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Cardiac Issues in Neuromuscular Disorders:

Monitoring, Prevention and Treatment

Disclosures

- Off label use of drugs
- No financial relationships

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History of CV care in dystrophinopathies

- No CV care → reactive care → present
- Present:
  - Part of multi-disciplinary neuromuscular team
  - My role: Heart Failure Cardiologist

Standards of care in dystrophinopathies

- 2003: European Neuromuscular Center
  - Neuromusc Disorders 13: 166-72.
- 2005: American Academy of Pediatrics
  - Pediatrics 116(6): 1569-73.

Standards of care: initial evaluation

- At time of diagnosis or 6 yo
- Includes:
  - Cardiologist
  - EKG
  - Non-invasive assessment of heart function

Standards of care: CV symptoms

- Gen: Malaise, fatigue, depression, decreased activity, sleep disturbances
- GI: abdominal pain, weight loss or gain, diarrhea, vomiting, anorexia (no desire to eat)
- Headache
- Palpitations
- Symptoms often easily missed in NM disease
- *Chest pain not very common even when severe heart dysfunction*

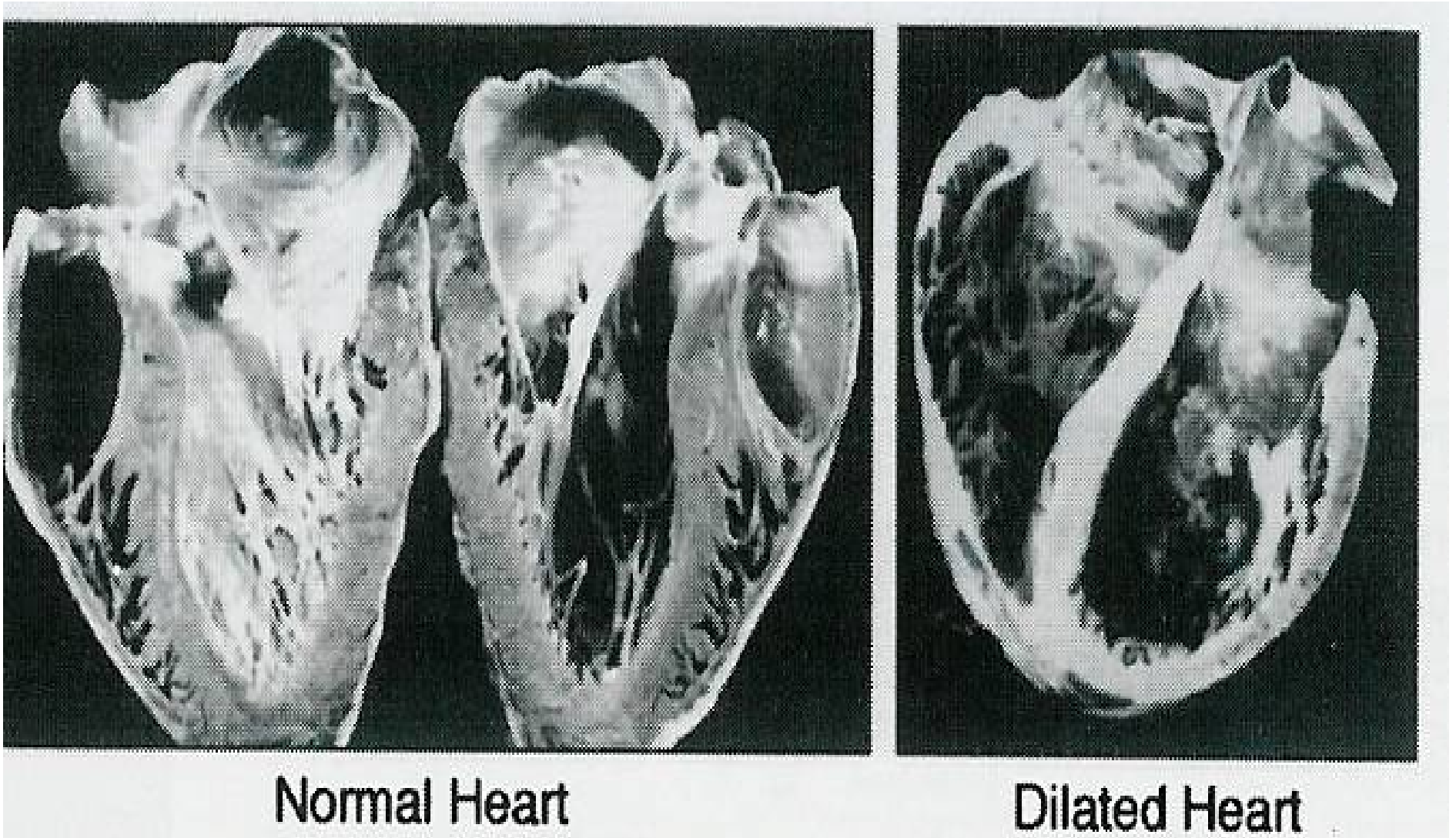
Standards of care: CV exam

- Increased heart rate (may be subtle)
- Increased rate of breathing
- May be harder to breath
  - May prefer to sleep sitting up
- Ascites: fluid in tummy area
- Hepatomegaly: liver enlarges
- Ankle swelling
- Poor pulses– weaker, harder to feel

Standards of care: monitoring

- After initial diagnosis, every other year <10 yo
- 10 yo → every 6-12mo
- Evaluation:
  - Cardiologist listening
  - EKG
  - Non-invasive assess function
    - Echo
    - MRI?
  - Holter monitoring of rhythm as needed

Dilated cardiomyopathy



Standards of care: therapy—special circumstances

- Heart rhythm abnormalities not uncommon
  - EKG, Event Monitor, Holter
  - EP study in cath lab?
  - ICD? Intracardiac defibrillator
- Thorough cardiac eval <6 mo of surgery
- Around time of initiation of steroids or if steroid-induced high blood pressure
- Blood thinners may be needed with severe dysfunction

Standards of care: prevention and therapy

- Avoid obesity, diabetes and high blood pressure
- Heart healthy diet/watch cholesterol & fat
- At earliest sign of heart dysfunction
  - ACE Inhibitor is first line
  - Beta blockers second line
  - Gentle use of diuretics
  - {Digoxin is controversial, but has been used}
  - Spironolactone and Co-enzyme Q also used
  - Blood thinners

Medication descriptions

- ACE Inhibitors:
  - Angiotensin Converting Enzyme Inhibitors
  - ACE binds to blood vessels walls and make them constrict
    - Inhibiting ACE allows vessels to relax
  - Enalapril, Lisinopril
  - Losartan: ACE receptor blocker
  - Relaxes blood vessels, lowers BP, easier to pump
  - Can cause dizziness

Medication descriptions

- Beta Blockers: (Coreg, atenolol)
  - Relaxes blood vessels, lowers BP, easier to pump
  - Slows heart rate
  - Can cause dizziness, fatigue
- Diuretics (water pills)
  - Help to rid of excess fluid retained due to heart issues.
  - Can relieve ankle and belly fluid

Medication descriptions

- Digoxin
  - Helps heart squeeze with more force
  - In some heart problems can make things worse
  - Can cause rhythm problems

Medication descriptions

- Spironolactone
  - A weak diuretic
  - Helps in muscle "remodeling" (in a good way) in some types of cardiomyopathy
- Co-Enzyme Q (anti-oxidant)
  - In theory, would help minimize damage from free-radicals
  - Controversial as to if really works

Medication descriptions

- Blood thinners:
  - Aspirin, low dose
    - Makes platelets not function normally
    - Platelets are like bits of cells that help blood clot
  - Coumadin/Warfarin
    - Old blood thinner
    - Oral med
    - Lots of challenges/blood tests
  - Heparin (injection only)

Emerging & Controversial Therapies

- Home milrinone use
- Advanced CHF therapies
  - Resynchronization therapy
  - Mechanical support?
    - Ventricular support devices (VADs)
  - Heart transplant?
    - Unwise if lung issues

Future directions

- Improved monitoring of cardiac effects
- General preventive strategies/therapies*
- Individualized diagnosis
  - Genotype + environment + ? = Phenotype
  - Link gene to best cardiac therapy
- CV specific prevention***