Myotonic Dystrophy and The Heart

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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 absorption micro CT scanner

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ECG/EKG

Electrocardiogram (ECG)
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
Intervals
FORCES

- Ventricular hypertrophy

left sided
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 function
Echocardiography function
MRI for cardiac function/morphology

Free breathing cine imaging LV short axis stack
Catheterization

Werner Forssman 1929
Catheterization
Electrophysiology testing
MD: Cardiac Manifestations

- Pump related: Dilated Cardiomyopathy-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 tachycardia (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 rhythm
Fast Cardiac Rhythms

Nonperfusional ventricular rhythm
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 impairment.
  - No association with number of CTG repeats
Pacemaker & AICD

Pacemaker - slow rhythms and block

Defibrillator - slow rhythms/block & fast rhythms
• 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.
• Is an invasive strategy with electrophysiological studies and prophylactic permanent pacing in MD1 patients with infranodal conduction delays superior to a noninvasive strategy?

• 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 occurrence of an initial event
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??

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.
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
Rate
Fast Cardiac Rhythms

- atrial flutter with 2:1 block

Repeat after emesis
Slow Cardiac rhythms

- Complete heart block