What is Amyloidosis?

Amyloidosis is a disease that results from a buildup of abnormal protein, called amyloid.

- The body is unable to remove this abnormal protein, which builds up in different body organs, and in turn interferes with their normal function.
- Abnormalities in organ function can be varied and may consist of hand, nerve, stomach and heart problems, among others.
- Symptoms of amyloidosis depend on the type of protein, its location and the amount of amyloid buildup in the body organs.

What are the common symptoms of Amyloidosis?

- Amyloidosis may affect several organ systems of the body.
- While the heart is commonly involved, several other features may occur several years before heart involvement (heart failure, slow heart rate, other) such as:
  - carpal tunnel syndrome in both hands
  - back pain from pinching of the spinal cord (spinal stenosis)
  - numbness in fingers and toes

These signs and symptoms are often referred to as “Red Flags”. Symptoms of amyloidosis are dependent on its type.
There are Two Common Types of Amyloidosis

1. **Light-chain or AL amyloidosis:**
   - Light chains are pieces of antibodies normally made in the bone marrow.
   - Excessive amounts of light chain proteins are produced due to a type of bone marrow cancer.
   - Excessive proteins clump together, called amyloid protein that cannot be broken down by the body. These get deposited in various organs, especially the heart and kidney, causing poor function.
   - If left untreated, light chain amyloidosis can result in heart failure, with a life expectancy of about 6 months.
   - Chemotherapy, provided by a blood or cancer specialist, can slow the disease process and may extend life by many years.

2. **Transthyretin or ATTR amyloidosis:**
   - Transthyretin protein is produced by the liver to support certain body functions.
   - In ATTR amyloidosis, this protein forms clumps of abnormal protein (amyloid protein) that cannot be broken down by the body.
   - These clumps get deposited in various body organs. The heart is most commonly involved in ATTR amyloidosis. It frequently co-exists with other organ function abnormalities.

Scan this QR code to learn more about amyloidosis treatments and approved therapies.
There are two types of ATTR Amyloidosis

1) Hereditary ATTR amyloidosis:
   • Results from abnormalities in the body’s DNA, known as mutations.
   • Hereditary ATTR amyloidosis can be transferred from generation to generation. See family tree below.
   • The most common mutation in the United States (called the V142I mutation) is present in 3.5-4% of black populations – African American and Afro-Caribbean people.
   • Hereditary ATTR amyloidosis effects both men and women almost equally and usually results in symptoms between 55-75 years of age.

2) Wild-type ATTR amyloidosis:
   • Results from changes in the body’s DNA with aging.
   • It is NOT transferred from generation to generation and typically shows up later in life, 65-95 years of age.
   • Wild-type ATTR amyloidosis is more common than the hereditary form.
   • Seen most commonly among the elderly and among men of all ages.
Call your doctor if you experience one or more of these symptoms.
When suspected, several tests can be performed that can help confirm the diagnosis of amyloidosis, and specifically amyloidosis of the heart. These may include:

✓ Blood and urine tests
✓ ECG and ultrasound (echocardiogram) of the heart
✓ MRI and nuclear scan (PYP scan) of the heart
✓ Nerve and muscle tests
✓ Testing of DNA abnormalities (mutations) for patient and family members
✓ In rare instances, a biopsy may be performed

How is Amyloidosis Diagnosed?

How is Amyloidosis Treated?

Management of amyloidosis may require input from a cardiologist, neurologist, orthopedic surgeon and other specialists.

Treatment is based on the type of amyloidosis and aims to reduce symptoms and delay disease progression.

Treatment of AL amyloidosis: Chemotherapy is generally required and prescribed by blood or cancer specialists.

Treatment of ATTR amyloidosis: Therapy is focused on controlling symptoms from the heart, and other organ systems. Several drugs have been approved by the FDA for management of ATTR amyloidosis. Choice of the drug depends upon whether ATTR amyloidosis is a hereditary or wild-type. These therapies work in the following fashion:

• **Silencers**: Reduce production of the abnormal protein by acting on the liver.
• **Stabilizers**: Limit the ability of the protein to cause further harm by preventing it from depositing in the body organs causing more damage.
• **Disrupters**: Remove amyloid protein by helping to break-up the abnormal protein already deposited in the body.

These ATTR amyloidosis therapies are administered under the supervision of an amyloidosis specialist.

To learn more about amyloidosis treatments and approved therapies, visit ASNC.org/CardiacAmyloidosis.
Early diagnosis is important. Talk with your doctor if you may be at risk or have symptoms of amyloidosis, so you may be referred directly to a specialist.

REFER DIRECTLY

This Think Amyloid resource is part of the Refer Directly program.

Visit ASNC for additional resources at ASNC.org/CardiacAmyloidosis

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