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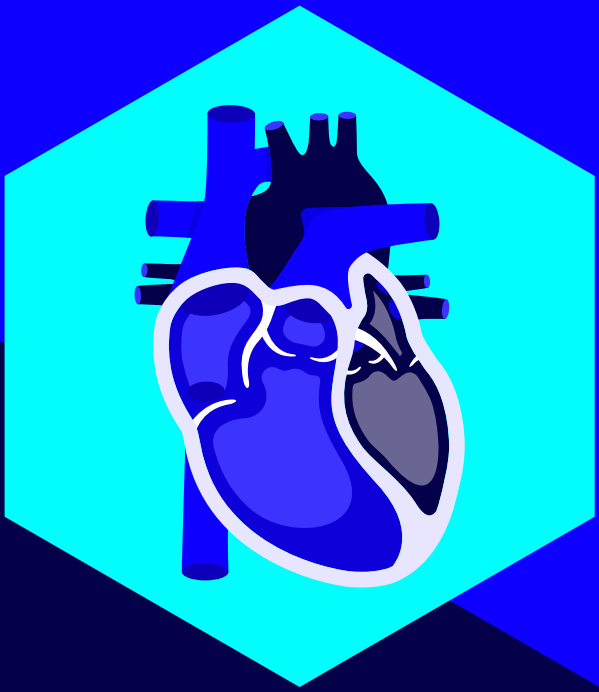
Information for patients and families

Cardiac care for Duchenne muscular dystrophy



Duchenne UK

dmdcareuk.org

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Cardiac care for Duchenne muscular dystrophy

DMD Care UK has brought together experts in neuromuscular diseases and cardiologists to agree the best standards of cardiac (heart) care for people with Duchenne muscular dystrophy in the UK. These are the clinical recommendations, which are based on the latest evidence, expert-opinions, and the patient perspective.

Full clinical recommendations can be accessed at **dmdcareuk.org/clinical-recommendations**

This booklet is designed to help DMD patients, carriers, and their families understand the clinical recommendations for their cardiac care.

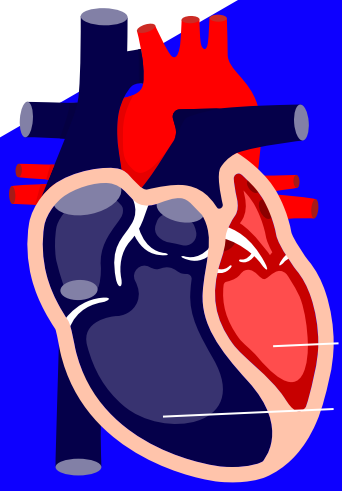
How the heart is affected in DMD

The heart muscle (also called the **myocardium**) needs **dystrophin** to function efficiently. In DMD, a lack of dystrophin means that heart muscles weaken over time. Heart cells are replaced by scar and fatty tissue, which leads to a type of heart muscle disease called **dilated cardiomyopathy**

The age when cardiomyopathy happens in DMD can vary from person to person, from early childhood to adulthood.

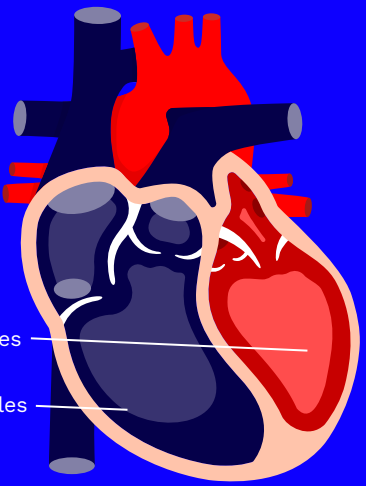
Children with DMD don't usually have cardiac symptoms. However, weakening heart muscles are part of the natural progression of the disease. Therefore, early monitoring, preventative treatment, and ongoing management are important for keeping the heart healthy into adulthood.

Normal heart



Chambers relax and fill, then contract and pump.

Heart with dilated cardiomyopathy



Muscle fibres have stretched
Heart chambers enlarge

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How do we monitor heart function?

The heart can be assessed in several ways. In DMD, heart assessment should always include an **electrocardiogram or ECG** (a test used to check the heart's rhythm) and an **echocardiogram or ECHO** (a type of **ultrasound** scan).

Echocardiogram (ECHO)

An ECHO shows how the heart is working as a muscle to pump blood around the body. It is used to monitor and measure the main heart-pumping chamber, **the left ventricle.**

You may be given measurements such as the **left ventricular ejection fraction (LVEF%)**, or **the left ventricular fractional shortening (LVFS%)**. These summarise how well the heart muscle is contracting (squeezing) and sending blood to the body.

If these tests show any cause for concern, your child may be offered a cardiac MRI. This will provide a more detailed assessment of the heart than an ECHO.

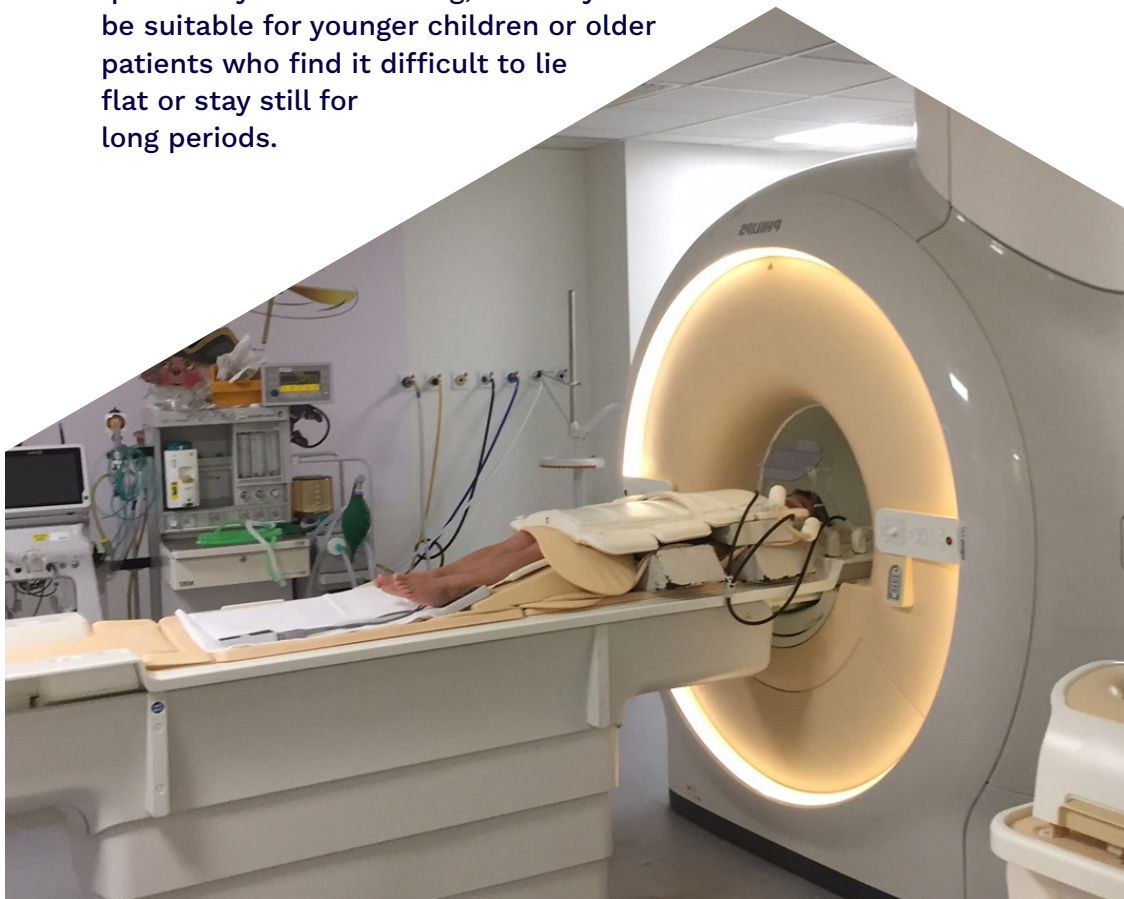
Cardiac MRI

MRIs are helpful when doctors need more information about whether changes are taking place in the heart, or if they need to identify whether the heart muscle is being replaced by scar tissue. This process is called fibrosis.

To detect fibrosis on an MRI, a substance called gadolinium must be injected into an arm vein. This is normally very safe. However, very rarely, it can cause serious reactions. The other heart assessments by MRI can take place without this injection.

Some people cannot be offered an MRI scan safely. For example, if they have implanted metalwork.

An MRI takes longer than an ECHO (around 45 minutes). It is quite noisy when scanning, and may not be suitable for younger children or older patients who find it difficult to lie flat or stay still for long periods.



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Blood pressure

Blood pressure should be checked regularly, especially in people on long term treatment with steroids, as these can cause high blood pressure **(hypertension)**. If untreated, hypertension can speed up heart damage, and increase the risk of other heart diseases.

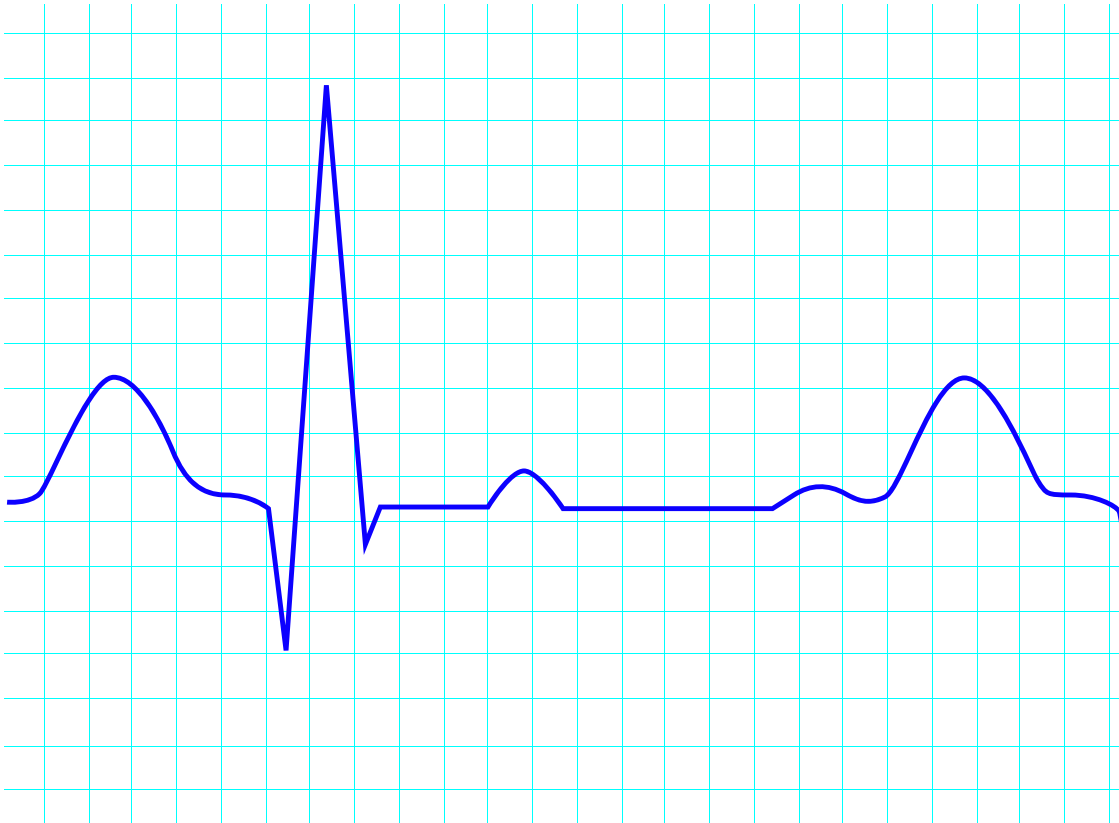
The heart medications used in DMD also lower blood pressure. Sometimes, this can cause blood pressure to become too low **(hypotension)**, which can cause symptoms such as dizziness or feeling sick. However, if these symptoms are minimal, lower than normal blood pressure is helpful for the heart.

If your child becomes seriously unwell for other reasons (such as a serious chest infection or dehydration) the doses of some heart medications may be reduced temporarily to avoid them lowering blood pressure further. The full doses can be restarted once they recover.

Other tests

When the heart has become weakened by DMD, its beating can become irregular, either all the time or on and off. Your child may not be aware that this is happening, or they may have momentary feelings of the heart 'skipping and jumping'.

A standard **electrocardiogram (ECG)** – a bit like a 'snapshot' heart recording – will show if the heart is out of rhythm at that time. However, a continuous longer-duration recording – a 24–48-hour Holter ECG – is often needed to investigate symptoms that come and go, or to see what the heart 'gets up to' over a longer period of time.



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When should the heart be checked in DMD?

When your child is diagnosed with DMD, your neuromuscular team should discuss how DMD affects the heart with you.

Your child should be seen by a paediatric **cardiologist** by age 6 for a more detailed discussion and to have baseline heart tests.

Following this, their heart should ideally be checked yearly. This is particularly important from age 10, when changes in the heart are more likely to be seen on an ECHO.

Your medical team should prescribe heart medication early, while the heart is still strong, to protect the heart for as long as possible.

Yearly checks should continue throughout adolescence and adulthood. These will monitor changes in heart function, provide a 'running commentary' on how the heart is doing, and allow doses of heart medication to be changed if needed.

How can we delay cardiomyopathy in DMD?

It is not currently possible to prevent heart weakness in DMD completely. However, steroids and heart medications can slow the decline in heart function.

This means your child's heart should not cause symptoms or affect their quality of life until late stages of DMD.

To get the most benefit from heart treatments, they should be started **no later than age 10**, often taken in combinations and in doses recommended by your medical team.

AGE 6
baseline tests

AGES 6+
yearly checks

BY AGE 10
heart medication prescribed

THROUGHOUT ADOLESCENCE AND ADULTHOOD
yearly checks continue, additional medications prescribed or doses adjusted

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How do we treat cardiomyopathy in DMD?

What medications might be prescribed

There are several different 'families' of drugs that protect the heart in DMD. Your medical team will discuss benefits and side-effects with you and monitor your child closely when they are first introduced. They will also check and adjust dosage according to your child's weight and age.

Angiotensin converting enzyme inhibitors (ACEis or ACE inhibitors)

These relax the blood vessels, reduce blood pressure and support heart function. ACE inhibitors allow the heart to work more effectively, even when damaged by DMD.

The names of some ACE inhibitors are: **enalapril, lisinopril, perindopril, ramipril** – but there are others.

ACE inhibitors are the first line of treatment for DMD. They should be introduced **no later than age 10** to prevent future heart decline, even though heart function will usually still be normal at this age.

ACE inhibitors are usually well tolerated even by young children. However, rarely they can cause side effects. This can include a persistent dry cough. If side effects occur, most people can be changed to an **angiotensin-receptor blocking agent (ARB)** medication.

Angiotensin-receptor blockers (ARBs)

This group of medications have similar actions to ACE inhibitor drugs but work slightly differently. Your child may be prescribed ARBs if they experience side effects with ACE inhibitors.

The names of some ARBs are: **losartan, irbesartan, candesartan** – but there are others.

Beta-blockers (BBs)

Beta-blockers slow heart rate and reduce blood-pressure by reducing the effects of the hormone adrenaline. They may be prescribed if your child's heart rate is persistently too fast (sinus tachycardia).

The names of some beta-blocking drugs are: **metoprolol, bisoprolol, carvedilol** – but there are others.

Beta-blockers can cause cold extremities (toes and fingers) especially in older, less mobile adults with DMD.

Mineralocorticoid receptor antagonists (MRAs):

MRAs are a type of diuretic. These lower blood pressure and reduce excessive fluid around the body by increasing the amount of urine (wee) you pass.

At more advanced stages of heart weakness, the body retains excessive water, so MRAs help to clear this. However, MRAs also help to prevent scarring in the heart, so can sometimes be prescribed in early stages of DMD in combination with an ACE inhibitor too.

Two currently available MRAs are **spironolactone** and **eplerenone**.

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Corticosteroids (also known as steroids)
Steroids are prescribed to most children diagnosed with DMD on a long-term basis and from an early age, to help slow the progression of muscle weakness.

They also have a protective effect on heart muscle.

However, long term use can also cause hypertension and weight gain which can have a negative effect on heart function, therefore their use needs to be carefully monitored and managed.

The two steroid medications used currently in DMD are prednisolone and deflazacort.

When should heart medication be started?

ACE inhibitors (or ARBs) should be prescribed no later than age 10 as a preventative measure to protect the heart in DMD, even if the heart seems completely normal on ECHO-scanning.

If an ECHO or MRI shows any signs of heart weakness, or your cardiologist recommends it, combinations of heart medication may be prescribed to keep the heart healthy for longer. For example, an ACE inhibitor, an MRA and a BB may all be used together.

Noticeable heart symptoms do not usually happen until very late in DMD-related cardiomyopathy. These symptoms should not be used as a guide for when to start or increase medication.

Warning signs and what to do in an emergency

You should have an emergency plan for your child, discussed with your medical team.

We recommend installing the Duchenne UK In Case of Emergency App onto your phone (and your child's, if they have one). You can upload information about your child, their DMD and the latest clinical assessments, including for their heart, so that you can share it with medical professionals in an emergency.

You can find it at **dmdcareuk.org/emergency-resources**

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If any of the following symptoms occur for the first time or have not been previously investigated, they may indicate a problem with the heart and you should call 999 and seek urgent medical attention:

- **faintness or feeling like you might collapse**
- **loss of consciousness or 'blackout'**
- **palpitations or awareness that the heart is 'racing' for no understandable reason**
- **deterioration in usual breathing pattern or feeling continuously short of breath when you wouldn't have been normally. Struggling to breathe, especially if this is worse when lying down**
- **Onset of weakness on one side of the body, even if resolves within minutes, e.g: face, arm or leg, slurred speech or unable to speak**
- **loss of vision or being unable to see in a particular direction, even if it resolves within minutes**

Women and girls who are carriers

Women and girls with a DMD-gene mutation are known as 'DMD carriers'. They are at increased risk of developing heart problems. As in boys and men with DMD, symptoms of heart failure in female carriers will only occur when the heart has already lost most of its pumping function.

DMD carriers should have regular heart checks about every 3-5 years, from the point of their diagnosis onwards. If heart problems are identified, combination treatment (typically ACE inhibitor and MRA) can be started to protect the heart. After starting these treatments, DMD carriers should have yearly heart checks.

Women who are carriers and are planning pregnancy should have pre-conception genetic counselling, so that they understand the risks of passing on the DMD gene to their children. If the DMD carrier is on heart medication, they should discuss pregnancy plans with their cardiologist.

Not all women who have a child with DMD are DMD carriers. When a child is diagnosed with DMD, genetic counselling and assessment should check if the mother is a carrier. If this is confirmed, testing can be offered to other female family members (daughters, maternal aunts and grandmothers).

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FAQs

Why aren't protective medicines (like ACE inhibitors) started even earlier?

Whilst ACE inhibitor drugs are generally well-tolerated in boys with DMD, they do have side effects. The evidence and agreed expert opinion is that 'by the age of 10' is the best balance between starting medication to protect the heart in time and not starting unnecessarily early, when it is not yet of benefit.

You might choose to talk to your cardiologist about beginning these medicines before your child is 10. This decision should be based on their judgement, experience, and your preferences.

Why isn't my child offered a cardiac MRI every year?

Usually, your cardiologist will be able to monitor your child's heart using ECGs and ECHOs alone. A cardiac MRI might only be needed if ECHO results do not give enough information.

Preventative medicines should be started by the age of 10, before an ECHO or MRI detects heart problems. This means that your cardiologist doesn't need the results of a cardiac MRI to decide whether to begin this therapy.

Some children find it difficult to be in an MRI scanner, so need a general anaesthetic beforehand. Therefore, it is important to only use an MRI where this could benefit treatment decisions. The need for sedation or general anaesthesia to perform a heart MRI scan is rarely justified.

If my child's heart is affected by DMD, is it safe for them to exercise?

Yes, it is important that your child exercises and does so safely. You should speak to your physiotherapist about preferred and safe forms of exercise. Some activities should be avoided for people with DMD – especially scooters, bouncy castles, trampolines, and rugby.

You can find some more guidance at **duchenneuk.org/sports-and-activities-with-dmd**

About DMD Care UK

DMD Care UK is a nationwide initiative to ensure every person living with Duchenne muscular dystrophy (DMD) in the UK has access to the best care.

This project is funded by Duchenne UK, Joining Jack and the Duchenne Research Fund. They work closely with the John Walton Muscular Dystrophy Research Centre in Newcastle and in collaboration with the North Star Network, funded by MDUK.

DMD Care UK has produced a series of information resources for UK DMD patients, families and other non-specialists on the recommended standards of care for DMD.

Find out more at **dmdcareuk.org**

Do you have questions or feedback about this booklet? Get in touch with **support@duchenneuk.org**

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Notes

You may find it useful to jot down questions, thoughts or useful information here. We have included a few suggestions below.

You can also find a glossary of terms here:
dmdcareuk.org/glossary

Name and contact details of cardiologist:

Cardiac medications prescribed (including dates and doses):

Details of next appointment/s:

This booklet has been developed by DMD Care UK and reviewed by clinicians and the family focus group within the project. It is based on the DMD Care UK cardiac care recommendations endorsed by the British Cardiovascular Society.

This booklet is for informational and educational purposes only. You should always discuss your medical care with your clinical team.

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