (PDA)
Ebstein malformation
Transposition of Great Vessels (TOGV)
Tetralogy of Fallot (TOF)
Pulmonary Atresia/Stenosis (PA/PS)
Tricuspid Atresia/Stenosis (TA/TS)
Truncus Arteriosus
Total Anomalous Pulmonary Venous Return (TAPVR)
Double Outlet Richt Ventricle
Hypoplastic Left Heart Syndrome (HLHS)
Aortic Arch Abnormalities

Coronary Artery Disease

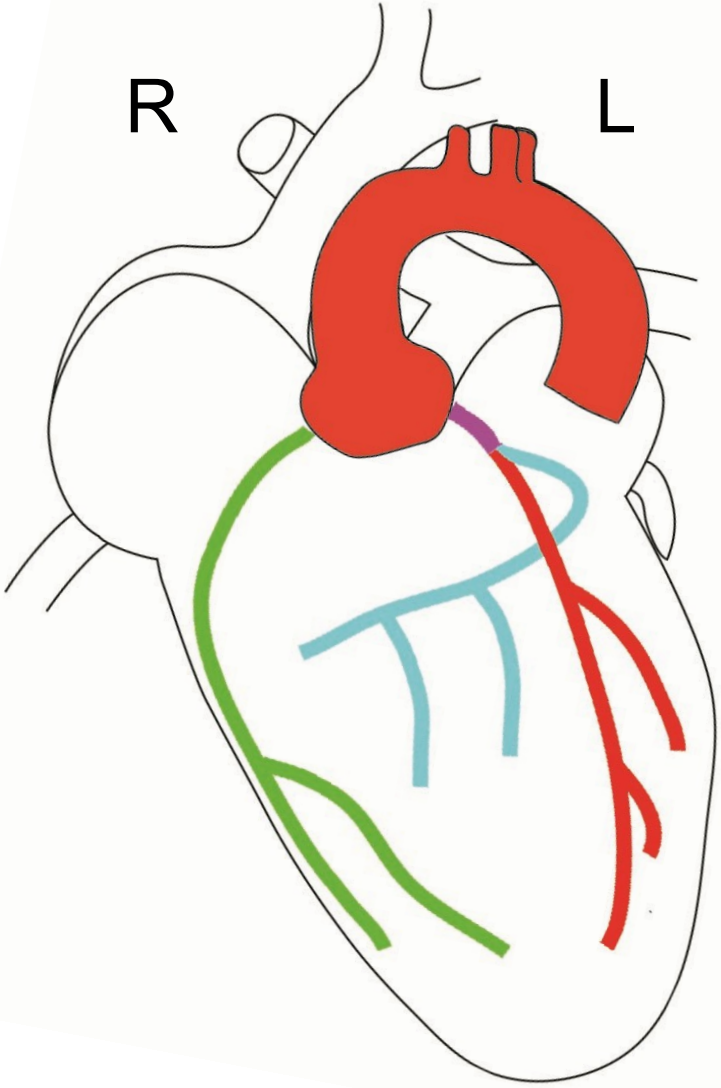
Coronary artery disease (CAD)

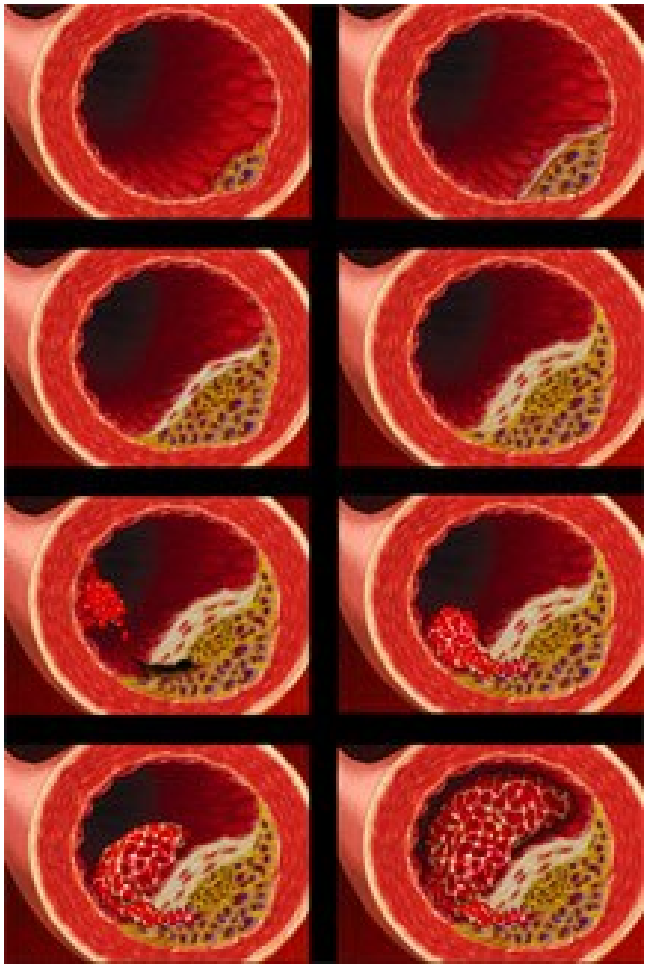




Coronary artery disease (CAD)

Coronary clogging due atheromatosis

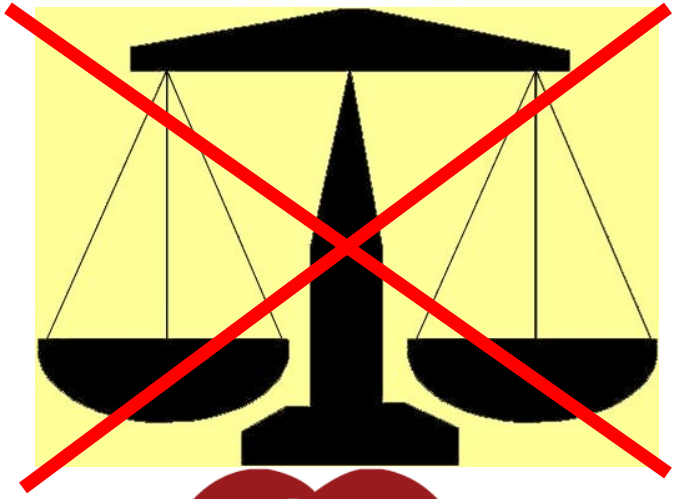




Coronary artery disease (CAD)

Ischemia arises from oxygen mismatch

↓ Oxygen supply < Oxygen demand



Myocardial infarction

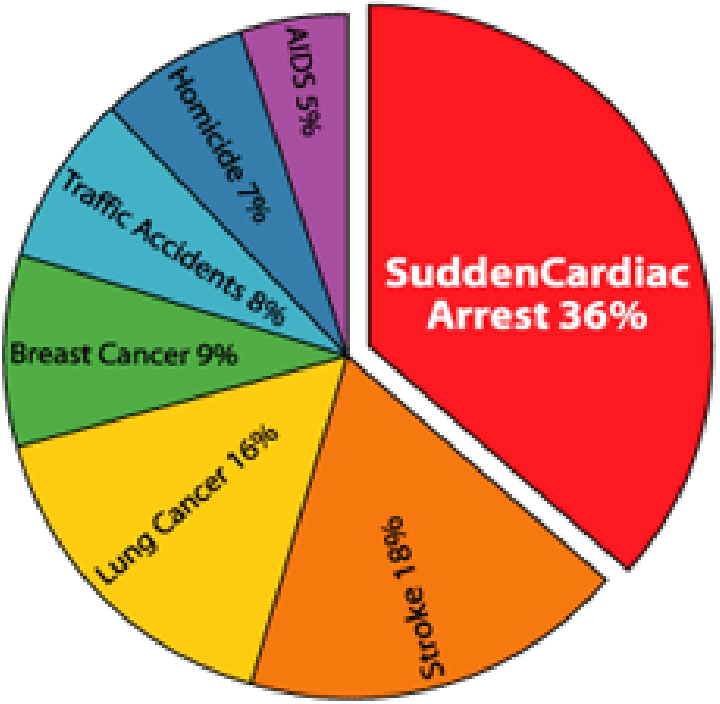
Sudden Cardiac Death

Pump failure
Dyspnoea, edema

Coronary artery disease (CAD)

Sudden cardiac death is a leading cause of death in the Western world

Estimated incidence of 1-2/1000/year
60-80% of SCD due to CAD

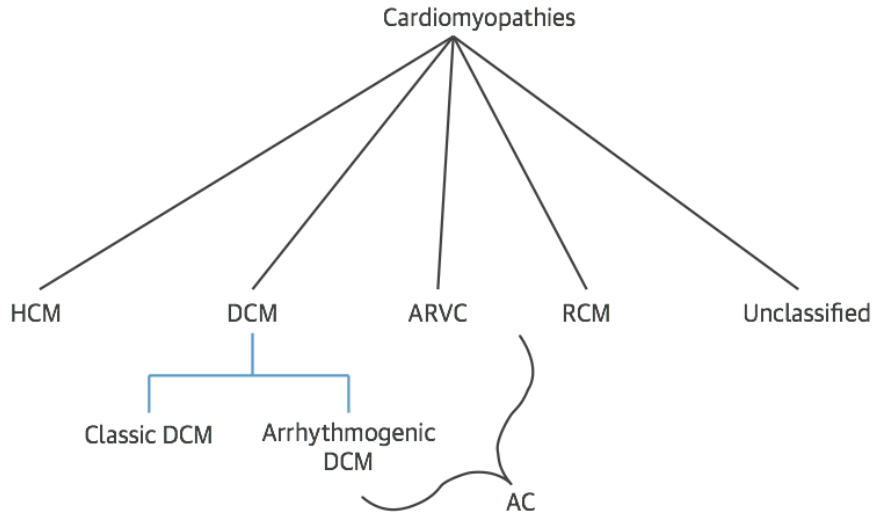


Sudden cardiac death 36%
Lung Cancer 16%
Stroke 16%
Breast Cancer 9%
Traffic accidents 9%

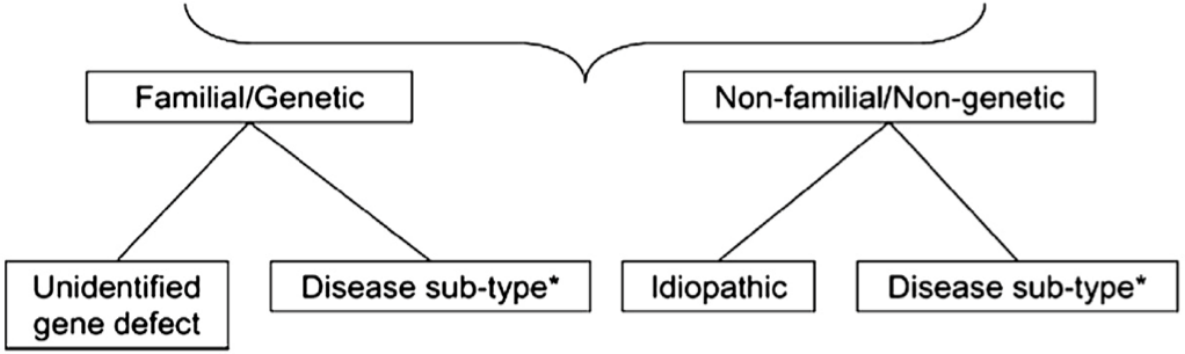
Non ischemic Cardiomyopathy

Non ischemic cardiomyopathies

Morphological classification

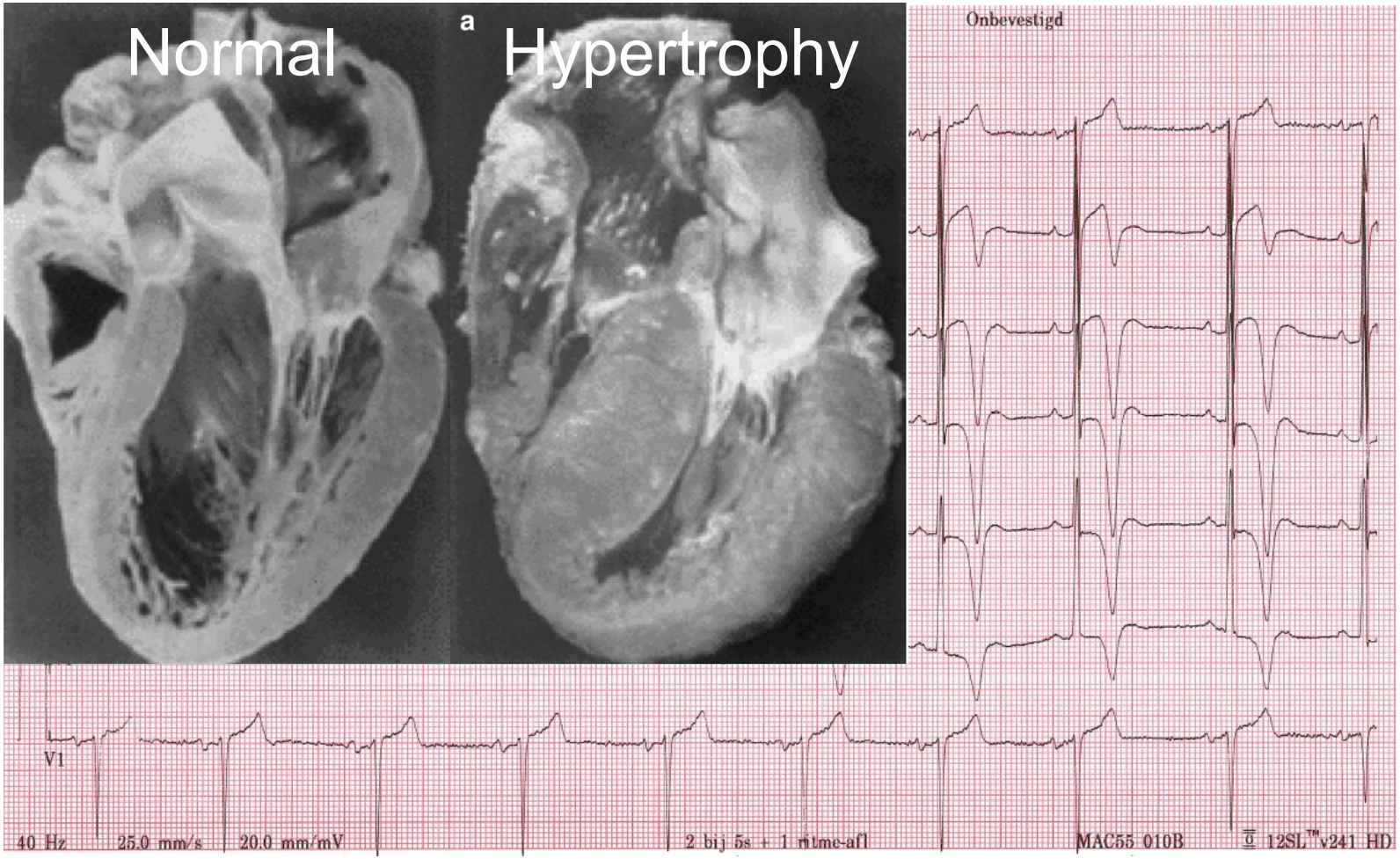


Genetic Classification



Non ischemic cardiomyopathies

Hypertrophic cardiomyopathy - HCM

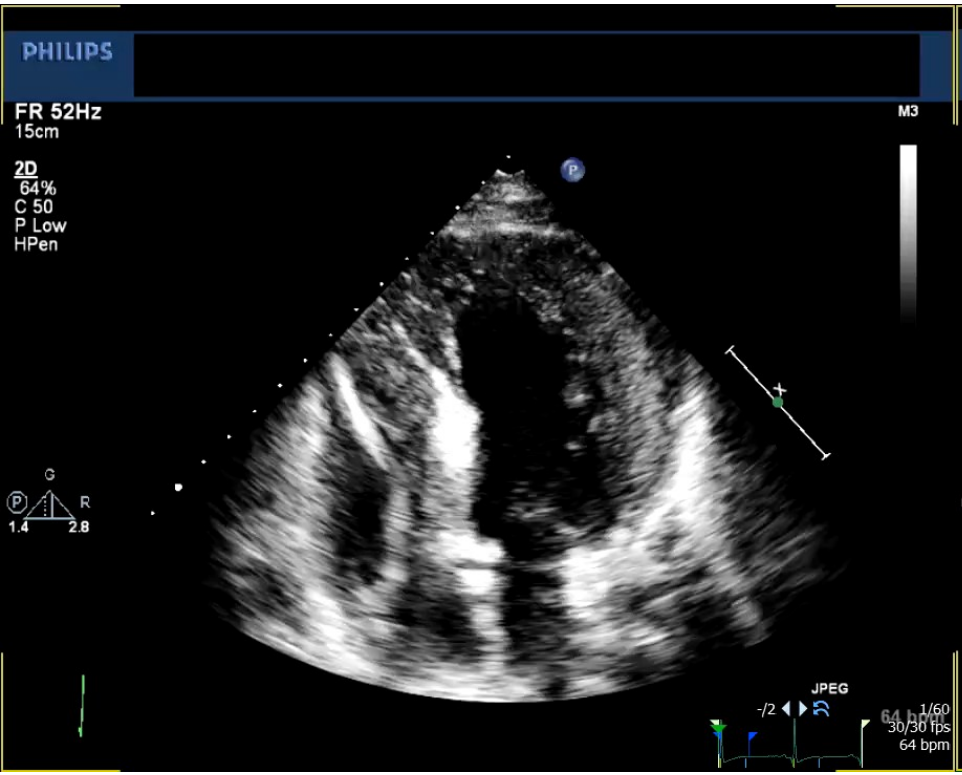


Non ischemic cardiomyopathies

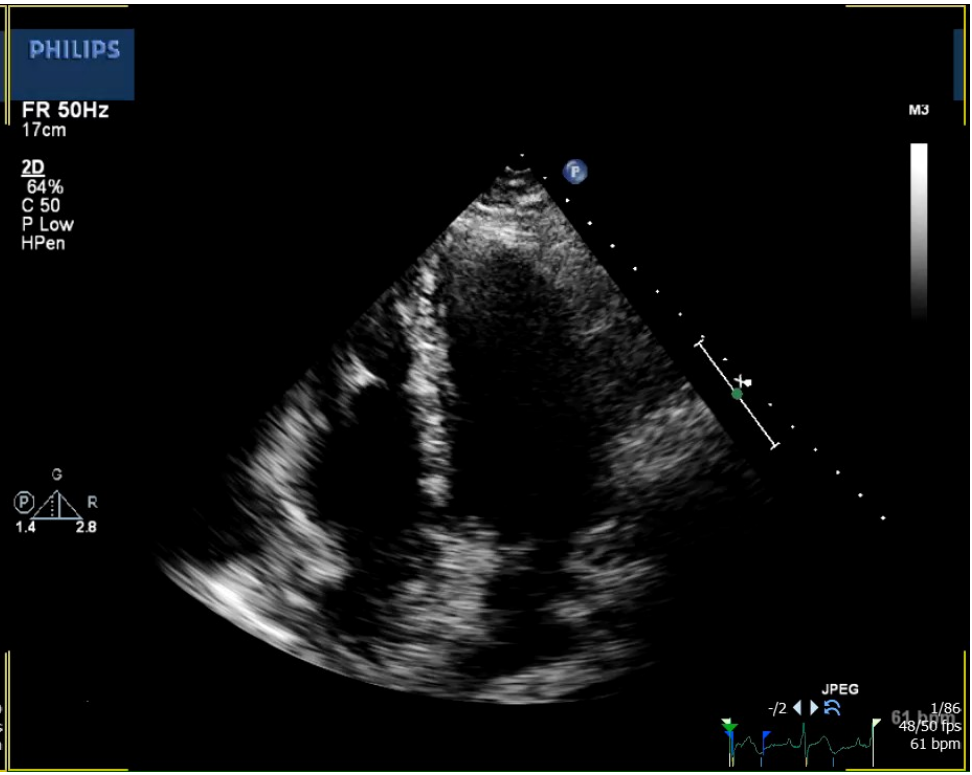
Hypertrophic cardiomyopathy - HCM

Hereditary HCM
Incidence = 1:250-500

HCM



NORMAL TTE



Non ischemic cardiomyopathies

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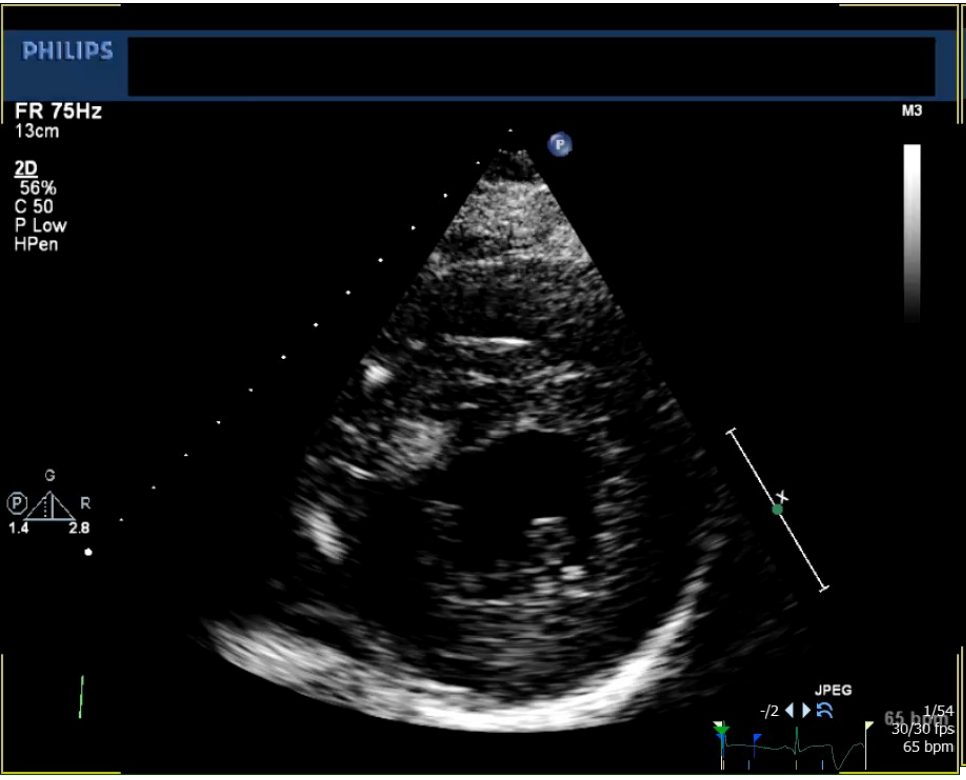
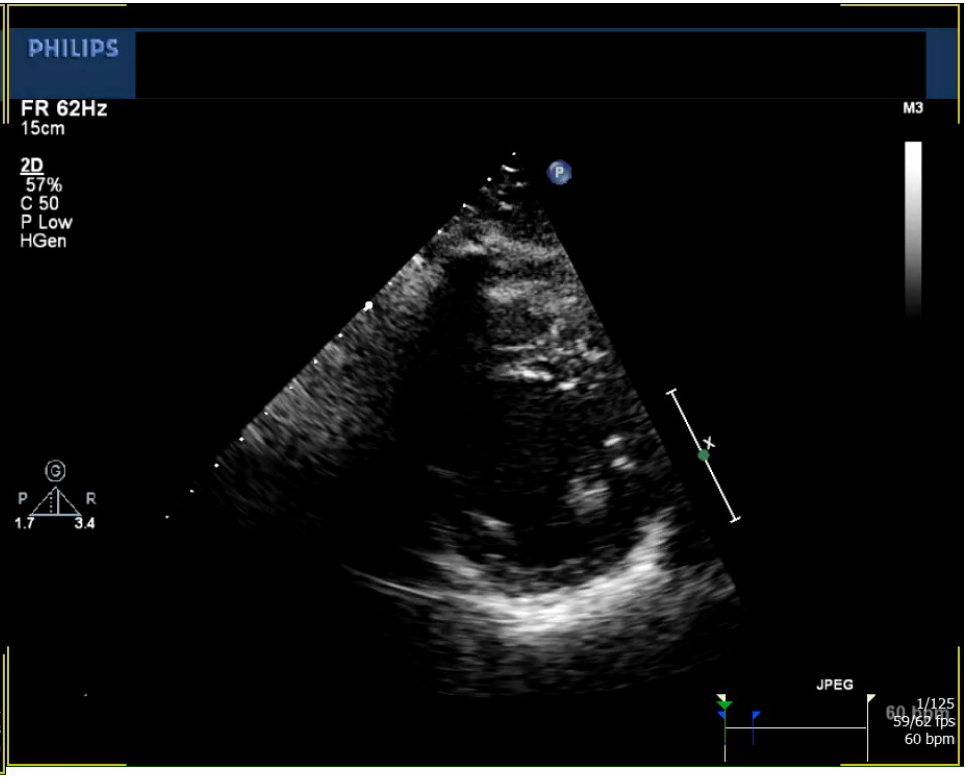


NORMAL TTE



Non ischemic cardiomyopathies

Hypertrophic cardiomyopathy - HCM

Risk of sudden cardiac death during exercise

A genetic killer in the family

When Karen Falconer's brother collapsed, it was thought he'd had a heart attack. The cause was more sinister: a genetic condition that causes cardiac failure in the young. Suddenly, early deaths in previous generations began to make sense

Karen Falconer | Tuesday 18 March 2014 | 0 comments

24 shares

Tim Nicot dead: Second Belgian footballer dies from cardiac arrest less than two weeks after death of Gregory Mertens

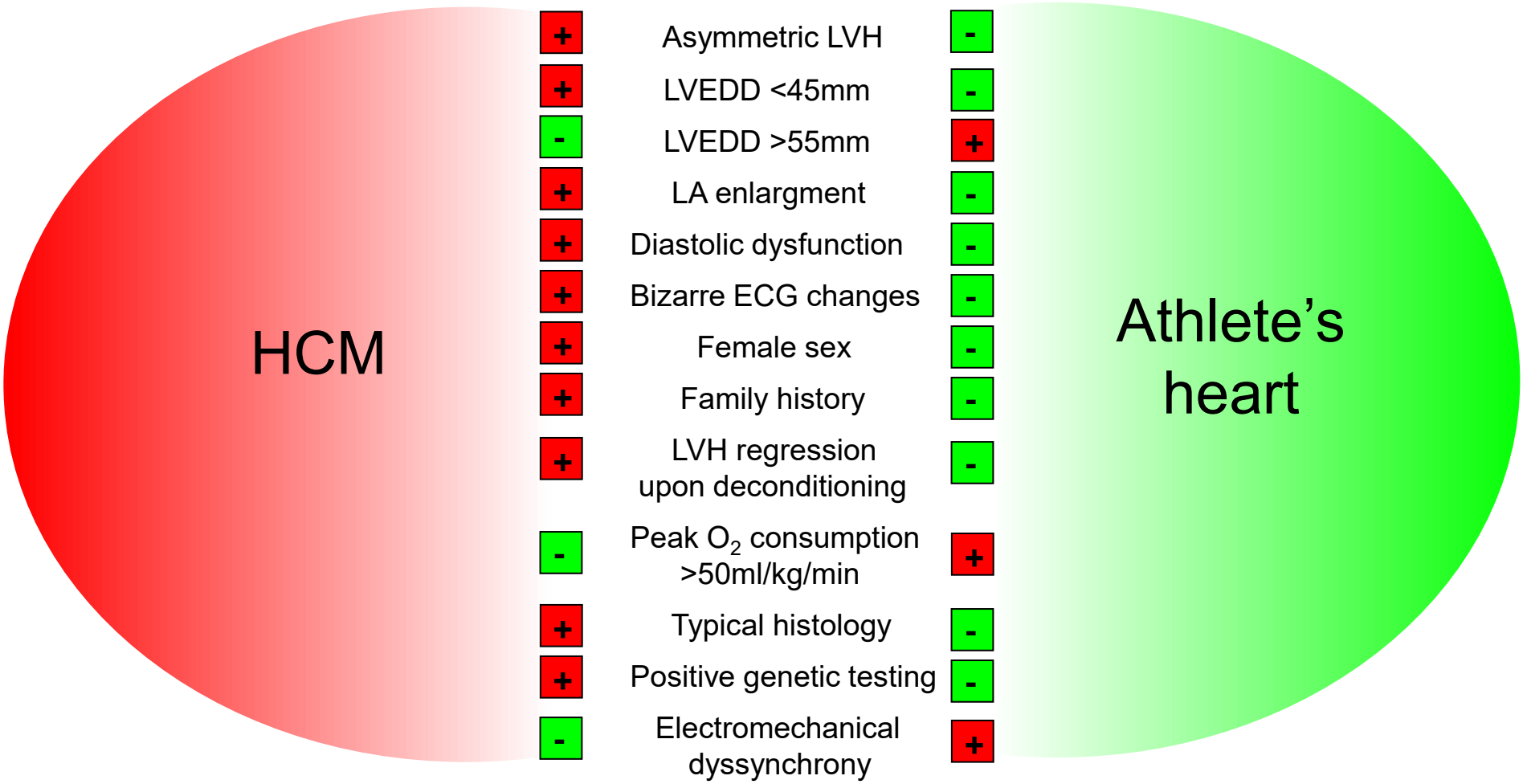
Sacramento youth basketball commun… shaken by on-court death of Elk Grove boy

Surviving sudden cardiac arrest: One teen athlete's story

Non ischemic cardiomyopathies

Athletes' heart = Adaptive wall thickening

not associated with SCD

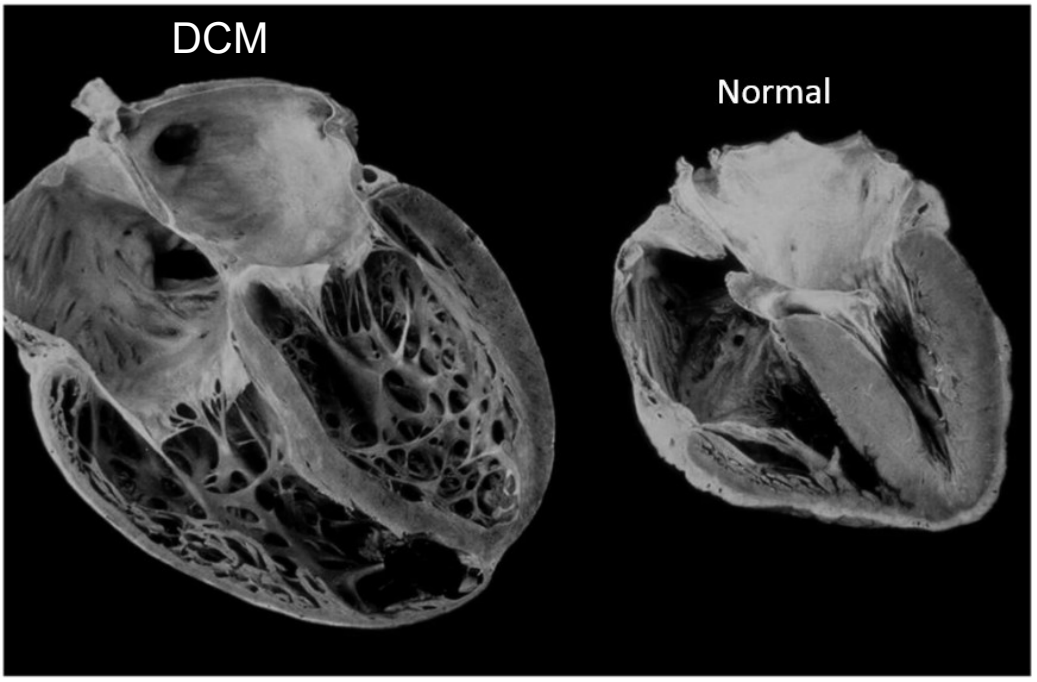


Non ischemic cardiomyopathies

Dilated cardiomyopathy - DCM

Often acquired

- Ischemic
- Toxic: ethyl, cocaine, amfetamines, anthracyclin chemotherapy
- Viral: HIV, myocarditis
- Tachycardiomyopathy
- Unknown origin



Think hereditary DCM:

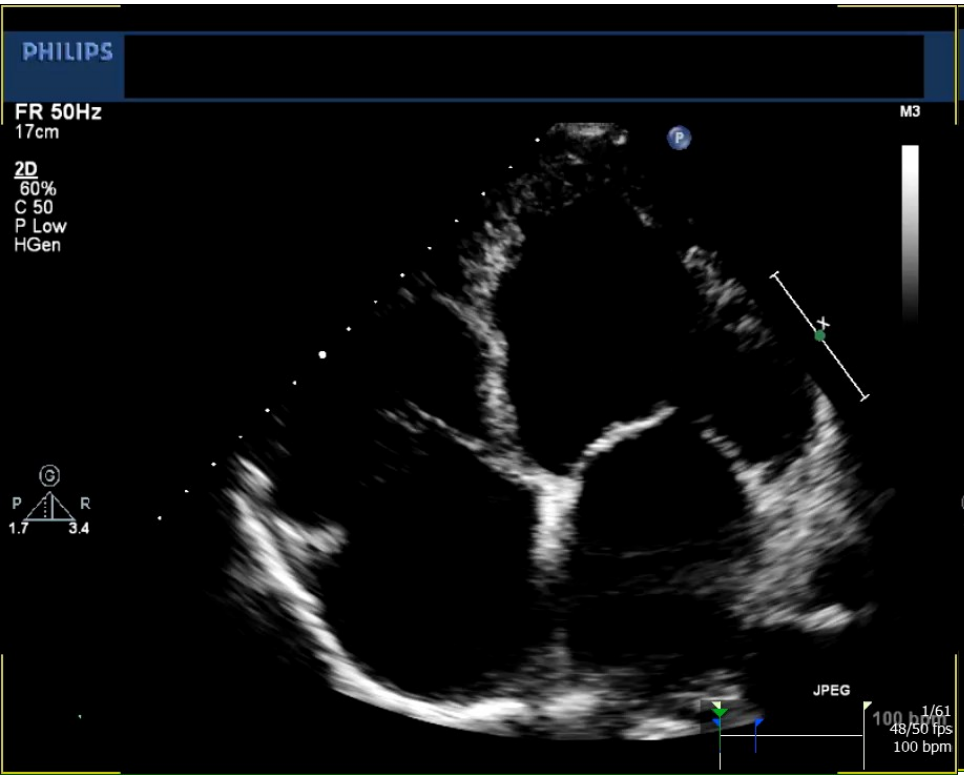
- Early onset
- AV–block
- SCD
- Relatives affected

Non ischemic cardiomyopathies

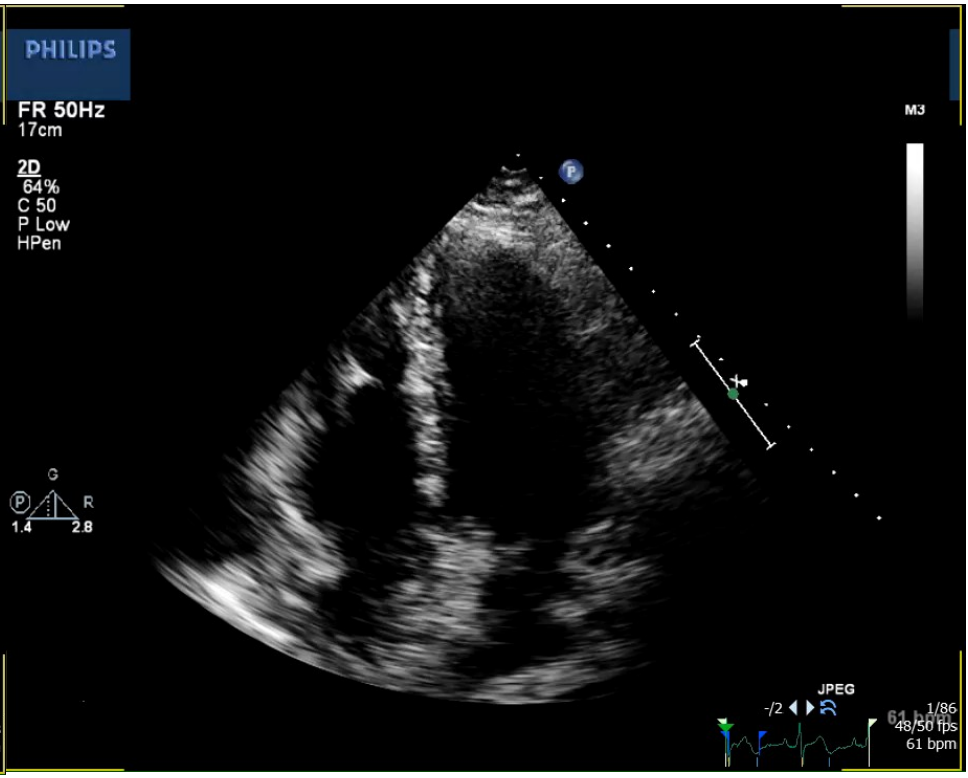
Dilated cardiomyopathy - DCM

DCM and reduced pump function = Risk for SCD

DCM



NORMAL TTE

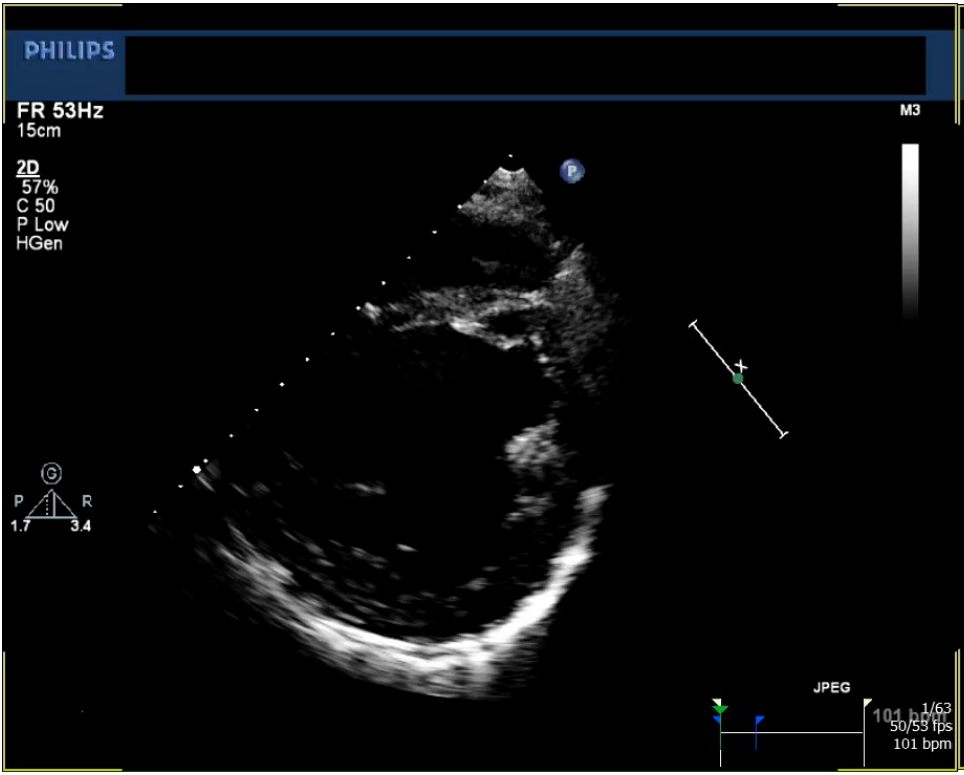


Non ischemic cardiomyopathies

Dilated cardiomyopathy - DCM

DCM and reduced pump function = Risk for SCD

DCM



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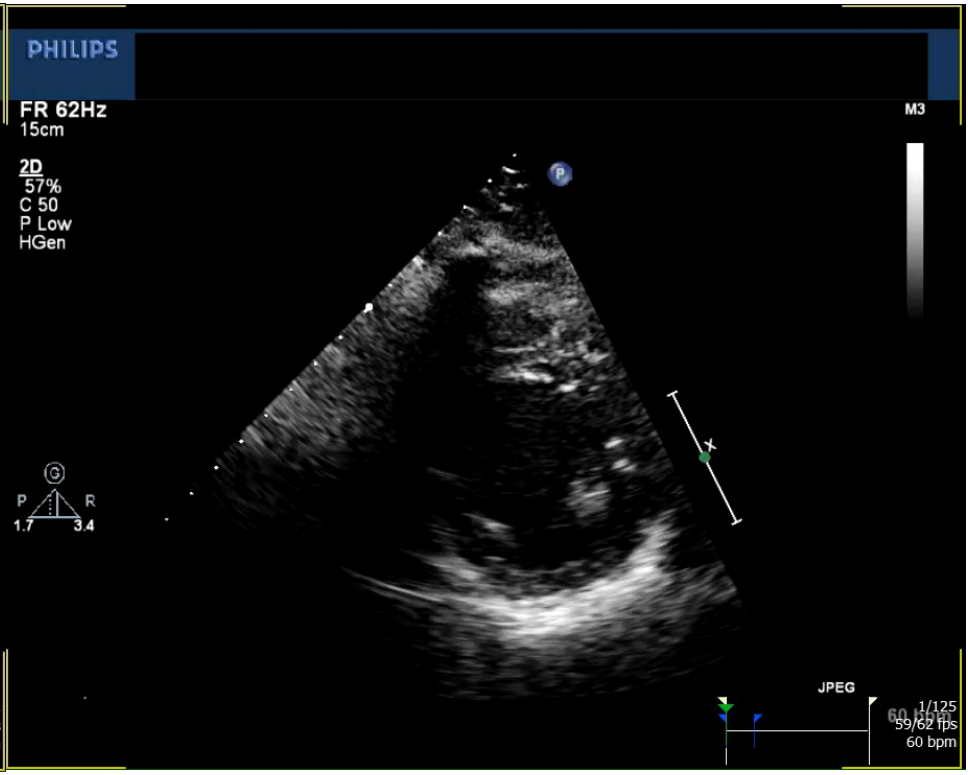
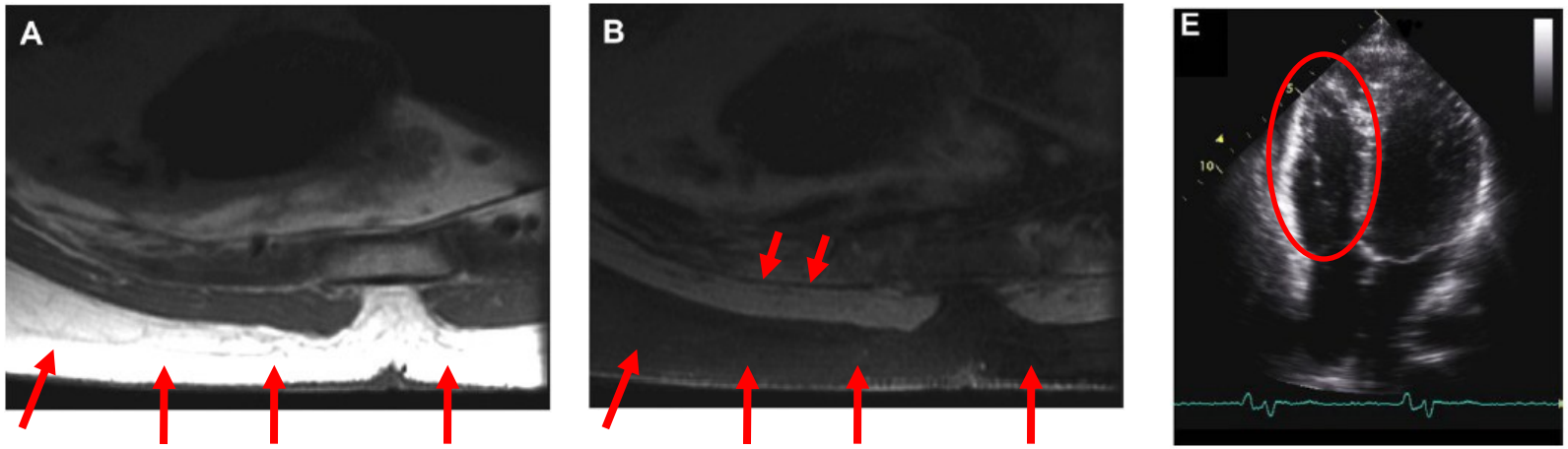


Arrhythmogenic Cardiomyopathy – ACM

Fibrofatty replacement of the cardiac wall
Progressive disease exacerbated by exercise



Ellinor et al. Heart Failure Clin. 2010;6:161-177

Clinical features

- Asymptomatic phase
- Arrhythmogenic phase: no detectable structural abnormalities
- Right ventricular heart failure
- **30% of SCD in athletes**

Non ischemic cardiomyopathies

Arrhythmogenic Cardiomyopathy – ACM

1994 Task force criteria

1994 Task Force Criteria

2 major, 1 major + 2 minor, or 4 minor

I. Global/regional dysfunction/structural alterations

Major

- Severe dilatation and reduction of RVEF w/no (or only mild) LV impairment
- Localized RV aneurysms (akinetic or dyskinetic areas w/diastolic bulging)
- Severe segmental dilatation of the RV

Minor

- Mild global RV dilatation and/or EF reduction with normal LV
- Mild segmental dilatation of the RV
- Regional RV hypokinesia

II. Tissue characterization of wall

Major

- Fibrofatty replacement of myocardium on endomyocardial biopsy

Minor

III. Repolarization abnormalities

Major

Minor

- TWI in right precordial leads (V_2 and V_3) (people age >12 yrs, in absence of RBBB)

IV. Depolarization/conduction abnormalities

Major

- Epsilon waves or localized prolongation (>110ms) of QRS complex in right precordial leads (V_1 to V_3)

Minor

- late potentials (SAECG)

V. Arrhythmias

Major

Minor

- LBBB sustained or NSVT (ECG, Holter, ETT)
- >1000 ventricular extrasystoles per 24 hours (Holter)

VI. Family History

Major

- Familial disease confirmed at necropsy or surgery

Minor

- Fam hx of SD (<35yrs) due to suspected ARVC/D
- Familial hx (clinical dx based on present criteria)

2010 Task force criteria

2010 Task Force Criteria

Definite = 2 major OR 1 major + 2 minor
Borderline = 1 major + 1 minor OR 3 minor
Possible = 1 major OR 2 minor

By 2D Echo
- Regional RV akinesis, dyskinesia, or aneurysm
- *and* 1 of the following (end diastole):

___PLAX RVOT ≥32 mm (correct for body size [PLAX/BSA] ≥19 mm/m^2
___PSAX RVOT ≥36 mm (correct for body size [PSAX/BSA] ≥21 mm/m^2)
___or fractional area change ≤33%

By MRI:
- Regional RV akinesia or dyskinesia or dyssynchronous RV contraction
- *and* 1 of the following :

___Ratio of RV end-diast vol to BSA ≥110 mL/m^2 (male) or ≥100 mL/m^2 (female)
___or RV ejection fraction ≤40%

By RV Angiography:
- Regional RV akinesia, dyskinesia, or aneurysm

By 2D Echo:
- Regional RV akinesia or dyskinesia
- *and* 1 of the following (end diastole):

___PLAX RVOT ≥29 to <32 mm (correct body size PLAX/BSA ≥16 to <19 mm/m^2)
___PSAX RVOT ≥32 to <36 mm (correct body size [PSAX/BSA] ≥18 to <21 mm/m^2)
___or fractional area change >33% to ≤40%

By MRI:
- Regional RV akinesia or dyskinesia or dyssynchronous RV contraction
- *and* 1 of the following:

___Ratio of RVEDV to BSA ≥100 to <110mL/m^2 (male) or ≥90 to <100mL/m^2 (fem)
___or RV EF >40% to ≤45%

- Residual myocytes < 60% by morphometric analysis (or <50% if estimated), w/fibrosis replacement of RV free wall myocardium in ≥1 sample, w/ or w/o fatty replacement of tissue on endomyocardial biopsy

- Residual myocytes 60% to 75% by morphometric analysis (or 50% to 60% if est.) w/fibrous replacement of the RV free wall in ≥1 sample, w/ or w/o fatty replacement of tissue on endomyocardial biopsy

- TWI (V_1, V_2, V_3) or beyond; >14 yrs; in absence of complete RBBB QRS ≥120 ms

- TWI in V_1 and V_2; >14 yrs; in absence of complete RBBB or in V_4, V_5, or V_6
- TWI in V_1 -V_4; >14 yrs; in presence of complete RBBB

- Epsilon wave (reproducible low-amp signals btn end of QRS complex to onset of T wave) in right precordial leads (V_1-V_3)

- LP by SAECG in ≥1 of 3 parameters in absence of QRS duration of ≥110ms on ECG
___Filtered QRS duration (fQRS) ≥114ms
___Duration of terminal QRS <40μV (LAS duration) ≥38ms
___RMS voltage of terminal 40 ms ≤20μV
- TAD of QRS ≥55ms measured from nadir of S wave to end of QRS, including R', in V_1, V_2, or V_3, in absence of complete RBBB

- LBS NSVT or sustained VT (neg or indet QRS in II, III, and aVF and pos in aVL)
- NSVT or sustained VT of RV outflow configuration, LBI (pos QRS in II, III, and aVF and neg in aVL) or of unknown axis
- >500 ventricular extrasystoles per 24 hours (Holter)

- ARVC/D confirmed in FDR who meets TFC
- ARVC/D confirmed pathologically at autopsy or surgery in FDR
- Pathogenic mutation (assoc or probably assoc w/ ARVC/D) in pt under eval

- Hx of ARVC in FDR in whom not poss or pract to determine if FM meets TFC
- Premature SD (<35 yrs) due to suspected ARVC/D in FDR
- ARVC/D confirmed pathologically or by current TFC in 2ndDR

Minor and maior criteria

Probability of diagnosis:

'Possible' diagnosis

'Borderline' diagnosis

'Definite' diagnosis

Non ischemic cardiomyopathies

Arrhythmogenic Cardiomyopathy – ACM

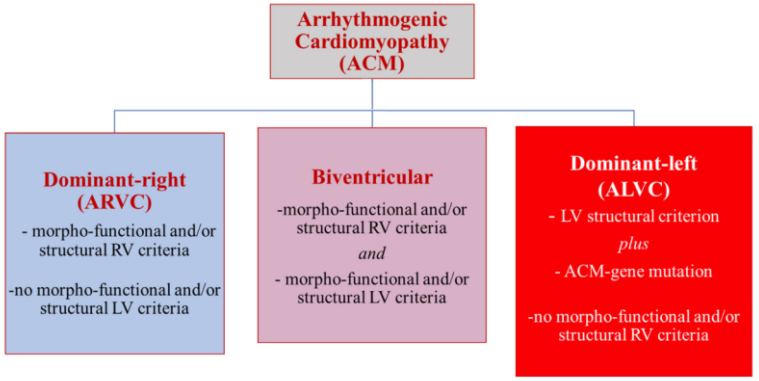
2020 Proposed Padua criteria

"Padua criteria" for diagnosis of Arrhythmogenic Cardiomyopathy.

Category	Right ventricle (upgraded 2010 ITF diagnostic criteria)	Left ventricle (new diagnostic criteria)
I. Morpho-functional ventricular abnormalities	*By echocardiography, CMR or angiography:* *Major* • Regional RV akinesia, dyskinesia, or bulging *plus* one of the following: - global RV dilatation (increase of RV EDV according to the imaging test specific nomograms) - global RV systolic dysfunction (reduction of RV EF according to the imaging test specific nomograms) *Minor* • Regional RV akinesia, dyskinesia or aneurysm of RV free wall	*By echocardiography, CMR or angiography:Minor* • Global LV systolic dysfunction (depression of LV EF or reduction of echo-cardiographic global longitudinal strain), with or without LV dilatation (increase of LV EDV according to the imaging test specific nomograms for age, sex, and BSA) *Minor* • Regional LV hypokinesia or akinesia of LV free wall, septum, or both
II. Structural myocardial abnormalities	*By CE-CMR:Major* • Transmural LGE (stria pattern) of ≥1 RV region(s) (inlet, outlet, and apex in 2 orthogonal views) *By EMB (limited indications):Major* • Fibrous replacement of the myocardium in ≥1 sample, with or without fatty tissue	*By CE-CMR:Major* • LV LGE (stria pattern) of ≥1 Bull's Eye segment(s) (in 2 orthogonal views) of the LV wall (subepicardial or midmyocardial), septum, or both (excluding septal junctional LGE)
III. Repolarization abnormalities	*Major* • Inverted T waves in right precordial leads (V_1,V_2, and V_3) or beyond in individuals with complete pubertal development and the presence of complete RBBB) *Minor* • Inverted T waves in leads V1 and V2 in individuals with completed pubertal development (in the absence of complete RBBB) • Inverted T waves in V1,V2,V3 and V4 in individuals with completed pubertal development in the presence of complete RBBB.	*Minor* • Inverted T waves in left precordial leads (V_4-V_6) (in the absence of complete LBBB)
IV. Depolarization abnormalities	*Minor* • Epsilon waves (reproducible low-amplitude signalsbetween end of QRS complex to onset of the T wave) in the right precordial leads (V1 to V3) • Terminal activation duration of QRS ≥55 ms measured from the nadir of the S wave to the end of the QRS, including R', in V1, V2, or V3 (in the absence of complete RBBB)	*Minor* • Low QRS voltages (<0.5 mV peak to peak) in limb leads (in the absence of obesity, emphysema, or pericardial effusion)
V. Ventricular arrhythmias	*Major* • Frequent ventricular extrasystoles (>500 per 24 h), non-sustained or sustained ventricular tachycardia of LBBB morphology *Minor* • Frequent ventricular extrasystoles (>500 per 24 h), non-sustained or sustained ventricular tachycardia of LBBB morphology with inferior axis ("RVOT pattern")	*Minor* • Frequent ventricular extrasystoles (>500 per 24 h), non-sustained or sustained ventricular tachycardia with a RBBB morphology (excluding the "fascicular pattern")
VI. Family history/genetics	*Major* • ACM confirmed in a first-degree relative who meets diagnostic criteria • ACM confirmed pathologically at autopsy or surgery in a first degree relative • Identification of a pathogenic or likely pathogenetic ACM mutation in the patient under evaluation *Minor* • History of ACM in a first-degree relative in whom it is not possible or practical to determine whether the family member meets diagnostic criteria • Premature sudden death (<35 years of age) due to suspected ACM in a first-degree relative • ACM confirmed pathologically or by diagnostic criteria in a second-degree relative	

ACM = arrhythmogenic cardiomyopathy; BSA = body surface area; EDV = end diastolic volume; EF = ejection fraction; ITF = International Task Force; LBBB = left bundle-branch block; LGE = late gadolinium enhancement; LV = left ventricle; RBBB = right bundle-branch block; RV = right ventricle; RVOT = right ventricular outflow tract.

Highly problematic



Super complex and probabilistic

'Is this the epsilon wave ?'

Not sensitive

Not useful in early disease

Not useful in forme fruste disease

Cannot be used to exclude ACM

Corrado et al. PMID PMID: **32561223**

Arrhythmogenic Cardiomyopathy – ACM

Non-dilated Left Venticular Cardiomyopathy (NDLVC)

"DCM without LV dilation"

"ALVC or Left dominant ARVC"

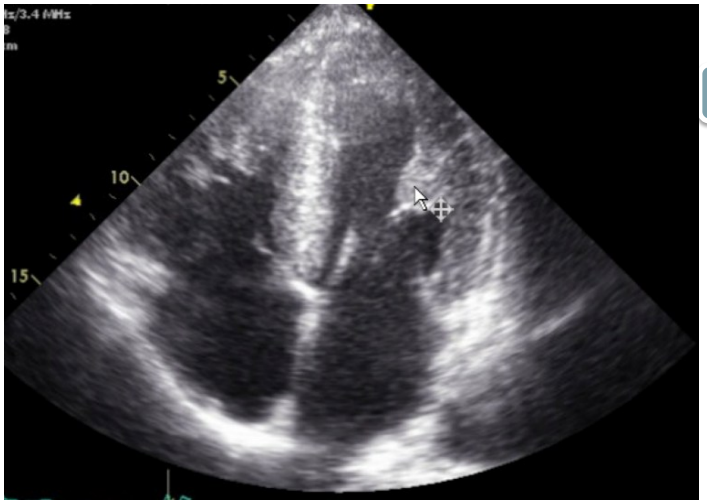
"Arrhythmogenic DCM not fullfilling ARVC criteria"

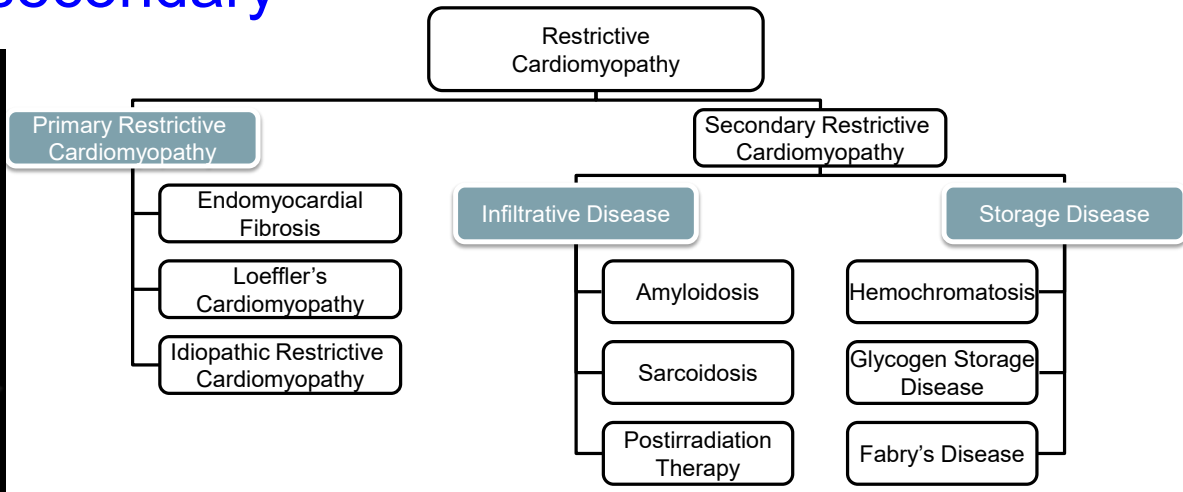
Arbelo et al. PMID: **37622657**

Non ischemic cardiomyopathies

Restrictive cardiomyopathy - RCM

Least common CMP (5% of all CMP)
Etiology: primary versus secondary





Clinical features

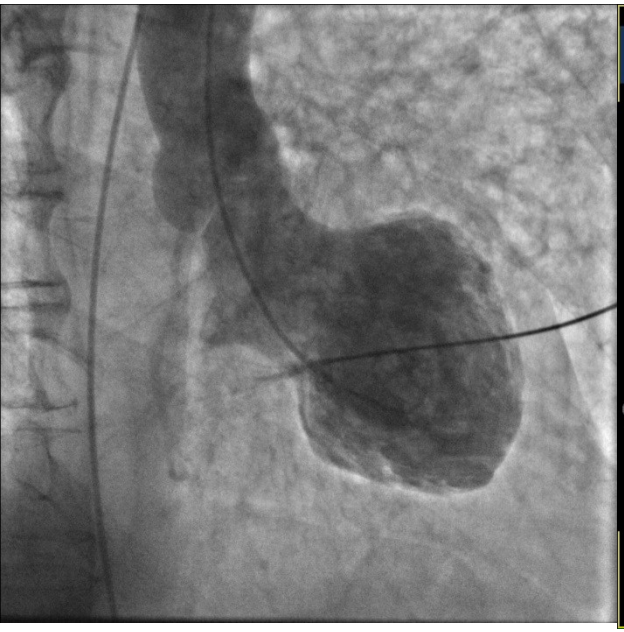
- Abnormal diastolic filling -> diastolic dysfunction
- Electro mechanical dissociation and conduction problems
- DD pericarditis constrictiva
- Atrial dilatation+++

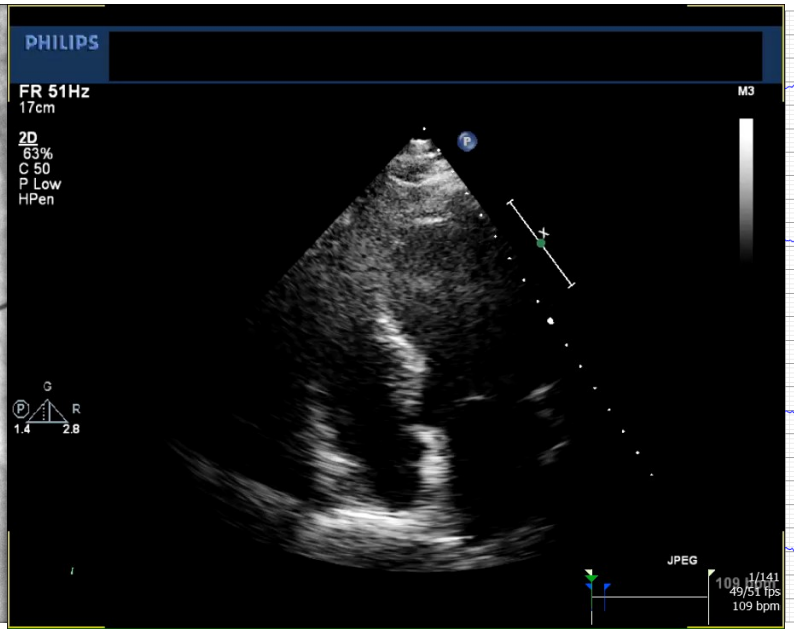
Non ischemic cardiomyopathies

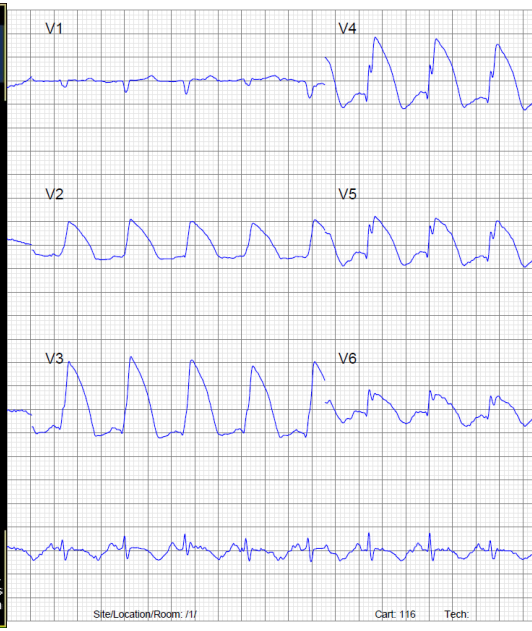
Unclassified

- Familial:
  - Left ventricular non compaction CMP (NCCM or LVNC)

- Acquired:
  - Takotsubo CMP







5. Electrical heart disease

5. Electrical disease - Arrhythmias

= Disruption of the normal impulse conduction

Too slow

Too fast

Loss of regularity

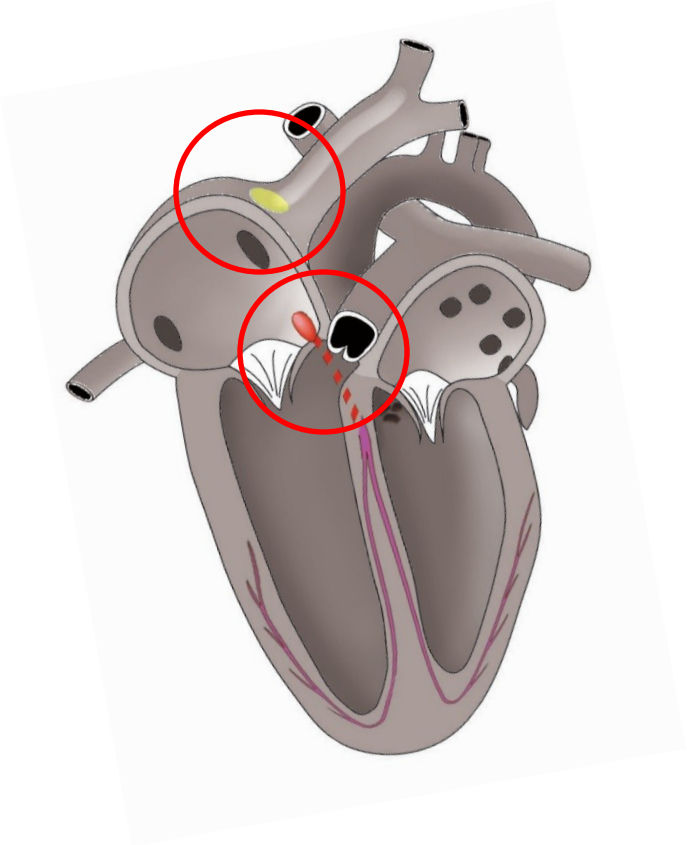
Abnormal origin/trajectory

5. Electrical disease - Arrhythmias

= Disruption of the normal impulse conduction

Too slow

- Sinus node disease
- AV nodal disease
- Combination



5. Electrical disease - Arrhythmias

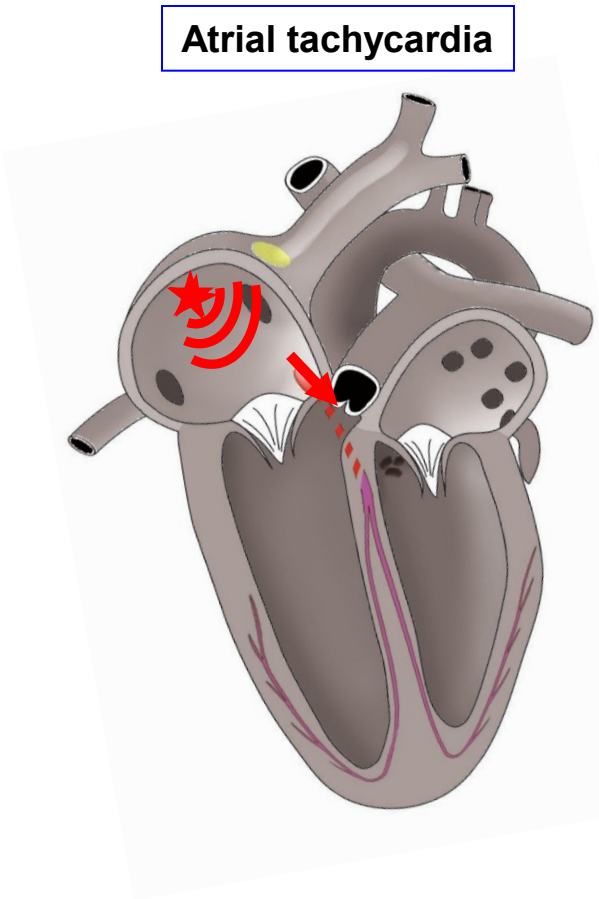
= Disruption of the normal impulse conduction

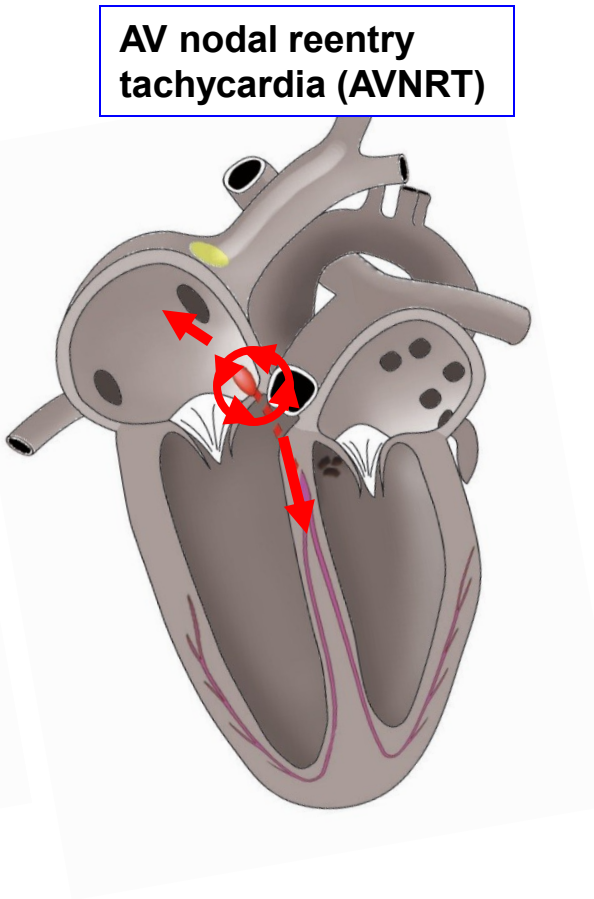
Too slow

- Sinus node disease
- AV nodal disease
- Combination

Too fast

- SVT





5. Electrical disease - Arrhythmias

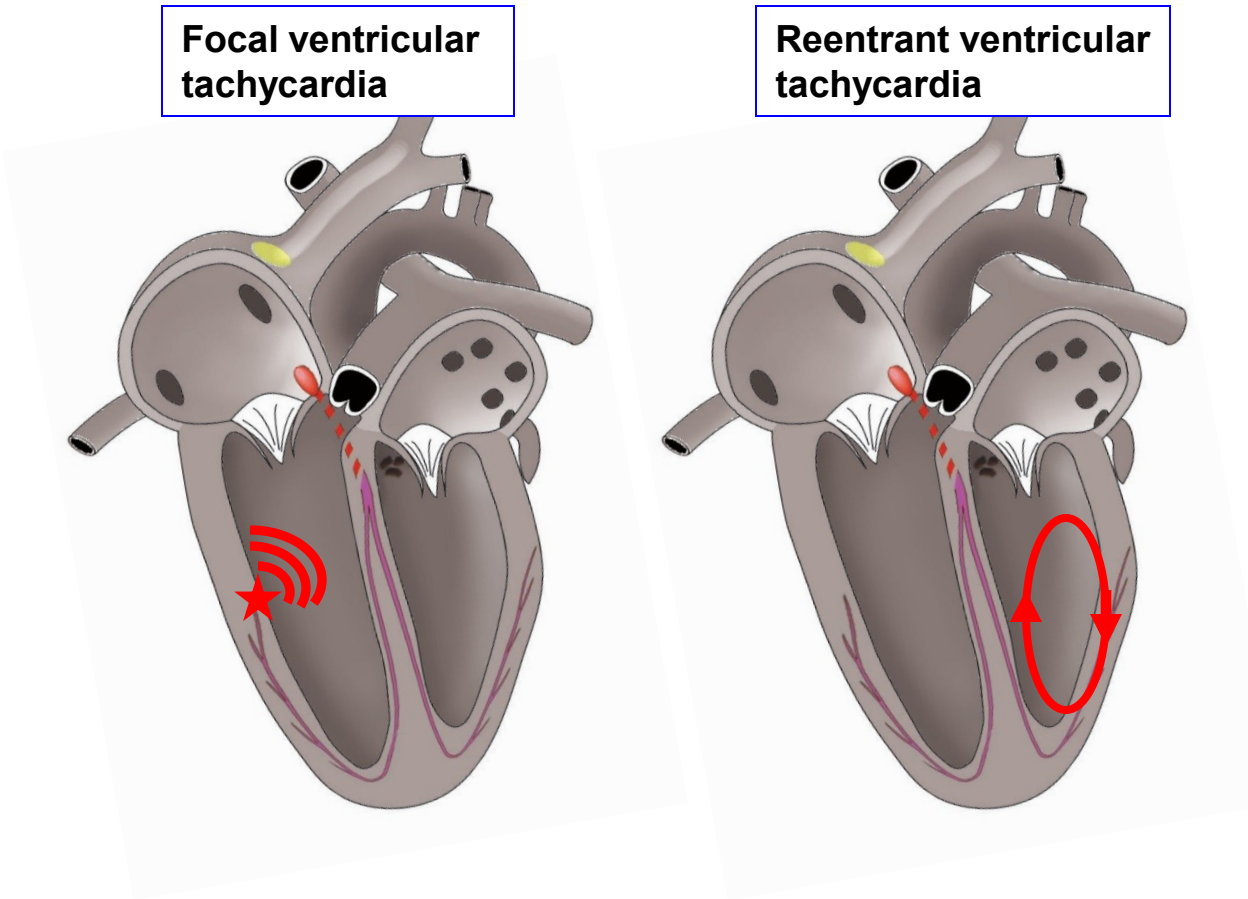
= Disruption of the normal impulse conduction

Too slow

- Sinus node disease
- AV nodal disease
- Combination

Too fast

- SVT
- VT



5. Electrical disease - Arrhythmias

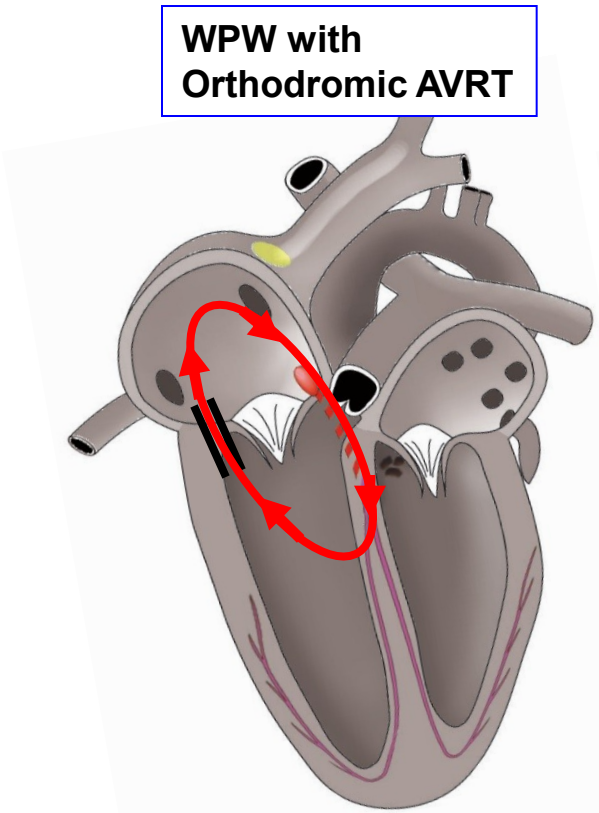
= Disruption of the normal impulse conduction

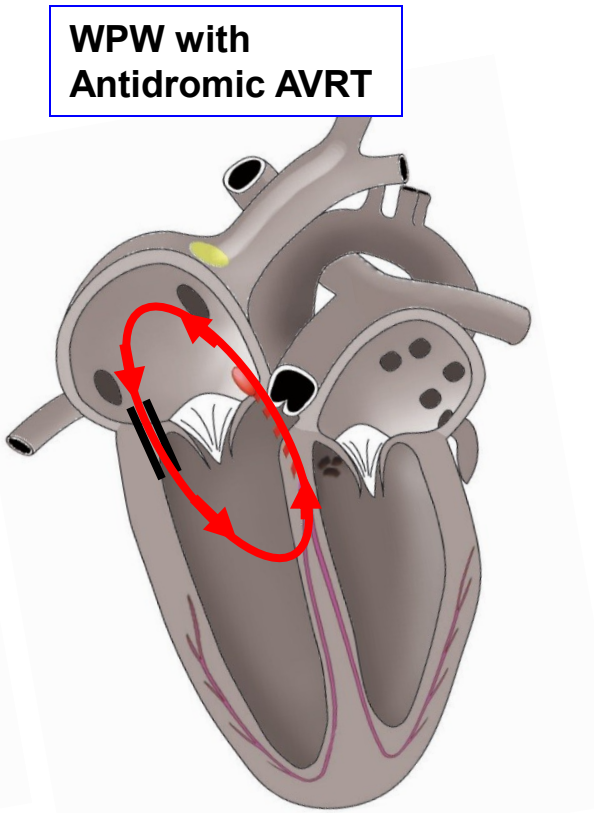
Too slow

- Sinus node disease
- AV nodal disease
- Combination

Too fast

- SVT
- VT
- AVRT





5. Electrical disease - Arrhythmias

= Disruption of the normal impulse conduction

Too slow

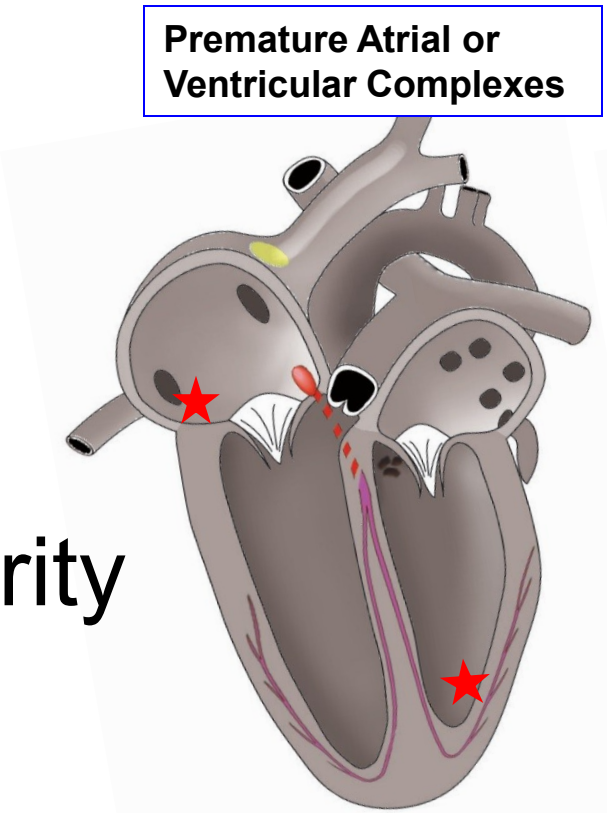
- Sinus node disease
- AV nodal disease
- Combination

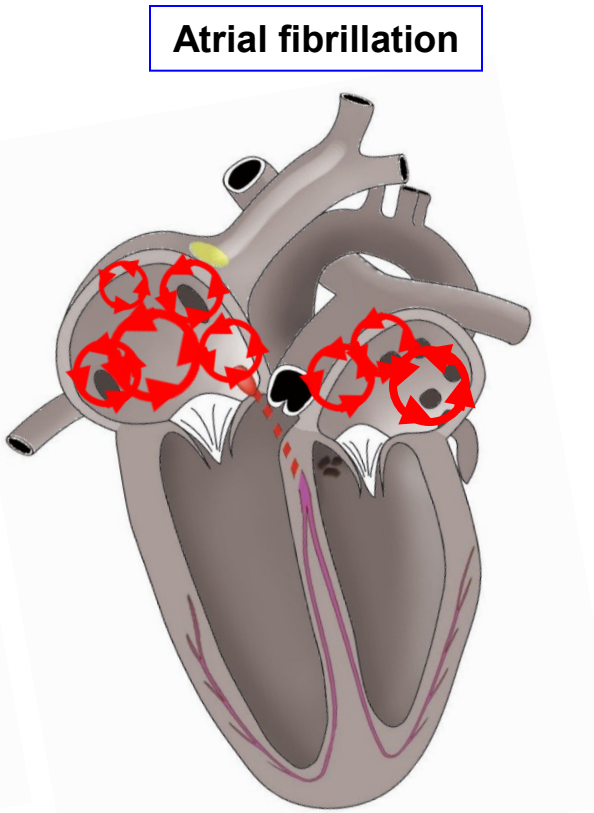
Too fast

- SVT
- VT
- AVRT

Loss of regularity

- PAC's / PVC's
- AF





5. Electrical disease - Arrhythmias

= Disruption of the normal impulse conduction

Too slow

- Sinus node disease
- AV nodal disease
- Combination

Too fast

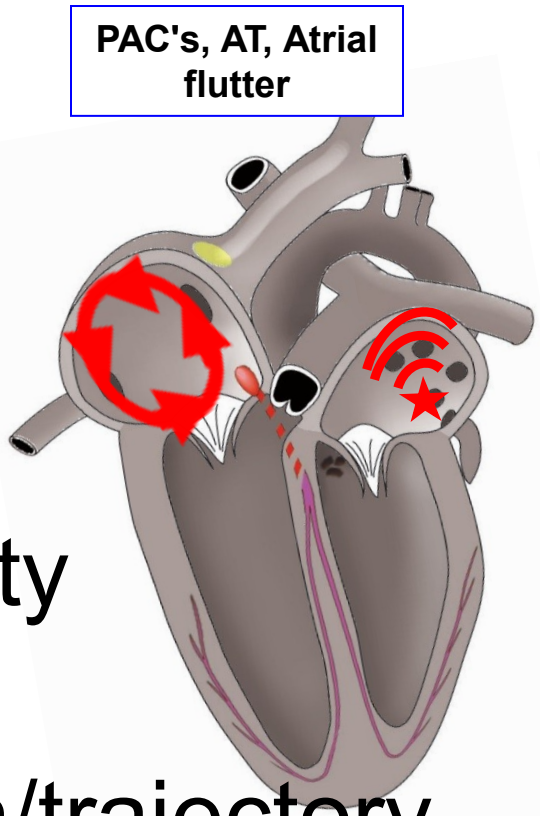
- SVT
- VT
- AVRT

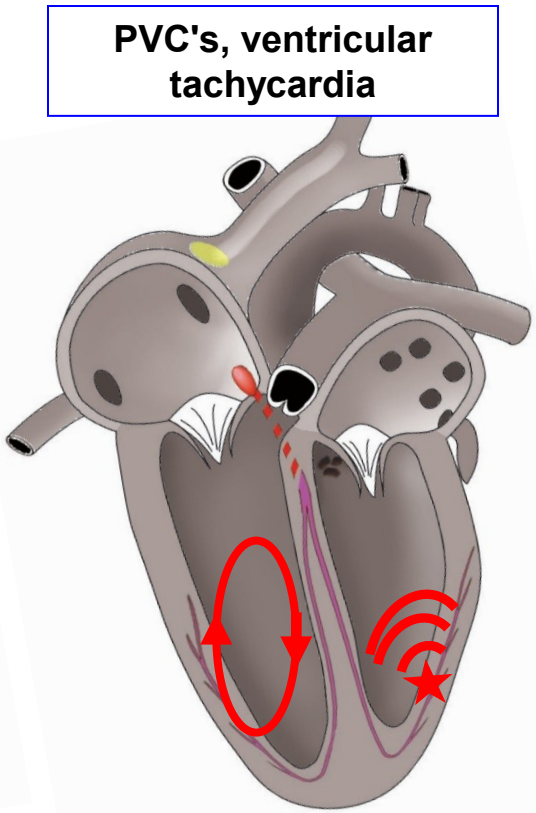
Loss of regularity

- PAC's / PVC's
- AF

Abnormal origin/trajectory

- PAC's, Atrial tachycardia, Atrial flutter
- AVRT
- PVC's, Ventricular tachycardia





5. Electrical disease - Arrhythmias

PED: LQTS, SQTS, BrS, ERS, CPVT, WPW, PCCD, IVF

Hereditary / Acquired / Both

Primary Electrical Diseases (PED):

- Long QT Syndrome (LQTS)
- Short QT Syndrome (SQTS)
- Brugada Syndrome (Brs)
- Early Repolarization Syndrome (ERS)
- Catecholaminergic Polymorphic VT (CPVT)
- Calcium Release Deficiency Syndrome (CRDS)
- Wolf-Parkinson-White (WPW)
- Progressive Cardiac Conduction Defect (PCCD)
- Idiopathic VF (IVF)
- Multifocal Ectopic Purkinje-Related Premature Contractions (MEPPC)

Thanks for your attention !

Morphologic classification is problematic

Morphologic classification is problematic

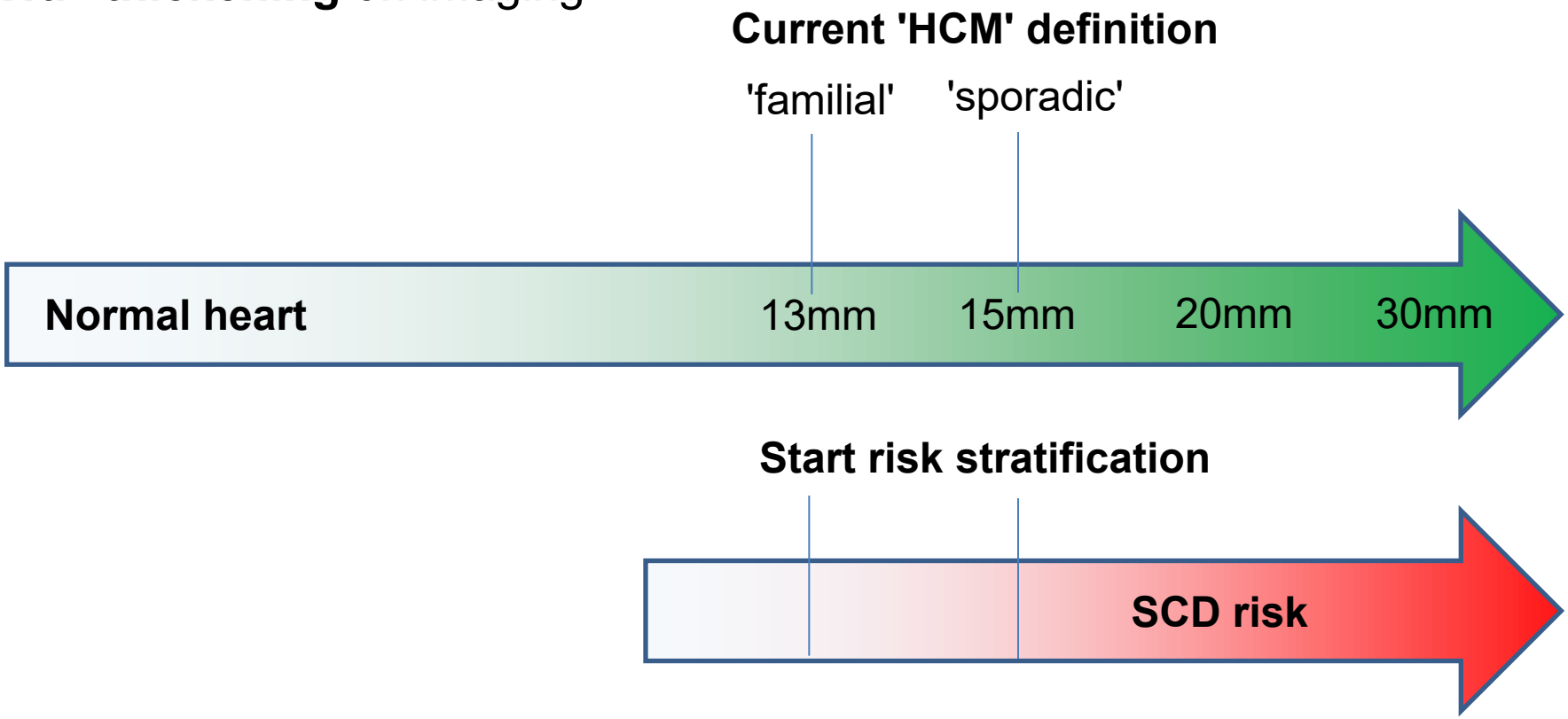
Hypertrophic cardiomyopahy:

*"End-diastolic **wall thickening** ≥13mm in familial or ≥15mm in sporadic cases, and in the absence of abnormal loading conditions (hypertensive, valvular)"*

Morphologic classification is problematic

Hypertrophic cardiomyopahy:

Wall thickening on imaging

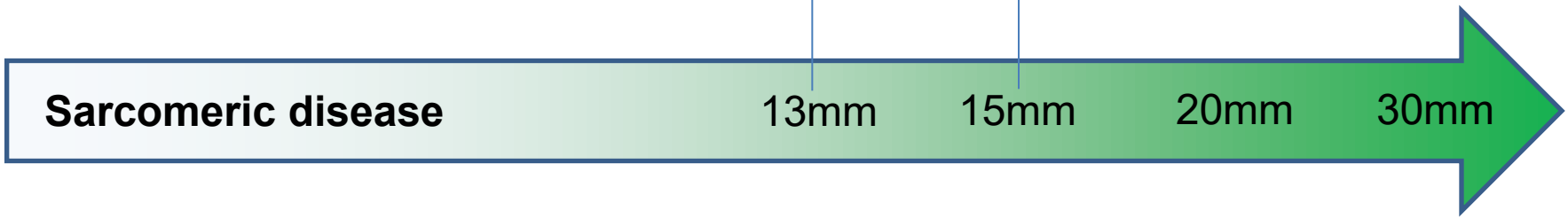


Morphologic classification is problematic

Hypertrophic cardiomyopahy:

Molecular disease of the sarcomere
Cellular hypertrophy
Cellular disaray
Interstitial fibrosis

Wall thickening on imaging



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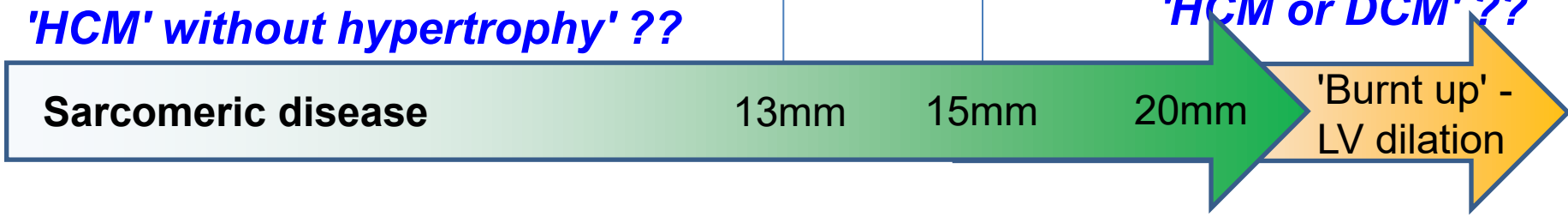

Morphologic classification is problematic

Hypertrophic cardiomyopahy:

Molecular disease of the sarcomere
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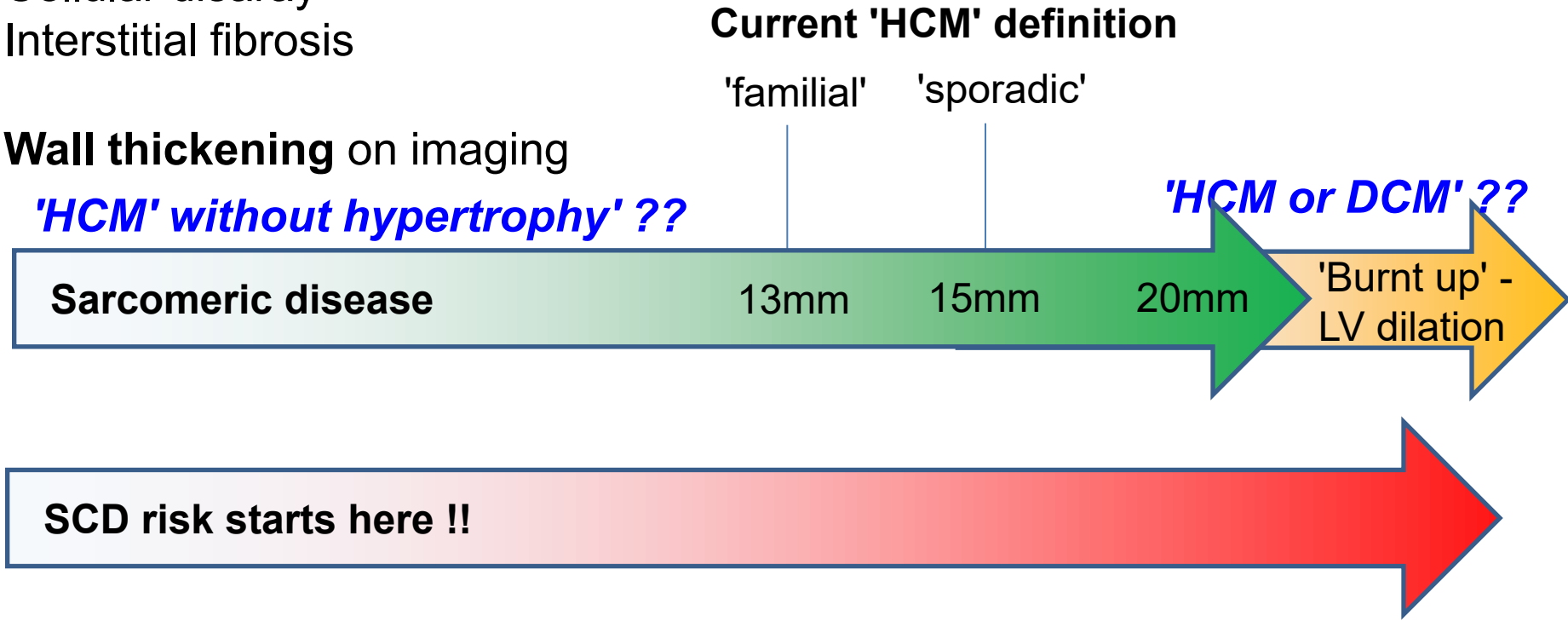
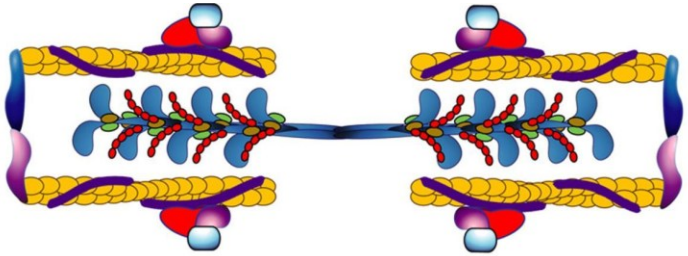
Wall thickening on imaging

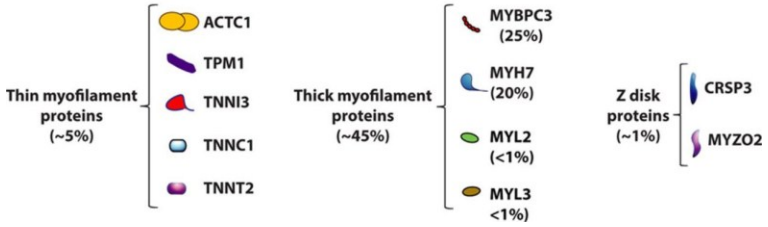


Morphologic classification is problematic

1. Molecular disease

Sarcomeric disease
Cellular hypertrophy
Cellular disaray
Interstitial fibrosis





2. Fenotype

Structural changes
Hypertrophy
Dilation
Restriction
Fibrosis

Electrical changes
Conduction disease
Dysrhythmia

3. Risk profile

Disease specific
Gene-specific
Mutation-specific

Family-specific

Follow-up
ILR
ICD

Marian et al, PMID: **28912181**