

Amyloidosis

Amyloidosis is a serious and often underdiagnosed rare disease that can cause permanent damage to the heart, nerves, kidneys and other organs. Each year, approximately 1,275 to 3,200 new cases are identified. However, this estimate may be low because of [misdiagnosis](#) and underreporting.

Delayed diagnosis, along with high treatment costs, can create significant challenges for patients and their families. The disease is attributed to proteins known as amyloids that build up in the heart, nervous system, digestive tract, kidneys and eyes. The condition can be passed down genetically or occur during aging. Symptoms vary based on where amyloid deposits are present in the body but may include numbness, weakness, swelling, glaucoma, kidney failure and diarrhea.

Three types of cardiac amyloidosis make up 95% of all cases:



AL (Light Chain) Amyloidosis:

Caused by a bone marrow disorder and may affect the heart, kidneys, liver, nerves or intestines.



Hereditary (ATTR)

Amyloidosis (hATTR): An inherited form caused by a genetic mutation, often affecting the nervous system and heart.



Wild-type ATTR Amyloidosis

(wtATTR): A non-hereditary form seen in older adults, mostly affecting the heart.

Diagnosing whether or not someone has amyloidosis, and what type they have, includes reviewing their signs, symptoms and family history and testing: echocardiogram, cardiac MRI, PET nuclear heart scan and a PYP scan. A genetic test using a blood or saliva sample may also help confirm the diagnosis.

Although amyloidosis can occur in anyone, it does not affect all groups equally. In the United States, the most common form of hATTR amyloidosis is more prevalent in African Americans and men. It typically affects middle-aged and older adults, but it can also develop in people in their 20s and 30s.

CHALLENGES FACING PATIENTS



Amyloidosis symptoms often resemble those of other conditions, which may lead to delayed or misdiagnoses and postpone access to treatment.



The cost of amyloidosis treatment can be substantial, particularly for patients managing other medical conditions at the same time. Common medications for amyloidosis can cost over \$225,000 per year without insurance.



In one study, 43% of patients reported using savings, borrowing money or making difficult financial trade-offs to afford treatment.

Further, some providers might have limited knowledge about amyloidosis, which might affect patients' quality of life and even survival rates.

Both patients and providers need the necessary information to treat amyloidosis quickly.

Policymakers can improve outcomes for people with amyloidosis by promoting disease awareness, supporting earlier diagnosis and expanding access to appropriate testing and treatment. Addressing these barriers can help patients receive care before irreversible organ damage occurs.