Patient Education

UNDERSTANDING LIGHT CHAIN AMYLOIDOSIS

Amyloidosis is a rare disease that occurs due to a buildup of protein in different vital organs throughout the body. The disease is often misdiagnosed and not well understood, so there is a growing need for education. Light chain amyloidosis is the most common type of amyloidosis. Read below for more information about the diagnosis and treatment of this disease.

This "Patient Education" tear sheet was produced in collaboration with the Amyloidosis Foundation (*amyloidosis.org*).

What is Amyloidosis?

Amyloidosis refers to several different types of diseases where abnormal proteins are produced. These protein fibers can attach and deposit into organs, tissues, nerves, and other places in the body, which affects normal function of the area. As the amyloid protein increases, health problems and organ damage may occur. When amyloid clusters together, it can occur in several places throughout the body (*systemic*) or gather in one specific area (*localized*).

The various types of amyloidosis have different treatment courses; therefore, the correct diagnosis is extremely important.

Understanding Light Chain Amyloidosis

In the United States, light chain, or AL, amyloidosis is the most common type, with approximately 4,500 new cases diagnosed each year. It usually affects individuals between 50 and 80 years old.

AL amyloidosis is caused by a bone marrow disorder. The marrow in the center of bones produces cells in the blood system, including plasma cells. In AL amyloidosis, these plasma cells produce an abnormal antibody, immunoglobulin protein.

Immunoglobulin proteins are composed of four protein chains: two light chains (either kappa or lambda light chains) and two heavy chains. In AL amyloidosis, the light chains become misfolded, and these misfolded amyloid proteins are deposited in and around tissues, nerves, and organs. As the amyloid builds up, it gradually causes damage and affects the function of the area where it accumulates. AL often affects more than one organ, though it does not affect the brain.

How Is AL Amyloidosis Diagnosed?

Diagnostic testing for AL amyloidosis involves blood tests, urine tests, and biopsies. Blood and/or urine tests can indicate signs of the amyloid protein and organ damage, but only bone marrow tests or other small biopsy samples of tissue or organs can confirm the diagnosis of amyloidosis.

Symptoms of AL Amyloidosis

Symptoms of AL amyloidosis vary by patient, though initially they can be minor. Fatigue, weight loss, and swelling are the most common symptoms. Less commonly, people may have bleeding or clotting problems.

For each patient, symptoms will depend on which organs are affected by the amyloid deposits and the degree to which that organ function is impaired. The most common organs affected include:

- **Kidneys:** Chronic kidney disease may occur, as amyloid deposits in the kidneys can affect how they filter proteins in the blood and dispose of toxins, and may result in a condition called nephrotic syndrome, in which there is excess protein in the urine (*proteinuria*) and the lower legs can become swollen (*edema*).
- **Heart:** Amyloid proteins make the heart unable to function efficiently, and deposits in the heart can cause it to become unusually thickened and stiff, which can result in shortness of breath and can affect the heartbeat (*arrhythmia*).
- **Digestive system:** Amyloid deposits in the gastrointestinal tract can cause nausea, diarrhea, or constipation; weight loss; loss of appetite; or a feeling of fullness in the stomach after eating small amounts of food.
- **Nervous system:** Amyloid deposits can affect the nerves of the hands, feet, and lower legs and may cause pain, numbness, and tingling. Nerves that control blood pressure, heart rate, bowel motility, erectile function, and other body functions can also be affected.

How Is AL Amyloidosis Treated?

Treatment for AL amyloidosis should be tailored to each patient. The type of treatment should be based on disease progression and seriousness of the patient's organ, tissue, or nerve involvement. Treatment plans are two-fold. **Supportive treatment** includes treating your symptoms and organ damage, while **source treatment** includes slowing down or stopping the overproduction of amyloid at the disease source.

Reversing any damage to the organs and other parts of the body is difficult to achieve, though if treatment begins earlier, the overall success rate is higher.

In the United States, a stem cell transplant is often the preferred therapy, as it can provide long-term control of the underlying disease. However, only a minority of AL patients are eligible to receive a transplant due to contraindications and risks of transplant-related morbidity and mortality. Other chemotherapy-based treatments are considered for the majority of AL amyloidosis patients. Patients with AL amyloidosis are benefiting from the recent development of new drugs for multiple myeloma – including proteasome inhibitors like bortezomib and carfilzomib and immunomodulators like thalidomide and lenalidomide – many of which work effectively on the plasma cells that cause AL amyloidosis.

Patient Resources from the Amyloidosis Foundation

The Amyloidosis Foundation was founded in 2003 to support medical and scientific research for amyloidosis and to raise awareness in hopes of leading to earlier diagnoses, to encourage research through grants, and to empower patients through a range of services, including up-to-date information on the disease.

For more information on Patient Toolkits, educational webinars, treatment centers, and patient support portals, visit *amyloidosis.org/resources*.

Cut out and give to a patient