



## Best Practices for Managing Cardiac Involvement with NMD

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In addition to affecting skeletal muscle, some neuromuscular diseases (NMDs) can affect cardiac muscle. Yet as knowledge of the genes causing these conditions grows and leads to new treatments, understanding of how to approach cardiac management has not kept pace.

Elizabeth McNally, MD, Director, Center for Genetic Medicine at the Feinberg School of Medicine at Northwestern University, says best practices for cardiac management vary by disease.

“The first thing with a neuromuscular disease is to know what the genetic diagnosis is,” she says. “With a muscular dystrophy, knowing the subtype tells us about possible heart issues. It also tells a cardiologist how to monitor the patient and how often to look at an echocardiogram and MRIs for details on heart muscle, and how often to run electrocardiograms.”

### Diseases with cardiac involvement

The most common cardiac issues associated with NMDs are cardiomyopathy and irregular heart rhythms (arrhythmias). The severity and onset of these cardiac complications vary significantly across types of NMDs.

Duchenne muscular dystrophy (DMD) and Becker muscular dystrophy (BMD) often involve cardiomyopathy. In these muscular dystrophies, “You also can have arrhythmias, which are usually later in the course of cardiomyopathy,” Dr. McNally says. “It’s important to start heart medications early in DMD to slow heart involvement.”

Both type 1 and type 2 myotonic dystrophy (DM1 and DM2) are known to have heart involvement in terms of both irregular rhythms and cardiomyopathy, although the DM2 heart involvement usually is later in life.

Limb-girdle muscular dystrophy (LGMD) is a broad group of muscular dystrophies. “Some types can have a great deal of heart involvement,” Dr. McNally says. Accurate genetic diagnosis is important for determining the appropriate cardiac evaluation and follow-up.

“With nearly all cardiac complications of neuromuscular disease, early treatment and monitoring can be very effective,” she says.

## **Cardiac management and NMD therapies**

“As we’ve seen more relevant treatments become available for neuromuscular diseases, like exon skipping for DMD, we need to know how they will affect the heart,” Dr. McNally says. “We don’t have a lot of information on certain treatments, and we are learning as we go in some ways.”

Because of this, multidisciplinary care, such as that offered at MDA Care Centers, is important. “In addition to the neurologist, a cardiologist has to be part of the team, working with the pulmonologist, the occupational therapist, the genetic counselor, and the nutritionist,” Dr. McNally says, adding that the cardiologist, if not well-versed in NMDs, should be willing to educate themselves on the topic to contribute effectively.

She also points out that telehealth allows more outreach to patients who do not live near an MDA Care Center or have access to other NMD-specialized care.

## **Detecting cardiac involvement**

A care team, including a cardiologist, that is well-versed in NMDs also is important for determining if there is cardiac involvement.

A cardiac diagnosis can look different and be harder to detect for very young children. For example, they might not grow at an appropriate rate because they’re not getting enough energy.

Heart involvement might go undetected for adults with mobility limitations due to their lack of physical activity. “Because heart symptoms may look different in a person living with neuromuscular disease, we rely much more on imaging studies like echocardiograms and cardiac MRI, and we don’t wait for symptoms,” Dr. McNally says. “We are much more proactive. We also use blood markers to check for heart involvement, and we favor earlier treatment to keep the heart from getting worse.”