

# MD

Myotonic Dystrophy  
SUPPORT GROUP

## The Heart and Myotonic Dystrophy

by

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# Myotonic Dystrophy

Heart involvement in Myotonic Dystrophy is one of the most important aspects of the condition because of its potentially serious consequences. Many cardiac complications can be treated if detected early.

## **How does the heart normally work ?**

- The heart has four chambers - the left and right “receiving chambers” or “atria” and the left and right “pumping chambers” or “ventricles”
- Each heart contraction pumps blood to the body, from the left ventricle, and to the lungs, from the right ventricle. This is coordinated by an electrical signal from the right atrium
- The electrical signal then passes throughout special heart cells to the rest of the heart. This electrical system is called the “conduction system”.

## **How is the heart affected in Myotonic Dystrophy?**

- Abnormal slow and fast heart rhythms are called "arrhythmias"
- Arrhythmias are a common complication in myotonic dystrophy
- Arrhythmias contribute significantly to early ill health and even death.

## **Arrhythmias and the problems they cause**

- Arrhythmias reduce the amount of blood that is pumped to the body and if not enough blood reaches the brain, patients can "blackout"
- If enough blood is pumped to keep patients conscious but not enough to exercise, this causes shortness of breath on exercise or sometimes at rest

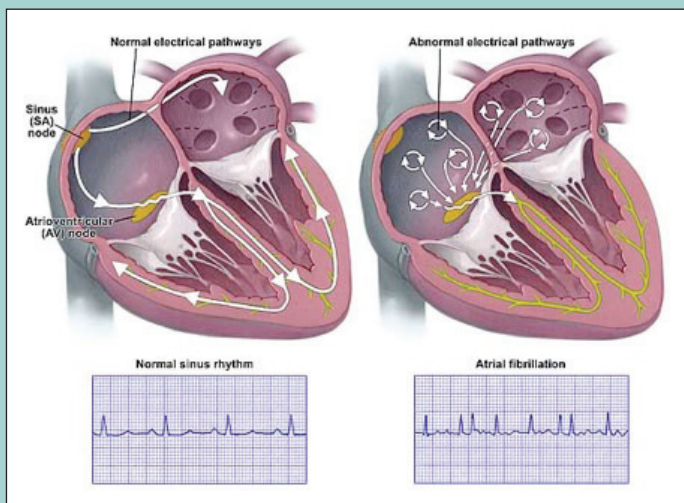
- Arrhythmias can cause palpitations, where a patient feels a heavy, irregular or very slow and forceful heart beat
- Lastly, some arrhythmias can cause clots to form, especially on the inner surface of the left atrium. These can “break off” or “embolise” to the brain and other organs potentially causing a stroke.

## Types of arrhythmias

There are three important types of arrhythmia in myotonic dystrophy. Two of these cause fast arrhythmias and one causes slow arrhythmias.

**1) Fast arrhythmia - Atrial Fibrillation** causes the receiving chambers of the heart or “atria” to beat in an incoordinated way that makes the pulse irregular and fast (see Figure 1).

Figure 1. Atrial Fibrillation



**2) Fast arrhythmia - Ventricular Tachycardia** is when the ventricles beat very rapidly and independently from the atria. If prolonged it can cause the blood pressure to fall and may result in loss of consciousness (see Figure 2). Ventricular fibrillation is when the ventricles beat in a completely chaotic way - this kind of rhythm usually causes a “cardiac arrest” and is potentially life threatening (see Figure 3).

**Figure 2. Ventricular Tachycardia**

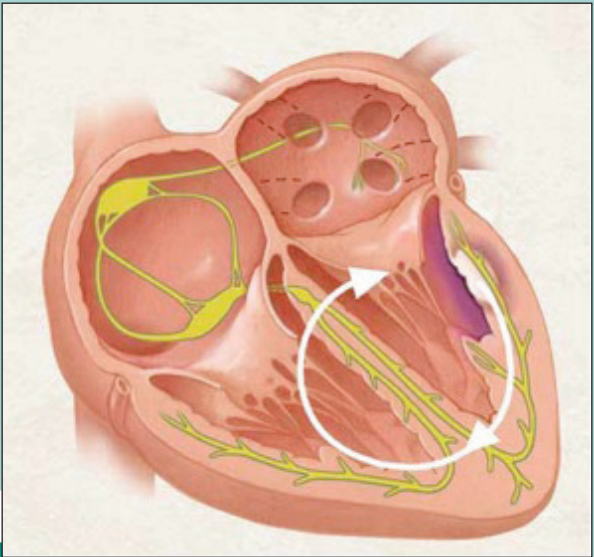
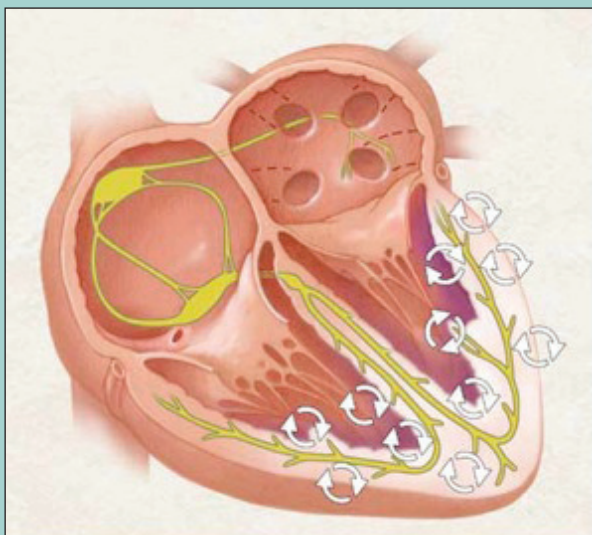
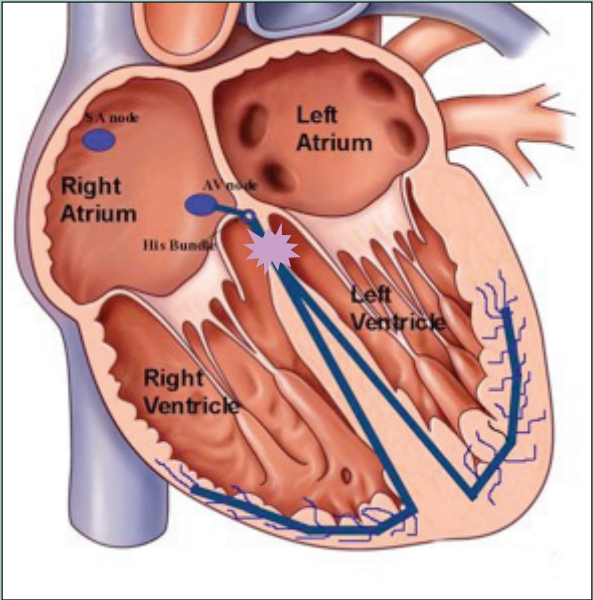


Figure 3. Ventricular Fibrillation



**3) Slow arrhythmia - Heart block** refers to a delay or complete blockage to the electrical signals as they pass from the right atrium to the ventricles. This can cause patients to have poor exercise tolerance due to shortness of breath or to lose consciousness (see Figure 4).

Figure 4. Heart block at the AV node



**Why do patients with Myotonic Dystrophy develop arrhythmias?**

- If the heart of patients with myotonic dystrophy is examined under the microscope there is often selective and



extensive wearing out or “degeneration” and scarring or “fibrosis” of the electrical (conduction) system.

### **How common are arrhythmias in Myotonic Dystrophy?**

- A 10 year follow-up study by Mathieu et al. (1999) demonstrated that out of 367 myotonic dystrophy patients studied, there was a 7 times greater mortality than in the general population. This was mostly caused by complications affecting the heart and breathing/swallowing muscles
- A mild delay in the conduction of electricity from the atria to the ventricles or “prolonged PR interval” occurs in 40% of patients and a delay in the electricity travelling through the ventricles or “increased QRS interval” occurs in 25% of patients. The true incidence of more severe heart rhythm problems is uncertain.

## Investigation for arrhythmias

- There are several tests which can be performed by your doctor to monitor your heart function
- The commonest test is an electrical tracing of the heart or electrocardiogram or “ECG”. This can be performed over 24 or 48 hours with a “Holter monitor”
- An echocardiogram or “echo” looks at the structure of the heart. The structure of the heart in myotonic dystrophy is usually normal but it is important to make sure that other problems are not affecting the heart as well as those caused by myotonic dystrophy.

More complex heart tests are increasingly performed and include:

- An electrophysiological study or “EPS” where a special wire is fed from a vein in the groin or elbow up to the heart so that specialist heart doctors can measure the electricity of the heart conduction system directly

- Magnetic Resonance Imaging or “MRI” is being increasingly performed on the heart. It provides detailed information on the heart structure and function and can show areas of scarring in the heart muscle.
- The role of EPS and MRI in the management of myotonic dystrophy is still being determined by doctors.

### **Treatment of slow or ‘brady’ arrhythmias**

- If the electrical impulse from the atria does not reach the ventricles, this is called “heart block”.
- This requires a pacemaker to be inserted which is a small box usually placed under the skin below the left collar bone which sends wires to the ventricles so that the heart rhythm can be monitored and, if needed, the pacemaker can “pace” the heart if it beats too slowly.

## **Treatment of fast or ‘tachy’ arrhythmias**

- Atrial fibrillation (AF) is usually treated with tablets to slow the fast heart rate down and to thin the blood to prevent clots from forming in the heart
- Sometimes doctors apply an electrical shock to the heart to “cardiovert” or electrical change the heart rhythm back to normal
- However, even if this is successful, the heart can often revert back to atrial fibrillation and tablet treatments will be needed
- Ventricular tachycardia or fibrillation may require an “implantable cardioverter defibrillator” or “ICD”.
- This is also a small electrical box that is fitted under the skin below the left collar bone with wires that pass to the heart. If the heart has an episode of ventricular arrhythmia, then the electrical box shocks

the heart back to a normal heart rhythm

- ICDs can be uncomfortable because they can accidentally deliver an electrical shock when it was not necessarily needed and therefore inserting an ICD requires careful consideration.

## **What do we still need to know?**

### **1. Why does myotonic dystrophy affect the conduction system?**

- We do not understand why the conduction system is particularly affected in myotonic dystrophy.

### **2. How should doctors monitor for heart problems? How often should doctors monitor the heart? When do we treat arrhythmias?**

- Most doctors perform at least an ECG every year
- There are regional differences in the UK

and throughout the world as to what further investigations are best to perform

- At UCLH/Queen Square we perform an ECHO and 24 hour Holter monitor approximately every 2 years
- If there are any suggestions from these basic tests that the heart rhythm is abnormal we would offer patients an EPS and/or an MRI of the heart and this is under review to see if it helps patients.

### **3) Which patients would benefit from a pacemaker and/or ICD? Does inserting a pacemaker and/or an ICD affect morbidity and mortality?**

- These questions remain unanswered and there are currently clinical trials and studies being conducted.

## Summary

- The conduction system of the heart is frequently involved in myotonic dystrophy
- Serious complications can arise from heart problems but many of these can be treated
- Patients with myotonic dystrophy need a proactive approach to management of their heart with a multidisciplinary team involving at least a neurologist and cardiologist
- Future trials are required to inform doctors as to which patients need treatment and when this should be started

## References:

1) Mathieu J, Allard P, Potvin L, et al. 10-year study of mortality in a cohort of patients with myotonic dystrophy. *Neurology*. 1999;52:1658-62.

## Other publications available from the Myotonic Dystrophy Support Group:

- Anaesthesia and Sedation for patients with Myotonic Dystrophy
- Basic Information for Midwives
- Bowel Problems in Myotonic Dystrophy
- Congenital Myotonic Dystrophy
- Excessive Daytime Sleepiness and Myotonic Dystrophy
- Facts for patients, family members and professionals
- Myotonic Dystrophy and the Eye
- Myotonic Dystrophy Support Group
- Relatives Information
- Why do we get new families with Myotonic Dystrophy?



**Myotonic Dystrophy**  
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