APOLIPOPROTEIN AMYLOIDOSIS

What You Need to Know

Amyloidosis Research Consortium

WHAT IS IT?

Apolipoprotein amyloidosis is a type of hereditary amyloidosis belonging to a family of proteins called apolipoproteins. It is caused by the misfolding of a specific apolipoprotein subtype. Apolipoproteins play important roles in the transport and metabolism of cholesterol and fat in the body. There are currently five types of apolipoprotein amyloidosis known to form, and researchers believe additional rarer types may exist. Each type is named after the specific protein that causes it, such as AApoA-I, AApoA-II, etc.

WHO DOES IT AFFECT?

Apolipoprotein amyloidosis can affect both men and women. Most types are hereditary, passed down in families. If a parent carries the genetic mutation, each child has a 50% chance of inheriting it. Not everyone who inherits the mutation develops symptoms, and symptoms may not appear until adulthood or later.

WHAT CAUSES APOLIPOPROTEIN AMYLOIDOSIS?

Apolipoprotein amyloidosis is caused by a change or mutation in one of the known apolipoprotein genes. Most types are autosomal dominant, meaning just one copy of the changed gene is enough to cause disease.

SIGNS AND SYMPTOMS

Symptoms vary depending on which type of apolipoprotein amyloidosis a person has and which organs are affected. The disease generally moves slowly, and it may be many years before problems are noticed.

Type	Organs affected	Common symptoms
AApoA-I (most common)	Kidneys, liver, heart, voice box (larynx), nerves	Protein in urine, high blood pressure, swelling, enlarged liver, shortness of breath, hoarse voice, numbness or tingling in hands or feet
AApoA-II	Kidneys	Protein in urine, worsening kidney function
AApoA-IV	Kidneys, heart	Worsening kidney function, shortness of breath, fatigue
AApoC-II	Kidneys	Protein in urine, declining kidney function
AApoC-III	Kidneys	Protein in urine, declining kidney function

Have Questions?
(617) 467-5170 or support@arci.org

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DIAGNOSIS

It is incredibly important to ensure an accurate diagnosis, as each type of amyloidosis is treated differently and a delay in treatment can lead to worsening symptoms. Organ biopsy with Congo Red staining and laser dissection mass spectrometry are necessary to identify the specific apolipoprotein causing the disease. Since apolipoprotein amyloidosis is a genetic condition, it is important to obtain genetic testing to look for a mutation in the apolipoprotein genes. Blood tests can that show low HDL ("good") cholesterol and low apolipoprotein levels can sometimes provide a clue that one of these conditions may be present.

PREVENTION AND TREATMENT

Apolipoprotein amyloidosis tends to progress slowly. There is currently no specific treatment for apolipoprotein amyloidosis. Most care focuses on managing symptoms and protecting the organs that are affected.

- **Kidney disease:** treating high blood pressure and protein in the urine can help slow kidney damage. Dialysis or a kidney transplant may be needed if the kidneys fail. Transplanted kidneys can work well for many years.
- **Liver transplant (AApoA-I):** because the liver produces the abnormal protein, replacing it with a healthy liver stops new amyloid deposits from forming. Patients who have had this procedure have shown decrease of amyloid deposits in other organs over time.Should my family members be tested for a hereditary form?
- **Heart and other organ transplants:** have been performed successfully in some patients and can provide long-term benefit.
- **Genetic counseling:** is recommended for patients and their families to understand how the condition is inherited and what testing options are available for relatives.

This information is not intended to provide medical advice. It is merely an educational tool. Patients should speak with their care team when making any treatment decisions. ARC gratefully acknowledges Mary O'Sullivan, RN, MHI, PMP for her contributions to this document.

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