



Hearts

Neuromuscular Information and Research Day

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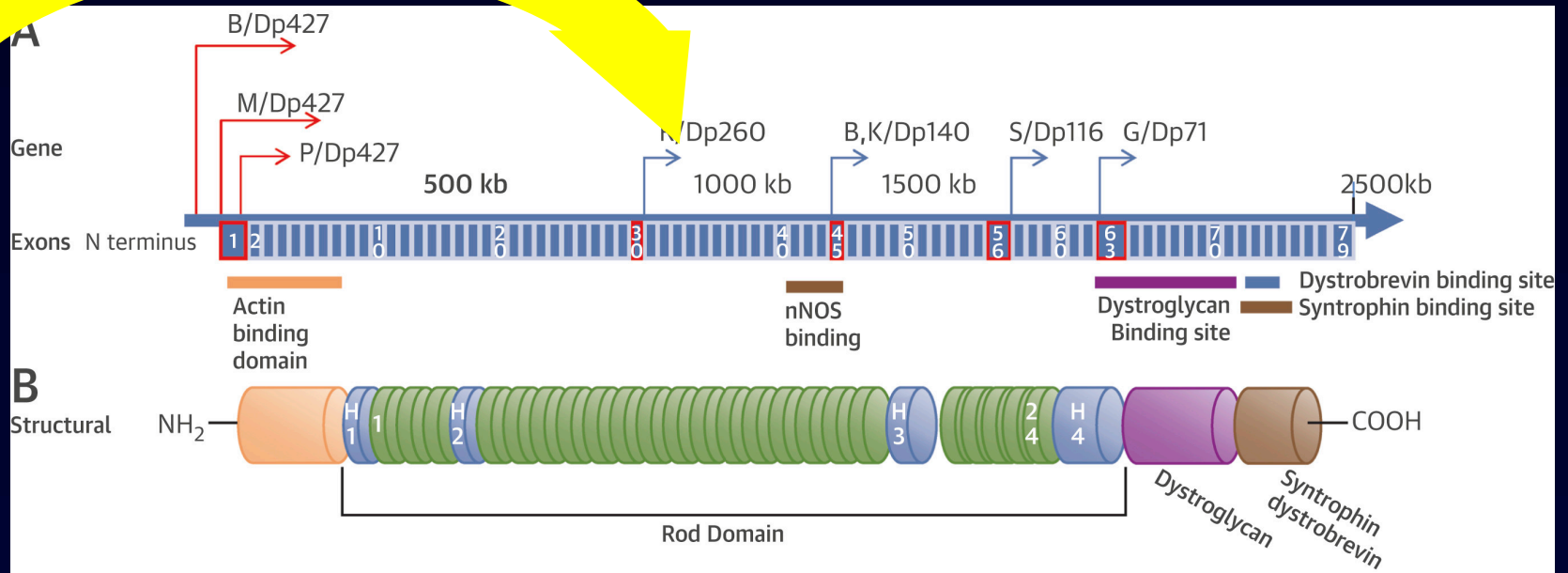
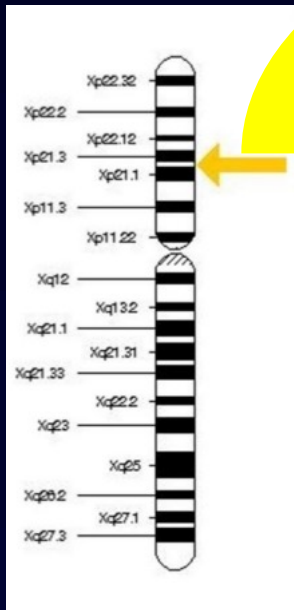


Summary

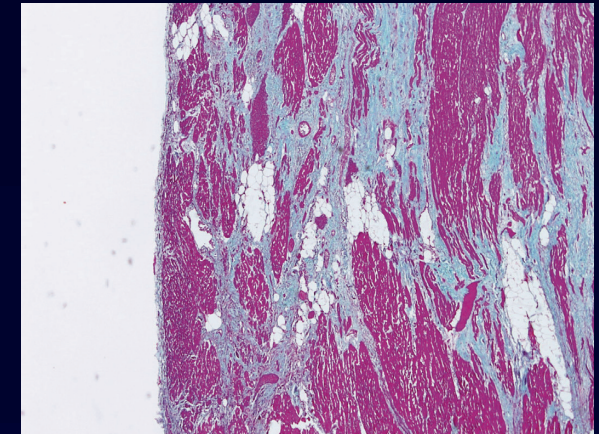
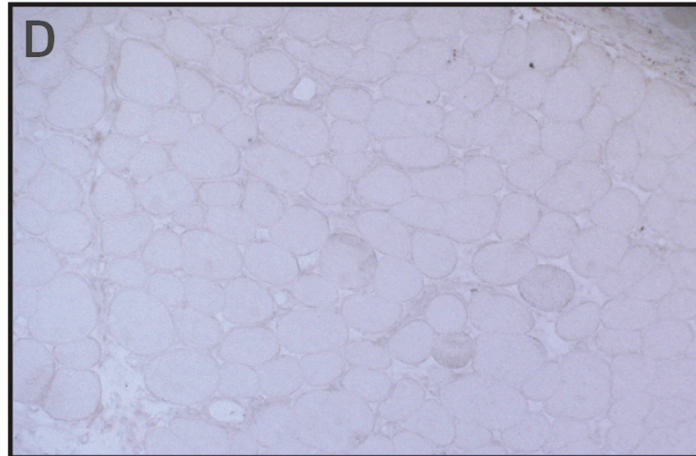
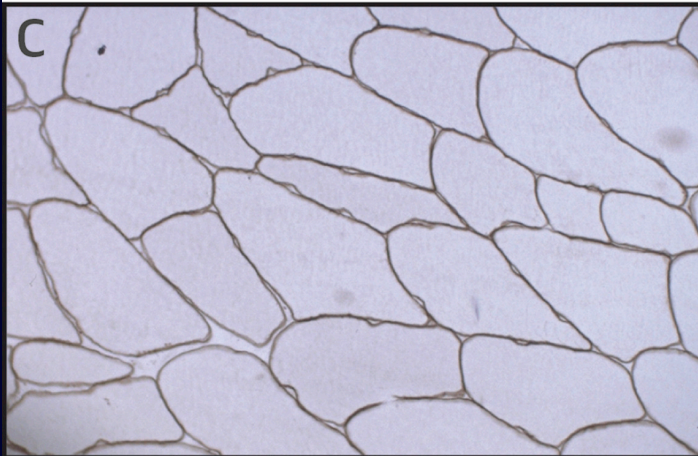
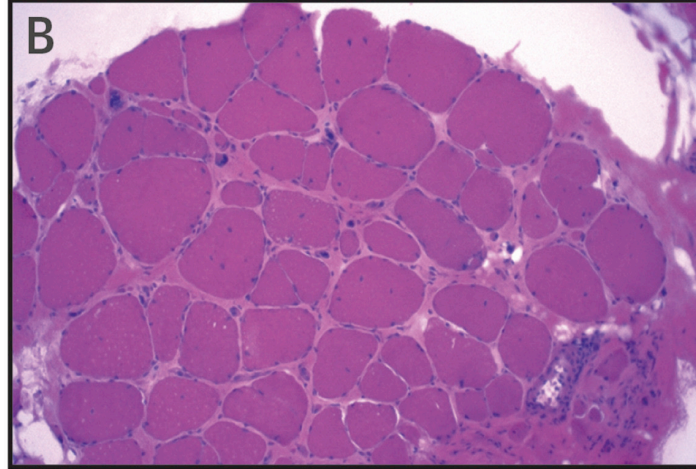
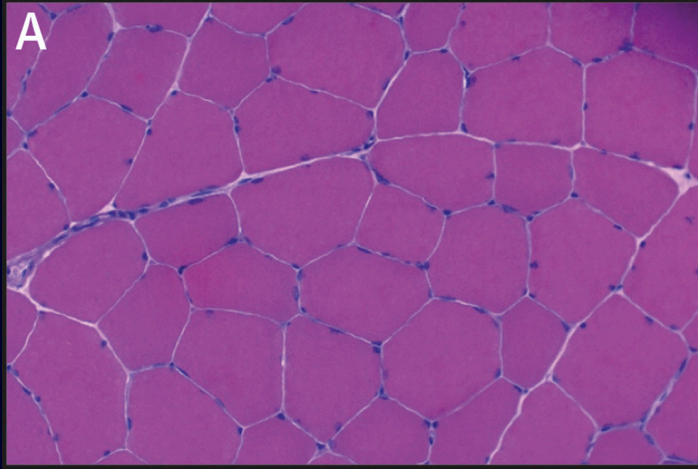
- Mechanism and Nature of Cardiac Problems
- Surveillance
- Management
- Future directions



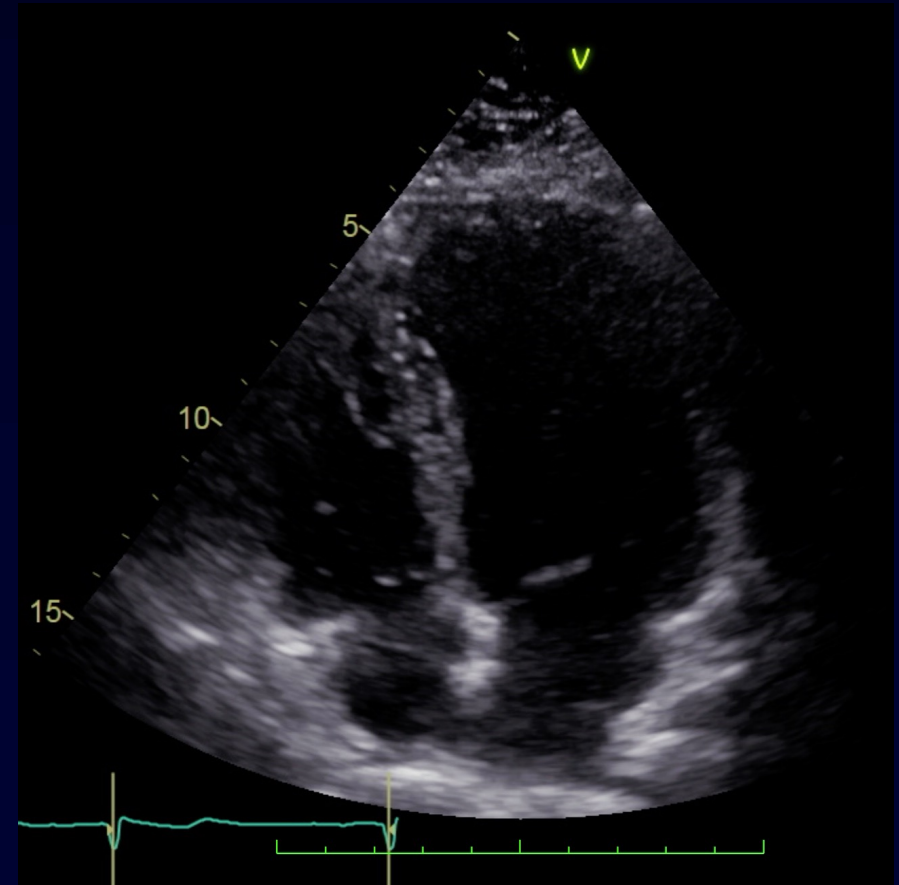
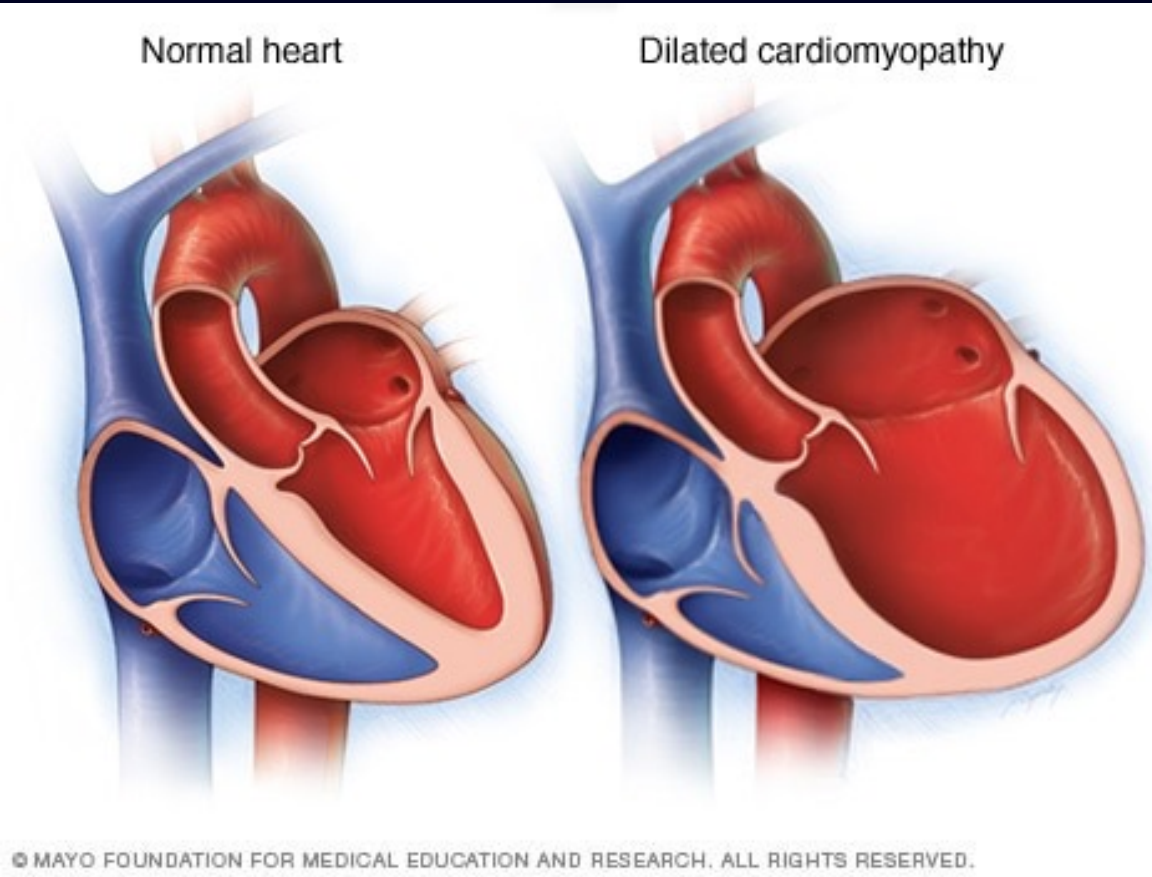
Mechanism and Manifestations



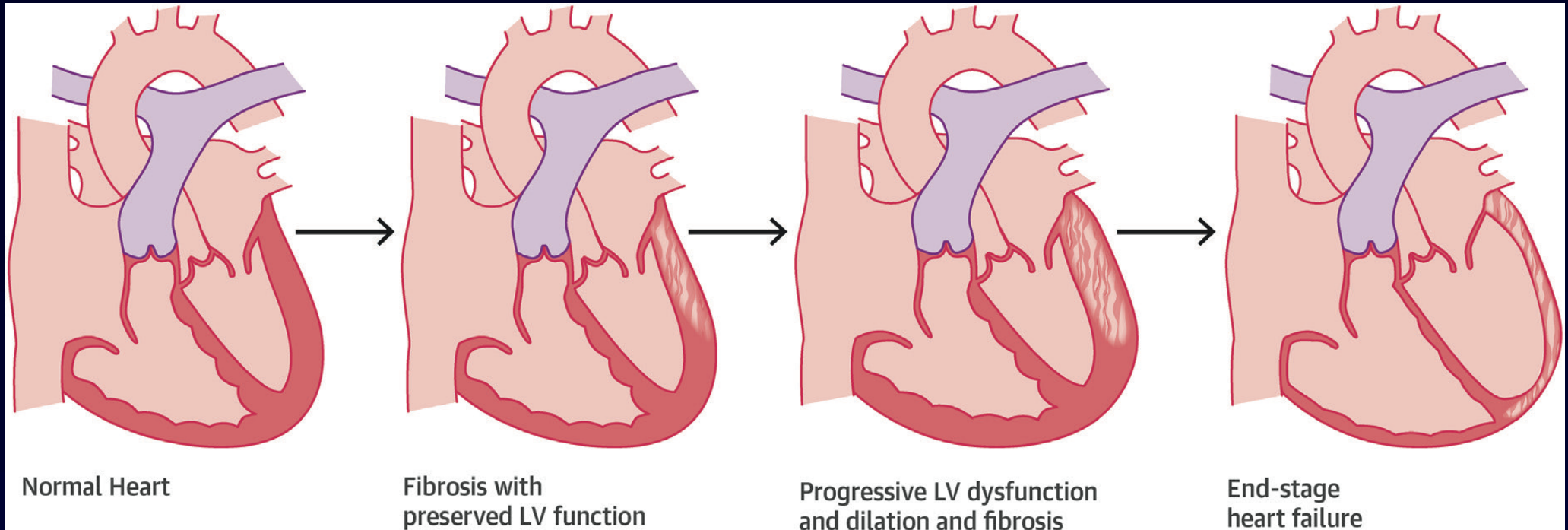
Mechanism and Manifestations



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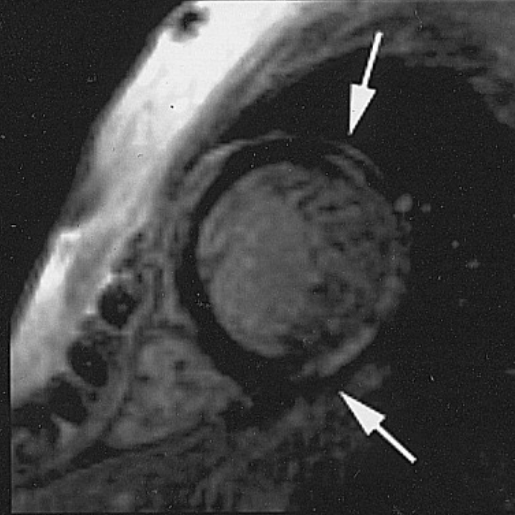
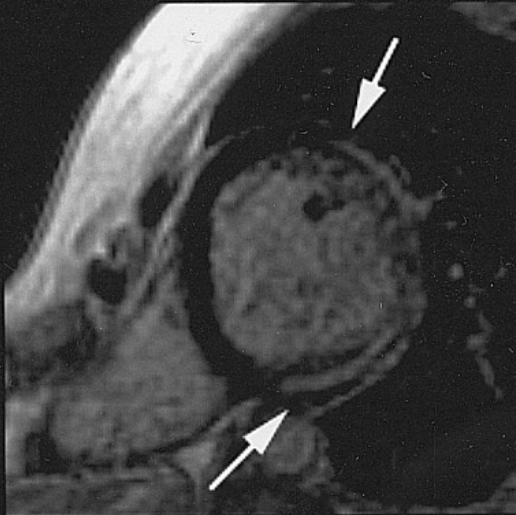
Mechanism and Manifestations

MYOCARDIAL DELAYED ENHANCED CMR

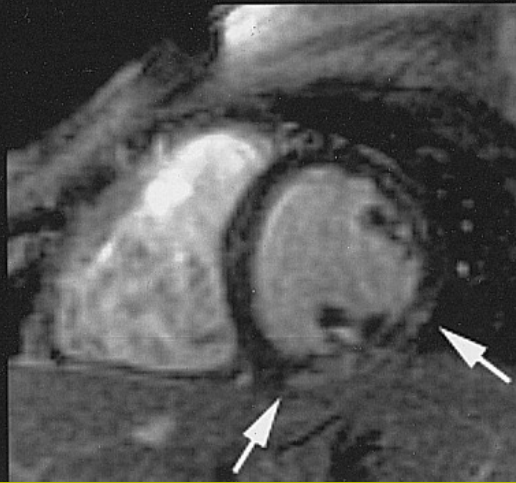
SA

LA

DUCHENNE



BECKER



Mechanism and Manifestations

- Heart failure symptoms:
 - Breathlessness on exertion
 - Breathlessness when lying down
 - Breathlessness at rest
 - But other things cause these symptoms too
 - Not being active may mask symptoms
- Heart rhythm issues: palpitations
- Rarely chest pain (more of a tightness)



Surveillance

- As management of lung problems improve, heart issues are becoming more noticeable
- As many with muscular dystrophy are non-ambulatory, symptoms may not be evident

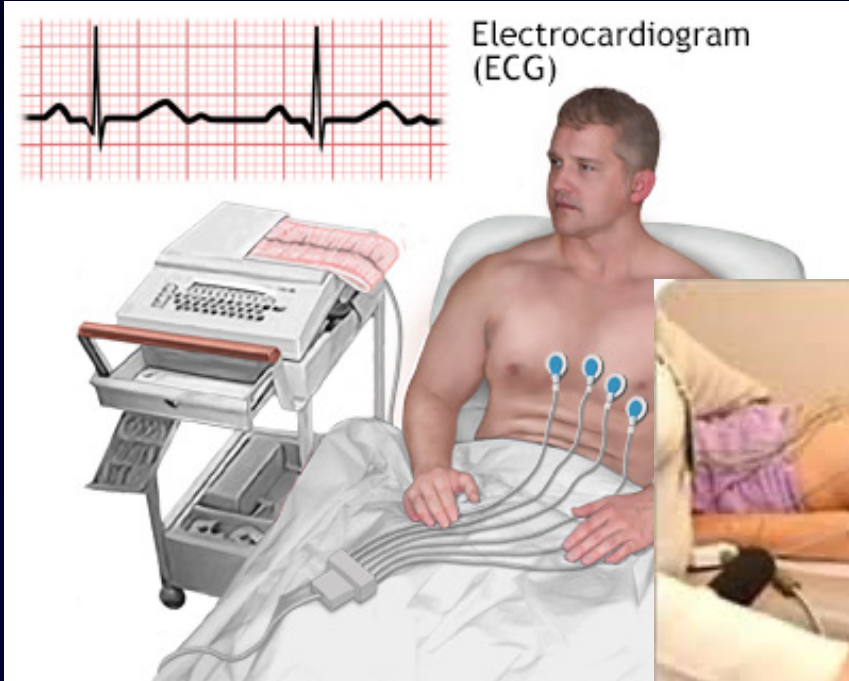


Surveillance-Cardiology Assessment

- Cardiology Review (recent change in frequency)
- ECG
- Echocardiogram
- ?MRI
- ?Holter monitor
- ?blood testing



Surveillance-ECG testing



Surveillance-Echocardiogram



How long does it take?
When to start?
How often?



Surveillance-MRI scanning?



When?

Not necessarily rebatable

How long? 40 mins-2 hours

Might need anaesthetic if young

Cost++



Surveillance-other testing

- Holter monitoring
- Blood testing (cardiac markers)



Management

- Steroids
- Other medications
 - (all heart failure medications also lower blood pressure)
 - ACE inhibitors/Angiotensin 2 inhibitors
 - Beta-blockers
 - Aldosterone antagonists
- Other medications used in non-MD cardiomyopathy



Management

- Steroids
 - Lower inflammation in muscles
 - Improve mobility
 - Early study showed less cardiomyopathy at 3 years with deflazacort compared to placebo
 - Another study showed better heart function (subtle)



Management

- ACE inhibitors/Angiotensin 2 inhibitors
 - Based on study with perindopril:
 - Compared to placebo, no difference in heart function after 3 years
 - After 5 and 10 years, there were less people with poor heart function in the perindopril group
 - Ideally prefer to start by age 10 (in reality, not many have started by that age)
 - Lower blood pressure, so start at low dose, gradually increase
 - Sometimes cause cough, so Angiotensin II antagonists are an alternative

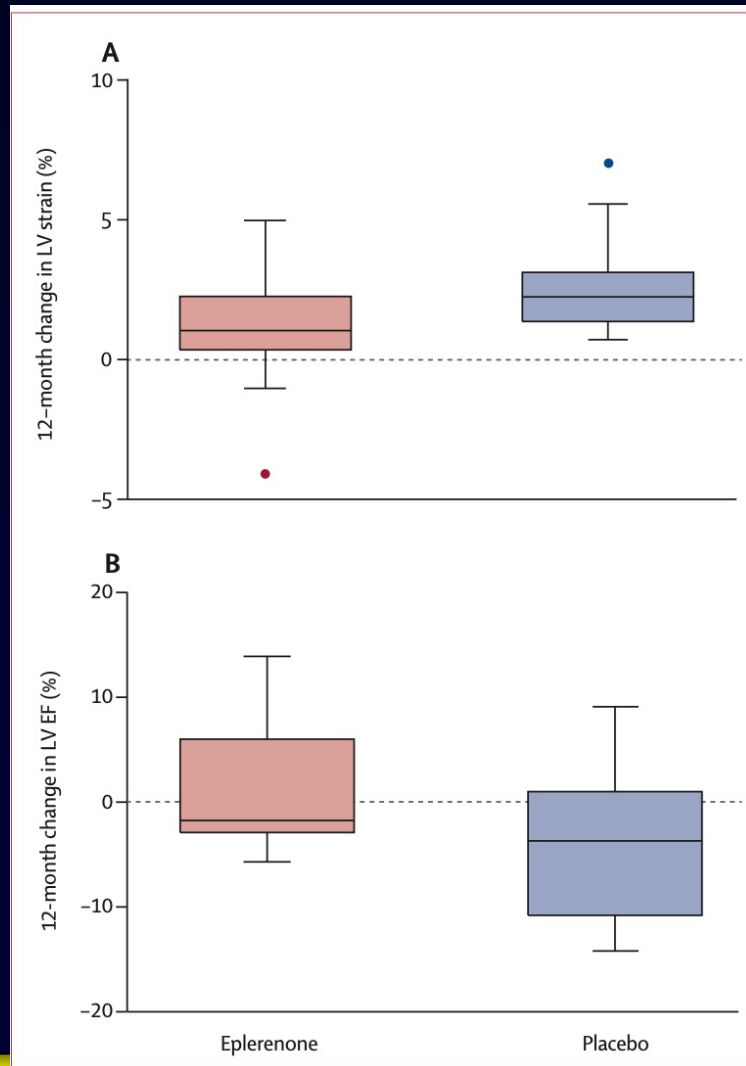


Management

- Beta-blockers
 - Lower heart rate and “work” of heart
 - Used in non-MD heart failure with clear benefit
 - Studies in Duchenne show similar benefit when added on to ACE inhibitors when heart rate is fast
 - Not as well proven, but advised



Management-Aldosterone antagonists



- Eplerenone 25 mg daily
- ?spironolactone 25 mg



Management

- Other medications used in non-MD cardiomyopathy
 - Sacubitril/valsartan (Entresto)
 - Ivabradine
- Non-medical therapy
 - Defibrillators
 - Assist devices
 - Heart transplantation



Future Directions

- Gene Modification
 - Exon skipping drugs (e.g. etepliresin)
 - Gene therapy



Female carriers

- Manifestations-usually mild, minority of women, rarely more severe
- Surveillance:
 - Ideally MRI and Echocardiogram in early adulthood
 - 3-5 yearly



Summary

- Mechanism and Manifestations
- Surveillance becoming more frequent, though limitations based on Medicare
- Management options improving
- New gene therapies may be helpful in future
- Female carriers need surveillance

