





the BRIDGE
hATTR amyloidosis

BRIDGE THE GAP

Personal experiences, feelings
and hopes from patients and carers
living with hereditary ATTR amyloidosis

This booklet is designed to support patients and
caregivers living with hereditary ATTR amyloidosis.




AMYLOIDOSIS ALLIANCE
THE VOICE OF PATIENTS

FAMY
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Hereditary ATTR (hATTR) amyloidosis is an inherited disease caused by a gene change (mutation). The mutation causes a protein in the blood called transthyretin (TTR) to misfold (take on an abnormal shape), which interferes with its function. When misfolded,TTR builds up and can form deposits in multiple organs and tissue, such as the nerves, heart and digestive system.









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We have come together as a group of individuals living with, or caring for someone with, hATTR amyloidosis to help increase awareness and understanding of the condition among newly diagnosed patients, their families and the general public.

As disease awareness 'Ambassadors' we hope that by sharing our own journeys, we can help individuals positively navigate their own illness. Our aim is to provide patients and families with access to high-quality, accurate disease information. We hope that our experiences can support productive conversations with healthcare professionals and support patients with the tools that can enable important conversations within the family to take place.

Coming to terms with a rare genetic disease is unique to each person diagnosed, as well as the family and the support network. Current limited awareness in society about the condition may provoke a lack of hope and lead to fear for the future. What we want is for patients to feel supported, listened to, understood and not alone, to build hope for all.

This booklet brings together a collection of our experiences and feelings at key points in our own journeys. To view our stories in more detail, please go to **https://www.hattrbridge.eu**

JEAN-CHRISTOPHE FIDALGO
BOARD OF FOUNDERS AND DIRECTORS
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What pre-diagnosis symptoms of your hATTR amyloidosis did you experience?

— VITOR

AMBASSADOR, PORTUGAL

I began to feel strange changes in my body. My ankles were weak, and I had numbness in my toes. I felt as if I was walking like a duck as my feet flopped. I was also losing weight and strength.

I made excuses for my inability to lift or move things. I was only in my 20s! I wasn't ready to face what was starting to happen to me yet.

— DAVID

AMBASSADOR, UK

One of my loves was hill and mountain walking. My first recollection that something wasn't quite right was when I began having difficulty walking, particularly uphill and over long distances. I had chest pains, tightness in my chest and palpitations. I knew it wasn't because I was unfit. Over the next year, there were other things going on in my body that I was noticing.

Work was sometimes quite physical and I struggled to keep up with the other guys. Occasionally I had to take time out and go and do some paperwork, just to get the chest tightness that I was experiencing down.

— CATILENA

AMBASSADOR, SPAIN

As I watched my mother die of this disease—and how she was dying—my own symptoms were quickly growing worse. More and more, I just wanted to disappear from the world and pretend that nothing was happening.

My initial symptoms kept worsening. Plus, I could no longer differentiate between hot and cold water. My health continued to decline, and I began to experience digestive problems—alternating between periods of constipation and diarrhea.

How did you find out that your symptoms were in fact hATTR amyloidosis?

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— JOSÉ

AMBASSADOR, PORTUGAL

My brother decided to get tested at the same time, so both of us went together to get the results. My little brother went in first…

We were both diagnosed at that time. It never crossed my mind that both of us could have the disease. I always thought it might be one of us but never both. The world didn't end that day, but it felt like it nearly did…

— ROLAND

AMBASSADOR, GERMANY

I was sent to the hospital for observation and more tests. There, they did a foot biopsy to gather tissue samples to study for the amyloid deposits, or so it was explained later. I was sent back to the cardiologist, who sent me on to the electrophysiologist where, finally, all of the results were summarized for us.

Believe it or not, the doctors finally had a name for what had been causing over 12 years of mysterious symptoms – cardio hATTR amyloidosis with fainting events due to falling blood pressure. I had never heard of hATTR amyloidosis.

— JEAN-CHRISTOPHE

AMBASSADOR, FRANCE

I was worried and frustrated because I felt powerless against what was happening to me. When the doctor told me I had neuropathy, I found myself thinking of my dad. He had neuropathy, too. *Could it be passed from one generation to another? Why me? Why my family? Is there a curse?*

I decided to change neurologists because I was angry after the three years lost since the first visit. The new one gave me four life-changing pieces of information, including my official hATTR amyloidosis diagnosis.

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How did you feel when you received your diagnosis?

— DAVID

AMBASSADOR, UK

"Well, in came the consultant. He walked in and quietly shut the door behind him, his face was confident and professional. Then he said it. He calmly explained to me that I have hATTR amyloidosis, an inherited gene mutation that accumulates amyloid deposits in the tissues. I was shocked, and I think it needed time to sink in. My wife looked over at me and I could see her eyes watering, which is what affected me the most."

— VITOR

AMBASSADOR, PORTUGAL

"I already knew in my heart the result would be positive. I was in a state of shock. I had inherited the condition that had killed my father and I was only in my mid-20s. I panicked. I wanted to run away."

— JOSÉ

AMBASSADOR, PORTUGAL

"Of course, having this disease wasn't news to me, but the reality of developing symptoms wasn't great.

Memories of my mother and how hard it was for her to live with amyloidosis came back to me. But I knew it was necessary to keep my head up and search for a solution."

When you got diagnosed, how did you approach your family with the news?

— JEAN-CHRISTOPHE

AMBASSADOR, FRANCE

"For me, it was quite difficult to talk about it with my mother since she had looked after my father for almost 10 years and, unfortunately, he wasn't getting any treatment. So there was a progression until he weighed 45 kilos, was confined to bed and couldn't move anymore.

I tried to protect my mother and I told her only when I knew I was being treated. I was certain it would be a shock for her, to think that she would repeat the experience she had lived through with her husband, with her son. When I told her the news, it was a shock for her, it was difficult, but she became really invested to support me."

— CATILENA

AMBASSADOR, SPAIN

"In the year I had the first symptoms, my mother was having the final symptoms of the illness. I decided not to tell her what I was going through because I didn't want to worry her. I regret not having done that. When I got my diagnosis, I felt very sad and scared. It coincided with the fact that the day before we had been at my mother's funeral.

After the consultation I went straight to my father's house and we hugged each other very tightly. I told him, "Dad, Mum has gone but the illness hasn't. I'm the patient now and it's time to take care of myself."

— ROLAND

AMBASSADOR, GERMANY

"After a life spent doing exciting things I was being confronted with this strange diagnosis. I cannot explain well enough what it felt like to learn about what was happening to me, and about what the future might bring.

I was also worried for my wife Nicky more and more. Her burden was heavy, and both of our emotions were raw. It was a very difficult period."

What was the reaction of your family members?

— DAVID

AMBASSADOR, UK

My two oldest children did not take the news well. They were really upset and concerned that they were going to see their father deteriorate, especially since they'd already seen how the disease affected their aunt. I tried to keep a positive outlook and reassure them that I was in great hands.

— MARGARIDA

WIFE OF VICTOR, PORTUGAL

I love my husband, and I hate amyloidosis. It makes me angry, tired and frustrated. I yearn for a cure. I cry from a deep, dark place when I'm alone. I don't want to go through this, but if I must, I know what to expect. I also know I must be strong.

— CATILENA

AMBASSADOR, SPAIN

We embraced very tightly and cried together for at least an hour. He (my father) said, "Catilena, I know this condition very well, but now you have to be strong, and I know you will be." Of course, my father is always worried about me. But from that moment on, I stopped living with the disease in silence, and I decided to tell my friends, family and co-workers.

How did your diagnosis impact you initially?

— CATILENA

AMBASSADOR, SPAIN

At first, I socially isolated myself. My family was angry and sad, but my husband and my father have been my biggest support. After a while, I decided to talk about what was happening to me, with my friends, co-workers, bosses, everyone. I stopped being silent about my illness and told them how I was feeling and what I was living with.

The people who truly love you end up understanding you. Those people respected that I needed time to get myself together.

— PHILIP

AMBASSADOR, NETHERLANDS

Life is about choices. Suddenly, I could choose to stop my stressful job in business and switch to something that I might actually enjoy. I could choose to read books just for fun.

I could choose to cook dinners at home for my family and actually be there to share with them. I could choose to change so many diapers, take a deep sniff every time, and be thankful to enjoy the presence of my kids.

— JEAN-CHRISTOPHE

AMBASSADOR, FRANCE

I remember very well when the cardiologist visited me in my room the evening before the pacemaker implantation. He looked at me, in the eyes, telling me that I'd never be able to dive again. *Never dive again?!* I was devastated. *How could this happen? This was my passion!*

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What are the symptoms that bother you the most?

— DAVID

AMBASSADOR, UK

"It's been four years since my diagnosis and, since then, a number of scans have confirmed that the amyloid in my heart has increased and signs of it going into the peripheral nervous system are now occurring. Due to my symptoms, I took a period of time off work. My role in the company required physical demands and long hours of traveling, so I often had to stop in the car to have a sleep. For this reason, I made the difficult decision to retire from the job I loved."

— JOSÉ

AMBASSADOR, PORTUGAL

"Of course, nothing about this is easy. I still have symptoms, and every morning this disease wears on me. As soon as I wake up, I walk out to our garden and vomit so that my family cannot hear me. Minutes later, I'll walk back into the kitchen with a smile on my face, greet my wife, and say "good morning" to my daughters. It is hard to start every day this way, but I try to forget what happens in the morning and focus on the people I love."

— VITOR

AMBASSADOR, PORTUGAL

"This condition can have a dramatic impact on the physical relationships between a man and woman. Men struggle with erectile dysfunction. Intimacy is important to Margarida and me, and we've found ways to continue to nurture this closeness."

What is your greatest fear about having hATTR amyloidosis?

— PHILIP

AMBASSADOR, NETHERLANDS

"My biggest anxiety concerns my children. Having seen the drastic and unexpected effects of the disease on other patients, just the sheer chance they might have to suffer to this extent fills me with dread. I feel responsible for them possibly getting the disease. They may face the prospect of a severe debilitating disease.

How can I as a parent protect them from that kind of harm? What choices will they have?"

— DAVID

AMBASSADOR, UK

"For me, the most frightening part of it all is wondering how it will affect my children and my other relatives. Two of my children, as well as at least a dozen nieces and nephews, could be living with this disease.

That plays on your mind. I hate to think of passing something like this on to my children and knowing it could even pass on to my grandchildren."

— JEAN-CHRISTOPHE

AMBASSADOR, FRANCE

"When you have amyloidosis, you tend to feel like you're not *quite* human anymore. So much is taken away from you: your mobility, your dignity. It's hard to keep a positive outlook on life or your situation but be positive you must! You must rally your support systems, prepare for battle and work out your strategy to survive every day."

What is your greatest wish for yourself or others who have hATTR amyloidosis?

— DAVID

AMBASSADOR, UK

My wish is for a greater understanding of this disease. It feels as if so many health professionals aren't familiar with it, but the quicker people can receive a diagnosis, the quicker they can receive treatment. I'm provided with great comfort and hope when I see the current research and treatment efforts.

Now, I feel the future is much brighter for not only my biological family but my entire amyloidosis family, as well.

— CATILENA

AMBASSADOR, SPAIN

I hope by sharing my own story I have brought honour to my mother. Many times, my mother wanted to talk about her disease and share her own story with me, but I didn't want to hear anything about it—I was too scared.

Sadly, she passed away before we were ever able to share our feelings. My hope is that by giving a voice to this disease, I will help other people feel loved and bring an awareness to amyloidosis.

— ROLAND

AMBASSADOR, GERMANY

I received an invitation to participate in a medical study. This gave me great pleasure. Not only might I possibly be able to help my own symptoms, but also my participation could help the researchers understand more and help other patients, as well. After being so held back by sickness, this made me feel like I could be useful again.

What advice would you give to other people concerned about or recently diagnosed with hATTR amyloidosis?

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— CATILENA

AMBASSADOR, SPAIN

Coming to terms with this disease is not easy. In fact, I always say that I haven't accepted this disease but instead have learned to live with it. Fortunately, I had a great example in my mother. She taught me three of the most important things when it comes to living with this condition.

First, to smile and to share your smile with others. Second, to always pack a survival kit with your essentials, like spare clothes and medication. And third, to enjoy every possible moment, especially the simple things, such as waking up each morning feeling grateful to be alive.

— JOSÉ

AMBASSADOR, PORTUGAL

Since learning I have this disease, my way of seeing the world has changed because I have to think about how much time I have left.

I feel like there is a need to live faster, to take advantage of my time, and to stop giving importance to things that don't matter. I am more focused on what and who I like. Of course, sometimes my way of thinking has a price because not everyone likes this attitude, but it is the way I choose to live my life.

— VITOR

AMBASSADOR, PORTUGAL

Margarida and I make the most out of our life. We take trips. We eat at restaurants that we love. We drive into the hills. We still go to the association where we met. At medical appointments, Margarida is there making certain I hear everything they say, and then that I do everything they say. My doctors treat us both more like family members than patient and caregiver. They really pay attention and listen to both of us.

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As both partner and carer, how have you coped with the reality of daily life?

— MARGARIDA

WIFE OF VITOR, PORTUGAL

"We both had a deep understanding of how amyloidosis is a progressive disease, and we both recognized the signs when the symptoms grew worse.

But my Vitor, he's a fighter. He goes to physio three times a week to help maintain his muscle mass. He works with weights to strengthen his arms and legs. When chronic nausea and diarrhea strike him, he'll commit to eating one more bite of his meal. We work as a team to help him transfer from bed to chairs to standing."

— NICKY

WIFE OF ROLAND, GERMANY

"The stress of not being able to do anything to take away his pain was devastating and giving all my loving care and making him laugh didn't seem to be enough. Nothing prepares one for such worries.

It's not easy to take care of someone else while also trying to manage one's own needs and health concerns. It's not easy to be present and active with the grandchildren. It's not easy to accept that life as you knew it will never be the same. Just when you think you cannot endure anything more, you find a way."
















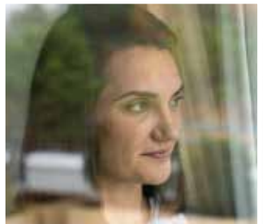


> At the start of this I said I did not think I would make a good disease awareness ambassador. I did not think I had anything worthwhile to say. But I was wrong.
>
> So here I am, deeply grateful for this gift of life and deeply grateful to those who have worked incessantly for more than 10 years on treatments for this disease.
>
> I am thankful for my new mission which leaves me where I started: What will you do with the coming 10 years ahead of you?

— PHILIP

AMBASSADOR, NETHERLANDS

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FAMY NORRBOTTEN

Sources of support

the BRIDGE hATTR amyloidosis

https://www.hattrbridge.eu

The Bridge™ is a website developed and produced by Alnylam Pharmaceuticals; it is designed to help raise awareness of hereditary ATTR (hATTR) amyloidosis and promote education on the condition for patients and their families.

AMYLOIDOSIS ALLIANCE
THE VOICE OF PATIENTS

Amyloidosis Alliance is an international umbrella patient advocacy organisation, aiming to raise awareness and improve the quality of care of amyloidosis patients.

FAMY NORRBOTTEN

FAMY Norrbotten is a Patient Association in the endemic region of Sweden that works to increase disease awareness and support research into ATTR Amyloidosis (hATTR) while also helping to improve the quality of life of amyloidosis patients and their carers.

Notes

Find your path to better health with **STAR: The Symptom Tracker** app for people with **ATTR amyloidosis**.

Download on the App Store

GET IT ON Google Play

For more information, please refer to the sources of support listed within this leaflet.

STAR: The Symptom Tracker app does not pursue a medical purpose. It is designed to keep a record of users' symptoms, how they change over time and their impact on users' well-being and lifestyle. The app is not intended as a disease monitoring tool or as a symptoms checker. To have symptoms checked, users are advised to talk to their healthcare professional. Furthermore, **STAR: The Symptom Tracker** app does not make any medical diagnoses. Users are advised to seek the advice of a medical professional if they are concerned about their health.

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