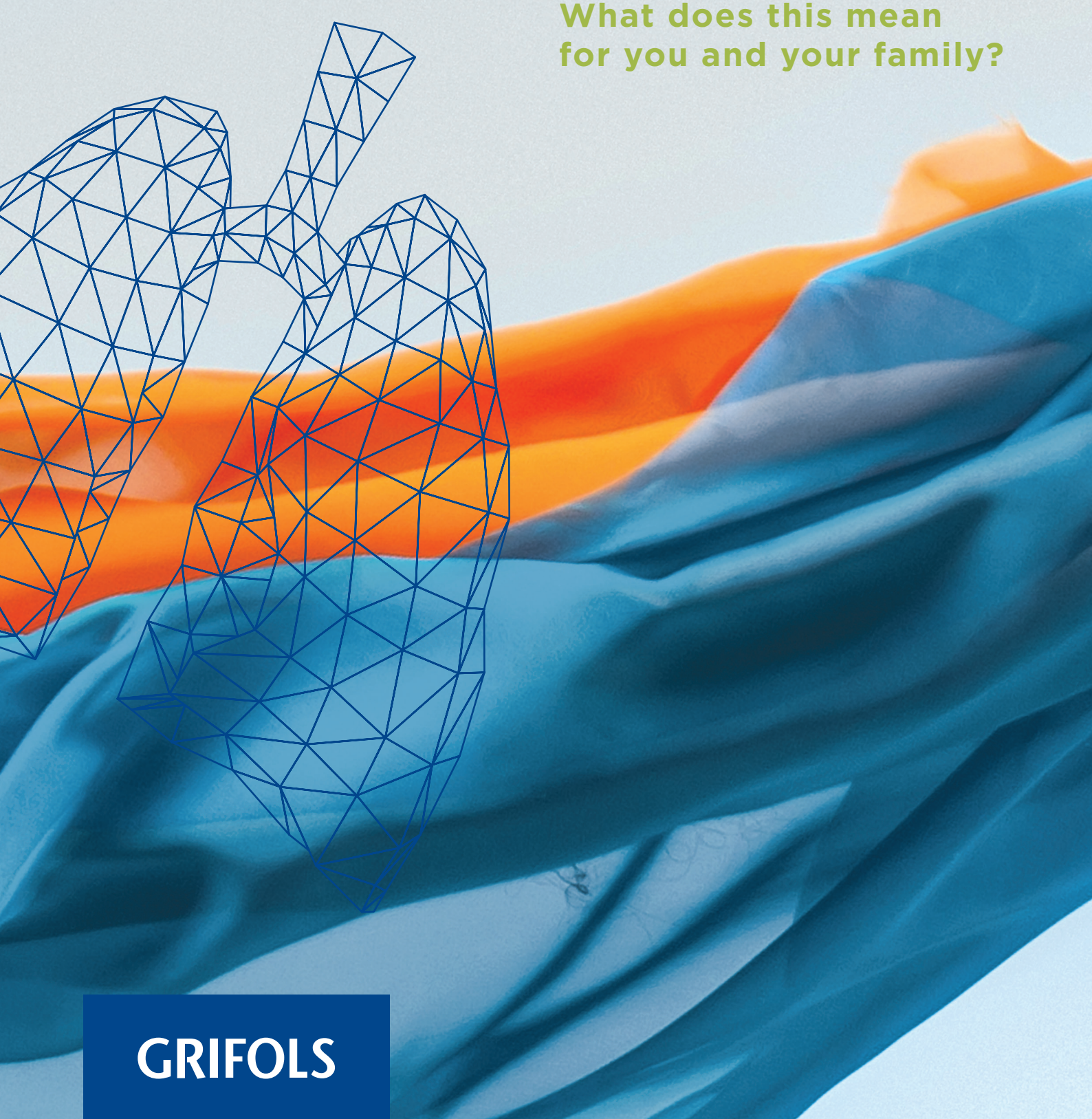
YOU HAVE BEEN DIAGNOSED WITH **ALPHA-1**

What does this mean for you and your family?



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WHY HAVE I RECEIVED THIS BROCHURE?

You have received this brochure because you have been **recently diagnosed** with **alpha-1 antitrypsin deficiency**, also known as **AATD** or **alpha-1**.

This brochure is intended to clarify questions and concerns you may have about the disease. Just remember that you are not alone, your physician is here to help you and there are also various support groups available.

WHAT IS ALPHA-1?

Alpha-1 is a hereditary condition that is passed on from parents to their children through genes. It can cause a severe lack of a protein in the blood called **alpha-1 antitrypsin** (AAT). This is an important protein mainly produced by the **liver**. The main function of AAT is to protect your **lungs** from inflammation caused by infection and inhaled irritants such as tobacco smoke.

Alpha-1 patients produce an abnormal protein which cannot be released from the liver at the proper rate. This may lead to an accumulation of the abnormal AAT protein in the liver, which can cause **liver disease**.

Some people with alpha-1 remain without symptoms throughout their lives. Early diagnosis and avoiding risk factors, such as cigarette smoking, can help prevent alpha-1, from causing disease symptoms.

When you have alpha-1 the lungs and liver can be impacted.

HOW DOES ALPHA-1 AFFECT MY BODY?

LUNGS

The main role of AAT is to protect the lungs from inflammation caused by infection and inhaled irritants. When our lungs are exposed to an irritant or even an infection, it can damage them. So, our body sends out an enzyme called neutrophil elastase to destroy the irritants.

The problem is that neutrophil elastase, if left unchecked, can also damage healthy lung tissue. That is where AAT comes in. AAT eliminates the excess neutrophil elastase in order to protect the lungs.

If you have alpha-1, there is no or not enough AAT reaching your lungs to stop the neutrophil elastase.

This means that healthy lung tissue slowly becomes compromised over time, which increases your risk of **emphysema.**

LIVER

When you have alpha-1, your liver either produces no AAT or creates it in the wrong shape. If it isn't the right shape, the AAT protein has trouble leaving the liver. So not only is there less AAT in your lungs to control the neutrophil elastase, but there is an excess of it being trapped in your liver.

Over time, this trapped AAT protein can cause irreversible damage to liver tissue, thereby increasing the risk of chronic liver disease.

However, there is a type of alpha-1 where no AAT is produced. In that case, there is no damage to the liver.

THERE ARE DIFFERENT TYPES OF ALPHA-1. WHAT DOES MINE MEAN TO ME?

Alpha-1 is a genetic disease that is passed from parents to children through two genes, also known as **alleles**, that are involved in the production of the AAT protein. The **normal** AAT protein is made from the **M allele**, but there are a range of different types of **defective alleles**. The most common ones are called **S and Z**. You may have a different allele not mentioned here.

Individuals with **one normal** allele and **one defective** allele are called **carriers**. Whether you are S or Z is directly related to how much AAT reaches your lungs, and, therefore, how severe your disease is considered. The severity depends on the **combination of alleles** a 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	**Mild to moderate AAT deficiency** — 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	**Severe AAT deficiency** — likely to get disease symptoms.
	ZZ	Alpha-1	**Severe AAT deficiency** — likely to get disease symptoms.

Being aware of your allele combination will help you to better understand your disease.

WHY AM I HAVING THESE SYMPTOMS AND WHAT ELSE SHOULD I EXPECT?

If you have been diagnosed with alpha-1, you probably have started to show some of the most common symptoms. **Alpha-1** is a disease that people are born with, but as the symptoms develop slowly over time it is often only identified once it has caused problems to their lungs and/or liver. Here we list the different symptoms and conditions that can be observed in alpha-1 patients. You may develop some them.

LUNGS

Symptoms and effects include:

- Shortness of breath
- Wheezing
- Chronic cough and sputum (phlegm) production
- Recurring chest colds
- Decreased exercise tolerance
- Year-round allergies
- Chronic bronchitis
- Bronchiectasis
- Emphysema

You might be familiar with some of these clinical manifestations because they are commonly grouped under the term **chronic obstructive pulmonary disease** (COPD).

LIVER

Symptoms and conditions include:

- Eyes and skin turning yellow (jaundice)
- Swelling of the abdomen (ascites)
- Vomiting blood or passing blood in the stool
- Unexplained liver problems or elevated liver enzymes
- Neonatal hepatitis
- Chronic liver disease
- Cirrhosis
- Liver cancer

In addition to lung and liver issues, some patients also experience **panniculitis**, an inflammatory skin disease created by an excess of white blood cells. Panniculitis frequently causes painful lumps under or on the surface of the skin.

If you have been recently tested and diagnosed due to another family member's diagnosis, it could be that you are not showing any symptoms. Bear in mind that alpha-1 may already be affecting your lungs and/or liver — be attentive to the signs and symptoms indicated above so you can notify your physician if you notice any warning signs.

Knowing the symptoms will help you and your doctor better control your illness.

IS THERE A TREATMENT FOR MY LUNG AND/OR LIVER DISEASE?

Currently, alpha-1 has no cure. But, while the damage caused to your lungs and/or liver is not reversible, it is possible to slow the progression of the disease. People with AATD **can live healthy and productive lives**.

Your physician will determine the course of your treatment and give you guidance on lifestyle measures to prevent or reduce the associated risks of alpha-1.

Some recommendations to better manage alpha-1 are:

ENVIRONMENTAL AWARENESS	INCREASED DOCTOR VISITS
Avoid repeated exposure to dust, fumes or gases	Get fully vaccinated for COVID-19, pneumococcal disease, hepatitis A and B and have yearly flu shots
CHANGES IN LIFESTYLE	
Do not smoke, and avoid second-hand smoke	Quit or cut back on drinking alcohol
Exercise regularly	Eat well

There are therapies that can be appropriate for relieving alpha-1 symptoms in some individuals, including antibiotics, bronchodilators, corticosteroids, supplemental oxygen, surgery, lung volume reduction, as well as lung or liver transplantation.

If eligible, a more specific treatment to slow the progression of alpha-1-related lung disease is **augmentation therapy**, also called replacement therapy. This therapy is based on the intravenous administration of AAT from the blood plasma of healthy human donors to individuals diagnosed with severe AAT deficiency and emphysema, to augment their alpha-1 levels in the lungs.

There are treatments and preventive measures that can help you take better control of your health.

WHERE TO FIND ADDITIONAL SUPPORT

Just remember, you may have alpha-1, but you are not alone.

In fact, there is more helpful information available and a large alpha-1 community to support you. The area where you live may have support groups and/or patient organisations that can offer you guidance and assistance on this journey. Patient organisations are the best place to share experiences and to learn more about living with alpha-1.

You can find out if there is a support organisation in your country here: **www.alpha-1global.org**

Additional information and resources about alpha-1 can be found here: **www.alpha1.org**

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