

THE
2ND DECADE



MYOTONIC
DYSTROPHY
FOUNDATION



2017 MDF Annual Conference

Sept. 8–9, 2017 San Francisco



MYOTONIC
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FOUNDATION

Care and a Cure

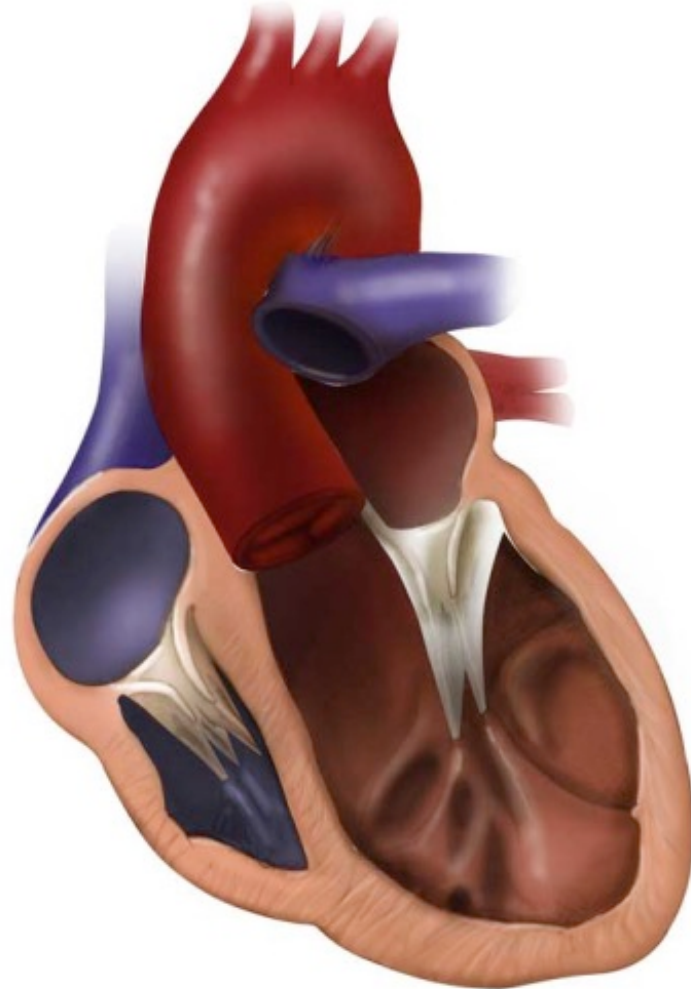
CARDIAC INVOLVEMENT IN MYOTONIC DYSTROPHY

Matthew Wheeler, MD PhD

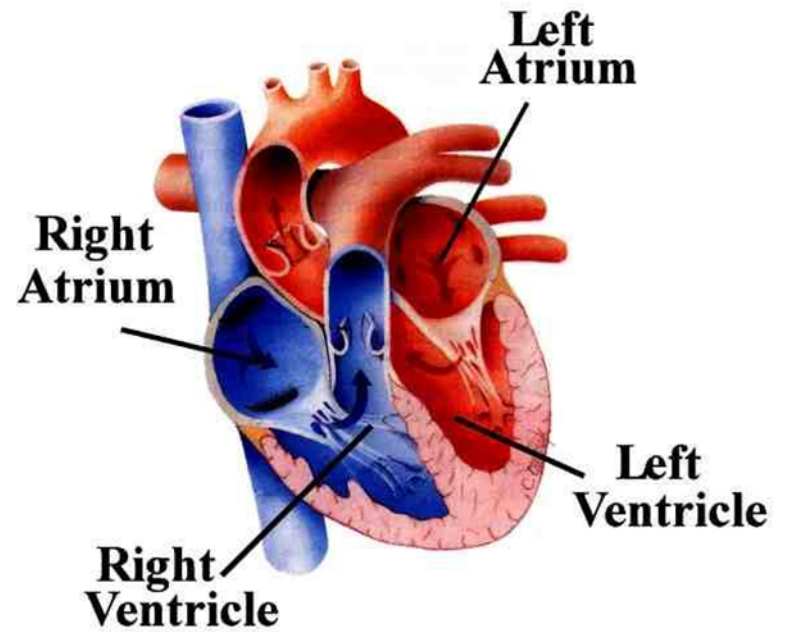
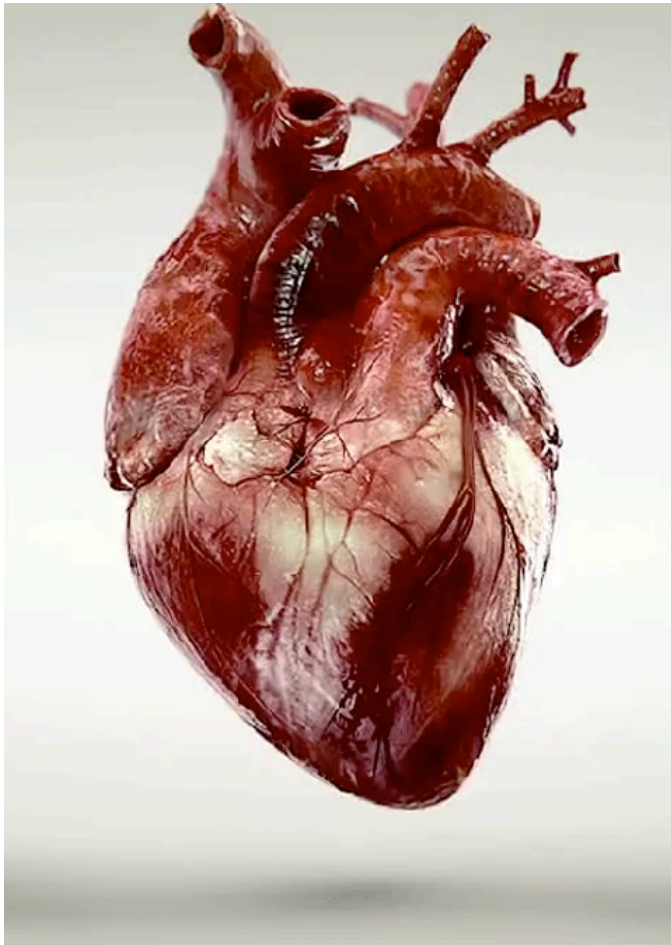


STANFORD CENTER
FOR INHERITED
CARDIOVASCULAR DISEASE

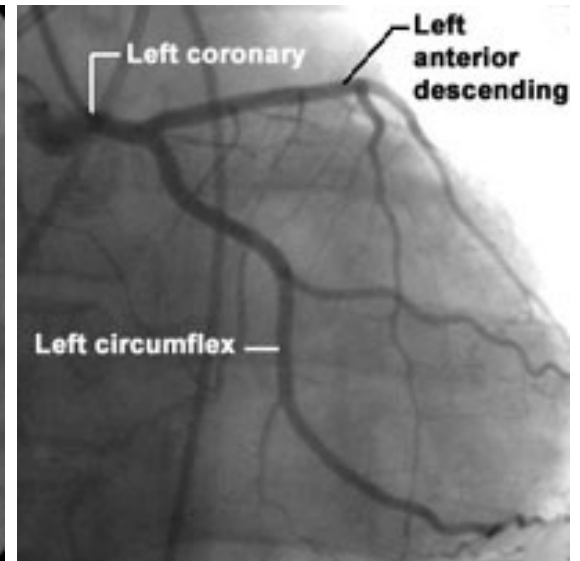
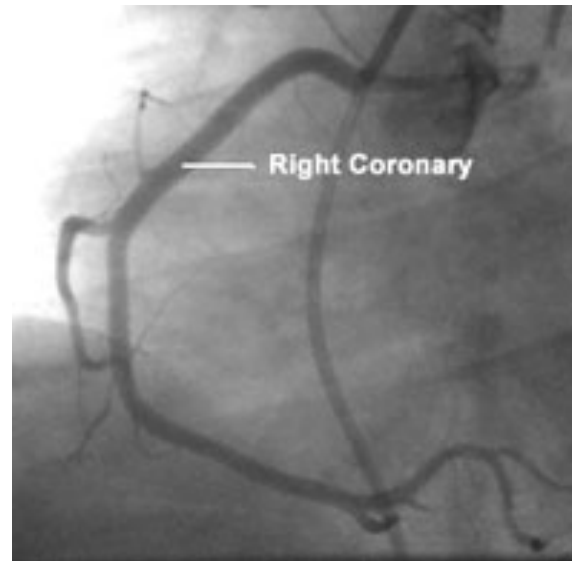
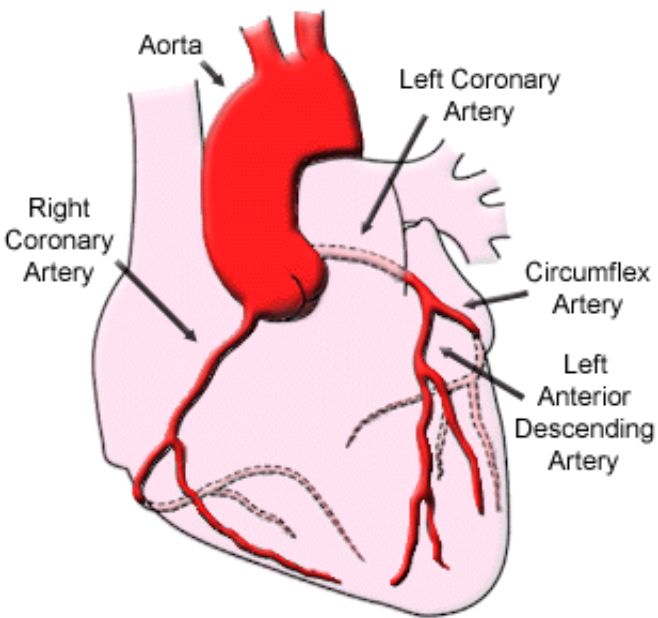
The Heart



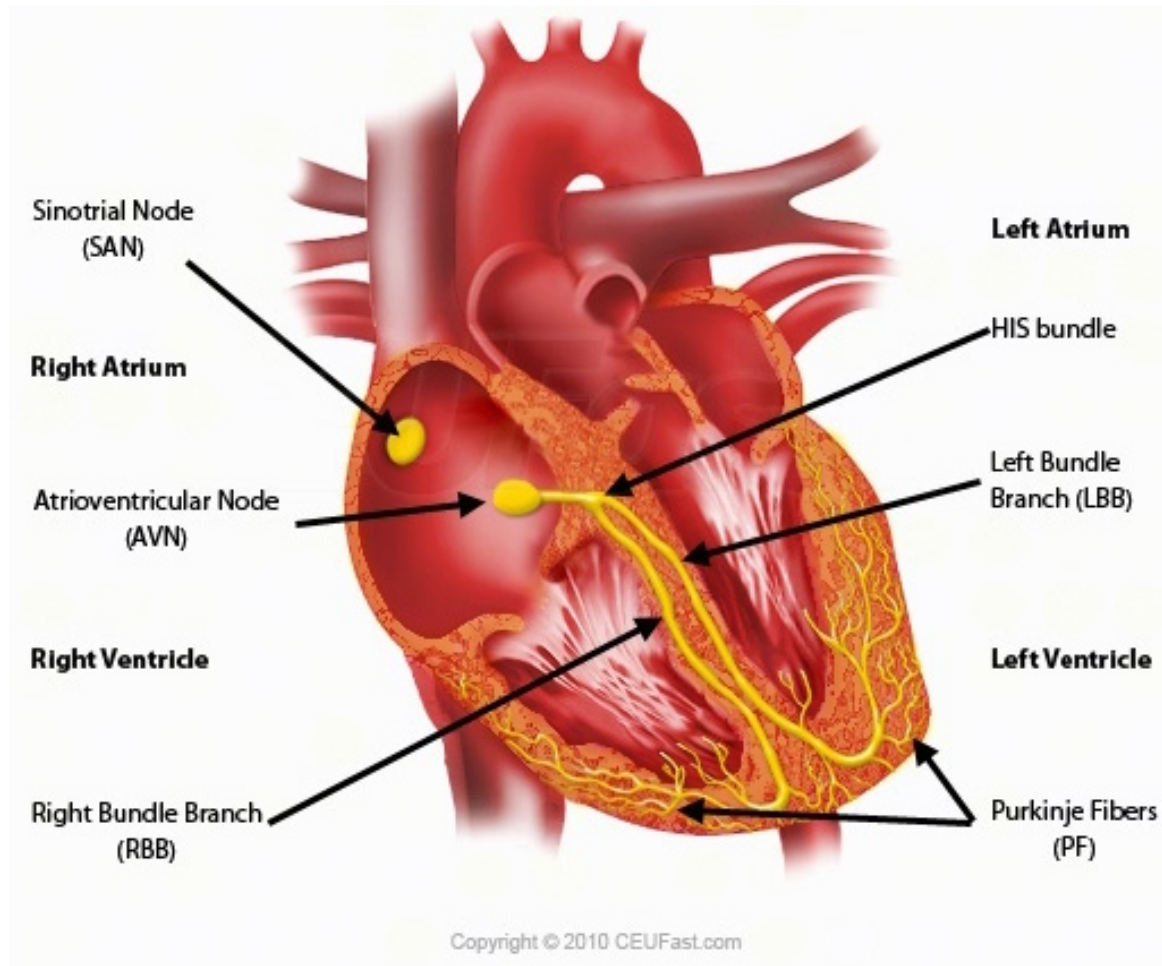
Pump



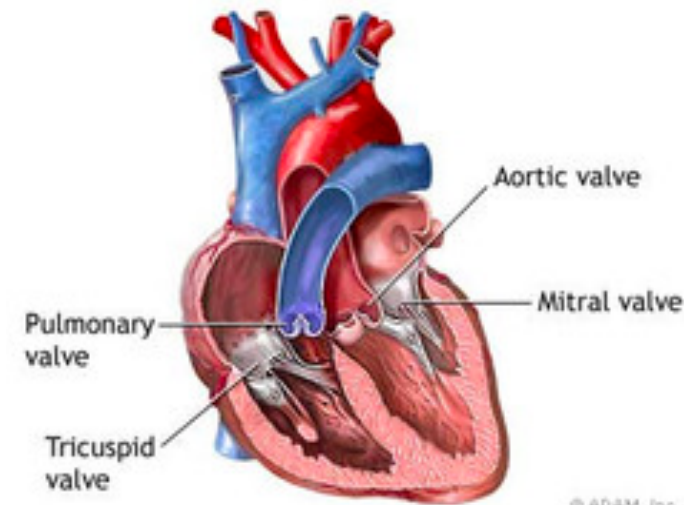
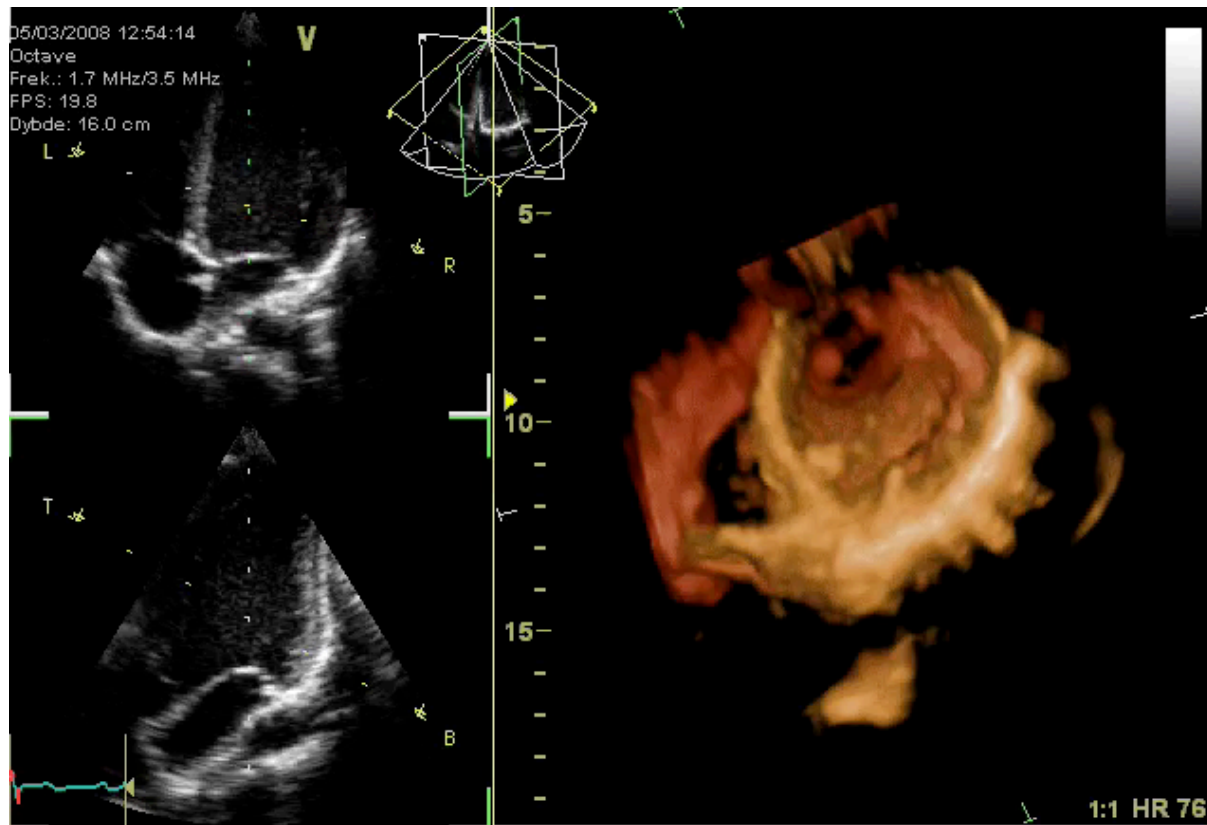
Fuel supply



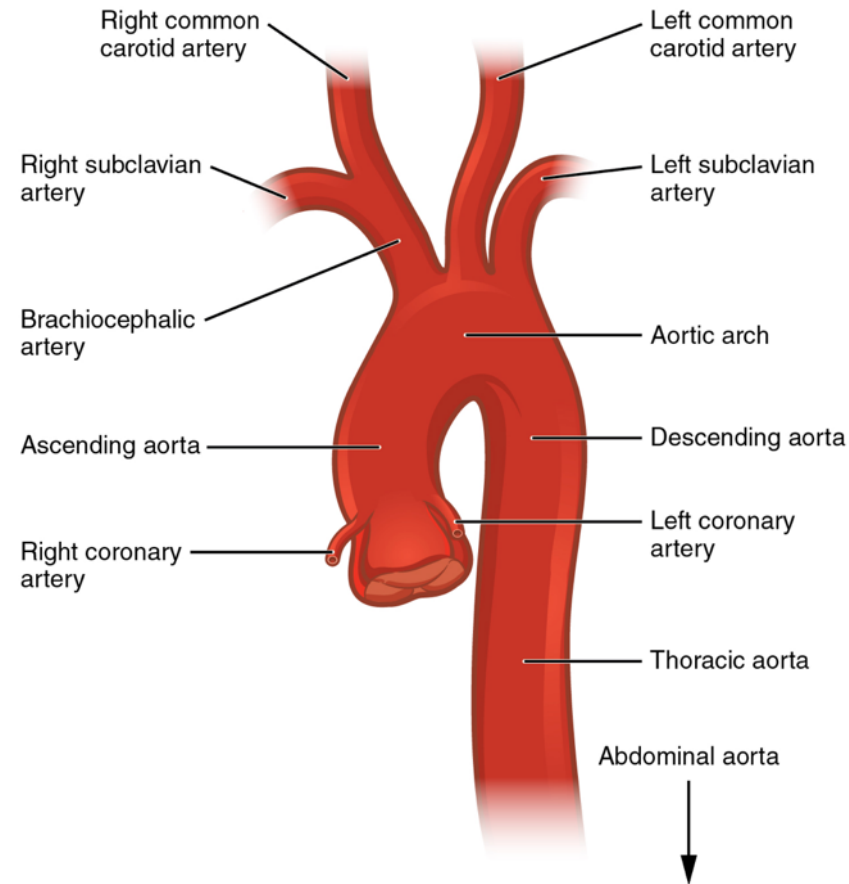
Electrical system



Valves



Vessels



Symptoms that may be the heart

- ❑ Skipped beats
- ❑ Lightheadedness
- ❑ Chest pain
- ❑ Nausea
- ❑ Exertional intolerance
- ❑ Shortness of breath
- ❑ Cough
- ❑ Fatigue

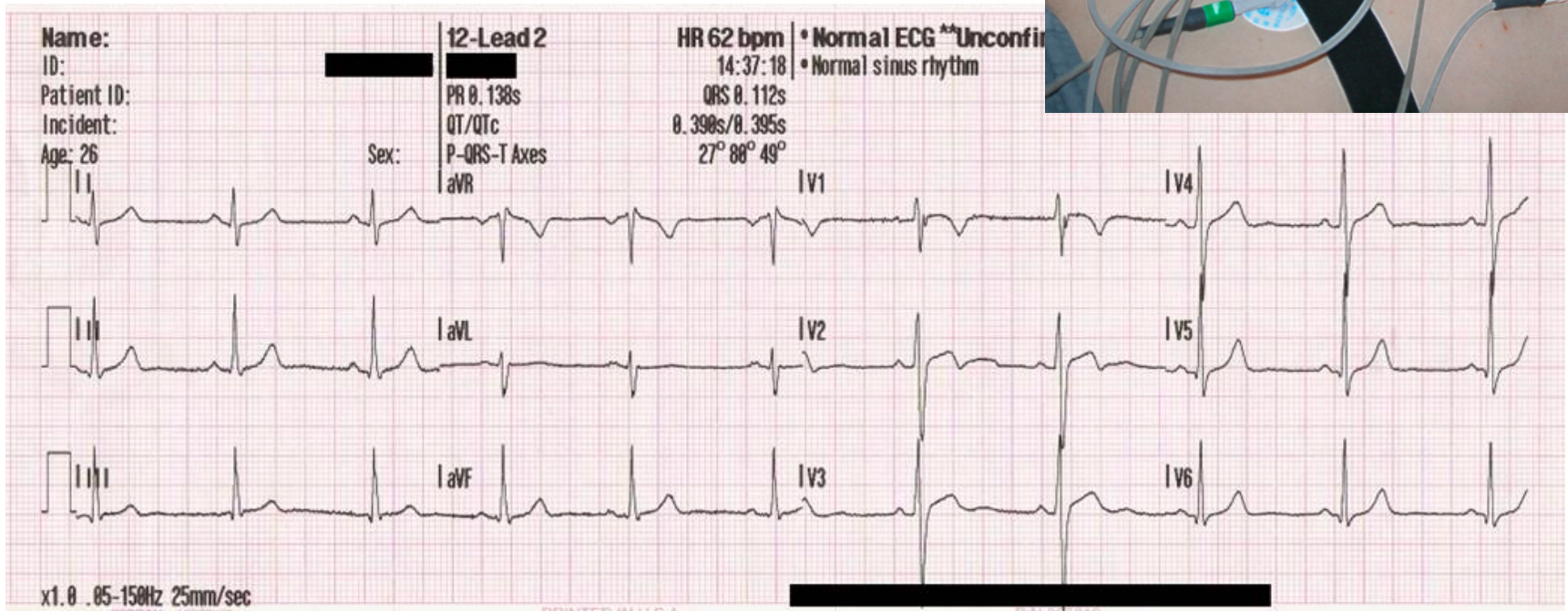


Signs

- ❑ Slow heart beat
- ❑ Fast heart beat
- ❑ Irregular heart beat
- ❑ Abnormal blood pressure
- ❑ Coarse breath sounds
- ❑ Swelling in legs
- ❑ Pallor
- ❑ Clammy skin



Tests - ECG



ECG = electrocardiogram

aka EKG, 12 lead electrocardiogram

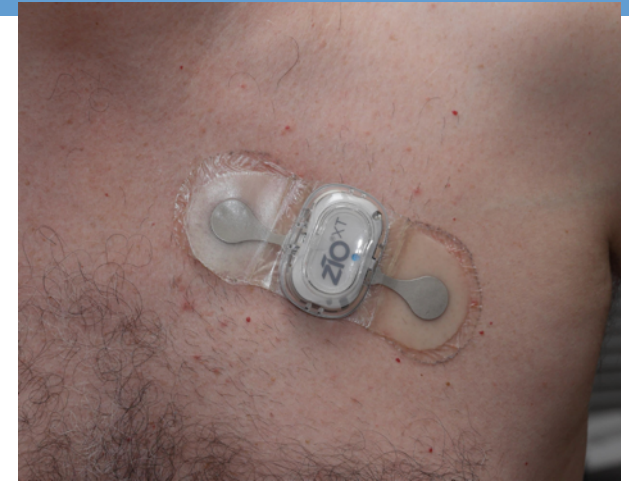
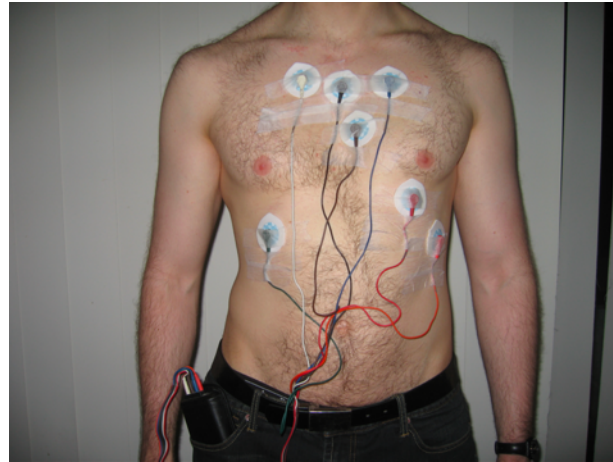
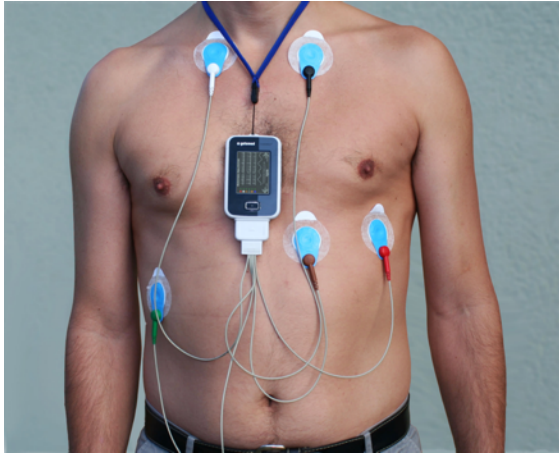
Tests - Echo



echo = echocardiogram

aka TTE = rest echocardiogram = ultrasound of heart

Tests - Rhythm monitor



Holter ~ rhythm monitor ~ event monitor ~ patch monitor ~ ambulatory telemetry

Additional tests

Routine

- Lipid/cholesterol panel
- Blood pressure
- Sleep study

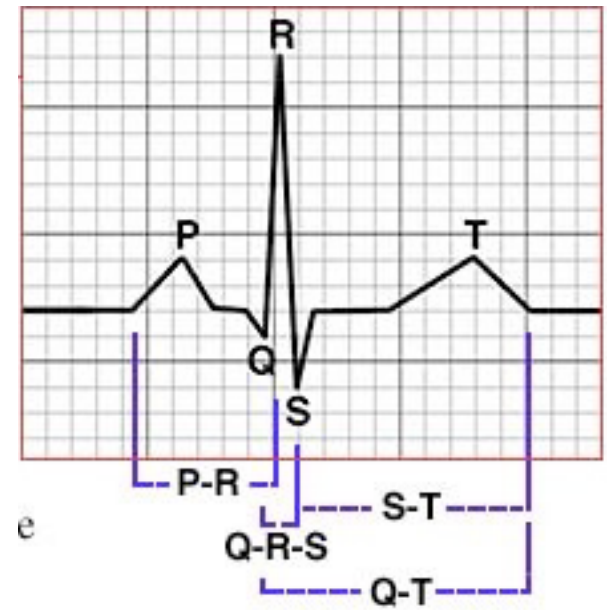
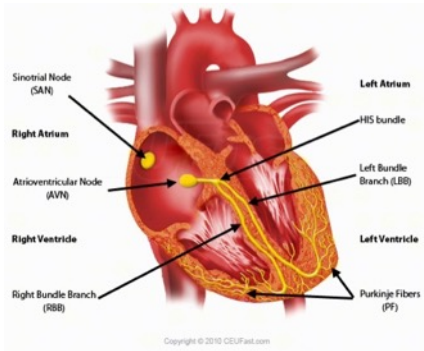
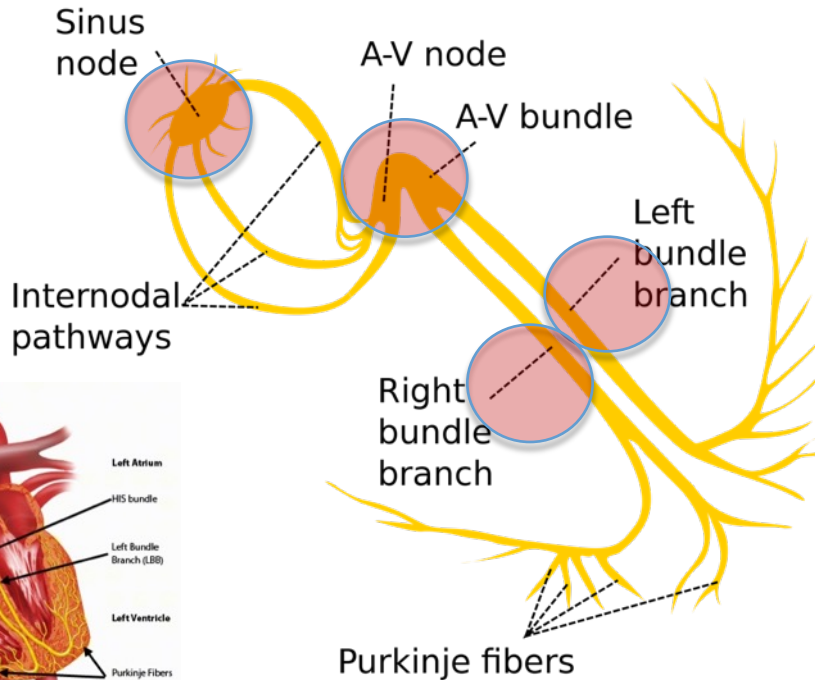
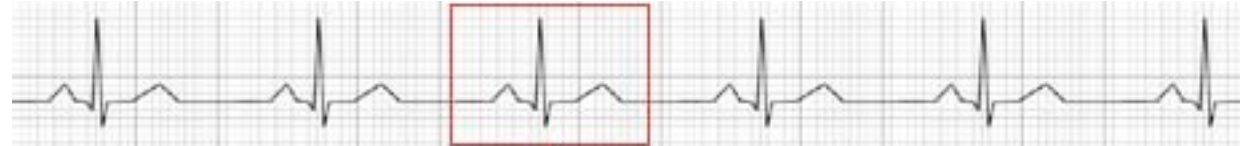
In some

- Stress test
- Cardiac MRI
- Electrophysiology study

Common cardiac manifestations in myotonic dystrophy

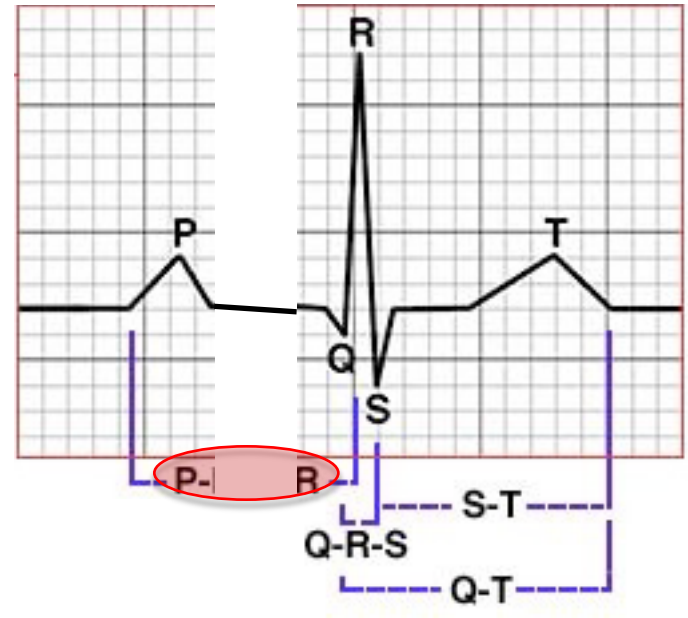
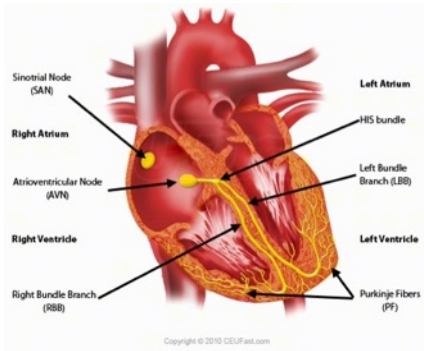
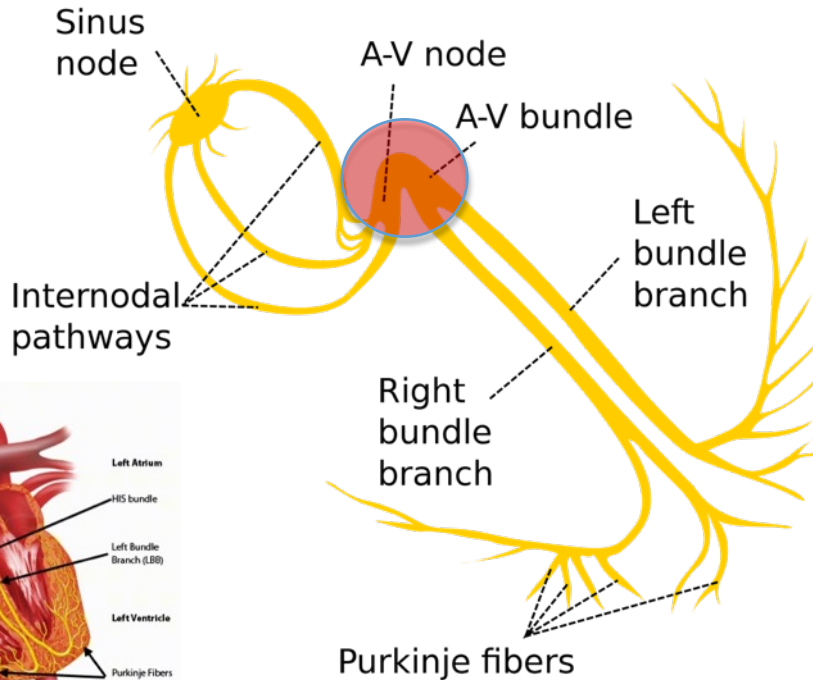
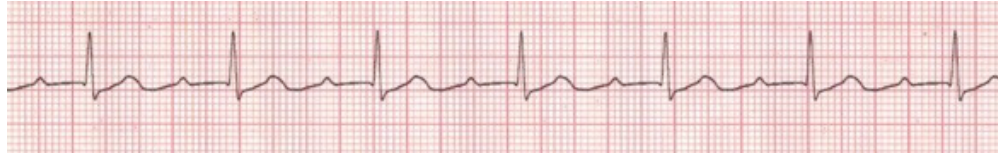
- Conduction delay
- Arrhythmias
- Pump dysfunction

Conduction delay



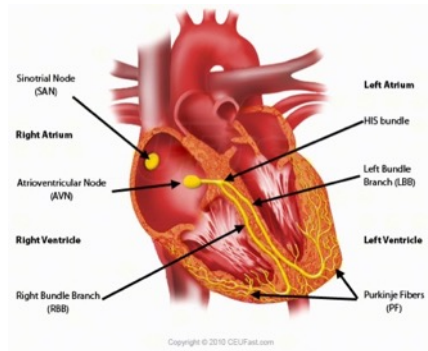
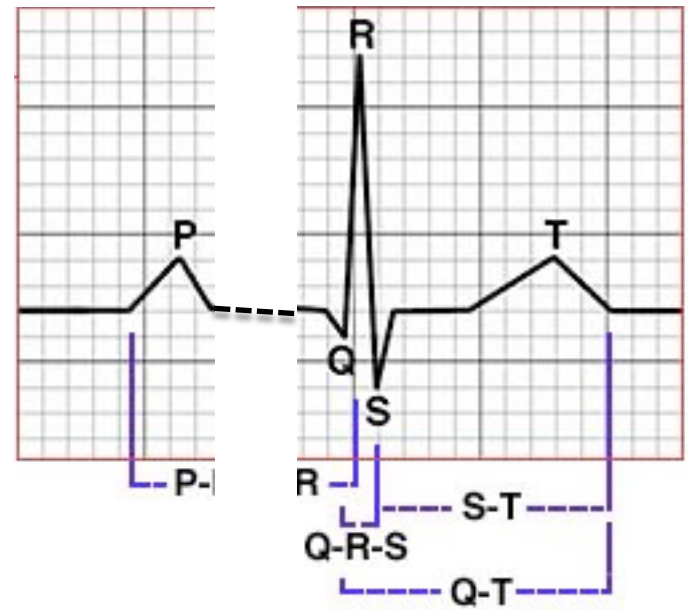
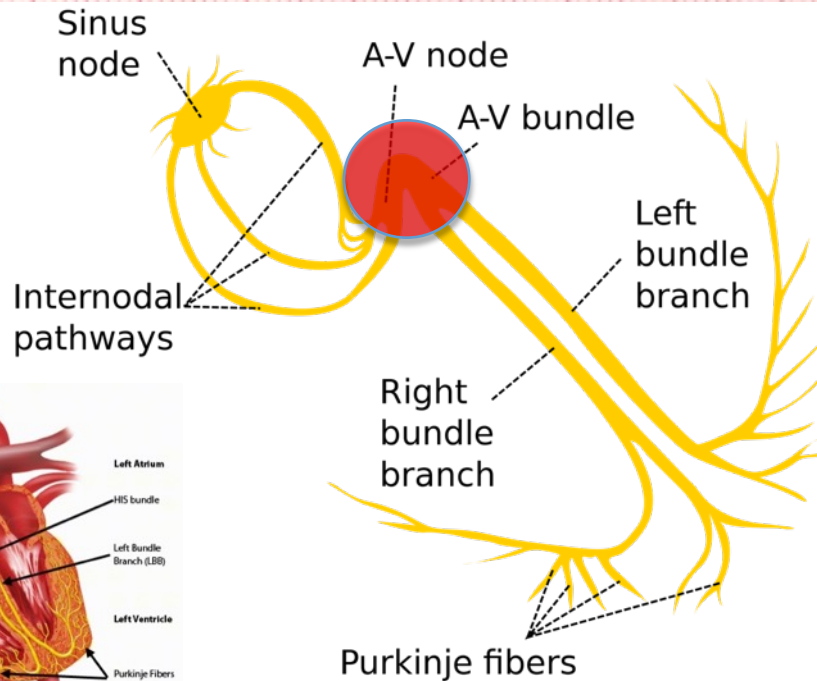
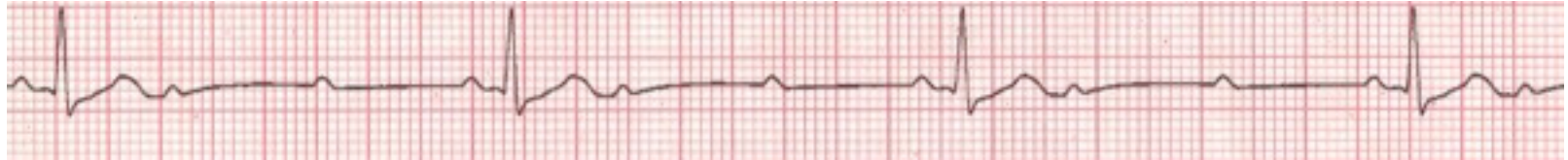
Significant AV delay

First degree AV block



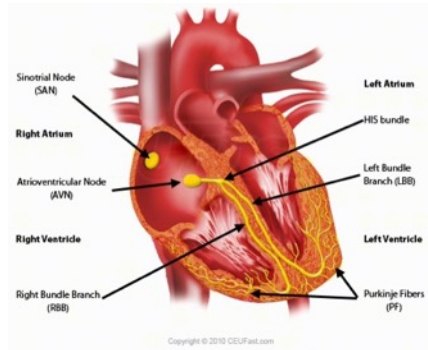
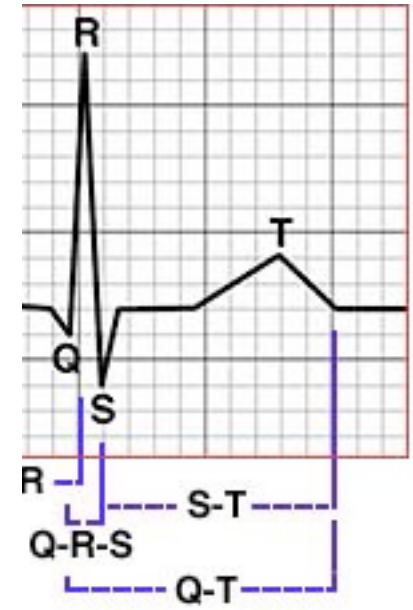
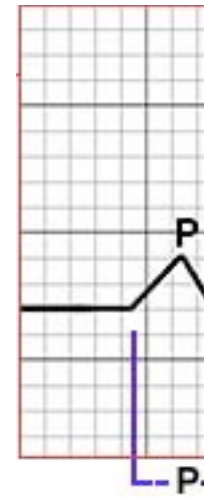
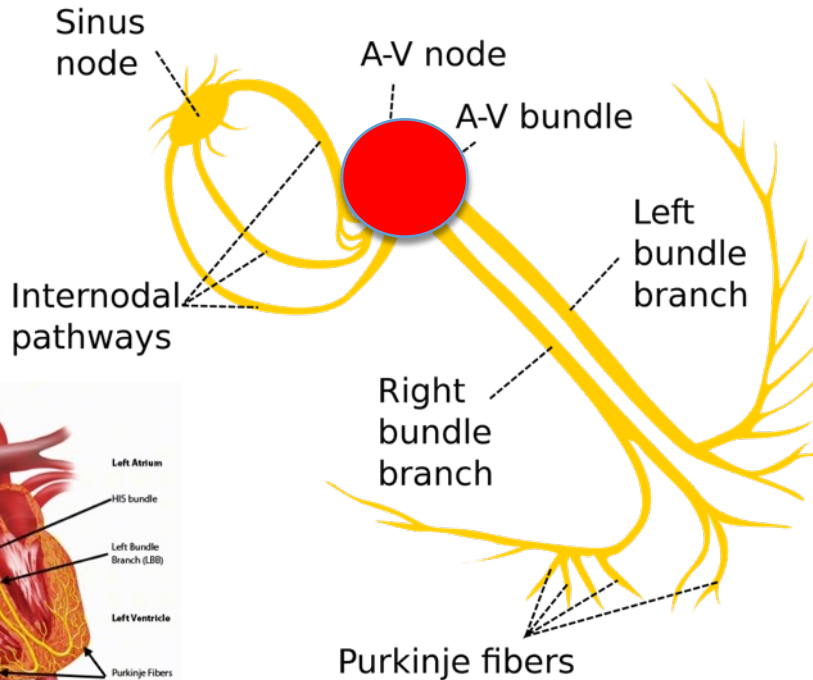
Significant AV delay

Second degree AV block



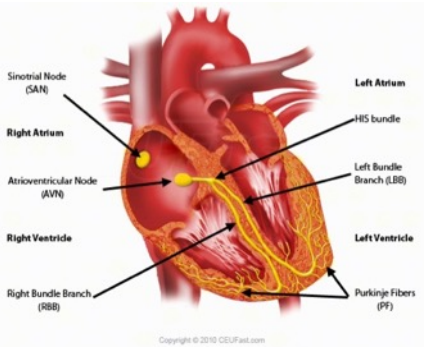
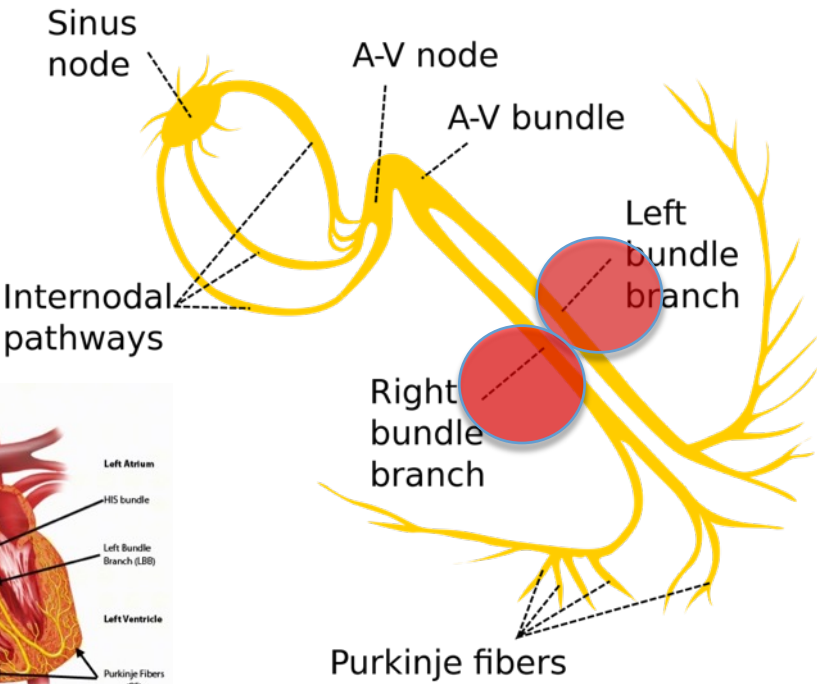
Significant AV delay

Complete AV block

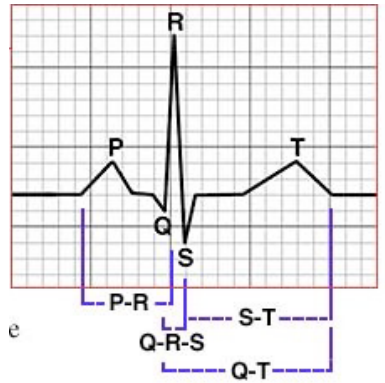


Significant intraventricular conduction delay

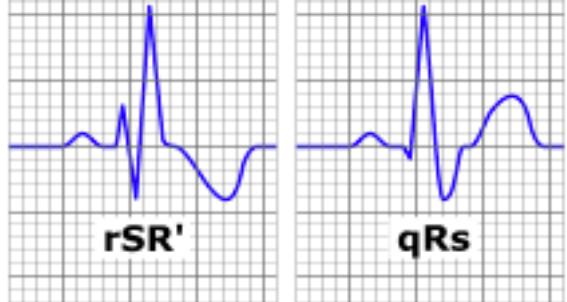
Right or left bundle branch block (RBBB or LBBB)



normal



RBBB

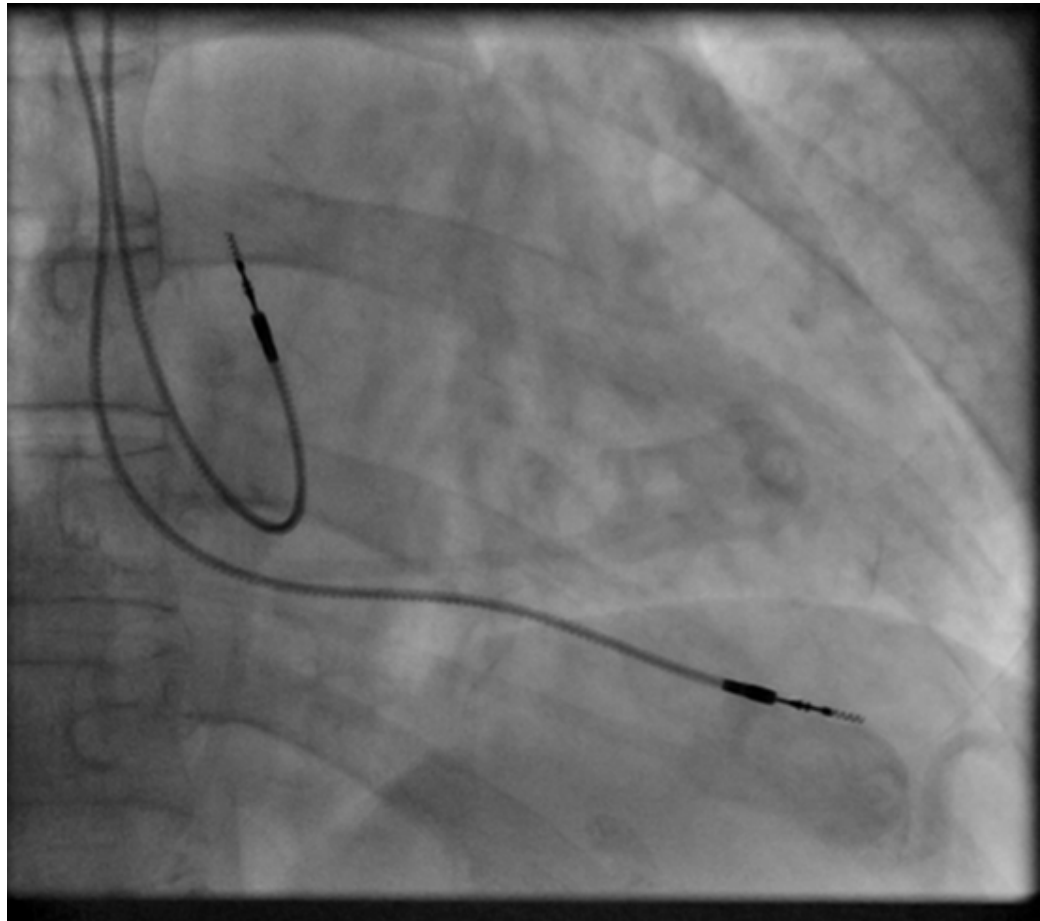


LBBB



Significant conduction delay treatment

Pacemaker



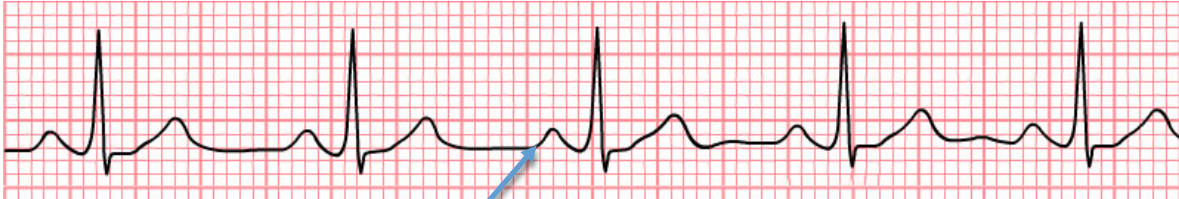
Conduction delay is common in Myotonic Dystrophy

- Most registries ~50% lifetime risk of developing significant conduction disease
- Predictors of significant conduction disease
 - Age (older)
 - Age of onset (younger)
 - Severity of muscular disease
 - Mutation size
 - Nonsignificant conduction disease

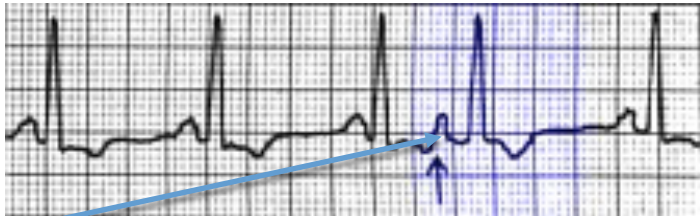
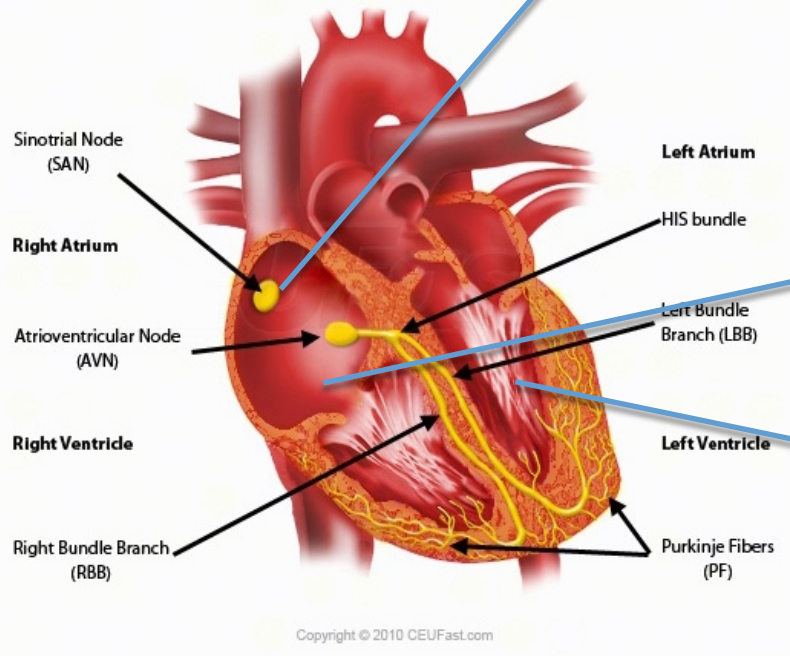
Conduction delay in DM1

- Study by Groh and colleagues New England Journal of Medicine 2008
- Conduction delay (even asymptomatic) associated with risk of cardiac death
 - PR interval >240 ms
 - QRS interval >120 ms
 - 2nd or 3rd degree AB block
- Implication: recommend device placement in DM1 patients with conduction delay using above thresholds

Benign rhythms



normal sinus rhythm

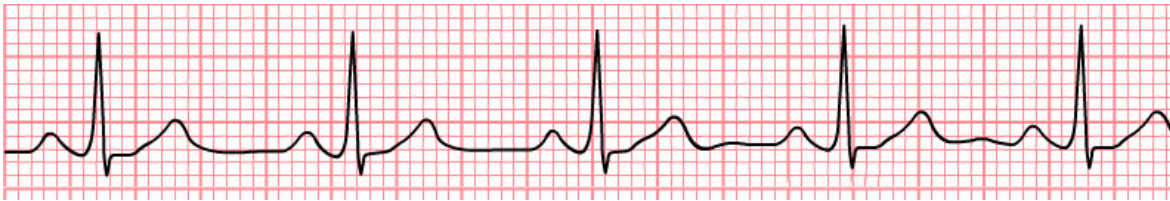


PAC

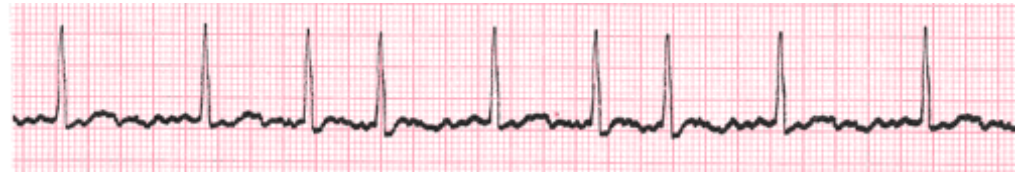
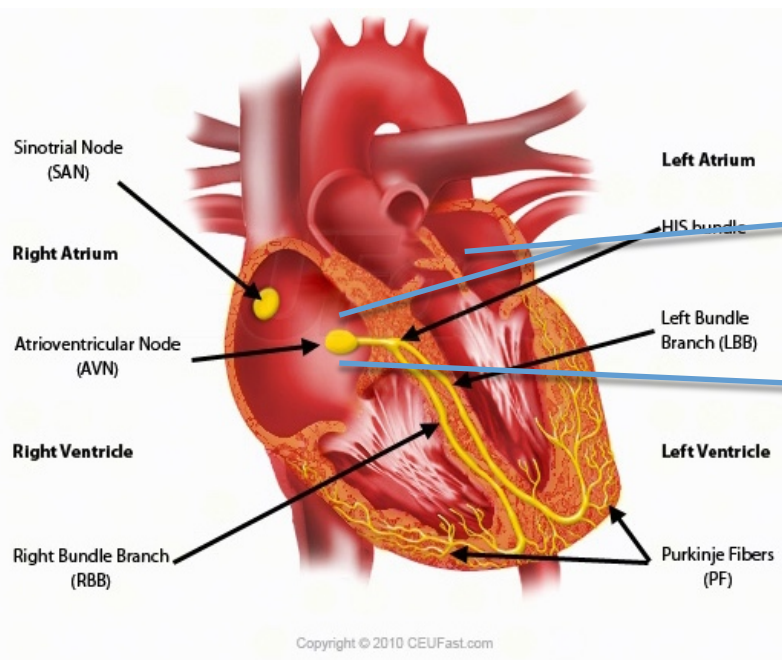


PVC

Atrial arrhythmias



normal sinus rhythm



Atrial fibrillation

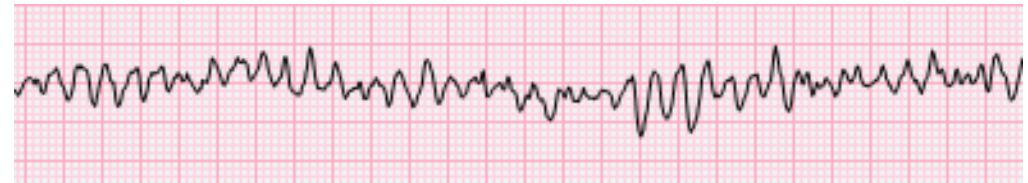
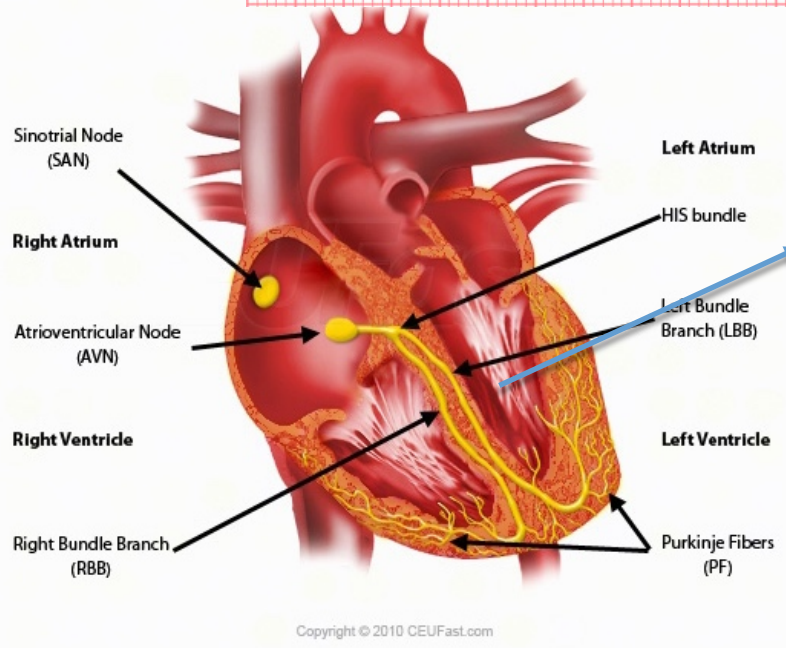


Atrial flutter

Ventricular arrhythmias



normal sinus rhythm



Ventricular fibrillation



Ventricular tachycardia

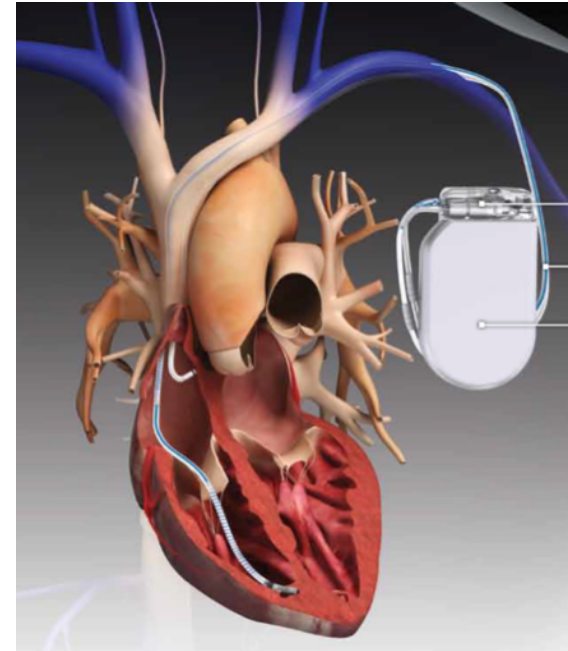
Treatment of atrial arrhythmias

- Anticoagulation
- Rate control (caution if no pacemaker)
- Rhythm control
- Ablation
- Device placement

Treatment of ventricular arrhythmias

- Cardioversion/CPR
- Emergency medical services
- Device placement –
implantable cardioverter defibrillator

In some: antiarrhythmics, ablation

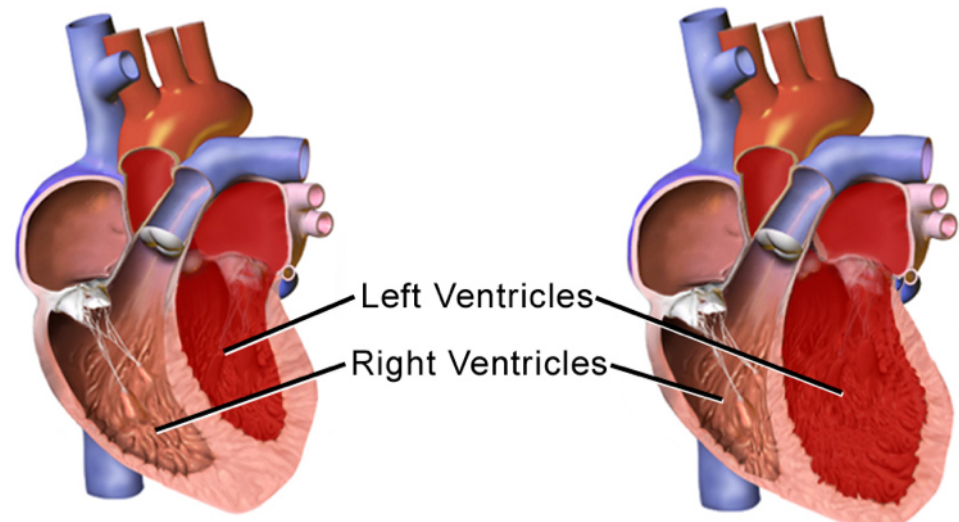


Arrhythmias in DM1

- Study by Groh and colleagues New England Journal of Medicine 2008; others since
- Arrhythmias(even asymptomatic) associated with risk of cardiac death
- Implication: recommend device placement in DM1 patients with arrhythmia

Pump dysfunction in DM1

- Up to 20% of patients will have pump dysfunction
- Most asymptomatic
- Severe pump dysfunction – symptomatic heart failure



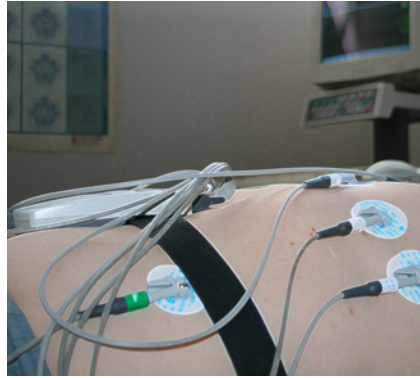
Treatment of pump dysfunction in DM1

- Medications known to be effective in other forms of pump dysfunction
- Main limitations are treatment effects
 - Lower blood pressure
 - Slow heart rate
 - Slow conduction
- Biventricular pacemaker may help reduce pump dysfunction

Cardiac screening in DM1

Recommendations:

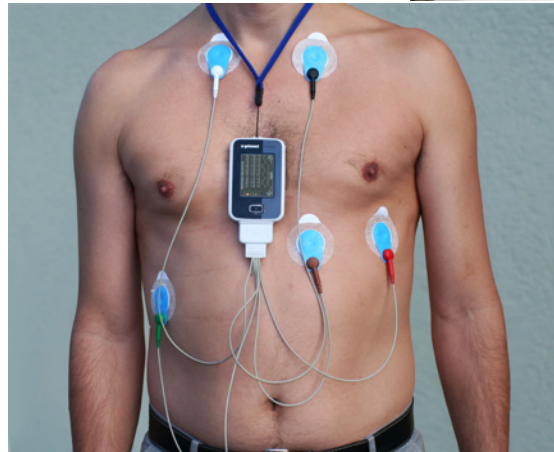
Electrocardiogram



Echocardiogram



Rhythm monitoring



ECG screening

- Evaluation for conduction system disease
 - Atrio-ventricular delay
 - Intraventricular conduction delay
- Rhythm changes
 - Atrial rhythm
 - Atrial fibrillation



Rhythm monitoring

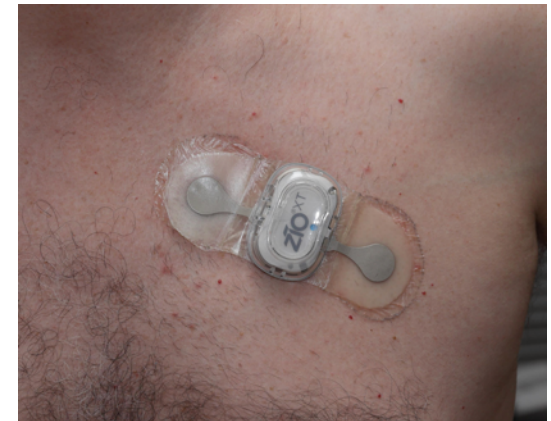
At least 24 hour rhythm monitor with ECG

We use extended rhythm monitoring, typically 14 days

Evaluate for evidence of intermittent heart block, atrial fibrillation, ventricular arrhythmias

Rhythm monitoring at minimum every 3 years

Indicated sooner if symptoms of passing out, syncope, severe palpitations



Structural evaluation

Rest echocardiogram at minimum every 3 years

Evaluation of ventricular size, thickness, function

Increase frequency of screening if decrement in function or significant intraventricular conduction delay

Cardiac MRI may provide incremental information



Screening results

- ECG
 - Normal
 - Borderline
 - Abnormal, not meeting criteria for pacemaker
 - Abnormal, meeting criteria for pacemaker/defibrillator
 -
- Result informs
 - treatment plan
 - repeat screening interval



Screening results

- Echocardiogram
 - Normal
 - Borderline, consider medication
 - Abnormal, medication recommended
 - Abnormal, meeting criteria for pacemaker and medication
- Result informs
 - treatment plan
 - repeat screening interval
 - additional testing



Open questions in DM1

- What is best approach to treating hyperlipidemia?
- What is risk of coronary artery disease, how best to screen?
- What is best timing of pacemaker/defibrillator?
- Can mexiletine or other sodium channel blockers be used safely for neuromuscular symptom management?
 - With ICD in place?
 - Without ICD in place?