Amyloid Cardiomyopathy

CARDIOMYOPATHY
Australia New Zealand
The Big-Hearted Community

A guide for people with amyloid cardiomyopathy, families, and carers in Australia and New Zealand.

Amyloid cardiomyopathy, also called cardiac amyloidosis, is a condition where incorrectly shaped proteins (due to misfolding) build up in the heart. These misfolded proteins collect outside the heart's cells and form clumps called amyloids, which make the heart stiff and less able to pump blood properly. This is a serious condition, but treatment is available – finding it early can make a big difference.

This information is for people living in Australia or New Zealand who are affected by amyloid cardiomyopathy. It provides general information and should not replace the advice of your doctor or health care team.

What is Amyloid Cardiomyopathy?

Amyloidosis is where the body's normal proteins change shape and stick together, forming clumps called amyloids. These clumps can build up in organs such as the heart, kidneys or nerves and interfere with how they work.

Cardiomyopathy means a disease affecting the heart muscle. So, when amyloid clumps collect in the heart, it's called amyloid cardiomyopathy. The build-up of amyloids makes the heart's walls thicker and stiffer – not because the muscle grows, but because abnormal protein is gathering in the tissue. This can make it harder for the heart to pump blood.

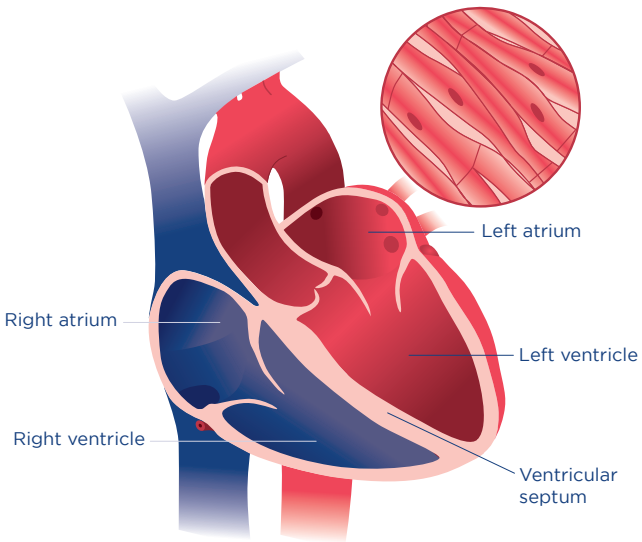
There are different types of amyloidosis that can affect the heart, and treatment depends on which type is present.

What are the types of Amyloid Cardiomyopathy?

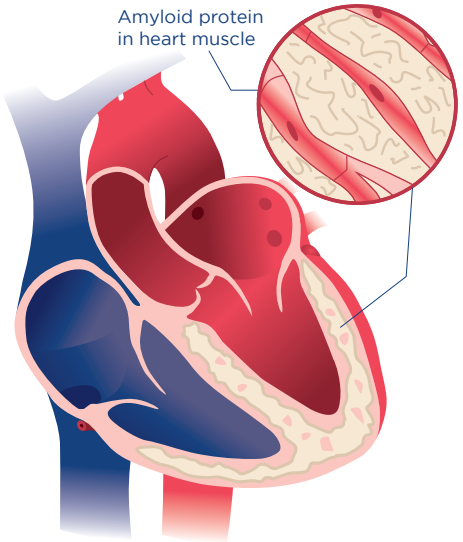
There are two main types of amyloid cardiomyopathy, caused by two different proteins.

1. **AL Amyloidosis** happens when **light chain proteins** – normally part of antibodies (proteins that help the body fight germs) – change shape (misfold) and clump together. The bone marrow, where these proteins are made, sometimes makes too many light chains because of a problem with some of its cells. When these proteins clump together, they form amyloid, which can build up in the heart and reduce how well it can pump blood.
2. **Transthyretin cardiomyopathy (ATTR-CM)** happens when **transthyretin** (TTR) – a protein that helps transport vitamin A and thyroid hormones in blood – changes shape and becomes unstable. When this happens, TTR forms sticky clumps called amyloid which can build up in the heart and reduce how well it can pump blood. There are two types of ATTR-CM:

Heart without Amyloid Cardiomyopathy



Heart with Amyloid Cardiomyopathy



- **Hereditary (hATTR-CM)** is caused by a genetic variant in the transthyretin gene. This change makes the transthyretin protein in the body less stable, so it's more likely to stick together and form amyloid deposits. This can affect different parts of the body, especially the heart and nerves, causing problems like tingling or numbness in the hands and feet, and issues with autonomic functions such as blood pressure or digestion.
- **Wild Type (wATTR-CM)** is a condition of older age and is not inherited or passed down through families. In this condition, the normal transthyretin protein slowly becomes unstable as we age. This allows it to form amyloid clumps, which build up mostly in the heart. Because the change happens gradually, symptoms often come on slowly.

What are the symptoms?

People with amyloid cardiomyopathy may notice a range of symptoms, which can develop gradually and get worse over time. Some people may also be asymptomatic, which means they are not experiencing any symptoms of the condition.

Common symptoms include:

- Feeling short of breath (especially while resting or lying down)
- Swelling in the legs, ankles, or belly
- Feeling dizzy or faint when standing up
- Irregular heartbeat or heart flutters (palpitations)
- Feeling unusually tired or having low energy
- Numbness or tingling in the hands and feet
- Losing weight without trying
- Feeling sick in the stomach or losing appetite

Many symptoms of amyloid cardiomyopathy are shared with other heart conditions, but some additional signs may help doctors suspect amyloid cardiomyopathy, especially if they appear together. These can include:

- Carpal tunnel syndrome in both hands *(more common in ATTR amyloidosis)*
- Narrowing in the spine (spinal stenosis)
- Swelling of the tongue or around the eyes *(more typical of AL amyloidosis)*

Not everyone will have these symptoms, and they differ between amyloidosis types. Having these signs does not mean a person definitely has amyloid cardiomyopathy, but they are clues doctors can watch out for.

How is Amyloid Cardiomyopathy diagnosed?

To find out if someone has amyloid cardiomyopathy, doctors use a range of tests. No single test can confirm the condition on its own, but each helps build a clearer picture. Some of the tests used are:

- **Echocardiogram (an ultrasound of the heart):** shows if the heart is an unusual shape or size, particularly if the walls of the heart are thickened, and if it is filling up and emptying blood normally.
- **ECG (electrocardiogram):** measures heart rhythm and can show if the electrical signals controlling the rhythm are normal.
- **Cardiac magnetic resonance imaging (MRI):** provides detailed images of the heart muscle that can show changes in shape and size, as well as how well the heart is pumping blood. Some types of MRI which use contrast agents can also highlight areas where amyloids have accumulated.
- **Nuclear imaging (bone scan):** uses 'tracers' which are molecules that bind to amyloids and make them visible on a scan. It is highly sensitive for diagnosing ATTR-CM, but not AL amyloidosis as the tracers don't bind well to the light chain protein-based amyloids.
- **Blood and urine tests:** can detect the light chain proteins which are overproduced in AL amyloidosis. They can also detect other proteins, such as cardiac troponin, which is released by heart tissue when it is damaged.
- **Genetic testing:** looks for the changes in the TTR gene that can lead to inherited types of amyloid cardiomyopathy.
- **Heart biopsy:** takes a small piece of heart tissue that can be assessed with a microscope to look for amyloids. This is an invasive test, but sometimes needed to confirm the diagnosis if other results are unclear.

Doctors use these tests together to make a diagnosis and to guide treatment.

How is it treated or managed?

Treatment aims to slow or stop further amyloid build-up, protect organ function, and maintain quality of life. It's important that doctors confirm which type of amyloidosis is present before starting therapy, as management depends on the type of amyloid cardiomyopathy a person has. Different approaches can be used to help manage the disease and improve symptoms.

AL amyloidosis:

AL amyloidosis is usually treated with chemotherapy, which helps stop the bone marrow from making the light chain proteins that clump to form amyloids. This chemotherapy often includes a combination of special medicines that can bind to bone marrow cells (daratumumab), suppress the immune system (cyclophosphamide), reduce inflammation (dexamethasone) and kill abnormal cells (bortezomib). Some people may also be suitable for a stem cell transplant, which replaces the abnormal bone marrow cells with normal ones.

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ATTR-CM:

There is no cure yet, but treatments can help control the disease and make life better. Treatments that aim to stop the progression of the condition include:

- **TTR synthesis suppressors:** medicines that reduce the amount of TTR that is made (most commonly patisiran or inotersen).
- **TTR stabilisers:** medicines that help keep TTR stable and prevent more amyloid from forming. In Australia, tafamidis is available through the Pharmaceutical Benefits Scheme, but in New Zealand, these medicines are not generally offered through the public system. Diflunisal is a non-steroidal anti-inflammatory drug (NSAID) that is also used off-label as a TTR stabiliser and is available in Australia through the Special Access Scheme at some major hospitals and amyloidosis centres, but not routinely available in New Zealand. This medicine isn't like over-the-counter NSAIDs so always speak to a doctor or trusted pharmacist before using diflunisal or any NSAID.
- **Fibril disruptors:** medicines that help to break down amyloids.
- **Liver transplant:** is occasionally considered for hereditary ATTR-CM, as most TTR is made in the liver. This approach is less common now because of newer, effective treatments.

Treatments that ease symptoms and conditions caused by cardiac amyloidosis include:

- **Diuretics:** help the body get rid of extra fluid, which can ease swelling and breathlessness. These are widely used and available in both Australia and New Zealand.
- **Beta-blockers and antiarrhythmic drugs:** used to correct abnormal heart rhythms. These are common treatments, but care is needed as some people are sensitive to drops in blood pressure or fluid levels.
- **Anticoagulants:** lower the risk of stroke in people with an irregular heartbeat.
- **Pacemakers or implantable cardioverter-defibrillators (ICDs):** may be recommended for people with significant rhythm problems or a high risk for sudden cardiac arrest. Not everyone is suitable for these devices.

Supportive care:

Support for people with amyloid cardiomyopathy combines many areas, including advice about diet, physical rehabilitation, and mental health care. Working with health professionals with a variety of specialties is the recommended approach. This team usually includes heart specialists (cardiologists), and in the case of AL amyloidosis, blood cancer doctors (haematologists). It is important to have regular check-ups with this specialist team, as they can help manage symptoms and track your progress.

Where to get support

Help is available for people affected by amyloid cardiomyopathy in both Australia and New Zealand. Your GP and cardiologist are the main contacts for discussing symptoms, treatment choices, and ongoing support.

You can also get information and connect with others by visiting **Cardiomyopathy Australia New Zealand**, joining our private Facebook group, and following our social media channels.

Additional supports:

Australia:

- Visit the Australian Amyloidosis Network **www.aan.org.au**
- Visit the Australian Heart Foundation **www.heartfoundation.org.au**

New Zealand:

- Ask your cardiologist about a Heart Failure Nurse Specialist in your area
- Visit the New Zealand Amyloidosis Patient Association **www.amyloidosis.org.nz**
- New Zealand Organisation for Rare Disorders: **www.nzord.org.nz**
- Visit the New Zealand Heart Foundation **www.heartfoundation.org.nz**

Scan here to visit the CMANZ website

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Created in partnership with Heart Foundation

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