

## WHAT IS AMYLOIDOSIS?

Amyloidosis was first described in the 19th Century, but only in recent years have significant advances been made in understanding the disease.

Amyloidosis is defined as a group of diseases in which one or more organ systems in the body accumulate amyloid proteins.

Amyloidosis is not classified as a cancer, however it is a haematological (blood) malignancy, and may be caused by a cancer.

There are three major types of amyloidosis that are all very different from each other.

## SIGNS AND SYMPTOMS OF AMYLOIDOSIS

-Each patient has a different set of symptoms depending on the type of amyloidosis and the organ systems involved.

-Many of these symptoms can occur in other diseases and it is because of this that diagnosis is frequently difficult.

-The progress of the disease may be slow, over many years, or rapid, with sudden onset of major medical crises.

-General symptoms can include:

Extreme weakness and tiredness;  
Weight loss; Digestion problems;  
Irritable bowel syndrome; Snoring;  
Impotence; Skin problems;  
Chronic pain in the wrist, legs and feet.

## DIAGNOSIS

-Only bone marrow tests or biopsies of tissue, and certain specialist blood and urine tests can positively establish the diagnosis of amyloidosis and classify the type.

-New scans are being developed to show doctors where the amyloid is concentrated, but these are not yet available in Australia.

## TREATMENT OPTIONS

Treatment depends on which type of amyloid protein is present in the body, where it is being deposited and how it is affecting organ(s) and other tissues. It may range from tablets to injected chemotherapy, to Stem Cell transplants, with and without heart, liver and kidney transplants, to palliative care.

Several medical specialists may be involved in decision making, but overall care is usually directed by a Senior Haematologist.

## PRIMARY AMYLOIDOSIS (AL)

-Primary amyloidosis is a plasma cell disorder and occasionally occurs with multiple myeloma.

-The deposits in this type of the disease are made up of immunoglobulin light chain proteins, so it is coded AL for amyloid light chain. These light chain proteins are created in the bone marrow by dysfunctional plasma cells.

-In primary (AL) amyloidosis, the organs most often involved include the heart, kidneys, nervous system, and gastrointestinal tract. Amyloid deposits in these organs can cause shortness of breath, fatigue, oedema (swelling of ankles and legs), dizziness upon standing, a feeling of fullness in the stomach (especially after eating), diarrhoea, weight loss, enlarged tongue, numbness of the legs and arms, and protein in the urine, and kidney failure.

-This is the most common type of amyloidosis and is usually treated with chemotherapy, to slow the production of the amyloid fibres which form when the light chains accumulate.

## SECONDARY AMYLOIDOSIS (AA)

-Caused by a chronic infection or inflammatory disease such as rheumatoid arthritis, and Familial Mediterranean Fever. The deposits in this type of the disease are made up of a protein called the AA protein. Medical or surgical treatment of the underlying chronic infection or inflammatory disease can slow down or stop the progression of this type of amyloid. In secondary (AA) amyloidosis, symptoms caused by the underlying chronic infection or inflammatory disease are frequently joined by the development of amyloid deposits in the kidney. This may cause protein in the urine, oedema, and fatigue.

## **HEREDITARY OR FAMILIAL AMYLOIDOSIS (ATTR)**

- This type of amyloidosis is inherited.
- It is a rare form of the disease and is found in families of nearly every ethnic background.
- The deposits in one type are made up of the transthyretin or TTR protein, giving this type the code ATTR. The protein is manufactured in the liver, and it is a mutation of this protein that causes this hereditary amyloidosis. Liver transplant may extend life.
- Some specific variants of the more than 50 variants are the Irish, Portuguese, Swedish, Japanese and Afro-American types.
- Each family has its own pattern of organ involvement and associated symptoms.
- In ATTR Amyloidosis, the nervous system and gastrointestinal tract are often involved. This can cause numbness and tingling in the arms and legs, dizziness upon standing, and diarrhoea.
- In this type of amyloidosis each child has a 50% chance of inheriting the disease.
- DNA testing may be necessary.
- If a child does not inherit the gene, he/she cannot pass it on to future generations.
- There are other proteins with mutations which can cause familial amyloidosis, but these are very rare.

## **AMYLOIDOSIS AUSTRALIA INC.**

The first organization to be formed in Australia and New Zealand with the sole purpose of providing support for Amyloidosis patients and their carers, families and friends.

Sponsored by generous donations from the Australian Stock Exchange – Reuters Foundation, and the families and friends of Amyloidosis patients.

The support we provide includes information resources for the medical professions involved in research and treatment of Amyloidosis.

Our website has extensive information with links to other websites providing further information:

[www.amyloidosisaustralia.org](http://www.amyloidosisaustralia.org)

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# **AMYLOIDOSIS**

**Amyloidosis  
(pronounced am-ee-loy-dosis)  
results when enough amyloid  
protein fibrils build up in the  
tissues in one or more organs  
causing organ malfunction and  
finally failure.**

**The heart, kidneys, nervous  
system and gastrointestinal tract  
are most often affected.**

**At present there is no known  
permanent cure for Amyloidosis.  
However, treatment which  
targets the cells that make the  
amyloid fibrils may limit further  
amyloid production and produce  
improvement in overall organ  
function, and the quality of life of  
the patient.**