Cardiac Care in Neuromuscular Disease: Considerations & Resources

A Resource for Clinicians

Background

- Cardiac disease is a major cause of morbidity and mortality in patients with some forms of neuromuscular disease (NMD)
- Early recognition of patients at risk for cardiac disease, integration of a cardiologist, and strong coordination of multidisciplinary care can optimize outcomes
- This document highlights key points from an MDA webinar with a cardiologist with NMD care expertise. View companion webinar here

NMDs with Increased Risk of Cardiac Disease*

Duchenne MD
- Cardiomyopathy as early as 9 years of age
- 90% affected by age 18 years
- **Cardiac disease is the leading cause of death in current era**

Becker MD
- Less severe than DMD, though more variable
- 70% affected by age 40 years
- Cardiac impact can be more serious than skeletal

Limb-girdle MD
- Variable cardiac involvement (25%-55% of patients) due to clinical heterogeneity
- Cardiac involvement may include arrythmias and cardiomyopathies

Myotonic dystrophy type 1
- Cardiac involvement in 80% of patients
- AV conduction abnormalities and arrhythmias most common; risk for sudden cardiac death
- Pacemakers for heart block

Emery-Dreifuss MD
- Arrythmias and conduction disturbances
- Cardiomyopathy is rare

Friedreich's ataxia
- Cardiac involvement in 90% of patients
- Often life-threatening
- 40% have supraventricular tachycardia

*NMDs presented are not inclusive of all NMDs.
AV, atrioventricular; DMD, Duchenne muscular dystrophy; MD, muscular dystrophy; NMD, neuromuscular disease.
1. Mavrogeni S, et al. World J Cardiol. 2015,26;7(7):410-414. 2. Andrews JG, Wahl RA. Adolesc Health Med Ther. 2018;9:53-63. 3. Michal M, et al. Folia Cardiol. 2020;15(3):243-249. 4. Giuliani L, et al. Eur Heart J Suppl. 2020;22(suppl E):E13-E19. 5. Sveen ML, et al. Arch Neurol. 2008;65(9):1196-1201. 6. Poppe M, et al. Ann Neurol. 2004;56(5):738-741.

Cardiac Manifestations of Common Genetic NMDs

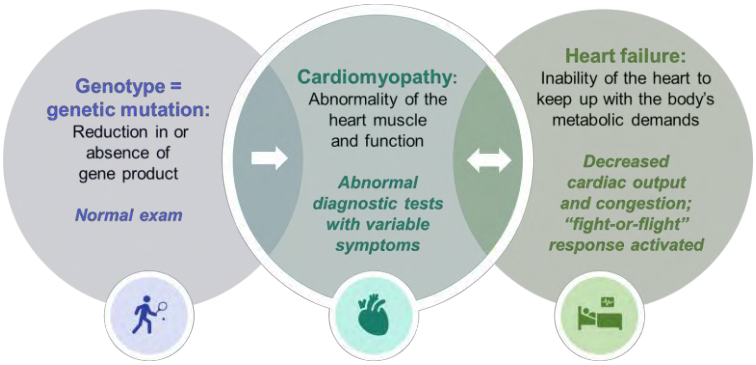
NMD Type	Molecular Defect	Mode of Inheritance	Cardiomyopathy	AV Conduction Disturbances	Arrhythmia
DMD	Dystrophin	XR	DCM	Rare	Common, mild
BMD	Dystrophin	XR	DCM	Rare	Common
EDMD	Emerin	XR	Rare	Common	Common
DM1	Protein kinase	AD	Rarely HCM/DCM	Common	Common
LGMD 1B	Lamin A/C	AD	DCM	Common	Common
LGMD 2E	β-sarcoglycan	AR	DCM	Common	Common
FA	Frataxin	AR	HCM	Rare	Common

AD, autosomal dominant; AR, autosomal recessive; AV, atrioventricular; BMD, Becker muscular dystrophy; DCM, dilated cardiomyopathy; DM1, myotonic dystrophy type 1; DMD, Duchenne muscular dystrophy; EDMD, Emery-Dreifuss muscular dystrophy; FA, Friedreich's ataxia; HCM, hypertrophic cardiomyopathy; LGMD, limb-girdle muscular dystrophy; XR, X-linked.

Developed with the expertise of Beth D. Kaufman, MD (Stanford University School of Medicine)
Last Reviewed: December 2023

MDA MEDICAL EDUCATION

Progression of cardiac disease varies based on type of NMD and individual variation



Signs and Symptoms: Cardiomyopathy and Conduction Abnormalities

Cardiomyopathy

- Commonly, none
- Increased heart rate for age
  - Palpitations
- Poor weight gain
  - Increased caloric requirements
- Increased work of breathing with exertion
- Wheezing ("asthma")

Conduction system abnormality

- Exercise intolerance
  - Limited increase in heart rate
- Fainting
- Dizzy spells
- Fatigue

Progression from Cardiomyopathy to Heart Failure

Decreased cardiac output causes inadequate blood flow to meet organs' needs.
Body compensates by activating "fight-or-flight" response.

Symptoms

General:
- Dizziness
- Fatigue

Cardiovascular
- Chest pain
- Palpitations

Respiratory
- Cough
- Shortness of breath

Gastrointestinal
- Loss of appetite
- Abdominal pain
- Vomiting

Signs

General:
- Failure to thrive/weight loss
- Cool extremities
- Swelling of face, abdomen, sacrum, legs

Cardiovascular
- Fast heart rate
- Lower blood pressure

Respiratory
- Labored or rapid breathing at rest

Renal
- Decreased urination, electrolyte abnormalities
- Fluid retention

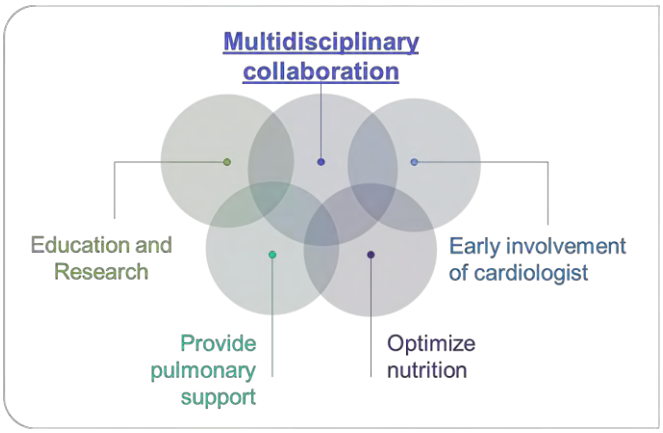
Developed with the expertise of Beth D. Kaufman, MD (Stanford University School of Medicine)
Last Reviewed: December 2023

MDA® MEDICAL EDUCATION

Management Requires Strong Multidisciplinary Coordination

Successful management =

- Multidisciplinary collaboration
- Patient and provider education
- Proactive serial screening for early detection of cardiac involvement
- Initiating cardiac therapies at early stages of cardiomyopathy to improve patient outcomes
- Treating modifiers of cardiac disease (respiratory support, nutrition)



NMD, neuromuscular disease.
Feingold B, et al. Circulation. 2017;136(13):e200-e231.

Cardiomyopathy Treatment Goal: Prevent Cardiac Remodeling and Decompensation

Guideline-directed medical therapies

Medications to decrease harmful changes in heart muscle that occur over time

- ACEI (enalapril, lisinopril)
- β-adrenergic blockers (carvedilol)
- Aldosterone antagonist

Treat modifiers of disease

- Respiratory support, avoid OSA
- Hypertension
- Weight management – avoid obesity, provide adequate nutrition
- Steroids (in DMD)

Preemptive planning

- Preoperative cardiac risk assessment
- Anesthesia strategy
- Postoperative management plan (fluid sensitivity, arrhythmia risk)
- Cardiology consultation

Acute Heart Failure Therapy Goal: Balance Needs of Body With Available Circulation…to Relieve Symptoms and Support Organs

Metabolic demands

Energy required for:

- Breathing
- Movement
- Eating
- Surgical healing
- Fighting infection/fever

Cardiac supply

Optimize blood flow to vital organs and tissues

- Medications
- Fluid management
- Nutrition
- Mechanical support

Developed with the expertise of Beth D. Kaufman, MD (Stanford University School of Medicine)
Last Reviewed: December 2023

MDA® MEDICAL EDUCATION

AHA Recommendations to Approach Cardiac Evaluation in NMD*

01 Providers/patient organizations are encouraged to promote education on the importance of screening, diagnosis, and management of CV complications ***(Class I; Level of Evidence B)***

02 Neurologists are encouraged to collaborate with cardiologists, electrophysiologists, or HF specialists depending on the condition being evaluated ***(Class I; Level of Evidence B)***

03 For cardiac conditions diagnosed in childhood warrant referral to a pediatric HF specialist ***(Class IIa; Level of Evidence B)***

04 Perform cardiac evaluation, ideally 3-6 months prior to anesthesia/sedation ***(Class I; Level of Evidence C)***

05 Cardiac monitoring by an anesthesiologist experienced in caring for patients with NMD for patients requiring surgery; facility should have appropriate intensive care facilities ***(Class I; Level of Evidence C)***

*the AHA recommendations presented here are not exact and have been summarized for this presentation; please review the publication for full recommendations. Feingold B, et al. Circulation. 2017;136(13):e200-e231.

Resources

2022 HRS Expert Consensus Statement on Evaluation and Management of Arrhythmic Risk in Neuromuscular Disorders.
Groh WJ. *Heart Rhythm.* 2022;19(10):e61-e120

Management of Cardiac Involvement Associated With Neuromuscular Diseases: A Scientific Statement From the American Heart Association. Feingold B. Circulation. 2017;136(13):e200-e231

Treatment Strategies for Cardiomyopathy in Children: A Scientific Statement From the American Heart Association. Bogle C,. Circulation. 2023;148(2):174-195

Pediatric heart failure collaborative with working group to improve treatments and outcomes of children with muscular dystrophy Villa C. Pediatr Cardiol. 2022;43(5):977-985

ISHLT Guidelines for Management of Pediatric Heart Failure 2014. Kirk R. ISHLT Monograph Series. Vol 8. 2014. Includes a chapter on HF in the setting of NMD

Use of advanced heart failure therapies in DMD. Wittlieb-Weber CA.. Prog Pediatr Cardiol. 2019;53:11-14.

Access companion MDA webinar here

Developed with the expertise of Beth D. Kaufman, MD (Stanford University School of Medicine)
Last Reviewed: December 2023

MDA MEDICAL EDUCATION