

# Primary Biliary Cholangitis (PBC)

## From the Patient's Perspective

### Webisode 2: The Diagnostic Journey



## CMEO Podcast Transcript

**Aliya Gulamhusein, MD, MPH:**

And why don't you just tell us a little bit about when you were diagnosed, how you were diagnosed, how that came to be, how long it took, those sorts of things?

**Maria Morais, RN:**

Okay. Well, I sort of progressed rather quickly. I'm not the typical primary biliary cholangitis (PBC) patient that often receive their diagnosis and go through their treatment, first-line treatment, and stabilize for many years. My story is one of a rather quick progression. So, my PBC journey really began in 2017 and I had my gallbladder removed for a large 3.5 centimeter stone. So, my initial symptoms were those of intestinal issues, gastro issues. And after a few months of having my gallbladder removed, my liver enzymes were not returning to normal. And so, the surgeon sent me back to my family doctor and then we then proceeded to go to a gastroenterologist who suspected PBC. He happened to be studying or had studied under a lead in PBC, Dr. Hirschfield, and so knew perhaps things to look for. And so he put me on ursodeoxycholic acid (urso), which is the first-line treatment as I understand it.

And then I received a Fibroscan who, that staged me in possibly stage one or two, as well as with blood work. So, from there I was told that I probably would have a transplant sometime in my life, but it would be later, which in and of itself was pretty shocking to hear the word transplant because I go from having a bit of diarrhea, not a bit, but cramping diarrhea, I didn't have an appetite, I was losing weight, I was losing hair, to the word transplant. So, my disease did progress rather quickly, as I said, with complex and concerning symptoms, that then resulted in a transplant in 2021. So, diagnosed in 2019, transplanted in 2021 with stage four decompensated end-stage liver disease and cirrhosis.

**Aliya Gulamhusein, MD, MPH:**

I wondered if you could comment a little bit on two things. Number one is the psychologic comorbidity or the psychologic impact of being diagnosed and dealing with a rare liver disease, right?

**Maria Morais, RN:**

Yeah.

**Aliya Gulamhusein, MD, MPH:**

It sounds like you were fortunate in that you were diagnosed by someone who had familiarity with, with PBC and with rare disease, but lots of patients aren't so fortunate, right? So, can you tell us a little bit about what that felt like? Number one, having a rare disease. And number two, just the anxiety of the unknown.

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#### **Maria Morais, RN:**

Yeah. So, I think anxiety is often an experience that most PBC patients will have to varying degrees, anxiety about the future of how this disease can progress. So, as humans, we research information. You're given a diagnosis, you go home and you start to read about it. Ideally, you're given information at your physician's office at the time of diagnosis, but there is lots of information on the website that is always, not always helpful. So, it does talk about that end stage in transplant, but there's lots of things in between. But not knowing where you're going to end up, anxiety is very worrying, frustration, right? Like all these symptoms or feelings and emotions that come up about having a chronic disease, the word rare and what that means for care. And do healthcare providers know what PBC is all about as it's categorized as a rare disease, a liver disease? The liver being a major organ that has many functions in the body also can be concerning for patients.

So, for me, the more I read, the more I became anxious, the more knowledge I gained as well. And anxious about just what it would mean to my quality of living, to my ability to be a mother, a wife, a social person, a professional. And so, it's like any stages of acceptance, you sort of go through that denial of, "This is not happening to me," or, "Why is this happening to me?" And that psychological impact that can happen when... feelings of guilt perhaps, "Did I do something?" I'm not a drinker, but knowing sort of the general public understanding of liver disease often comes with stigma of drinking. And sort of figuring out that this disease has nothing to do with alcohol. We are not doing this to ourselves.

So, for me, I did go on medication just try and help, and that was my family doctor suggesting that, just to try and calm myself down a bit, my mind, right? And for the first time I did also go through counseling, which as a healthcare professional myself, to know that that can be helpful is one thing, but then to take the steps to actually go for counseling is another, right, to admit that you need it, that I needed it. And that started to be helpful and just starting to talk about it with others because often if you have such a diagnosis, it can be stigmatizing to talk about it.

#### **Aliya Gulamhusein, MD, MPH:**

Sure. For sure. I mean, you bring up so many important points that when we sit in the clinic sort of see our patients go through and it's so difficult. It's really, I mean, as physicians, we sort of navigate towards treating the things that we know how to treat well, and sometimes the things that are harder but are super impactful, are left a little bit by the wayside. And so, that leads into a question that's actually something that I'm interested in. In your pre-transplant journey, what do you feel like your physicians did best and where could they have improved in your care? I mean, you obviously went through a lot from diagnosis to all of the manifestations of liver failure, which some patients may not experience. But what do you think that the healthcare system or your physicians did well and where could they do better?

#### **Maria Morais, RN:**

Yeah, that's a good question that I've certainly reflected on my journey to this point. And I'm really happy with where I'm at and the kind of care that I still am receiving as a post-transplant patient because there are other autoimmune issues that I've developed since PBC and it's all kind of co-happening. But in terms of my treatment, I started with sort of my family doctor. And that was a critical point to have trust in the family doctor to know that it's just not diarrhea that I'm having, that it's just not stomach upset and that I'm having problems keeping my food in my body. To have a trigger to say, "Okay, let's take it to a specialized service or care to try and figure

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out what is the best plan here, what's going on, and to actually have a diagnosis," because without a diagnosis, you sort of are flopping through the system and trying different things. So, for me to have a trusted medical provider to listen to me and my symptoms was really important.

And then from that to then have specialized care with a gastroenterologist that had some sort of knowledge, which I don't know how much PBC education non-specialized hepatologists are receiving, because in a practice, how many would you regularly see if you're not a hepatologist? So, I was grateful for that, and that he put me on ursodiol right away without even having a super formal PBC diagnosis at hand, just the suspected. And for whatever reason, my disease progressed quite quickly. So, had I had it for a number of years in a mild or that it was there and I never had the opportunity to have blood work to... there was really no symptom to say, "You need to be checked out." But I would say then my hepatologist that took care of my Fibroscan, confirmation of diagnosis, as well as then treatment of my ascites was very much a partner in my care and was listening to what I needed as I started to have severe itching and we started treatments on that, and nothing we could do with the fatigue other than just resting.

And certainly, that's not really a solution, but that's what you can do. And then the brain fog with other treatments. So, I do wish that I had earlier diagnosis and I think moving forward for other patients, that's what we're trying to do.

**Carolyn Legaspi, NP:**

Okay. Do you know what testing your gastroenterology (GI) doctor did?

**Tiffany Brown:**

The first time I saw him, he told me he was 90% sure that he knew what I had, and he did some blood work and the blood work came back and he diagnosed me just from blood work, but also ordered a Fibroscan.

**Carolyn Legaspi, NP:**

Okay, I see. So, your GI doctor had a pretty good inclination right off the bat that it was PBC?

**Tiffany Brown:**

Yes.

**Carolyn Legaspi, NP:**

Yeah, we see that a lot. Our GI doctors, our GI colleagues are pretty good, the ones that I get referrals from are really great at actually identifying and teasing out PBC from other autoimmune-related liver disease. And that's correct, I mean, you can diagnose PBC just from blood work. It's one of the more straightforward, can be one of the more straightforward diagnoses if you know what to look for, right?

**Tiffany Brown:**

Yes, because is it ANA or... I was positive.

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**Carolyn Legaspi, NP:**

The most specific, it's a very specific autoimmune marker, and it's the anti-mitochondrial antibody (AMA), right?

**Tiffany Brown:**

Yes, AMA.

**Carolyn Legaspi, NP:**

Your AMA. If your AMA is positive and the alkaline phosphatase is 1.67, the upper limit of normal, which yours was nine to 10 times that, that's diagnostic for PBC. Plus, you were having all of the symptoms related to PBC, which can be nonspecific sometimes if you go in and tell your doctor you're having some itching and you're having some fatigue, a lot of those things can be related to other autoimmune disorders.

**Tiffany Brown:**

Sure.

**Carolyn Legaspi, NP:**

But the workup itself is very specific for PBC. So, if your AMA is positive, then that's kind of a done deal.

**Tiffany Brown:**

Yes.

**Carolyn Legaspi, NP:**

So, I'm assuming your AMA was positive-

**Tiffany Brown:**

Yes.

**Carolyn Legaspi, NP:**

The alkaline phosphatase was positive as well, or elevated as well. Let's go back to that Fibroscan. So, the FibroScan is a tool that we use, it's an ultrasound, a bedside ultrasound that we use in the office all the time. I do it all the time on my patients. What did your Fibroscan look like when you were initially diagnosed?

**Tiffany Brown:**

I forget my score because I've had three Fibroscans. Each of them show consistent at a high F-3 to F-4.

**Carolyn Legaspi, NP:**

I gotcha. So, you were already diagnosed at the time of diagnosis, advanced scar tissue in the liver, it sounds like.

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**Tiffany Brown:**

Yes.

**Carolyn Legaspi, NP:**

Okay, F-3 to F-4, which can be quite scary.

**Tiffany Brown:**

It was scary.

**Carolyn Legaspi, NP:**

And then was there any overlap with fat content in the liver?

**Tiffany Brown:**

No.

**Carolyn Legaspi, NP:**

Okay. So, no fat content, just some liver stiffness, which we see a lot with our PBC patients. So, I'm assuming your doctors wanted to be quite aggressive with your treatment, given the fact that your Fibroscan was... that it showed that you had advanced scar tissue?

**Tiffany Brown:**

Yes, we monitored a lot. I saw doctors a lot. Well, I went from not seeing doctors to seeing doctors a lot.

**Carolyn Legaspi, NP:**

All the time. I know. How did you feel about, how were you feeling when all of this diagnosis came about? Did it start to make sense, your symptoms, or how did you feel once you got the diagnosis?

**Tiffany Brown:**

It did make sense, which was some grounding, but I will be very honest with you, it was overwhelming. I thank God for those support groups on Facebook that I found. I tripped over them. No one suggested them to me. I just tripped over them on Facebook and I joined them. And those people that experience same and some different symptoms than I do. I mean, some have had hospital stays and I haven't, but they educated me a whole bunch on how to talk to doctors when I would go in for appointments.

**Carolyn Legaspi, NP:**

Yeah, no, definitely. Because sometimes as a provider you don't know what you don't know. You don't know what the patients know or what they don't know, and you only have a finite of time there. So, it is great that when you have a diagnosis of a chronic disease, that you have kind of a network of resources and you seek those resources.

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**Tiffany Brown:**

Yes.

**Carolyn Legaspi, NP:**

So, you went to Facebook groups and then any other organizations that you look to, to kind of get education or support?

**Tiffany Brown:**

Not really. I will tell you, my Facebook groups, they are my support groups, they're the ones I go to. And in the beginning I would ask a lot of questions. Now I guess you could say I'm just a stalker of my support groups because I've learned how to search. You can search in the search bar for things that you're looking up and gain that information without starting a whole new thread.

**Carolyn Legaspi, NP:**

Okay, I see. Was a liver biopsy ever on the table for you for liver disease?

**Tiffany Brown:**

I had .... I went, I started seeing a hepatologist in 2020, and he discussed a few times doing a liver biopsy. My GI specialist, he said, "Look, we've already diagnosed, there's no need to do that, and you're already in cirrhosis," or at least he felt, "If you're not, you're on the doorstep of it. Why do we want to go in and complicate things and have something for you to get over to take unnecessary medication, to have an unnecessary procedure when we already know?" So, between the two of them, they kind of worked that out, and no, I've never had even one scheduled.

**Carolyn Legaspi, NP:**

Okay. Yeah, no. In the setting of PBC, it's not necessary. A liver biopsy is not necessary unless you don't have a positive AMA, right?

**Tiffany Brown:**

Yes.

**Carolyn Legaspi, NP:**

If your mitochondrial antibody is negative, there is a PBC that's AMA-negative.

**Tiffany Brown:**

Yes.

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**Carolyn Legaspi, NP:**

... and that's how we would utilize a liver biopsy, but was just wondering if it was ever on the table to tease out other overlap autoimmune diseases or to confirm that fibrosis scoring.

**Tiffany Brown:**

My hepatologist at Piedmont retired, and so I chose another one in the group that was younger, and he did a barrage of blood tests that hadn't been done in a while. And his strong feeling was, I was your perfect PBC candidate. I showed nothing else. Well, I have Sjogren's and I have alopecia.

**Carolyn Legaspi, NP:**

You have other autoimmune diseases ...

**Tiffany Brown:**

Yes.

**Carolyn Legaspi, NP:**

... and Sjogren's is quite common with PBC together. Yeah.

**Tiffany Brown:**

Yes.

**Carolyn Legaspi, NP:**

Were you diagnosed with Sjogren's first or diagnosed with PBC first?

**Tiffany Brown:**

PBC.

**Carolyn Legaspi, NP:**

Okay. And then they started testing for Sjogren's. Were you having the dry eyes or the other symptoms?

**Tiffany Brown:**

Yes. For the life of me, I didn't understand that. I didn't use eye drops, and I would have almost these blinding-like headaches where I would just try to nap. And then I realized the more I drank water, the better I felt because I pretty much drink water and coffee, and coffee's limited, but the more water I would drink, the better my eyes felt, and that was the only thing that would relieve it. And so yeah, the dry eyes, they were a significant problem for me, and still can be.

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#### **Alyson Robertson:**

When you're diagnosed with PBC, it's easy to try and hide up into a ball and want to run from it and not deal with it and kind of wish it all to go away. But you need to take the bull by the horn and find out all the information that you can, not necessarily from Google, but from reputable sources. Ask your doctor where you can go to, what their sources are. Go to places like the PBCers Organization or the Canadian PBC Group or the PBC Foundation. There, you're going to find, I don't want to say tried and true, but reliable, good resources with good information.

It is a lifestyle change. It's not just a change for a day or a week or a month or a year. This is a forever because PBC is with you forever now. And find your support group, that's really important too. I mean, it's good to have your family, and even people, you might decide whether or not you want to tell your coworkers about it. I personally chose to tell my coworkers, because I felt it was important for them to know why I might be off one day or why I needed to run to the bathroom and someone needed to watch my classroom because of those side effects. And it's scary, but most people understand and they want to help you, they want to do what's good for you, and they want to be there for you.

#### **Kate Parfrey, NP:**

Thank you, Alyson, so much for all of that valuable information. I think it's really important that the word gets out there for patients to kind of learn how to cope with their disease and what resources to turn to when they need them. But thank you so much for sharing your story.

#### **Alyson Robertson:**

Absolutely.

#### **Kate Parfrey, NP:**

It's been a pleasure meeting you today.

#### **Alyson Robertson:**

Thank you. It's been a good pleasure meeting you, too.