

# Haemochromatosis and Cardiomyopathy

## About cardiomyopathy caused by haemochromatosis

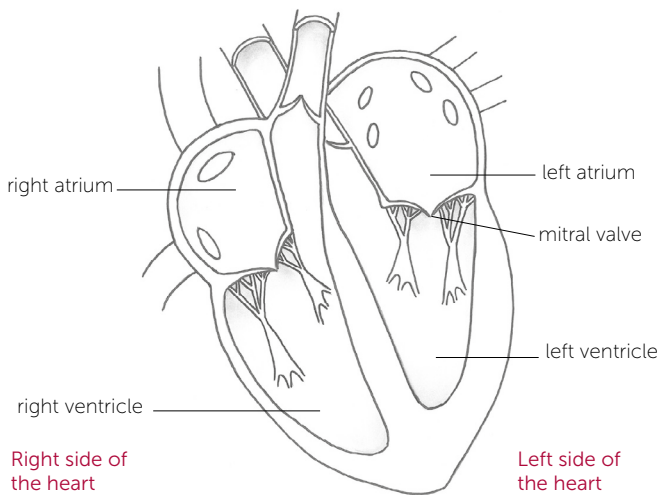
- Genetic Haemochromatosis is an inherited condition where a person absorbs too much iron from their diet, leading to iron overload.
- Iron overload can cause various symptoms, and can cause damage to the body's organs.
- Where haemochromatosis causes a build up of iron in the heart it can cause cardiomyopathy.

This information sheet is for people with haemochromatosis who have developed cardiomyopathy. For more information on haemochromatosis see the 'further information' section at the end of this information sheet.

## What is cardiomyopathy?

Cardiomyopathy is a disease of the heart muscle ('cardio' means heart, 'myo' means muscle and 'pathy' means disease). It isn't a single condition, but a group of conditions that affect the heart and reduce its ability to pump blood around the body.

It can affect the shape of the heart, or the size and thickness of the muscle walls, which reduces the heart's ability to function properly. There are different types of cardiomyopathy, which vary in how much and which parts of the heart they affect.



- **Hypertrophic cardiomyopathy** – the muscle wall of the left ventricle becomes thickened and stiff, so it has to work harder to pump blood.
- **Arrhythmogenic cardiomyopathy** – the heart muscle cells do not join together properly, and get replaced by scar tissue or fat. This makes the muscle stretched and weaker, and so less able to pump blood.

- **Left ventricular non-compaction (LVNC)** - the heart muscle does not develop normally in an unborn baby, but develops small indentations which appears spongy. This affects how the heart works.

See our information sheet 'What is cardiomyopathy'.

## What causes cardiomyopathy?

There are many different causes of cardiomyopathy. Although some causes are unknown, it is often a genetic condition (caused by a mutation in one of the genes). Cardiomyopathy can also be caused by haemochromatosis. When this happens, it is called a secondary (or 'acquired') condition, as it is caused by a primary condition (the haemochromatosis).

## What happens in haemochromatosis?

Iron from the diet is normally stored by the body in the bone marrow, with small amounts stored in the liver (to form new red blood cells). However, in haemochromatosis, an excessive amount of iron (more than the body needs) is absorbed by the body. The iron levels build up as the body is unable to get rid of it, and this is called 'iron overload'. Over time, this overload leads to a build up of iron in different parts of the body, including the liver, pancreas, skin, joints and pituitary gland. The overload damages the organs and causes a number of symptoms including arthritis, chronic fatigue, liver disorders and abdominal pain.

## How does haemochromatosis cause cardiomyopathy?

As well as building up in other organs of the body, iron levels can build up in the heart. This happens slowly, and the heart may continue to work well until the overload becomes quite advanced. This can cause restrictive or dilated cardiomyopathy.



In restrictive cardiomyopathy, the muscle walls of the ventricles (the lower, pumping chambers of the heart) become stiff, restricting the heart's movement. Although the heart can continue to contract to pump blood around the body, the stiffened muscle of the ventricles does not relax properly which means that the ventricles can't fill with blood. So the ventricles receive less blood than normal and blood flow around the body and the heart is reduced. Also, blood gets 'backed up' as it cannot enter the heart as normal. This causes a build-up of pressure in the top chambers of the heart, a backup of blood in the lungs, and increased pressure in the veins in the neck and the liver. Without treatment, the left ventricle of the heart can become enlarged and stretched, and develop into dilated cardiomyopathy.

In dilated cardiomyopathy, the enlargement of the left ventricle makes it weaker and less able to pump blood out of the heart, which reduces the amount of blood leaving the heart.

Knowing that iron overload can affect the heart can help to identify any early symptoms of this, and allows treatment to be started earlier. The sooner treatment is started the more likely it is to be effective in reducing and controlling symptoms.

### What are the symptoms of cardiomyopathy caused by haemochromatosis?

Cardiomyopathy caused by haemochromatosis usually causes symptoms that are collectively known as 'heart failure'. Heart failure happens when the heart isn't pumping well enough, either because the walls of the ventricles are enlarged, weakened, damaged or too stiff, or the heart's valves don't work properly.

This affects how well the heart pumps, how well it fills with blood between, or how well the blood flows through the heart. It also causes an increased pressure in blood vessels and a build up of fluid in the lungs and tissues.

Although the term can sound scary, heart failure just means that the heart is not working properly, and needs support in order to meet the demands of the body.

The symptoms of heart failure include the following.

- **Breathlessness** – fluid builds-up on the lungs, making it harder to breathe. It might be worse when lying down or at night, and can affect sleep.
- **Tiredness** – as the heart function is reduced, less energy is delivered to the tissues, which causes fatigue. This might happen all the time, and so doing any activities can be exhausting.

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- **Swollen ankles and tummy** – build up of fluid in the tissues, as the heart isn't pumping well enough to remove excess fluid, can cause swelling (oedema).




As well as heart failure, some people have arrhythmias. These are abnormal heart rhythms, where the heart beats too fast, too slow, or erratically, because the electrical messages which control the heart's rhythm are disrupted. Arrhythmias can be very serious, and include the following.

- **Atrial fibrillation (AF)** – this is a common arrhythmia caused by a disruption of the electrical messages that normally cause the heart muscle to contract. The atria (top chambers of the heart) beat very quickly and are uncoordinated. This can make the flow of blood around the atrium 'turbulent', and the heart less efficient at pumping out blood. These uncoordinated messages can also become transferred to the lower chambers of the heart (the ventricles). This can cause a fast and irregular heart rhythm. Although in itself it is not life-threatening, it can cause uncomfortable palpitations (feeling your heart beating). However, the turbulent flow of blood can increase the risk of blood clots forming, which can increase the risk of a stroke. Blood thinning drugs are used to reduce the risk of blood clots.
- **Ventricular fibrillation (VF)** – this is an arrhythmia where the contraction of the ventricles (lower chambers of the heart) is uncoordinated, and they 'quiver' rather than contract normally. This means that blood is not pumped out of the heart effectively. This is life-threatening as the heart suddenly stops beating (a cardiac arrest), and needs urgent medical attention.
- **Ventricular tachycardia (VT)** – this is an arrhythmia where the normal passage of electrical impulses in the heart is interrupted by an abnormal impulse coming from within the left ventricle. This can result in a fast heart rate. This can reduce the output from the heart and cause the person to feel dizzy or lose consciousness.

### How is cardiomyopathy diagnosed?

There are several tests that might be done to diagnose cardiomyopathy, which look at the structure and function of the heart. Tests include the following.

- **Physical exam and medical history** – to see what physical symptoms are happening and what may be possible causes of these symptoms.
- **ECG (electrocardiogram)** – this looks at the electrical activity of the heart and how the spread of electrical messages pass through the heart muscle. It is also used to see whether

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arrhythmias are happening. An ECG might be done during exercise (if exercise usually brings on the symptoms).

- **Chest X-ray** – this looks at the size and structure of the heart, and whether you have fluid on the lungs (a symptom of heart failure).
- **Echo (echocardiogram)** – this is a type of ultrasound scan, which uses sound waves to create echos when they hit different parts of the body. Echos look at the structure of the heart and how it is working.
- **MRI (magnetic resonance imaging) scan** – this scan produces high quality images and is used to look at the structure of the heart and blood flows through it. It can also show the iron deposits and fibrosis within the heart muscle.

## How is cardiomyopathy treated?

Cardiomyopathy itself cannot be cured, but the symptoms it causes can be. What treatment is used depends on what symptoms the person is having, and aims to reduce the workload on the heart and support it to function.

Treatment usually follows the NICE (the National Institute for Health and Care Excellence) guidance for heart failure, and includes the following.

- **Diuretics (water tablets)** – help to reduce water retention (which causes swelling in the ankles and around the lungs) by encouraging the kidneys to produce more urine.
- **ACE inhibitors (angiotensin-converting enzyme inhibitors)** – relax and open up blood vessels, making pumping blood easier and takes strain off the heart. They can control blood pressure, and control and prevent worsening of heart failure symptoms.
- **Pacemaker** – these send electrical impulses to the heart to stimulate the contraction of the heart muscle, and set a normal heart rhythm (if the heart is unable to set its own pace, or the heart rhythm is disrupted).
- **CRT (cardiac resynchronisation therapy device or biventricular pacemaker)** – this special type of pacemaker is used when the person has left bundle branch block (LBBB). LBBB is where there is a block in part of the electrical messaging pathway that normally cause the heart to contract. The pacemaker helps to resynchronise (coordinate) the electrical messages and improve how well the heart pumps.

- **Beta blockers** – slow down the heart rate and control arrhythmias.

*For more about heart failure see our information sheet 'About heart failure'.*

*For more about the NICE guidelines visit [www.nice.org.uk](http://www.nice.org.uk) and search 'heart failure'.*

If the person has arrhythmias they may have specific treatment for this, which may include the following:

- **Anticoagulants (blood thinners)** – may be used in people with arrhythmias to reduce the risk of blood clots forming, which could lead to a stroke.
- **Anti-arrhythmic medication** – used to control abnormal heart rhythms.
- **ICDs (implantable cardioverter defibrillator)** – which monitors for any arrhythmias in the ventricle (bottom chamber of the heart). Depending on the type of arrhythmia, the ICD will treat by delivering a therapy (small electrical shock).

## Treatment for haemochromatosis

As well as treating the symptoms of cardiomyopathy, treatment for the underlying haemochromatosis will also be given.

*For more information on treatment see [www.haemochromatosis.org.uk](http://www.haemochromatosis.org.uk)*

## Further information

The following have more information about haemochromatosis.

### NHS Choices

[www.nhs.uk](http://www.nhs.uk) and search 'haemochromatosis'.

### The Haemochromatosis Society

The charity for people with the condition, and those supporting them.

[www.haemochromatosis.org.uk](http://www.haemochromatosis.org.uk)

*Send your feedback to [contact@cardiomyopathy.org](mailto:contact@cardiomyopathy.org)*

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