

Amyloidosis: The Underdiagnosed Rare Disease

Amyloidosis, a group of rare diseases with different subtypes, is estimated to affect about 150,000 Americans.

Yet fewer than half of patients are diagnosed in time to prevent permanent organ damage. Delayed diagnosis remains one of the greatest challenges facing patients and clinicians alike.



What is amyloidosis?

Amyloidosis occurs when abnormal proteins accumulate in organs such as the heart, kidneys, liver and nerves. People with amyloidosis often experience fatigue, swelling and unintentional weight loss. Over time, amyloid deposits can interfere with normal organ function and lead to serious complications. Early diagnosis is critical, as treatment is most effective before significant damage occurs.



Why is amyloidosis often missed?

Amyloidosis is often difficult to recognize because its symptoms closely resemble those of other common conditions. As a result, patients are frequently misdiagnosed and may spend years seeking answers before reaching the appropriate specialist. On average, it takes one to three years for patients to receive an amyloidosis diagnosis. The condition also remains underrecognized among many clinicians, who may be unfamiliar with the disease, its warning signs or the specialists best equipped to manage it.



How do disparities impact diagnosis?

Disparities in diagnosis and care continue to affect patient outcomes. Women are less likely to be diagnosed compared to their male counterparts, as some testing tools do not adequately account for biological differences like thinner heart muscle walls. Black patients are more likely to present with advanced cardiac complications, such as heart failure, but are less likely to be referred for specialized symptom management. Rural patients also face a higher risk of misdiagnosis or delayed diagnosis, often because of limited access to specialists and advanced testing. These barriers can delay treatment and worsen outcomes.



How can policymakers help close gaps?

Policymakers can help improve diagnosis by expanding access to genetic testing and ensuring coverage for diagnostic tools such as tissue biopsies and blood tests. Supporting clinician education initiatives can improve awareness of amyloidosis and promote earlier recognition. Policymakers can also help ensure patients have timely access to emerging and existing treatments by reducing unnecessary coverage barriers.



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