

# DID YOU KNOW ALPHA-1 IS A GENETIC DISEASE?

**Don't let your family  
go untested**



**GRIFOLS**

## WHY HAVE I BEEN GIVEN THIS BROCHURE?



If you or someone close to you has been diagnosed with **alpha-1 antitrypsin deficiency**, also known as **AATD** or **alpha-1**, or if you or a family member is an alpha-1 carrier, this brochure includes important information, so please share it with your relatives.

Here you will find what alpha-1 is, how it passes on within a family, and how testing and early diagnosis can improve the health of people affected by alpha-1.

## WHY IS IT IMPORTANT THAT YOUR FAMILY IS AWARE OF ALPHA-1?



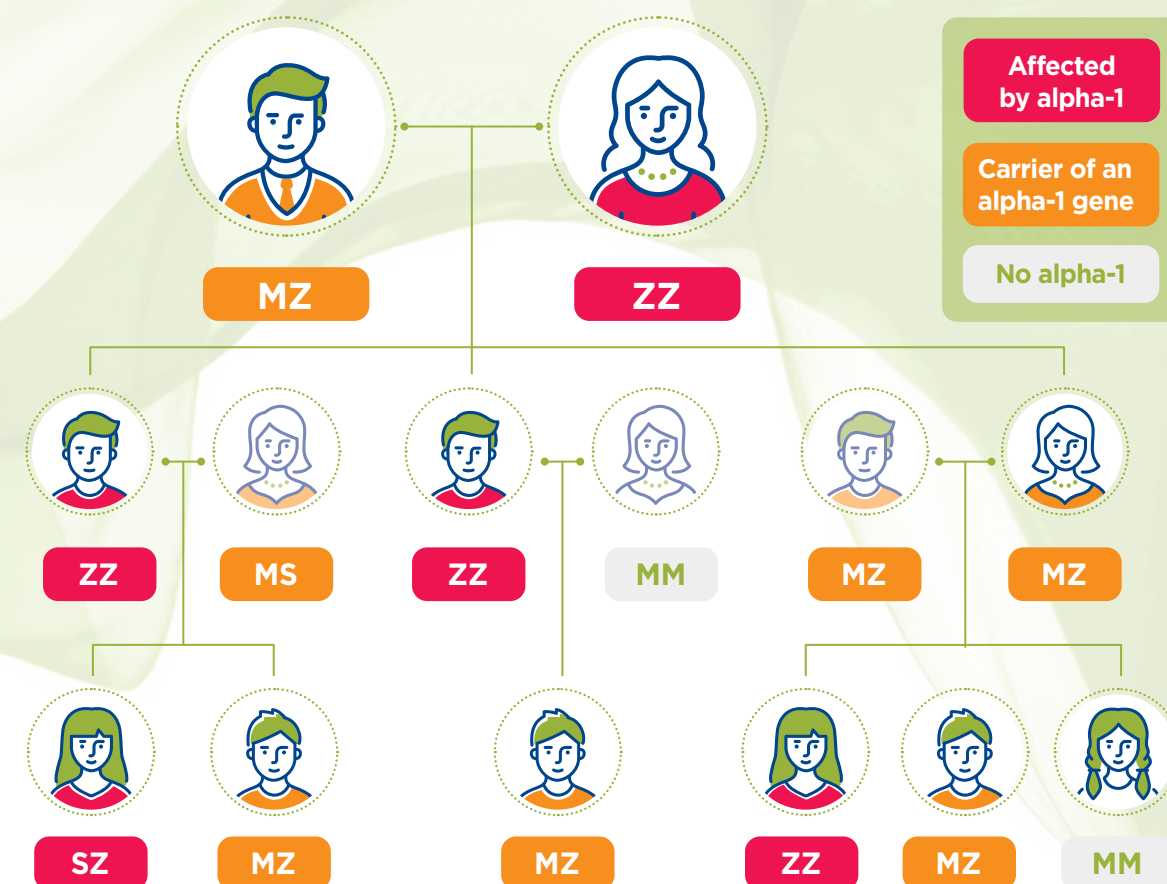
Alpha-1 is a genetic disease, which means **it is passed on within a family**.

As you have been diagnosed with **alpha-1**, it is possible that someone in your family has the disease as well.

At the very least, your parents, children and possibly other family members are carriers.

Remember, carriers are people who can pass on the disease to their children but may not have the disease themselves.

### Example of how alpha-1 is inherited



**If you have alpha-1, then it is possible that someone else in your family has it as well.**



# M, S AND Z ALLELES, WHAT DO THEY MEAN AND WHY DOES IT MATTER?



Everyone has two genes, also known as **alleles**, related to the production of the alpha-1 antitrypsin (AAT) protein. The **normal** AAT protein is made from the **M allele**. The most common **defective** alleles are called **S and Z**.

Whether someone **has the disease** or is a **carrier** depends on the combination of alleles that the person has.

ALLELE COMBINATION		PATIENT DESCRIPTION	RISK OF LUNG AND/OR LIVER DISEASE
Two normal alleles	MM	Normal	
One normal, one defective allele	MS	Carrier	It is unclear whether there is a risk of getting disease symptoms.
	MZ	Carrier	<b>Mild to moderate AAT deficiency</b> — may get disease symptoms.
Two defective alleles	SS	Alpha-1	It is unclear whether there is a risk of getting disease symptoms.
	SZ	Alpha-1	<b>Severe AAT deficiency</b> — likely to get disease symptoms.
	ZZ	Alpha-1	<b>Severe AAT deficiency</b> — likely to get disease symptoms.

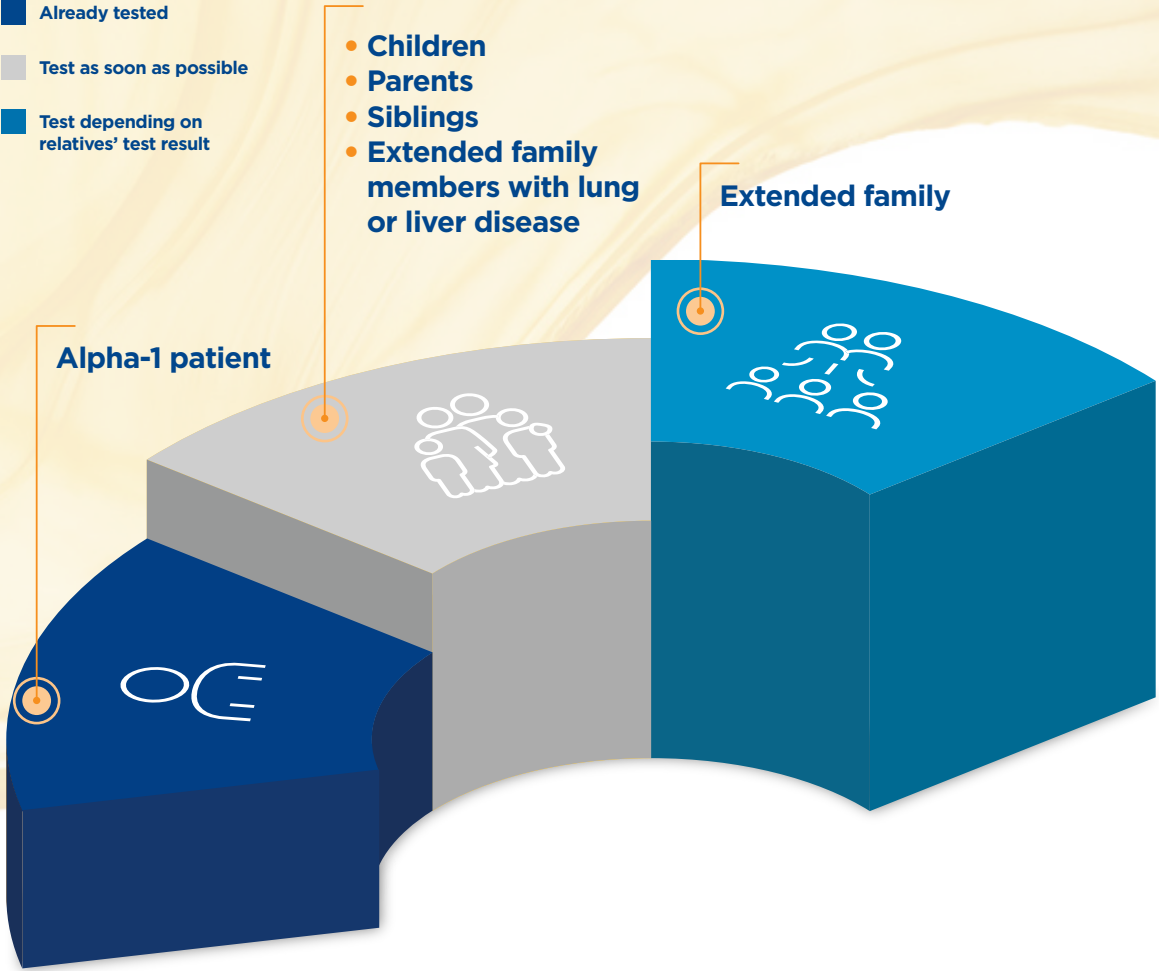
Knowing your allele combination will help you to better understand the risks.

# WHO SHOULD GET TESTED?



If you have alpha-1, your **immediate relatives** may also have the S or Z genes. So, now that you know your allele combination, you can help them know theirs too. Even carriers can develop disease symptoms, which is why it is recommended that family members are screened for alpha-1.

In addition to testing your close relatives, **extended family members with lung or liver disease** are considered at greater risk and should be considered candidates for testing. Based on the test results, your doctor may suggest whether to test more distant relatives.



## HOW DOES TESTING WORK?



Whether you have been diagnosed with alpha-1 or are a carrier, you are already aware that testing for alpha-1 is fairly **simple, quick** and highly **accurate**.

The diagnosis of alpha-1 is usually determined by a blood test; however, genetic testing can also be done through a mouth swab test.

Your doctor will help your family decide which test is right for them.

**Testing for alpha-1 is simple and will help your family know if they are at risk.**

## WHY IS TESTING SO IMPORTANT?



You already know that alpha-1 is a disease that people are born with and that it slowly damages the lungs and liver.

In fact, the disease progression is often so gradual that you **can only see the physical symptoms once enough damage has already occurred** to the liver and/or lungs. Many people have the condition but are unaware of it.

Testing relatives allows for an **early diagnosis**. If diagnosed, their doctor can take measures to slow the progression of the disease, with the goal of **preventing significant damage to the lungs and liver**.

The earlier the diagnosis, the sooner preventative measures can be taken. The main actions that can be taken include:



**Lifestyle** changes



If eligible, **treatment** to slow the progression of the disease

As well, if someone in your family already has any lung or liver issues, an alpha-1 diagnosis means that their doctor would know the underlying cause and can take measures to manage it as best as possible.

**Remember: early detection helps to slow down disease progression.**



# TESTING FOR ALPHA-1: THE ONLY WAY FOR YOUR FAMILY TO KNOW FOR SURE

## No disease

If you have alpha-1, then it is possible that someone else in your family has it as well.

## Carrier

What genetic risk does your sibling, child or parent have?

## Alpha-1 disease

**The only way to know is through testing.**

**Speak with your doctor to find out more about testing your family for alpha-1.**

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