CARDIAC INVOLVEMENT IN MYOTONIC DYSTROPHY

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The Heart
Pump
Fuel supply
Electrical system

- Sinotrial Node (SAN)
- Right Atrium
- Atrioventricular Node (AVN)
- Right Bundle Branch (RBB)
- Left Atrium
- HIS bundle
- Left Bundle Branch (LBB)
- Left Ventricle
- Purkinje Fibers (PF)
Valves
Vessels

- Right common carotid artery
- Left common carotid artery
- Right subclavian artery
- Left subclavian artery
- Brachiocephalic artery
- Aortic arch
- Ascending aorta
- Descending aorta
- Right coronary artery
- Left coronary artery
- Thoracic aorta
- Abdominal aorta
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

- Sinus node
- Internodal pathways
- A-V node
- A-V bundle
- Left bundle branch
- Right bundle branch
- Purkinje fibers
Significant AV delay
First degree AV block
Significant AV delay
Second degree AV block
Significant AV delay
Complete AV block
Intraventricular conduction delay

- Sinus node
- Internodal pathways
- A-V node
- A-V bundle
- Left bundle branch
- Right bundle branch
- Purkinje fibers
Significant intraventricular conduction delay
Right or left bundle branch block (RBBB or 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

Electrocardiographic Abnormalities and Sudden Death in Myotonic Dystrophy Type 1
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

Electrocardiographic Abnormalities and Sudden Death in Myotonic Dystrophy Type 1

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?