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Amyloidosis

Amyloidosis is a rare and serious illness. It occurs when normal proteins in the body become misshapen and clump together. These misshapen proteins are called amyloid deposits or fibrils. Over time, the amyloid deposits build up in organs and tissues causing symptoms or even organ failure.

Amyloid protein deposits can be found in specific organs, such as the heart, lung, skin and bladder, or they can be systemic found throughout the body. The latter one is the most common type of Amyloidosis.

Types of Amyloidosis

There are different types of amyloidosis, depending on which protein is abnormal. Among these can be found:

AL amyloidosis: This is the most common type in the United States. AL amyloidosis involves a type of protein called "light chains." Normal light chains are used to make antibodies, which help the body fight infections. In AL amyloidosis, cells in the bone marrow (the tissue inside of your bones that makes blood cells) make abnormal light chains. This disease can be associated with Multiple myeloma, which is a cancer of a type of white blood cell called "plasma cells".

AA amyloidosis: The most common form in resource-limited countries. In this condition, the amyloid protein that builds up in the tissues is called serum amyloid A. This is an acute-phase protein, meaning its levels in the blood increase in response to inflammation. AA amyloidosis is associated with chronic diseases, such as diabetes, tuberculosis, rheumatoid arthritis, and inflammatory bowel disease.

Hereditary or familial amyloidosis: Hereditary amyloidosis is rare. But some mutations in the transthyretin (TTR) gene can be passed down through generations and cause amyloid protein deposits to build up in peripheral nerves (Familial Amyloid Polyneuropathy) or the heart (Familial Amyloid Cardiomyopathy).

Other types: Amyloidosis can also occur due to sporadic mutations in different genes or due to prolonged periods of dialysis in patients with end-stage kidney disease. In these cases, the disease would not be hereditary.

Common Symptoms and Signs

Amyloidosis clinical manifestations are usually determined by the organ affected and the amount of amyloid protein buildup. It can cause renal disease, cardiomyopathy, pulmonary disease, GI disease, musculoskeletal disease, neurological and hematological abnormalities, or skin manifestations. Some signs and symptoms that you should look out for include:

Arrhythmias and feeling dizzy when changing positions.	Changes in urine and/or stools.
Unintentional weight loss and loss of appetite.	Joint pain
Severe fatigue due to anemia.	Skin changes, such as waxy or thickened skin, unexplained rashes and easy bruising.
Weak hand grip, which may arise from carpal tunnel syndrome.	An increase size of liver and/or spleen.

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Risk Factors

Most people who develop amyloidosis have no known risk factors. In most cases, the cause is unknown. But the following factors may raise a person's risk of developing the disease:

Age. The risk of amyloidosis increases as a person gets older. For AL amyloidosis, most people diagnosed are **older than 40.**

Gender. Amyloidosis is more common **in men than in women.**

Other diseases. As explained in the Introduction, amyloidosis is sometimes linked with another disease. For instance, research shows that **12% to 15% of people with multiple myeloma** also develop AL amyloidosis.

Family history. Hereditary amyloidosis can run in families. This may be due to a genetic change, called a mutation, **passed down from generation to generation.**

Diagnosis

The definitive method for diagnosis of amyloidosis is tissue biopsy, although the presence of amyloidosis may be suggested by the history and clinical manifestations. Some of the tests used in the process of diagnosis include:

- Laboratory tests
- Heart evaluation
- Radiologic imaging such as ultrasound
- Bone marrow aspiration
- Molecular testing
- Tissue biopsy

How Common is Amyloidosis?

Because amyloidosis is rare, the diagnosis is often delayed, or missed. Therefore, it is difficult to know exactly how many people are affected by this disease.

It is estimated that about **4,000 people** in the **United States** develop amyloid and light chain (AL) amyloidosis **each year.**

The disease is typically diagnosed between the ages of **50 and 65.** However, people **as young as 20** have also been diagnosed with AL amyloidosis.

Hereditary and autoimmune **amyloidosis (AA) are less common than AL amyloidosis.** Some research notes that the number of cases of AA amyloidosis are declining due to better treatments of the underlying inflammatory conditions.

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