The Heart Is A Muscle, Too! The Cardiomyopathy of Duchenne and Becker Muscular Dystrophy



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Why should we all be interested in the heart?



The heart is a muscle, TOO!!! (and a very very important muscle...)



Important heart vocabulary

- Cardiologist- a heart doctor
- Atria- the chambers that receive blood in the heart
 - There are two atria (left and the right)
- Ventricles- are pumping chambers of the heart
 - there are two ventricles (left and right)
- Cardiomyopathy- disease of the heart muscle
- Heart Failure- when the heart is no longer able to meet the body's demands
- **Ejection fraction (EF)-** is a measurement of the percentage of blood leaving the heart each time it contracts.
- Fibrosis- scar tissue



Blood returns from lower half of the body

A key concept in understanding DMD cardiomyopathy

Cardiomyopathy in Duchenne muscular dystrophy is characterized by extensive sub-epicardial fibrosis



How should we care for the DMD/BMD heart?







Diagnosis and management of Duchenne muscular dystrophy, part 2: respiratory, cardiac, bone health, and orthopaedic management

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Cardiac care-summary

- Should begin at diagnosis
- Establish relationship with family
- Family history is important
 - Maternal carriers are at risk
- Evaluation should include
 - Physical examination
 - Cardiac testing
 - ECG and non-invasive imaging (echo or CMR)
- Early years patient should be seen yearly
- Frequency increased based on clinical needs
- Cardiac evaluation before major surgeries
- ACE inhibitors started prior to the age of 10 years







Chest pain in the DMD patient

- FREQUENT
- Must be musculoskeletal?
- Often dismissed in a peds ER
- Cardiac evaluation rarely undertaken
 - ECG "maybe"
 - Troponin I (cTnI) "rarely"
 - cTnI sensitive and reliable marker of cardiac tissue injury
 - cTnI is normal or minimally elevated at baseline in DMD



Chest pain in DMD patient

- Hypothesize progression of DMD cardiomyopathy results from episodic myocardial injury rather than from continuous ongoing injury
 - Series of silent recurrent events leading to cumulative injury
 - Process similar to that in skeletal muscle??
 - Suggests a step wise model of disease progression as opposed to a linear one
- Unknown if there could there be external triggers
 - Viral infection
 - Physiological stress
 - Other illnesses

Cardiac imaging

- Historically it was believed that
 - "Cardiomyopathy did not manifest until late teen early adult years"
 - Disease was characterized when imaging was "less sophisticated"
 - If you can't see it must not be there
 - Better tools now allow us to see important new things
- Imaging allows us to redefine natural history of DMD cardiomyopathy
 - As imaging improves we see the disease with new eyes
 - Two imaging modalities are commonly employed
 - Echo
 - CMR





Echocardiography has limitations



- Image quality is poor in many DMD patients (especially in non-ambulatory patients)
- Sub-optimal images result in poor clinical decision making and poor clinical trial data

CMR offers imaging advantages

Advantages

- Lack of radiation exposure
- Detailed information:
 - LV and RV function
 - Fibrosis quantification
 - Myocardial strain

Disadvantages

- IV placement required for fibrosis quantification
- Longer scan times
- Sedation may be needed
- Lack of global availability



Summary

- The heart is an important muscle, too!
- DMD chest pain may signal acute cardiac injury
 - cTnI levels and ECG should be obtainedCMR is a valuable tool to evaluate DMD cardiomyopathy
- CMR is a valuable tool to evaluate DMD cardiomyopathy
- In conclusion...



The Heart is a muscle too!



