Cardiac Health and Myotonic Dystrophy: A Q&A with Elizabeth McNally, MD, PhD

Myotonic dystrophy (DM) is a multisystemic form of muscular dystrophy. The most common type, originally called Steinert's disease, is known as myotonic dystrophy type 1 (DM1). Another form, once known as proximal myotonic myopathy (PROMM), is now called myotonic dystrophy type 2 (DM2).

Both types of DM are characterized by progressive muscle loss and weakness, but they can present with symptoms in any organ system. Cardiac complications can occur in either type but are better described in DM1.

This variability can make it difficult to manage cardiac health in patients with DM. We spoke with Elizabeth McNally, MD, PhD, a cardiologist and geneticist at Northwestern University's Feinberg School of Medicine in Chicago, to learn more about treating and managing cardiac issues in DM.

Q: What cardiac issues are associated with DM?
A: The most common heart problems in DM include first-degree heart block and/or more advanced heart block (second/third degree), bradycardia, atrial fibrillation, ventricular fibrillation, and heart failure with reduced ejection fraction (HFrEF) and heart failure with preserved ejection fraction (HFpEF). Cardiac conduction system abnormalities are the most common cardiac abnormality in DM.

DM1 and DM2 share overlapping cardiac complications, although the age of onset of cardiac conditions is typically earlier in DM1, and the cardiac findings have been better detailed in the literature. Presentation in DM can be quite variable, and cardiac findings can be the first presentation of DM1 — prior to development of muscle weakness or hand cramping. In young people (under age 35) who present with atrial fibrillation, a careful neuromuscular history should be ascertained, and family history should inquire about early onset cataracts. DM genetic testing should be considered.

Once a patient has been genetically diagnosed, annual electrocardiograms (ECGs) and regular echocardiograms are typical. Heart rhythm can be effectively monitored using skin patch monitors. These monitors detect heart block, bradycardia, tachycardia, and chronotropic response, although this is highly dependent on how physically active the patient is. Patch monitoring of longer durations has a greater ability to detect abnormal rhythms.

The annual ECGs should monitor for progressive PR prolongation and QRS widening over time by comparing to prior tracings. If there is a marked shift in PR prolongation, this can be an indication for a pacemaker.

When placing a pacemaker, consider the placement of an internal cardioverter defibrillator (ICD) at the same time. QRS widening or advancing QRS widening on serial ECGs can help make this determination.

Q: What are the health consequences of these cardiac issues?
A: The degree to which DM patients will have symptomatic bradycardia depends on the heart rate. Many cardiac abnormalities may be asymptomatic but still present a risk.

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Fatigue is a very common finding in people with DM, and fatigue is multifactorial in nature, from both central and peripheral issues. Sleep disturbances are also common, as is neuromuscular respiratory weakness. The care team should engage a pulmonary sleep specialist who can monitor and treat these issues.

Fatigue in DM can be associated with daytime sleepiness, and patients are often treated with stimulants to reduce daytime sleepiness. However, stimulants can promote irregular heart rhythms, so the risk-benefit calculation should be carefully considered.

For DM patients experiencing atrial fibrillation, typical “CHADS-VASC2” calculators are insufficient for predicting stroke risk, since DM is a rare disorder where risk increases at younger ages. Stroke risk can also derive from components of atrial myopathy, and this can influence the choice to recommend anticoagulation. Fall risk can be increased due to neuromuscular weakness, which should also be taken into account in the decision to recommend anticoagulation.

Patients should be counseled to report symptoms like near syncope, syncope, sustained palpitations, or fast or slow heart rhythms. Smart watches can be of assistance.

Because it can be difficult to detect heart failure symptoms in people with reduced mobility, blood markers such as NT-proBNP can be very useful. Shortness of breath can be multifactorial, with components of heart failure and neuromuscular respiratory weakness. Peripheral edema may be present. Right heart failure can occur in those with untreated neuromuscular respiratory weakness.

Q: What tests and studies do you use to assess heart health in patients with DM?
A:

- Annual ECGs and routine skin patch monitors. Two-week monitors can provide more information.
- Routine echocardiograms to monitor LV function.
- NT-proBNP serum measurements; NT-proBNP has superior sensitivity to BNP and is favored for patients with neuromuscular disease.
- Cardiac MRI is useful for monitoring delayed enhancement and other indicators of increased arrhythmia risk.
- Coronary artery calcium scans can help risk stratify; this is relevant since DM patients have an increased risk for diabetes and may have premature coronary artery disease (CAD).
- Routine stress tests may be difficult, if not impossible, since patients may be unable to exercise on a treadmill and/or have difficulty grasping the bars on a routine exercise treadmill test. If suspicion for CAD is high, a CT angiogram or angiography should be considered. Patients with limited exercise capacity due to neuromuscular disease may not manifest routine symptoms for CAD yet still carry risk.

Q: How do you treat or manage patients’ cardiac issues?
A: The choice of pacemaker implantation and/or ICD placement should be made based on symptoms and findings from monitors and ECGs. Loop monitors can be helpful to gather more data to inform device placement or decisions regarding anticoagulation. It is very helpful to have access to an electrophysiologist familiar with the conduction system risks of myotonic dystrophy.

HFrEF is treated with beta blockers, ARNI, MRA, and SGLT2i. Diuretics are used as needed for edema. Beta blockers should be used with caution if there is evidence for bradycardia or other electrical conduction system slowing.

It is reasonable to avoid stimulant use for DM-associated fatigue in those who have experienced cardiac arrhythmias.

Q: Are there any new treatments or clinical best practices that help DM patients with cardiac involvement?
A: We are awaiting the results from ongoing trials using agents targeting DMPK or CUG repeats. These trials are aimed at treating the muscle symptoms in DM1. We do not fully know the effect of these drugs on the heart. Cardiac safety is being monitored in these trials. Cardiologists will need to become familiar with these agents, should the trials be successful and the drugs gain approval.

The use of GLP-1 agents has not been specifically examined in DM. It is likely these agents are being used to treat diabetes and obesity, which are known to occur in DM. There is a risk for sarcopenia with GLP-1 agents, so this should be considered.

The clinical care team should include neurology, cardiology, and pulmonary, and the team should be in regular communication with each other to address issues related to shortness of breath, fatigue, use of stimulants, and other issues that may be multifactorial in nature.

Q: What can DM patients do to support their heart health?
A: In addition to the DM-specific issues mentioned above, more common cardiovascular conditions can also occur. People with DM are at increased risk for diabetes, thus increasing cardiovascular risks. Statins can be used in DM. It is helpful to check serum CK and LFTs prior to initiating statins so that a baseline is available. Rosuvastatin has lower risks for muscle involvement (pain) and is therefore favored over other statins. PCSK9-targeted agents (antibodies or siRNA) can be used.

Resources

- MDA Grand Round webinar: Myotonic Dystrophy (DM): Core features, clinical subtypes, and outcome assessments
- MDA What's New in Neuromuscular Disease webinar: Updates in Myotonic Dystrophy
- MDA Quest Media content on myotonic dystrophy
- GeneReviews: Myotonic Dystrophy Type 1
- GeneReviews: Myotonic Dystrophy Type 2
- Myotonic Dystrophy Foundation: Myotonic Dystrophy Toolkits, Publications & Resources