Myotonic Dystrophy and the Heart

A COMMUNITY GUIDE

Myotonic Dystrophy FOUNDATION

Thank You

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Disclaimer: This guide was created to help educate you about myotonic dystrophy and the heart. It does not replace any advice from your doctor and is educational only. This guide is meant to accompany the Myotonic Dystrophy Foundation publication "Clinical Care Recommendations for Cardiologists[i]" designed specifically for cardiologists caring for people living with myotonic dystrophy. For additional clinical resources, refer to the Heart Rhythm Society which provides more information on evaluation and management of arrhythmic risk in neuromuscular disorders including a comprehensive review of myotonic dystrophy type 1 and 2.

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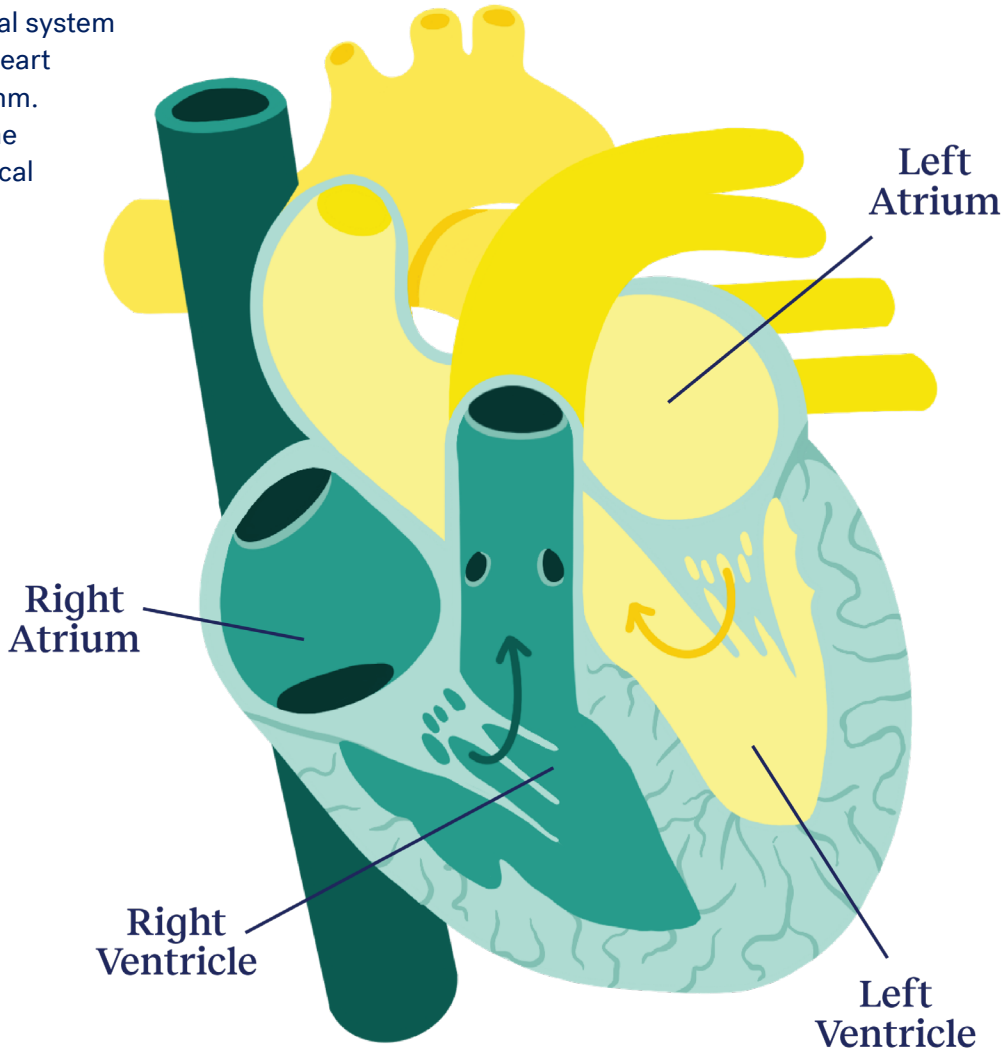
Myotonic Dystrophy and the Heart: a community guide

For individuals living with myotonic dystrophy (DM), heart or "cardiac" issues can pose a serious threat to health because the heart is itself a muscle and is affected by many of the muscular dystrophies. People living with myotonic dystrophy type 1 (DM1) or type 2 (DM2) can experience cardiac issues. In general, individuals with DM1 are at a higher risk of cardiac problems and at a younger age than individuals with DM2. However, because myotonic dystrophy is the most variable disease in medicine, there is variability in the likelihood of cardiac issues. It is important that care is adapted to the situation of DM1 and DM2. This resource is applicable to individuals with both myotonic dystrophy type 1 and type 2 and aims to help you understand heart risk and management.

Heart Anatomy

The heart is a muscle with four chambers that pump blood: two upper chambers called the right and left atria pump blood in, and two lower chambers called the left and right ventricle pump blood out to the rest of the body.

The heart has an electrical system made up of specialized heart cells that control its rhythm. The pumping action of the heart results from electrical signals spreading from these specialized cells to other cells that provide most of the heart's contraction.



Potential Heart Problems

Myotonic dystrophy can affect both the heart's *electrical system*

Problems with the heart's **electrical system**

- More common in myotonic dystrophy than problems with the pumping system.
- Electrical issues cause the heart to beat abnormally — this is also called an "arrhythmia."
    - Bradyarrhythmia or bradycardia = too slow
    - Tachyarrhythmia or tachycardia = too fast
- *Examples of arrhythmias are sinus bradycardia, atrial fibrillation, atrial flutter, ventricular tachycardia, ventricular fibrillation, and heart block. The latter three arrhythmias can be acutely dangerous including leading to sudden death.*

AND its *ability to pump*.

Problems with the heart's **pumping system** due to weak heart muscle

- Less common in myotonic dystrophy than problems with the electrical system.
- Risk of heart failure.
- *A weak heart muscle is a "cardiomyopathy."*

Someone with heart issues may experience a variety of symptoms:

- Feeling faint or temporarily losing consciousness (syncope)
- Fluttering or pounding heart (palpitations)
- Difficulty breathing (dyspnea)

However, heart issues are often asymptomatic (showing no symptoms) which is why it's important to be evaluated regularly even if you have no symptoms.

What Kinds of Doctors Treat Heart Issues?

Doctors specializing in evaluating and treating heart issues are called **cardiologists**. There are several different types of cardiologists; their knowledge and duties can overlap. The most common types of cardiologists that a primary care physician or neurologist might refer someone with myotonic dystrophy to include:

General Cardiologist
General cardiologists are experts in heart and blood vessel diseases; they can often deal with arrhythmias.

Cardiac Electrophysiologist
A cardiologist who specializes in the diagnoses and advanced treatment of arrhythmias.

How Will My Doctor Determine the Health of My Heart?

It is recommended to have a cardiac evaluation as soon as an individual is diagnosed with myotonic dystrophy. In some cases, this evaluation may be with your primary care physician. They will assess your medical history and symptoms, provide an examination, and perform simple tests such as an **electrocardiogram** (ECG, see below). Often, individuals will be referred to a cardiologist, who will perform specialized tests (like the ones listed below) early after diagnosis.

Electrocardiogram (ECG)/(EKG)

An ECG records the heart's electrical signals and identifies if there are electrical signal abnormalities. ECG's can diagnose if an arrhythmia is present.

- **12-Lead ECG:**
  Provides a short snapshot of the heart rhythm and can be done in most medical clinics.

- **Longer Term ECG Monitoring:**
  Something that may be worn, or in one case implanted under the skin, to monitor the heart over a longer period of time to detect arrhythmias that may be less frequent. Examples include ambulatory ECG, Holter Monitor, Event Monitor or Recorder, and cardiac monitor.

What to Expect During an Electrocardiogram

1.

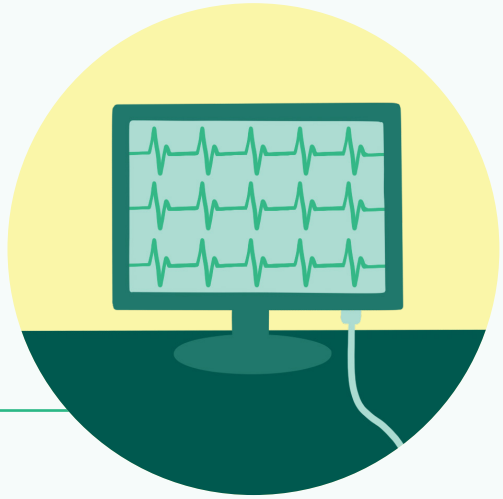
10 electrodes
are attached to you.

2.

Electrodes transmit heart's electrical activity to ECG machines to provide 12 different views of the heart.

3.

ECG machine creates wave pattern representing heart's rhythms.



Imaging Studies

Imaging Studies are used as follow up tests if ECG tests are abnormal, as well as to identify if there are pumping issues known as “cardiomyopathy.” These studies use an X-ray or an ultrasound to create an image of the heart.

Types of imaging studies include an **echocardiogram** (ECHO), **computerized tomography** (CT or CAT Scan), and **magnetic resonance imaging** (MRI).

What to Expect During an Echocardiogram

1. Gel is placed on your chest.

2. Transducer sends sound waves through chest to heart.

3. Sound waves bounce back from heart to transducer.

4. Computer processes soundwaves to make visual of beating heart.

What to Expect During a CT or MRI Scan

1.

You may need a contrast agent, which is a substance used to help internal body structures appear in images.

2.

Lie on table outside the scanner.

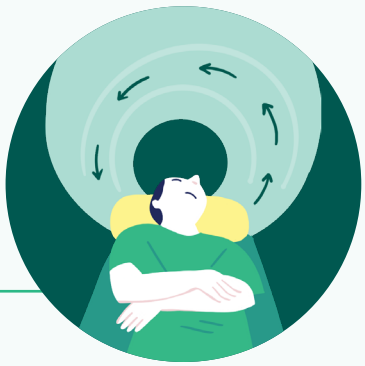
3.

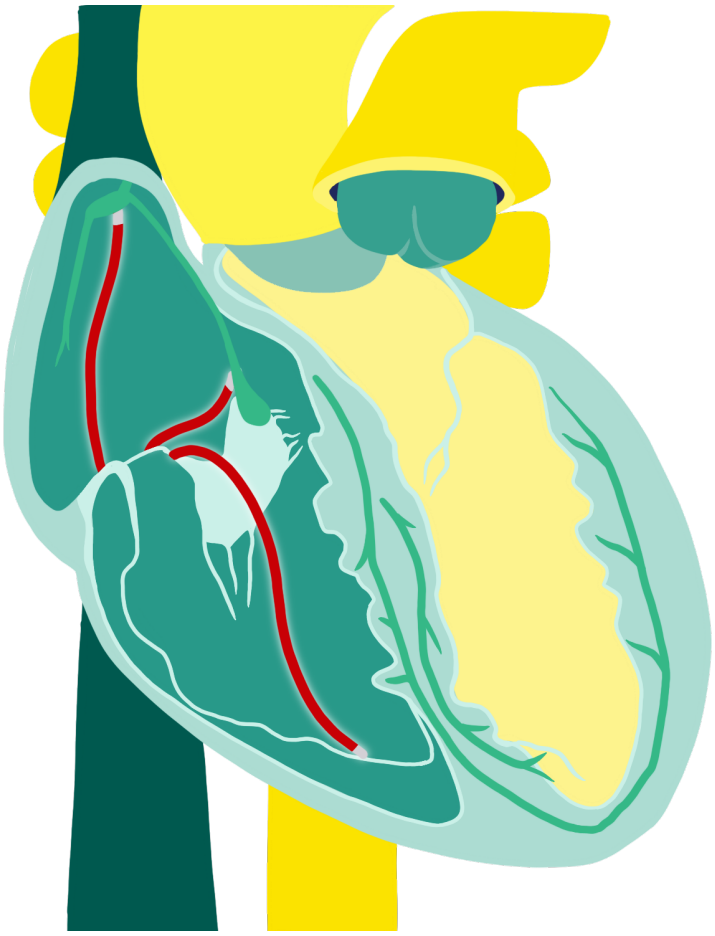
The table slides into scanner.

4.

During a CT Scan - the scanner spins around taking X-rays for approx 10-30 minutes .

During an MRI - the imaging equipment runs for 15-60 minutes.





Electrophysiology Study (EPS)

A test used to evaluate your heart's electrical system and check for abnormal heart rhythms. A thin coated wire electrode (a catheter) is inserted into a vein and threaded through to the heart. The catheter measures the heart's electrical signals. If an abnormal area of the heart is determined to be causing arrhythmias, that area can be treated with cold or heat energy in a process called catheter ablation.

See below for more information on catheter ablations.

Note: Heart issues are often asymptomatic (show no symptoms)—make sure to be evaluated regularly even if you have no symptoms.

How Will I Be Treated?

If your test results are normal, you may be treated by continual monitoring. Typically, the doctor will see you annually to assess your heart health. If your test results are abnormal, doctors may recommend further treatment. Listed below is information about medication, procedures, and devices used to address cardiac problems.

Medication

Talk to your physician regarding medications that may be used to treat heart issues for someone with myotonic dystrophy.

Catheter Ablations

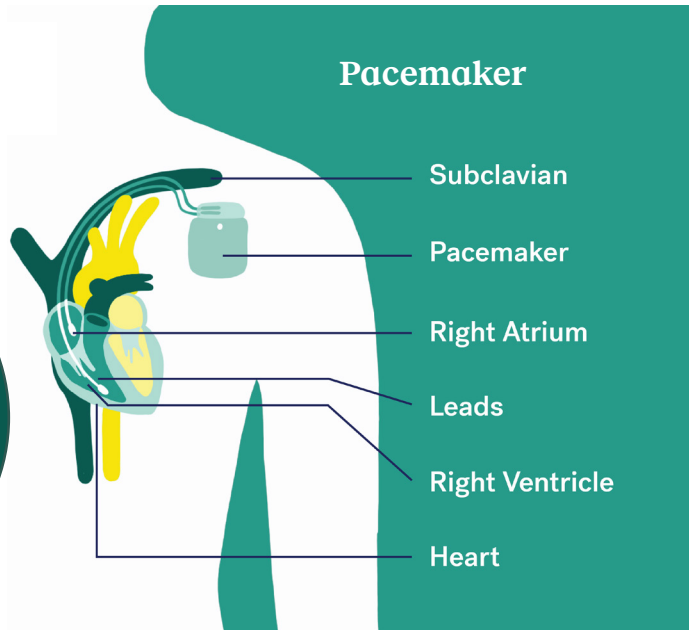
Invasive cardiac procedure where heat or cold energy is applied to the locations in the heart where abnormal electrical signals originate to treat arrhythmias. This is done as part of an electrophysiology study, as explained above.

Implantable Devices

Electronic devices that consist of a pulse generator and wires, called leads, provide electrical signals to the heart to maintain a normal heart rhythm. These devices are implanted via surgery. Most commonly, the doctor will make a small cut below the collarbone and the leads of the device are then guided through a vein into the heart and then connected to the generator. Examples of implantable devices include the following:

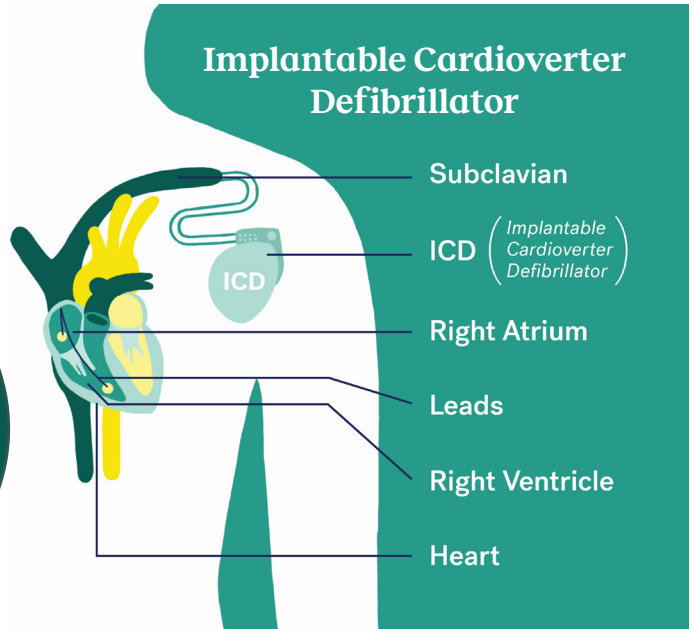
Pacemaker:
Treats a slow heart beat.

Note: Pacemakers can be implanted or there is a smaller version that is placed completely in the heart without additional leads.



Defibrillator or Implantable Cardioverter Defibrillator (ICD):
Can treat both a slow and fast heart beat and potentially interrupt dangerous arrhythmias.

Note: Some implantable devices can be placed either after a cardiac event or to help prevent a cardiac event.



Note: Anesthesia raises special risks to those living with Myotonic Dystrophy. *Anesthesia may be used during surgery for implantable devices and catheter ablation. Since the disease results in a heightened sensitivity to sedatives and analgesics, be sure to provide your doctor with the Myotonic Dystrophy Foundation's Anesthesia Guidelines(ii) and keep a Medical Alert Card(iii) on your person to ensure critical guidelines are followed. (See Appendix.)*

What Can I do to Support my Heart Health?

Be attentive and report your symptoms to your cardiologist or primary care physician.

- Tracking symptoms and reporting them back to your physician can be done through an implantable device, if you have one (e.g., pacemakers, ICDs, or insertable cardiac monitors).

Stay on top of testing

- Get an ECG every 6-24 (typically 12) months.
- Cardiac imaging every 2-4 years (even without symptoms).

Find a specialist familiar with both the heart and myotonic dystrophy and advocate for regular care.

- Use MDF's **Find a Doctor Map**[iv] to find specialists familiar with myotonic dystrophy near you.
- Give medical professionals MDF's comprehensive Clinical Care Recommendations for **Adults with DM1**[v].
- Give medical professionals MDF's comprehensive Clinical Care Recommendations for **Adults with DM2**[vi].
- Give your cardiologist MDF's Clinical Care Recommendations for **Cardiologists**[i].
- If you need help advocating for your care, you may consider utilizing resources provided by the **Patient Advocate Foundation**[vii], which provides case management.

Ask your cardiologist about how to exercise safely.

Stay informed about myotonic dystrophy and cardiovascular management using MDF's resources on myotonic.org, including:

- **Cardiac Issues Related to DM Video**[viii].
- **Heart Health Talk by Saman Nazarian MD, PhD**[ix].
- **Ask-the-Expert: Your Heart & Myotonic Dystrophy**[x].
- Summaries for **Congenital myotonic dystrophy**[xi], **DM1**[xii], & **DM2**[xiii].

Print and fill out a Myotonic Dystrophy Medical Alert Card[iii] to put in your wallet for emergencies.

Additional Information

[i] Clinical Care Recommendations for Cardiologists

Consensus-based recommendations for cardiologists treating adults with DM1.

https://www.myotonic.org/sites/default/files/pages/files/MDF_Consensus-basedCareRecsAdultsDM1_Cardiologists_1_21.pdf

[ii] Anesthesia Guidelines

Practical suggestions for providers managing anesthetics for those living with DM.

https://www.myotonic.org/sites/default/files/pages/files/MDF_PracticalSuggestionsDM1_Anesthesia2_17_21.pdf

[iii] Medical Alert Card

A wallet-sized card with critical guidelines for emergency responders.

https://www.myotonic.org/sites/default/files/pages/files/MDF-Medical-Alert-Card.pdf

[iv] MDF's Find a Doctor Map

A community-informed resource outlining medical professionals who understand and have experience with DM.

https://www.myotonic.org/find-a-doctor-map

[v] Clinical Care Recommendations for Adults with DM1

Evidence-based clinical care guidelines for adults living with DM1.

https://www.myotonic.org/sites/default/files/pages/files/MDF_Consensus-basedCareRecsAdultsDM1_1_21.pdf

[vi] Clinical Care Recommendations for Adults with DM2

Evidence-based clinical care guidelines for adults living with DM2.

https://www.myotonic.org/sites/default/files/pages/files/MDF_Consensus-basedCareRecsAdultsDM2_1_21.pdf

[vii] Patient Advocate Foundation

A foundation with a variety of patient services aimed to increase access to quality healthcare.

https://www.patientadvocate.org/

[viii] Cardiac Issues Related to DM Video

Learn more about the heart, how it may be impacted by DM, and preventative measures you can take.

https://www.myotonic.org/digital-academy/cardiac-issues-related-dm

[ix] Heart Health Talk by Saman Nazarian MD, PhD

Learn more about DM cardiac symptoms and how you can promote heart health.

https://www.myotonic.org/digital-academy/heart-health-2019-myotonic-annual-conference

[x] Ask-the-Expert: Your Heart & Myotonic Dystrophy

A webinar by Pradeep P.A. Mammen, MD, FACC, FAHA, FHFSA an expert in the DM field on DM and the heart.

https://www.myotonic.org/digital-academy/ask-expert-your-heart-dm

[xi] Cardiovascular System – CDM

An MDF webpage all about congenital myotonic dystrophy, including how it may impact the heart.

https://www.myotonic.org/cardiovascular-system-cdm

[xii] Cardiovascular System – DM1

An MDF webpage all about DM1 and the cardiovascular system.

https://www.myotonic.org/cardiovascular-system-dm1

[xiii] Cardiovascular System – DM2

An MDF webpage all about DM2 and the cardiovascular system.

https://www.myotonic.org/cardiovascular-system-dm2

The mission of the Myotonic Dystrophy Foundation is Community, Care, and a Cure.

We support and connect the myotonic dystrophy community.
We provide resources and advocate for care.
We accelerate research toward treatments and a cure.

Myotonic Dystrophy FOUNDATION

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