AL amyloidosis - An Introduction

International Myeloma Foundation (UK)

This introduction is written for people who have been diagnosed with AL amyloidosis. It is also for their families and friends. It provides a brief overview of AL amyloidosis and its treatments.

If you would like a more detailed overview, IMF (UK) produce a guide called 'AL amyloidosis - your essential guide'. If you would like to know more about any aspect of AL amyloidosis and its treatment please call the IMF (UK) Helpline on 0800 980 3332. This is a free and confidential service available from 9am to 5pm, Monday to Friday. It is manned by trained nurses and supported by medical and scientific advisors.

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What is amyloidosis?

The term amyloidosis describes conditions in which protein is deposited in an abnormal form, as amyloid fibres (fibrils), in the body's tissues and organs. Amyloid fibrils can gradually build up in tissues and organs because the body can only break them down very slowly. They can therefore disrupt the structure and function of the affected tissues and organs.

What is AL amyloidosis?

AL amyloidosis is one form of amyloidosis arising from abnormal plasma cells. Plasma cells are found in the bone marrow and protect the body against viruses and infections by producing antibodies. In AL amyloidosis a single population (or clone) of plasma cells grows excessively and produces abnormal proteins which are deposited as amyloid fibrils in tissues and organs. The heart, liver, kidneys, spleen, intestine, skin and the nervous system can be affected. AL amyloidosis does not affect the brain.

It is a rare disease, with approximately 600 new cases in the UK each year. It usually affects people between the ages of 50 and 70 years, but can occur in younger adults.

What causes AL amyloidosis?

The causes of AL amyloidosis are not known. It is not contagious or known to be inherited. AL amyloidosis can be related to myeloma, which is a bone marrow cancer.

AL amyloidosis and myeloma

Myeloma is a type of cancer affecting the plasma cells, which are normally found in bone marrow. In myeloma, one plasma cell becomes defective and multiplies rapidly to produce too many plasma cells. The abnormal cells produce an abnormal antibody known as paraprotein, which can be measured in blood and/or urine samples.
About 10 - 15% of patients who have been diagnosed with myeloma have co-existing AL amyloidosis, or will go on to develop it. It is rare for people with AL amyloidosis to go on to develop myeloma.

What are the signs and symptoms of AL amyloidosis?

Common signs and symptoms include:

- Tiredness.
- Weakness.
- Weight loss.
- Loss of appetite.

More specific signs and symptoms, which depend on where the amyloid fibrils have built up, include:

- Swollen ankles.
- Shortness of breath.
- Tingling fingers or numbness in the extremities.
- Diarrhoea.
- Feeling of fullness, even after a small meal.
- Bruising easily.
- A stiff, sore tongue.

How is it diagnosed?

There are a number of tests to confirm the diagnosis and investigate the extent of the disease. These include:

- Organ tissue biopsies (to check for presence of amyloid fibril deposits).
- Bone marrow samples (to check for presence of abnormal plasma cells).
- Blood and urine tests (to check for evidence of abnormal proteins).
- SAP scans (a scan which can detect amyloid fibril deposits in the body).
- Echocardiograms and ECGs (to check function of the heart).

Treatment

Treatment has two objectives:

- To treat the abnormal plasma cells, in order to reduce or eliminate their production of abnormal amyloid protein and amyloid fibril formation. This may allow affected tissues and organs to recover function.
- To alleviate the symptoms caused by the build up of amyloid fibrils in particular tissues and organs.

The aim of treatment is to make the patient feel better, prevent further tissue and organ damage and improve function. The disease may be controlled, slowed down or halted temporarily.

Treating the abnormal plasma cells

- Chemotherapy
  Cytotoxic chemotherapy drugs are given to destroy the amyloid- producing plasma cells. The drugs can be given in cycles by injections or tablets. There are many different combinations and doses of drugs allowing a best course of treatment for each individual patient. Normally, lower doses of chemotherapy will take longer to have an effect on the disease, but side effects will not be as great. Common side effects of chemotherapy include nausea, infections, sore mouth and hair loss. Less common side effects include numbness or tingling in the hands and feet, and sometimes fertility can be affected.
- High dose therapy and stem cell transplantation
  High dose therapy followed by autologous stem cell transplantation is a form of chemotherapy treatment that works well for some patients. It involves giving high doses of chemotherapy to destroy the bone marrow, followed by a transfusion of the patient's own stem cells which have been previously collected and stored. These stem cells repopulate the bone marrow and re-start production of blood cells. This high-dose treatment carries more risks than lower doses of chemotherapy, so its suitability will depend on factors such as the patient's age and extent of their disease. Stem cell transplant using a matched donor (allogeneic transplant) carries even higher risks and its use in the treatment of AL amyloidosis is not yet established.

- **Organ transplantation**
  If an organ is severely compromised, slowing the production of amyloid may not be sufficient in itself. When damage to an organ is considered to be permanent, a transplant may be possible. Kidney and liver transplants have taken place in a number of cases, but further treatment to stop the abnormal plasma cells affecting the new organ has also been required.

Alleviating the symptoms of AL amyloidosis

Supportive treatments may be needed to alleviate specific symptoms and these will vary from patient to patient. They are commonly used alongside, and often after, completion of the treatments to eliminate the underlying abnormal plasma cells. Supportive treatments include:

- Special diets (if kidneys or heart are affected).
- Diuretic tablets (to relieve water retention).
- Drugs to treat low blood pressure.
- Drugs to control diarrhoea.
- Pain management.
- Renal dialysis (if kidney function is very poor).

Specialist Centres

In the UK there is one specialist amyloid centre, the National Centre for Amyloidosis at the Royal Free Hospital in London. Many tests are carried out here that are specific to amyloidosis and may not be available elsewhere. A doctor can arrange for a patient to be referred to this centre where, after discussion with both the patient and his/her own doctor, the most appropriate treatment can be decided upon. The centre can also advise doctors on the management of their patients. Patients can be followed up at regular intervals at this centre, but the patient's own doctor will see the patient more frequently and administer and monitor any treatment locally.

The future

Experimental treatments (such as allogeneic transplants) aim at curing the disease, although none has yet been proven. At present all treatment aims to eliminate the abnormal plasma cell. Early reports show that thalidomide, a drug known to be effective in the treatment of myeloma, may have an effect, although further study is needed. It is hoped that eventually treatments will emerge that are able to promote the breakdown of amyloid protein and fibrils by the body, and much research continues in this area.

IMF (UK) Freephone Helpline

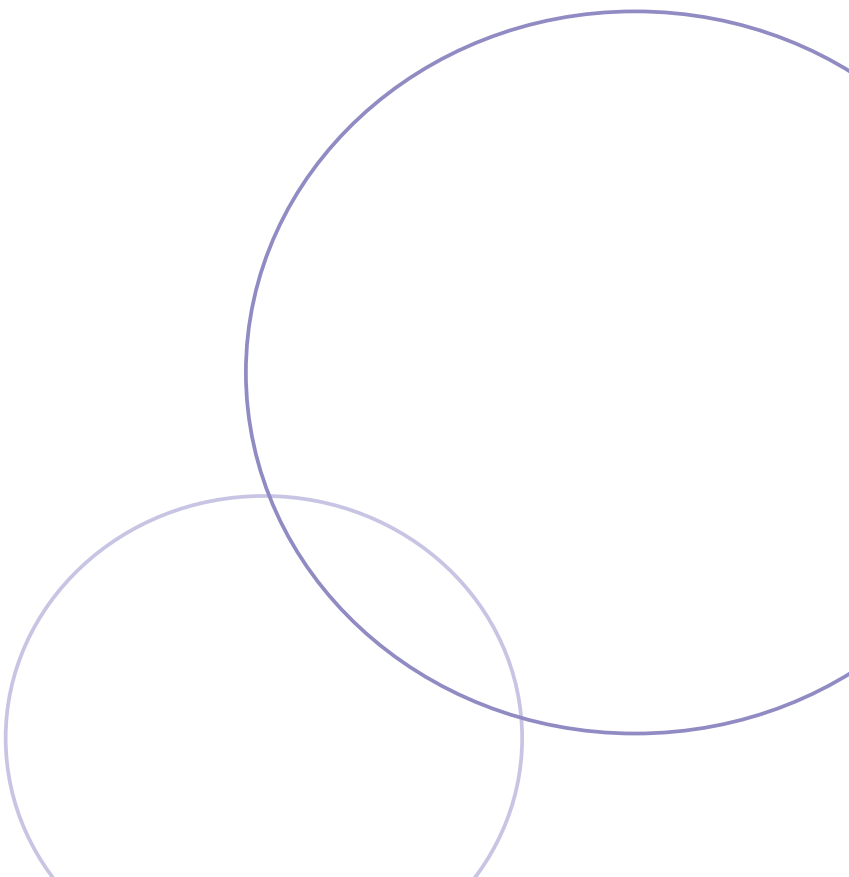
Helpline: 0800 980 3332 (UK only)

Lines are open Monday-Friday, 9am-5pm and an answerphone is in operation at all other times. If you are greeted by the answerphone, please leave your name and telephone number and we will contact you as soon as possible.

Our telephone lines do not use Caller Display Equipment and we use Permanent Number Withhold on all outgoing calls.

This booklet is one of a range of publcations covering many aspects of living with AL amyloidosis. Please contact the IMF (UK) for more details.

"Dedicated to improving the quality of life of myeloma patients while working towards prevention and a cure".



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