CONDITION

Primary amyloidosis

Primary amyloidosis is a condition in which abnormal antibody proteins build up and damage organs.

WHAT IT IS
Systemic buildup of abnormal antibody light-chain protein

BODY SYSTEM
Blood, kidneys, heart, nerves, liver, and digestive system

MAIN SYMPTOMS
Fatigue, swelling, breathlessness, numbness

WHAT CAUSES IT
Acquired abnormal bone marrow plasma cells

HOW IT IS DIAGNOSED
Tissue biopsy with amyloid typing

MAIN TREATMENT
Plasma-cell therapy and organ support

CAN IT BE CURED?
No - treatment suppresses amyloid production

IS IT AN EMERGENCY?
Sometimes - chest pain, fainting, or severe breathlessness

LONG-TERM OUTLOOK
Often serious; organ damage and treatment response shape outlook

Overview

Primary amyloidosis is a condition in which abnormal proteins called amyloid build up in tissues and organs. The buildup can make affected organs stiff or prevent them from working normally.

The term usually refers to systemic light-chain amyloidosis, often shortened to AL amyloidosis. "Systemic" means it can affect several parts of the body. Light chains are small parts of antibodies, which normally help the immune system fight infection.

In this condition, abnormal plasma cells in the bone marrow make too many light chains. Plasma cells are white blood cells that produce antibodies. The light chains fold into the wrong shape, join together, and form amyloid deposits.

The kidneys and heart are often affected. Amyloid can also build up in nerves, the liver, the digestive system, skin, soft tissues, and blood vessels. The pattern is different for each person.

Primary amyloidosis is not the same as inherited amyloidosis or AA amyloidosis caused by long-term inflammation. Identifying the exact amyloid protein is essential because these conditions require different treatments.

Symptoms

Symptoms often develop gradually and may seem unrelated. They depend on which organs contain amyloid and how much their function has changed.

Possible symptoms include:

Some people have only mild symptoms at first. Others are diagnosed after tests find kidney protein loss, changes in heart function, or an abnormal antibody protein.

These symptoms have many possible causes. Having one or more does not mean you have amyloidosis.

- Tiredness, weakness, or reduced stamina
- Shortness of breath, especially during activity or when lying flat
- Swelling in the feet, ankles, legs, or abdomen
- Unexplained weight loss or reduced appetite
- Foamy urine, caused by protein leaking through the kidneys
- Numbness, tingling, burning pain, or weakness in the hands or feet
- Dizziness when standing because blood pressure falls
- Diarrhoea, constipation, nausea, or feeling full quickly
- Easy bruising, including bruising around the eyes
- An enlarged tongue, sometimes with tooth marks along its sides
- A fast, irregular, or pounding heartbeat

Causes and risk factors

Primary amyloidosis begins with a small group of abnormal plasma cells in the bone marrow. These cells make unstable antibody light chains that travel in the blood and form deposits in tissues.

The change in plasma cells is acquired during life. It is not usually inherited, and it is not caused by something you ate, an infection, or a particular lifestyle.

The condition is related to other plasma-cell conditions. Some people have monoclonal gammopathy, in which the body makes one abnormal antibody protein. A smaller group also has multiple myeloma, a blood cancer involving plasma cells. Most people with monoclonal gammopathy do not develop amyloidosis.

Doctors often cannot identify why the abnormal plasma-cell group developed in a particular person. Having a known plasma-cell condition increases risk, but it does not make amyloidosis certain.

Diagnosis

Diagnosis usually requires a biopsy. This means removing a small sample of tissue and examining it in a laboratory. Samples may come from abdominal fat, bone marrow, or an affected organ such as the kidney or heart.

A special stain called Congo red helps show amyloid under a microscope. The amyloid must then be typed to identify the protein forming the deposits. Specialised methods, including mass spectrometry, can distinguish AL amyloid from inherited and other forms.

A negative fat or bone marrow biopsy does not always rule out amyloidosis. If suspicion remains high, a biopsy from an affected organ may be considered.

Blood and urine tests look for the abnormal light chains and plasma-cell group. These commonly include serum free light chains and tests called immunofixation. These tests support the diagnosis but do not, by themselves, prove that tissue contains amyloid.

Further tests assess which organs are involved. They may include:

Heart test results are especially important for assessing severity and planning treatment. Genetic testing may be needed if an inherited form remains possible.

- Blood and urine tests for kidney and liver function
- Heart blood markers, an electrocardiogram, and an echocardiogram
- Heart magnetic resonance imaging in selected cases
- Nerve tests when numbness or weakness is present
- A bone marrow biopsy to examine plasma cells

Treatment and management

Treatment has two linked goals: stop abnormal plasma cells from producing light chains and support organs already affected by amyloid.

Drug treatment usually combines medicines that target plasma cells. These may include an antibody treatment, a proteasome inhibitor, chemotherapy, and a corticosteroid like prednisone or hydrocortisone . The exact combination and intensity are chosen according to heart and kidney function, other health conditions, and how well treatment is tolerated.

Blood tests measure how quickly the abnormal light chains fall. A strong reduction is called a haematologic response. Stopping new light-chain production gives the body a chance to gradually clear some deposits and may prevent further organ damage.

Some people who are fit enough may have high-dose chemotherapy followed by an autologous stem cell transplant. This uses the person's own blood-forming stem cells to restore the bone marrow after treatment. It is suitable only for carefully selected people because amyloid-related heart or organ problems can increase treatment risks.

Supportive care is also important. It may include medicines to remove excess fluid, treatment for abnormal heart rhythms, kidney care, pain relief for nerve symptoms, and nutrition support. Dialysis may be needed if the kidneys can no longer filter the blood adequately.

A heart or kidney transplant may be considered for a small number of people with severe organ failure whose plasma-cell condition can also be controlled.

Some medicines commonly used for heart or blood pressure conditions may be poorly tolerated when amyloid affects blood pressure or heart function. Medication changes should be made with the clinical team rather than stopped without advice.

Complications

Amyloid deposits can cause lasting organ damage. Possible complications include:

Heart involvement is a major factor in long-term outlook. Early control of light-chain production may limit further damage, but recovery of organ function often takes longer than the blood response.

- Heart failure or abnormal heart rhythms
- Very low blood pressure, especially when standing
- Heavy protein loss in urine and kidney failure
- Nerve damage affecting feeling, movement, digestion, or blood pressure

- Liver enlargement and reduced liver function
- Poor nutrition from appetite or digestive problems
- Bleeding or bruising caused by blood-vessel or clotting problems

Prevention

Primary amyloidosis cannot currently be prevented. It results from an acquired abnormal plasma-cell group, and there is no proven lifestyle change, food, or supplement that prevents it.

If you already have a plasma-cell condition, attending the recommended monitoring appointments may help detect new organ changes. Monitoring does not guarantee prevention, but it can support earlier assessment.

Living with it and what to expect

Primary amyloidosis is usually a long-term condition requiring specialist follow-up. Treatment may control the abnormal plasma cells, but existing amyloid deposits do not disappear immediately.

Light-chain levels can improve within weeks or months when treatment works. Changes in kidney, heart, or nerve function usually take longer. Improvement may continue over many months, while some damage may remain permanent.

Follow-up commonly includes blood and urine tests, heart markers, and checks of symptoms, blood pressure, weight, and organ function. Treatment may be changed if the light chains do not fall enough, side effects are difficult, or the condition returns after a period of control.

Relapse is possible because small numbers of abnormal plasma cells may remain. Some people have prolonged remission, meaning there is no active progression for an extended period. Others need further courses of treatment.

Daily life is shaped mainly by the organs involved. Fluid retention, low blood pressure, tiredness, or nerve symptoms may affect work, exercise, travel, and household tasks. Activity can often continue within personal limits, with adjustments during treatment or periods of organ symptoms.

There is no single outlook that applies to everyone. People with limited organ damage and a deep treatment response generally do better than those with advanced heart involvement. Newer plasma-cell treatments have improved control of the condition, but primary amyloidosis

can still be serious.

When to get medical help

Call emergency services if you develop:

These symptoms can have causes other than amyloidosis, but they need immediate assessment.

Contact your treatment team promptly if you notice rapidly increasing swelling, a sudden weight increase from fluid, much less urine than usual, worsening dizziness, new palpitations, or increasing breathlessness.

If you are receiving treatment that affects the immune system, follow the instructions you were given about fever or signs of infection. Some infections need same-day assessment.

Mention gradual changes-such as increasing fatigue, numbness, bruising, digestive symptoms, or reduced activity tolerance-at routine follow-up. Contact the team sooner if they are progressing or interfering with eating, walking, sleep, or daily activities.

- Severe or rapidly worsening shortness of breath
- Chest pain, fainting, or loss of consciousness
- New weakness on one side, facial drooping, or difficulty speaking
- A very fast or irregular heartbeat with dizziness or chest discomfort
- Coughing or vomiting blood, or heavy uncontrolled bleeding

Common questions

Is primary amyloidosis a type of cancer?

It is caused by abnormal plasma cells and is treated with many of the same medicines used for plasma-cell cancers. However, the main harm comes from the light-chain deposits rather than from a large tumour. It can occur with multiple myeloma, but the two conditions are not the same.

Is primary amyloidosis inherited?

Usually not. AL amyloidosis is an acquired plasma-cell condition. Other forms of amyloidosis can be inherited, which is why accurate amyloid typing-and sometimes genetic testing-is important.

Can amyloid deposits go away?

Some deposits may gradually decrease after the abnormal light-chain production is controlled. This process can take months or years. Organ function may improve, remain stable, or stay impaired, depending on the amount and location of damage.

How quickly does primary amyloidosis progress?

There is no fixed rate. Progression is influenced by how much abnormal protein is being made and which organs are affected. Heart involvement can cause important changes sooner than deposits in some other tissues.

Is there a special diet for primary amyloidosis?

No diet removes amyloid deposits. Dietary advice is based on the organs involved. For example, some people with fluid retention or kidney problems may be advised to adjust salt, fluid, protein, or potassium. These changes should be personalised so that nutrition is not unnecessarily restricted.

How long can someone live with primary amyloidosis?

A single life-expectancy estimate is not reliable. Outlook is most strongly influenced by heart involvement, other organ damage, general health, and the response to plasma-cell treatment. Some people achieve long remissions, while advanced organ involvement can shorten life.

Sources

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