Diagnosis of Light Chain (AL) Amyloidosis: What You Should Know

What it covers

- Diagnosing light chain amyloidosis in individuals with suspected AL

Why it matters

- Amyloidosis is a rare group of disorders caused by protein misfolding. In North America, one of the common forms of amyloidosis is light chain amyloidosis (AL).
- AL can be associated with significant morbidity and mortality, especially with late diagnoses. Delays in diagnosis, often spanning months to greater than 2 years, are common in AL amyloidosis.
- Diagnostic delays may be related to wide-ranging symptomatology, overlapping with many other conditions—as a result, AL is frequently not even considered as a potential diagnosis.
- There is an urgent need to increase awareness of symptoms associated with light chain amyloidosis and for clear diagnostic pathways for this disease.

Who it affects

- Hematologists, cardiologists, hematopathologists, nephrologists, neurologists, internists, GI specialists, and additional specialties which may be involved in AL diagnostic process.

What are the highlights

- Awareness of red flag signs and symptoms of AL is crucial to avoiding diagnostic delays.
- The use of serum immunofixation, urine immunofixation and serum free light chains with ratio enhances the clinical suspicion of AL.
- The diagnosis of renal, nerve and multiorgan AL can be made effectively through surrogate biopsies, which requires a combination of bone marrow biopsy and fat pad sampling. Target organ biopsies maybe favoured in certain clinical situations.
- For individuals with suspected cardiac AL, the diagnosis of cardiac AL can be made effectively either through a combination of fat pad and bone marrow biopsy, or through endomyocardial biopsy. Proceeding with one approach versus the other relies on several factors including access, available expertise, clinical presentation, and cost.
- Existing gaps in evidence highlight the need for additional research into strategies and predictive models for earlier detection of AL and organ involvement, minimally invasive surrogate biopsies, and optimizing diagnostic pathways.

Total number of panel recommendations: 12

REFERENCE

Kukreti, V., Seftel M., Aguirre, M.A., Azzam, M., Boedicker, D., Bumma, N., Carroll, A., Comenzo, R., Cook, J., Dasgupta, N., De La Torre, A., Dispenzieri, A., Jamal, F., Kawtharany, H., Khouri, J., Leung, N., Nazzal, J., Picken, M., Raza, S., Sanchorawala, V., Sarswat, N., Shaikh, H., Singh, D., Mustafa, R. American Society of Hematology 2025 Guidelines on Diagnosis of Light Chain Amyloidosis. *Blood Advances*

For more information on the ASH Clinical Practice Guidelines on Diagnosis of Amyloidosis, **visit http://www.hematology.org/amyloidosis-guidelines**