Amyloidosis

Summary

- Amyloidosis is an umbrella term that describes diseases caused by abnormal deposits of the protein amyloid.
- The symptoms of amyloidosis vary widely, depending on which tissues and organs are affected.
- There is no cure for amyloidosis.
- The aims of treatment are to prevent further deposits of amyloid proteins and ease the symptoms.

Amyloidosis is an umbrella term that describes diseases caused by abnormal deposits in the body of the protein amyloid. These rare and incurable diseases are more common in people over the age of 40 years, although the reason for this is unknown.

Amyloidosis is now classified chemically according to the type of defective protein fibrils that are formed. This insoluble protein accumulates in tissue cells and causes damage or altered function. These proteins sometimes escape into the blood stream and cause damage elsewhere in the body (systemic).

The cause of amyloid protein formation can be:

- A hereditary form – commonly affecting eyes, heart, kidneys and brain
- amyloid triggered by a chronic disease, infection or cancer – AA amyloidosis
- mutations of antibody producing cells – AL amyloidosis
- rare infectious agents, such as Creutzfeldt-Jakob disease and Kuru.

Systemic amyloidosis can be serious or even life-threatening, depending on which parts of the body are affected. Research has established that there is no link to dietary proteins, but the exact cause of amyloidosis remains unknown.

Symptoms of amyloidosis

The symptoms of amyloidosis vary widely, depending on which tissues and organs are affected. In some cases, amyloidosis has no symptoms until extensive tissue damage has occurred.

Some of the symptoms may include:

- ankle and leg swelling (oedema)
- muscular weakness
- unexplained weight loss
- pins and needles or numbness in the extremities
- memory loss
- diarrhoea
- swollen tongue
- dizziness when standing up suddenly (orthostatic hypotension).

Primary, secondary and familial amyloidosis

The three classifications of amyloidosis include:

- **Primary amyloidosis** is now called light chain fragment amyloidosis (AL) – the disease develops by itself

without apparent cause. Parts of the body commonly affected include the heart, lung, skin, tongue, intestines, liver, kidney and spleen.

- **Secondary amyloidosis** – happens when the disease develops as a complication of another disease, especially multiple myeloma. Others include rheumatoid arthritis, tuberculosis, osteomyelitis or ankylosing spondylitis. Parts of the body commonly affected include the adrenal glands, lymph nodes, liver, kidney and spleen.
- **Familial amyloidosis** – this type is particularly rare. If a person has the flawed gene that causes familial amyloidosis, they have a 50 per cent chance of passing the same condition on to their offspring. Parts of the body commonly affected include the peripheral nerves, the nerves of the wrist and the eyes.

Effects of amyloidosis on heart, kidney or nervous system

The heart is a four-chambered pump that circulates blood around the body. Amyloid deposits within the heart muscle restrict its function, leading to heart failure (cardiomyopathy). Symptoms may include breathlessness and irregular heartbeat.

The kidneys filter wastes from the bloodstream and help to regulate the body's water balance. Amyloidosis damages the small filtering units of the kidney and causes them to leech protein into the urine. The drop in blood protein causes oedema (fluid retention) in the feet and ankles.

Ultimately, the damage to the kidneys impairs their function so that wastes build up in the bloodstream. For reasons unknown, kidneys affected by amyloidosis also tend to trigger higher levels of blood cholesterol.

The nervous system helps all the parts of the body to communicate with each other. It also reacts to changes both outside and inside the body. Around one in four people with amyloidosis experience carpal tunnel syndrome, which is a 'pinching' of the main nerve that services the hand as it runs through the wrist. Other generalised nervous system symptoms include pins and needles and numbness.

More specific symptoms depend on which nerves are affected. For example, if the nerves that service your bowel are affected, you may experience constipation or diarrhoea, or both.

Alzheimer's disease results from a build up of amyloid in the brain and can have a genetic basis.

Diagnosis of amyloidosis

Amyloidosis is diagnosed using a number of tests including:

- **General tests** – include blood and urine tests.
- **Bone marrow biopsy** – a small tag of bone marrow is removed and examined for abnormalities.
- **Tissue biopsy** – a small tag of abdominal fat or rectal tissue is removed with a slender needle and examined in a laboratory.

Treatment for amyloidosis

The aims of treatment are to prevent further deposits of amyloid proteins and ease the symptoms. Treatment options depend on the form of amyloidosis, what organs are affected and the associated symptoms.

Treatment may include:

- adequate rest
- treatment for any underlying disorder such as rheumatoid arthritis
- drugs to slow amyloid activity
- a special diet tailored to your needs – for example, a person with affected kidneys will benefit from a low-salt diet
- dialysis, if the kidneys are affected

- drugs to stabilise the heartbeat, if the heart is affected
- transplants of affected organs may be considered

Current trials include the use of particular chemotherapy medications and bone marrow transplants.

Where to get help

- Your doctor

Things to remember

- Amyloidosis is an umbrella term that describes diseases caused by abnormal deposits of the protein amyloid.
- The symptoms of amyloidosis vary widely, depending on which tissues and organs are affected.
- There is no cure for amyloidosis.
- The aims of treatment are to prevent further deposits of amyloid proteins and ease the symptoms.

This page has been produced in consultation with and approved by:

Better Health Channel - (need new cp)