American Heart Association.

AL Amyloidosis
Quick Reference Guide

Disease Overview

AL Amyloidosis (Light Chain) is a rare but life-threatening disease caused by abnormal plasma cells producing misfolded light chain proteins. These proteins form amyloid fibrils that deposit in organs - most commonly affecting: Heart, Kidneys, Liver, Gastrointestinal system, Nervous system. **This leads to progressive and often irreversible organ dysfunction.**

Early suspicion and testing are critical. AL Amyloidosis often presents with vague symptoms, and delays in diagnosis can result in severe organ failure.

Diagnosis should be treated as a medical emergency as rapid intervention is essential to prevent further damage. Treatment focuses on halting light chain production through: Chemotherapy, Monoclonal antibodies, Stem cell transplantation.

A high index of suspicion and prompt action can significantly improve patient outcomes.

Key Diagnostic Testing

Initial Screening Tests

Serum free light chains (kappa & lambda)
Serum electrophoresis with immunofixation
Urine electrophoresis with immunofixation
Troponin T & NT-proBNP
Electrocardiogram
Echo (with strain imaging)/Cardiac MRI (with and without contrast)

Further Diagnostic Testing

Abdominal fat pad biopsy
Bone marrow biopsy

Diagnosis confirmation requires a tissue biopsy with Congo red staining to detect amyloid deposits:

- Abdominal fat pad biopsy
- Bone marrow biopsy

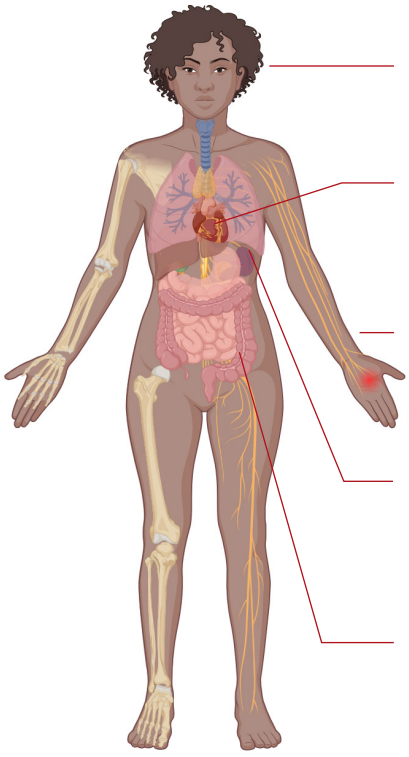
Note: If both biopsies are negative but clinical suspicion for AL Amyloidosis remains high, consider biopsy of the affected organ.

Different Diagnosis

Abnormal Monoclonal Testing Diagnosis Considerations

Monoclonal Gammopathy of undetermined significance
Multiple Myeloma, Smoldering Multiple Myeloma, or Light Chain Smoldering Multiple Myeloma
Waldenström Macroglobulinemia
POEMS Syndrome

Clinical Clues



General:
Weakness, unexplained fatigue, macroglossia

Hematologic:
Easy bruising, periorbital purpura (raccoon eyes)

Cardiac:
Dyspnea, hypotension, edema, arrhythmias, increased for L-Ventricularwall thickness

Neurologic:
Peripheral neuropathy, carpal tunnel syndrome, autonomic dysfunction

Renal:
Proteinuria, nephrotic syndrome, kidney dysfunction

Gastrointestinal:
Significant unintentional weight loss, diarrhea/ constipation, malabsorption, unexplained GI bleeding, hepatomegaly

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Diagnosing Amyloidosis

Disease Suspicion
See Clinical Clues

Initial Screening Tests
- Serum free light chains (kappa & lambda)
- Serum electrophoresis with immunofixation
- Urine electrophoresis with immunofixation

Abnormal Monoclonal Protein

Yes
AL Pathway

Urgent Referral to Hematology
Additional Testing:
- Abdominal fat pad biopsy
- Bone marrow biopsy
- Tissue biopsy confirmation requires detection of amyloid deposits using Congo red staining, with amyloid typing determined by mass spectrometry

Pathway Confirmation of AL Amyloidosis:
- Tissue biopsy (e.g., abdominal fat pad) Congo red-positive
- Amyloid typing confirms AL (via mass spectrometry)
- Clonal plasma cell disorder identified (bone marrow)

Initiate Hematology-directed Therapy Immediately

No
ATTR Pathway

Cardiac Scintigraphy Uptake- Grade 2/3

No
ATTR Cardiac Amyloidosis Unlikely

Yes
Genetic Mutation

No
ATTRwt
Wild Type

Yes
ATTRv
Variant

Referral Guidance

- Refer suspected AL Amyloidosis patients to an amyloidosis center when possible.
- If an amyloidosis center is unavailable, prompt consultation with hematology and cardiology is essential.
- A diagnosis of AL Amyloidosis is a medical emergency that demands timely evaluation and multidisciplinary collaboration to ensure the best possible outcomes.

Patient Advocacy and Support Resources

MACKENZIE'S MISSION
mm713.org

Amyloidosis Research Consortium
arci.org

AMYLOIDOSIS SUPPORT GROUPS
You Are Not Alone
www.amyloidosissupport.org
amyloidosissupport.org

ALEXION The AL Amyloidosis Toolkit is proudly supported by Alexion, AstraZeneca Rare Disease.