My Guide to Cardiac Amyloidosis

Check it out!

This booklet is for those with hTTR TTR and AL amyloidosis. This is a short introduction to amyloidosis. Please ask your heart team the specific type of amyloidosis you have. Ask them for more information, especially if you do not have AL or TTR amyloidosis.

What is amyloidosis?

The answer is complicated. Amyloid is an abnormal protein stored in the body, and the body cannot remove it. Proteins are large molecules that are needed for the body to function correctly. They are responsible for the structure, function, and regulation of body tissue and organs.

Proteins and amyloid proteins

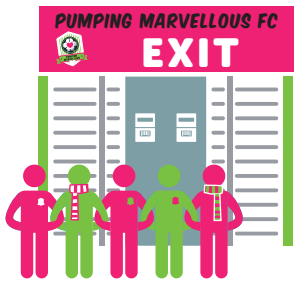
There are many different chemicals that are needed for the body to work normally. One family of these chemicals is called proteins. Proteins are remarkable and essential molecules that allow the body to function properly. Many types of proteins exist, each of which can have one or more functions in the body. For instance, the body contains proteins that build muscle, fight infections, act as regulators that switch another molecule in another part of the body on or off, or even function as transporters to carry other molecules around the body.

When the body has finished using a protein, it can break it down and eliminate it from the body using organs such as the liver, kidneys, and lungs.

For reasons we don't understand, some proteins clump together - 'misfold' - into large unbreakable sheets rather than being broken down in the usual way. Many different types of proteins can undergo this abnormal process, but the sheet they produce remains the same. This sheet is known as the amyloid protein. The various proteins that make up these sheets and the different diseases they are linked to indicate the type of amyloidosis present.

A QUICK ANALOGY...

Imagine football fans (the proteins) trying to leave the ground. By walking through the turnstiles one by one, they make it through; however, if the fans linked arms they would not be able to pass through the turnstiles - just like how the amyloid proteins can't pass through the body.



How does amyloid cause illness?

Amyloid protein can be laid down in any organ as the body cannot break it down.

The impact of amyloidosis depends on the amount, location, and organ in which it is deposited. Even a small amount of amyloid can have a large effect if it is in a critical site.

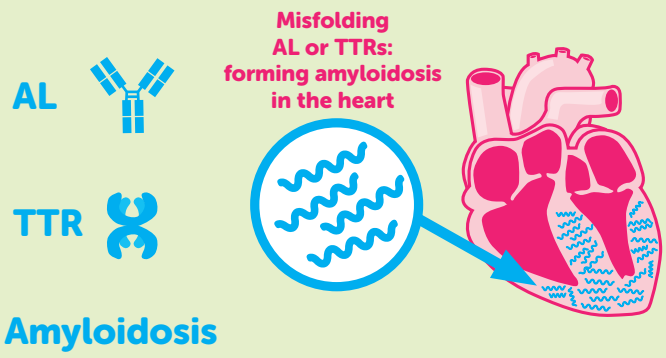
Types of amyloidosis:

There are numerous types of amyloidosis, often named after the protein that misfolds to cause it.

The two most common types of amyloid proteins that affect the heart are **AL** and **TTR amyloidosis**.

We will discuss these types of amyloidosis in greater detail. Please consult your heart team about the specific type of amyloidosis you have and enquire further about your condition, especially if you do not have AL or TTR amyloidosis.

This information guide will help you understand AL amyloidosis and two forms of TTR amyloidosis:
wild-type TTR amyloidosis and **hereditary amyloidosis**.



AL Amyloidosis

This occurs when plasma cells (which help fight infection) in the bone marrow produce abnormal proteins known as light chains – hence the name Amyloid Light or AL Amyloid. This condition shares some features with certain bone marrow plasma cell cancers like myeloma. Your team will tell you if this is relevant to you.

TTR Amyloidosis

This type of amyloid forms due to the abnormal breakdown of a protein that transports vitamin A or thyroid hormone from its production sites to where it is needed. This protein is known as transthyretin retinol-binding protein (TTR). There are two types of TTR amyloid:

- A genetic (or hereditary) abnormality that leads to the formation of amyloid from the TTR protein – hTTR **hereditary (familial) amyloidosis**:
- The second type occurs for unknown reasons when the normal (or wild type) TTR protein transforms into amyloid for presently unknown reasons — **wTTR amyloidosis**. As far as we know, this is not a genetic condition.

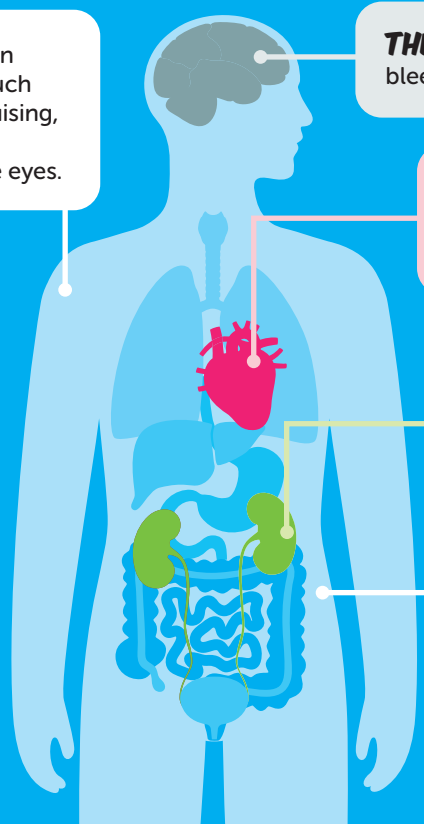
Symptoms:

Diagnosing amyloidosis is like piecing together clues in a detective story. Your doctor assembles clues from the symptoms that prompted your visit and other conditions that have influenced your health. Amyloid can affect any part of the body, including:

SKIN – skin changes such as easy bruising, especially around the eyes.

NERVES - this can involve amyloid being:

- Deposited in nerves, causing reduced nerve function such as feeling – this is called peripheral neuropathy (neuropathy means nerves that do not function well).
- Squashing nerves at crucial points in their journey by being deposited around them, possibly causing conditions in areas such as the hand (carpal tunnel syndrome) or in the back (spinal stenosis).
- Affect groups of nerves that are responsible for complex functions such as bowel, blood pressure, fight or flight responses and sexual function – autonomic nervous system problems.



THE BRAIN – causing strokes, dementia and bleeding in the brain.

HEART – causing Heart Failure, heart rhythm disturbances, heart valve issues, and heart muscle problems.

KIDNEYS – this can lead to abnormal blood and urine test results and may sometimes cause swelling due to fluid retention.

GASTROINTESTINAL (ABDOMINAL) – including causing diarrhoea, constipation, weight loss.

MUSCLES, TENDONS AND JOINTS – causing tendon ruptures and arthritis.

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Diagnosis of AL and TTR amyloidosis:

The diagnosis of amyloid may have been made because of:

- The numerous issues it has caused in various organs or bodily systems (such as the nervous system) over the years.
- A specific problem in one organ alone.
- A result of a biopsy of an affected organ.
- An unexpected finding during a scan.

The steps to diagnosing amyloidosis are:

1 Is there evidence of a body organ affected by amyloidosis?

This may be as a result of tests such as:

CT image scans, ultrasounds, or MRI scans capture pictures of the organ. An ultrasound of the heart is referred to as an echocardiogram.

A SAP (serum amyloid protein) scan can reveal substantial deposits of amyloid in the body (excluding the heart). This test is available only at a few highly specialised centres.

These tests suggest the presence of amyloid in the tissues, but they cannot identify the type of amyloid.

2 Is this AL amyloid or a different type of amyloidosis?

Your team would have determined whether you had AL amyloid or a different type of Amyloid. They did this with tests to confirm or exclude AL amyloid, such as:

Blood tests to see whether there was excess production of the protein causing AL amyloid.

Urine test to see whether there was excess production of the protein causing AL amyloid.

If these tests are positive, a biopsy of an organ affected by amyloidosis is required to confirm the presence of AL amyloid. If a biopsy confirms amyloidosis, the specific type of amyloid can be identified through the biopsy itself. If these tests are negative, a special nuclear medicine scan known as a DPD scan (99mTc-DPD scintigraphy) is performed. The scan is a non-invasive method for imaging cardiac amyloidosis. If it shows amyloid in the heart, the likely diagnosis is TTR amyloidosis. If TTR amyloidosis is confirmed, a genetic test (either from blood or a cheek swab) can determine whether your TTR amyloid has a genetic cause. If the type of amyloid protein remains unclear, similar to diagnosing AL amyloid, a biopsy of the affected organ is necessary. In some cases, a biopsy of organs that are more accessible but may not be causing symptoms (such as the lip, under the skin, or even from the bottom) are considered. The chances of detecting the amyloid protein may be slightly lower from biopsies taken from areas that are not obviously affected by amyloidosis.

Remember: treatments for amyloidosis are improving year on year. You must discuss all your options with your medical teams.

Treatment:

Amyloidosis is a chronic long-term illness.

The symptoms and life expectancy of individuals with amyloidosis are influenced by the organs affected and the degree of damage caused to those organs by the condition.

The principles of treatment for amyloidosis are:

- Specific treatments for AL and TTR amyloid proteins themselves.
- The symptoms associated with amyloidosis.

The specific treatment for amyloidosis depends on the underlying cause:

- AL amyloidosis: Treatment often involves chemotherapy to target the abnormal plasma cells that cause them.
- hTTR and wTTR: Treatments may include medications that reduce the production of the TTR protein, such as Patisiran, Inotersen, or Valtruisan, or stabilise the TTR, such as Tafamidis and Acoramidis.

Living with amyloidosis & symptomatic care

It is crucial to address the symptoms that arise from other parts of your body affected by amyloid. This may range from physiotherapy and basic pain relief to therapies that support failing organs, such as those for heart or kidney failure. In some individuals with amyloidosis, medications previously used for blood pressure or Heart Failure may be prescribed less frequently or may cause additional side effects. Consult your medical team to determine if you still require all the medications you were taking at the time of your amyloidosis diagnosis.

Simple steps taken to look after all of you (such as a healthy diet, lifestyle and exercise) will also help you feel the best you can with amyloidosis.

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Free Cardiac Rehab
pumpingmarvellous.org/heart-failure-guide/heart-failure-rehabilitation/

Educational support
pumpingmarvellous.org/community-hub/support-guides/

Amyloidosis information

Further information about Amyloidosis can be found at:
Royal Free website patient page for the National Amyloidosis Centre (NAC)
www.royalfree.nhs.uk/services/amyloidosis

Amyloid Association
www.amyloidosisuk.org

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