

# What is the medical and emotional journey for patients with a rare genetic disease infected with COVID-19 in the United States?

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The pandemic altered everyone's life, but especially high-risk populations such as the elderly, diabetics, and individuals with chronic diseases. Alpha-1 Antitrypsin Deficiency (AATD), for example, is a genetic condition that affects the lungs, liver, and other organs. The new SARS-CoV-2 (COVID-19) is a respiratory virus, which makes AATD patients a high-risk population, as ninety percent of these patients have lung disease.

This research focuses on learning about AATD patients. Five interviews were conducted to learn about the experience of AATD patients who were infected with COVID-19 and reside in the United States. To accurately reflect the patients' emotions, four design tools were used: empathy map, patient profiles (whose names have been changed for privacy purposes), customer journey, and a systemic map. The most important finding was that Alphas (people with AATD) need to take exaggerated measures to avoid contracting the virus, as they are considered a high-risk population. Nevertheless, the very discourse of preventive measures by everyone around them has caused those infected to feel ashamed and not want to share their story. Through these stories, we hope people can empathize with patients and motivate them to get tested so that undiagnosed Alphas can begin treatment.

## What is Alpha-1 Antitrypsin Deficiency?

Alpha-1 Antitrypsin Deficiency (AATD) is an inherited genetic disease of European origin characterized by an increased risk of illness in the lungs and liver, among other organs. Additionally, it's distinguished by a low percentage of the AAT protein that acts as an anti-inflammatory and protection source for the lungs. This protein is produced in the liver, and the liver of the patients doesn't release the protein due to low production levels. Consequently, as the protein accumulates, liver damage develops. People with AATD can develop chronic obstructive pulmonary emphysema (COPD), cirrhosis, and others.

The deficiency is not well known among the public, as there is little information available, and most people have never heard of it. Physicians lack knowledge of it, and they rarely test patients with pulmonary problems. Consequently, the data shown in this study is not entirely accurate since we are missing worldwide diagnostics.

There is no known cure for the disease, but there are treatments that help reduce lung damage, such as respiratory therapy, lung and liver transplantation, and augmentation therapy. Patients take this treatment throughout their entire life; it consists of receiving a weekly dose of AAT protein intravenously to help the lungs. There is currently no treatment, however, for liver damage.

## Objectives

There is a lot of literature on the measures that a person with AATD must enforce, but it fails to mention the emotional toll for the patients infected with COVID-19. The aim is to illustrate and fill the gap.

Through COVID-19, we can make people conscious of this deficiency and raise awareness of what to do and how to treat it.

## Methodology

To reflect the emotions of AATD patients and understand the reality they face, the methodology used in this research was qualitative.


The interviews were conducted with five patients with AATD who were infected with COVID-19 and resided in the United States. The intent was to learn about their journey. In addition, four physicians with expertise in the deficiency were surveyed to learn how they have treated the virus. Further interviews were also conducted with staff from Alpha Net, an organization that deals directly with AATD patients.

It's important to mention that this sample is not representative of this universe being the AATD infected with COVID-19 that live in the United States; as 629 patients were infected with COVID-19. The half universe is Alpha 1 patient in the world.

## Insights

People with AATD need to take exaggerated measures to avoid being infected with COVID-19, as they are considered a high-risk population. But the very discourse around preventive measures, promoted by everyone around them, such as doctors, families, and friends, has caused those who have been infected to feel ashamed and not want to share their stories.

### CUSTOMER JOURNEY



**SARA GREEN**  
Duration: 4 weeks  
Age: 57  
From Michigan, USA

Information is key. I had an awful experience at the hospital. I was not in my home state.

**Alpha information**  
Lung affected 45% lung FEV1  
Phenotype: ZZ  
Augmentation therapy and nurse at home.



**EVA WEST**  
Duration: 4 months  
Age: 58  
Wilmington, USA

Placed away on March 9, 2021

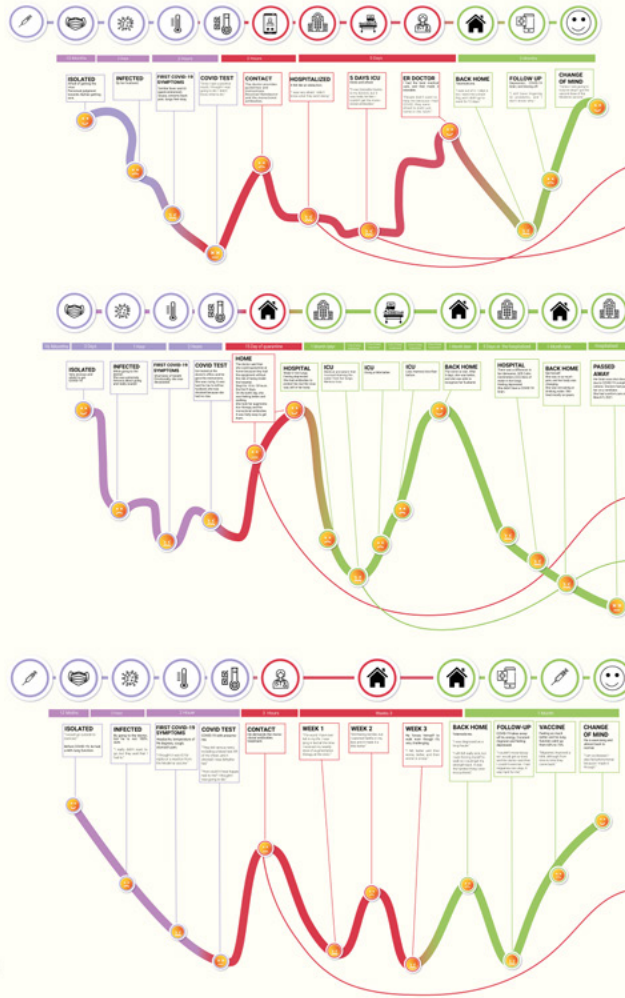
**Alpha information**  
Lung affected 23% lung FEV1  
Phenotype: ZZ  
Augmentation therapy self-infused  
Two liters of oxygen daily in a nebulator.



**LIAM HART**  
Duration: 1 month  
Age: 66  
Sydney, USA

To get a nurse and I changed my routine to make my wife happy.

**Alpha information**  
Lung affected 73% lung FEV1  
Phenotype: ZZ  
Augmentation therapy at a clinic.



## Results

Alpha-1 patients must be well-informed of the treatment they need if they get COVID-19, as many doctors aren't aware of the Monoclonal Antibodies Therapy or are unwilling to share this information because there is a limited number of medicines.

The research revealed that taking the augmentation therapy infusion treatment may help the patient and confirmed that the vaccine ensures a less aggressive illness. Furthermore, most of the subjects participating in this research had long-term sequelae (were long haulers). Another finding is that those infected with COVID-19 are treated with callousness in hospitals.

Experts anticipated that the Alpha-1 patients most affected by COVID-19 would be those who had transplants, since they are required to take a lot of medication and antibodies. However, in this study, we found that these patients were actually the ones who lived through the disease less aggressively because their lungs function at 90% (above that of the average Alpha-1 patient).

It is of the utmost importance to mention that every patient's story and reactions are different, as well as every disease.

## Conclusions

Through the COVID-19, we can learn about AATD, raise awareness, and identify carriers. For this purpose, the Alpha-1 Foundation offers a free test kit, so people can know if they carry this deficiency (www.alpha1.org).

Alphas are a high-risk population, and, as such, they live in fear of contracting the virus. They need to understand that if they are infected, they have nothing to be ashamed of. The COVID-19 is a community-spread virus, which means that anybody can fall ill.

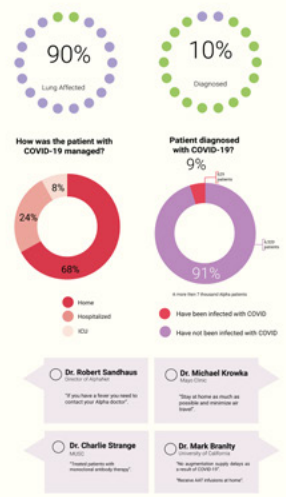
The research found that patients are looking to be understood and eager to share their knowledge of this disease.

## COVID alongside AATD

- i. A study in Italy argues that their high mortality rate is due to the increasing number of patients with Alpha-1 in their population. Since most of the patients that have been diagnosed with Alpha-1 reside in the northern part of the country, this is where the mortality rate is higher.
- ii. In a recent clinical trial in Ireland, Augmentation therapy is used to treat long-term COVID-19 patients (long haulers), specifically those with severe lung affections (Article, 2020).

253.404 Pi\*ZZ Worldwide

Alpha 1 anti-tryptic affects one in every 1,500 - 3,000 people of European descent, among which there are an estimated 100,000 Americans, while over 20 million from the USA are carriers (Moffitt, 2020).



### COMMON SYMPTOMS

Depression  
Treated with monoclonal antibodies  
They all work in AlphaNet as coordinators  
Ashamed of contracting COVID-19  
Lung damage  
Scared of COVID-19  
Limited pain medication to avoid liver damage  
Extreme lockdown  
Dream of having a healthy life  
Two doctors one in charge of the Alpha problems and the other of the COVID-19  
They are all religious  
Long haulers  
Hope for an AATD cure

