TRANSTHYRETIN AMYLOIDOSIS (ATTR)

RARE DISEASE PREVALENT IN THE PORTUGUESE COMMUNITY

ROSA NEEDS TO BREAK CULTURAL BARRIERS

DISCOVERING THE DISEASE

Rosa learns from her doctor that she has amyloidosis and there is a 50% chance that she passed on the gene that causes this disease to her children. Genes carry the information that determines your traits and can be passed down from parents to children.

BREAKING CULTURAL BARRIERS

Rosa is concerned. She is afraid to tell her children that they—and their own children— may have inherited the gene that causes amyloidosis.

In the Portuguese community, sharing personal information, even within families, is uncommon. This is especially true for personal health information. However, the older generation can play a significant role in influencing health behaviors, improving early diagnosis, and encouraging genetic testing in younger family members through modeling, encouraging, informing, and supporting.

Intergenerational influence usually occurs between women, from mother to daughter, and from older affected individuals to younger presymptomatic carriers.

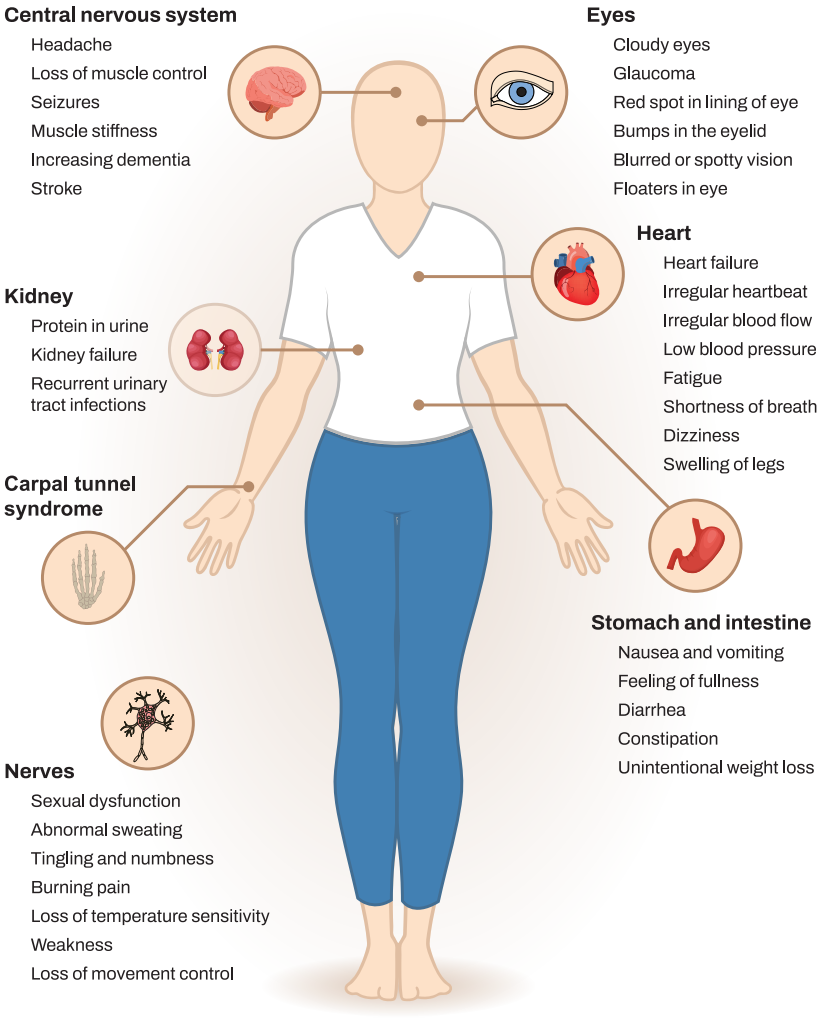
WHAT IS AMYLOIDOSIS AND WHY SHOULD I BE CONCERNED?

Amyloidosis is a group of rare and often fatal diseases caused when certain proteins in the body “misfold” and build up as fibrils in organs, damaging their ability to function properly. One type of inherited amyloidosis, called ATTRv, is caused by a defect in the gene that produces transthyretin (TTR), a common protein that transports thyroxin and retinol in the body. In ATTRv, the misfolding TTR protein builds up as fibrils in the heart and nerves. Heart and nerve damage from ATTRv can severely limit a patient’s quality of life and, if untreated, lead to death.

ATTRv IS MORE COMMON IN THE PORTUGUESE COMMUNITY

A particular type of ATTRv, called Val30Met, is more common in people of Portuguese and Brazilian descent. Approximately 1 in every 538 people in northern Portugal has this type of ATTRv, which mainly affects the nervous system. Problems include digestion, breathing, and heart rhythm issues.

There is no known cure for ATTRv Val30Met, but early intervention can help limit the effects of the disease, extend life expectancy, and improve patient quality of life.



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TELLING THE CHILDREN

Because it is important to treat the disease early, Rosa decides to tell her daughter, Isabelle, that she has amyloidosis and how it impacts her. They discuss genetic testing for Isabelle. Knowing whether Isabelle has the defective gene will help her be alert for early symptoms, prepare for the future, and get an early start on treatment if necessary.

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CONSULTING WITH A SPECIALIST

Following the discussion with her mother, Isabelle consults with a genetic counselor. The counselor helps Isabelle think through the issues about genetic testing and the potential benefits for her family. With the counselor's guidance, Isabelle decides to get tested and finds that she has inherited the defective gene that causes hereditary amyloidosis.

The genetic counselor also informs Isabelle of the Genetic Information Nondiscrimination Act (GINA) that protects her against discrimination by employers and health insurance companies.

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HAPPIER FAMILY

Now that Isabelle and her adult children have been tested, the family openly discusses how to best manage the disease and the best treatment options with their doctors. Importantly, everyone feels less stressed and much happier.

MORE INFO

Genetic Information Nondiscrimination Act (GINA)
www.eeoc.gov/genetic-information-discrimination

Free testing for US- and Canada-based families is available at:
www.invitae.com/en/sponsored-testing/alnylam-act-hattr-amyloidosis.

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THE DISCUSSION

Telling family members that you have been diagnosed with amyloidosis can be a difficult and emotional conversation.

Here are some suggestions to help you communicate with your family

CHOOSE THE RIGHT TIME AND PLACE

Find a quiet and comfortable setting where you can talk.

BE PREPARED

Before you talk with your family, educate yourself about amyloidosis. This will help you explain the condition, its prognosis, and treatment options.

STAY CALM AND COMPOSED

It is natural to feel a range of emotions when discussing a serious medical condition. Remaining calm can help you clearly share the information.

USE SIMPLE LANGUAGE

Explain amyloidosis in simple terms. Provide a basic understanding of the condition, its symptoms, and its impact on your life.

BE HONEST

It is important to be open and honest. Share your feelings, concerns, and any uncertainties you may have.

SHARE INFORMATION ABOUT YOUR TREATMENT PLAN

Let your family know about your treatment plan and prognosis. Reassure them that you are working with medical professionals to manage the condition.

ENCOURAGE QUESTIONS

Invite your family members to ask questions and share their thoughts and concerns.

ACCEPT SUPPORT

Let your family know how they can support you during your journey with amyloidosis, and that their support can make a difference.

SHARE RESOURCES

Provide information about reliable sources that can help them better understand the condition and its implications.

DISCUSS GENETIC TESTING

Discuss the benefits of genetic testing. Even if they decide against it, encourage them to share their family history with their physicians.

WRITE A LETTER

In the most difficult situations, write a letter to your relatives informing them of your situation and how you are coping.