

# Heart failure or cardiomyopathy

This can be quite common though is not always present. It can also switch from one type to another.

## Different types of cardiomyopathy and heart problems seen in Barth syndrome

- Dilated cardiomyopathy (DCM) is when the heart is larger than normal and does not pump as well as it should.
- Hypertrophic cardiomyopathy (HCM or HCOM) is when the heart muscle becomes too thick and stiff to pump properly.
- Endocardial fibroelastosis (EFE) is when there is dense white thickening of the inner lining of the heart.
- Left ventricular non-compaction (LVNC) is when there are deep pockets in the wall of the left ventricle, making it look like a sponge, instead of being nicely compacted.

Your baby might be affected whilst still in the womb or may be diagnosed after birth or during infancy. Some mothers have a history of miscarriages and stillbirths.

## Symptoms of cardiomyopathy

- Paleness
- Puffiness
- Feeling tired
- Crying weakly or faintly
- Struggling to feed
- Breathlessness or breathing faster than usual or grunting when breathing out
- Fainting
- Chest pain with a cold sweat
- Palpitations

In babies, heart failure can sometimes look like a chest infection that doesn't get better after a week or two.



## Diagnosing cardiomyopathy

Your doctor may listen to your baby's heart with a stethoscope, check his blood pressure, do a chest X-ray, an electrocardiogram (ECG) and an echocardiogram (Echo).

### ECG (electrocardiogram)

This is the most basic, non-invasive test. It involves taping electrical leads onto the chest, legs and arms and taking readings of the electrical activity of the heart. These are printed out onto paper for the doctor to examine. It only takes a few minutes and causes no discomfort

### Echocardiogram (echo)

This non-invasive test uses ultrasound waves to look at the structure of the heart. It produces a picture of the heart and allows doctors to measure the size of the chambers of the heart and how thick the heart muscle is and how well it is working. It also looks at the heart valves and can show if there is any regurgitation (when some of the blood leaks back through a valve in the wrong direction).

### Holter Monitor or 24 Hour Tape

This is like a 24 hour ECG where the child's heartbeat is recorded whilst he carries on with his normal activities. It is used to make sure there are no irregular heart rhythms.

### Blood Tests

These may be needed and a local pain-killing gel can be used on the skin to reduce discomfort.



## Common Cardiac Medications used in Barth syndrome

Different medications can help to take some strain off the heart and help it function better.

**ACE inhibitors:** captopril, enalapril, lisinopril etc. These medicines are good at controlling the symptoms of heart failure and preventing it from getting any worse.

**Beta blockers (carvedilol):** These slow the heart rate and reduce the amount of work the heart has to do, so that it needs less oxygen, blood and nutrients. Some beta-blockers can help control abnormal heart rhythms.

**Diuretics:** furosemide, spironolactone. These reduce the workload of the heart by making sure that the body does not hold onto too much salt or water.

**Anti-coagulants:** warfarin, aspirin. These prevent harmful blood clots from forming in the heart.

## Heart Function can improve and deteriorate

Many children do well on their heart medications but, for some reason which we don't yet understand, their heart function can sometimes get worse, requiring a heart transplant. About one in four boys with Barth syndrome in the UK and one in seven worldwide have needed a heart transplant.

On the other hand, we've seen a surprising number of dramatic improvements in the heart function of many children, to the point that heart scans (echocardiograms) return to normal. This improvement can either continue but it might also worsen again later.

## Arrhythmia

There is also a small risk of arrhythmia (seen in about 10% of patients) which is usually accompanied by the warning signs of nausea, pallor or dizziness.

Many of our families have found that a basic resuscitation course (CPR) is very helpful and puts their mind at rest since it gives them the training they might need to deal with an emergency.