Amyloidosis Ireland

Amyloidosis UK

the BRIDGE hATTR amyloidosis

ATTRv Amyloidosis

Is a genetic test right for me?

Alnylam® PHARMACEUTICALS

Developed and funded by Alnylam Pharmaceuticals

What is **ATTRv amyloidosis?**

ATTRv amyloidosis (v for variant) is also known as hereditary ATTR (hATTR) amyloidosis. This is an inherited, progressive condition that can cause symptoms affecting the heart, nerves and digestive system.

How is ATTRv amyloidosis inherited?

ATTRv amyloidosis is caused by an alteration in the **TTR gene** - when a gene becomes altered in this way, it is known as a mutated gene.

People living with ATTRv amyloidosis will have **inherited a mutated TTR gene from a parent.**

When someone with the altered TTR gene has a baby, there is a **50% chance** that they will pass the gene onto their child.

However, even if someone inherits the gene, that does not mean that they will definitely develop the condition or symptoms.

There are genetic tests that can tell you if you carry the gene. Knowing this information can be very useful as it could help your doctors to assess your risk of developing the condition, spot your symptoms early and ensure you receive treatment quickly.

What is **genetic testing?**

Genetic testing is a process that can identify mutations in genes that cause ATTRv amyloidosis.

It is useful for people with, or at risk of, ATTRv amyloidosis (for example, if your relatives are affected by the condition), to confirm a diagnosis or understand your risk.

Is genetic testing available in the UK and Ireland?

Yes, recently, leading experts in the UK and Ireland published recommendations outlining best practice for who should get a genetic test for ATTRv amyloidosis.

These recommendations are included within this booklet so that you know the options that are available to you.

What do these experts **recommend?**

The experts recommend three types of people for genetic testing

People with suspected symptoms (confirmatory genetic test)

- For an individual experiencing symptoms characteristic of ATTRv amyloidosis to confirm if they have the condition.
- Testing at this stage will allow your doctor to confirm your diagnosis quickly and effectively.

People without symptoms who may have ATTRv amyloidosis (pre-symptomatic genetic test)

- For people who have a family member with a confirmed diagnosis of ATTRv amyloidosis.
- The person having this test does not need to be experiencing symptoms in order to have a test.
- This test will allow your doctors to monitor your health and detect any signs and symptoms early.

People with amyloid deposits characteristic of ATTR amyloidosis (diagnostic genetic test)

- For people who have had a biopsy that has shown they have 'amyloid deposits' in their body. In this case, the genetic test is to confirm which form of amyloidosis they have.
- Testing at this stage will allow doctors to confirm the exact type of amyloidosis someone has, and inform the appropriate follow up and treatment.

How can I **get a genetic test?**

Having a genetic test is a big decision that could have implications for both you and your family, so it is important that you know what the test might mean for you.

If any of the recommendations in this booklet apply to you or a family member, you may feel unsure about what to do next.

Is there anyone I can speak to for more information about the test?

If genetic testing is an option you want to explore further, it is best to discuss this with your GP.

If you choose to consider having a test, your GP will refer you to a genetic counsellor for additional support. They will provide you with more information, such as:

- **What genetic testing is and involves**
- **Understanding the risks and benefits of testing**
- **The potential implications of a test for you and your family**
- **Support that you can access after receiving the results of a test**
- **How to speak to your family members about genetic testing**

Genetic testing is usually recommended for adults (18+) who can give informed consent to having a test and understanding its implications. If you have any questions about testing, your GP or clinical team will be able to answer these.

Where can I learn more **about these recommendations?**

Visit **www.hATTRbridge.eu** to hear David, who lives with ATTRv amyloidosis and has a number of family members affected by the condition, and Tootie, a cardiac genetics nurse, discuss the recommendations in more detail.

Where can I go for **more support?**

There are organisations that specialise in ATTR amyloidosis that can give you additional information:

Amyloidosis UK

- **https://amyloidosisuk.org/**
- **https://amyloidosisuk.org/useful-resources/**

Amyloidosis UK acts to provide extensive information and support to patients in the UK and is formed by a community of people living in the UK affected by ATTR amyloidosis.

Amyloidosis Ireland

- **https://amy.ie**
- **info@amy.ie**

Amyloidosis Ireland offers support to patients affected by ATTRv amyloidosis and ATTRwt amyloidosis (wt for wildtype). They are committed to raising awareness to ensure people are diagnosed and have access to available treatments at the earliest opportunity.

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Scan to find out more at www.hATTRbridge.eu

This material has been informed by the latest clinical consensus recommendations published by a multidisciplinary panel of experts in ATTR amyloidosis. This document is intended to support patients and families with, or at risk of, ATTRv amyloidosis to make informed decisions about their care.

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